Genetic Screening for Rare Diseases, Syndromes, and Traits

SEQUENCING

1. 17 Alpha-hydroxylase / 17,20-Lyase Deficiency, Combined Complete
2. 17 Beta-hydroxysteroid Dehydrogenase 3 Deficiency
3. 25-hydroxyvitamin D Deficiency
4. 2-methyl-3-hydroxybutyryl-CoA Dehydrogenase Deficiency
5. 3-methylcrotonyl-CoA Carboxylase 1 Deficiency, Biotin Responsive
6. 3-methylcrotonyl-CoA Carboxylase 2 Deficiency
7. 3-methylglutaconic Aciduria, Type III
8. Aarskog-Scott syndrome
9. Achromatopsia
10. Achromatopsia 3
11. Acquired Idiopathic Sideroblastic Anemia
12. Acromesomelic Dysplasia, Maroteaux Type
13. Actin Myopathy
14. Acute Hepatic Porphyria, Severe Infantile-onset
15. Acute Lymphoblastic Leukemia
16. Acute Myeloid Leukemia with Complex Karyotype
17. Adenine Phosphoribosyltransferase Deficiency
18. Adenosine Deaminase Deficiency, Partial
19. Adenosine Monophosphate Deaminase Deficiency
20. Adenylosuccinase Deficiency
21. Adiponectin Deficiency
22. Adrenal Hyperplasia
23. Adrenal Hyperplasia II, Deficiency of 3-Beta-hydroxysteroid Dehydrogenase, Type II
24. Adrenal Hypoplasia, Congenital
25. Adrenal Insufficiency, Congenital
26. Adrenoleukodystrophy, Neonatal
27. Agammaglobulinemia
28. Agammaglobulinemia and Isolated Growth Hormone Deficiency
29. Alagille Syndrome
30. Albinism, Ocular, Type I
31. Albinism, Oculocutaneous, Type I, Temperature Sensitive
32. Albinism, Oculocutaneous, Type IA
33. Albinism, Oculocutaneous, Type III
34. Alexander Disease
35. Alkaptonuria
36. Allan-Herndon-Dudley Syndrome
37. Alopecia Areata
38. Alopecia Universalis Congenita
39. Alpers Syndrome
40. Alpers-like Hepatocerebral Syndrome
41. Alpha-1-Antitrypsin Deficiency
42. Alpha-B Crystallinopathy with Cataract
43. Alport Syndrome
44. Alström Syndrome
45. Alzheimer's Disease, Early-onset (Familial)
46. Amish Infantile Epilepsy Syndrome
47. Amyloid Polyneuropathy, Andrade or Portuguese Type
48. Amyloidosis I
49. Amyloidosis I, Hereditary Neuropathic
50. Amyloidosis III
51. Amyloidosis, Familial, Finnish Type
52. Amyloidosis, Familial, Visceral
53. Amyloidosis, Reactive Amyloid Systemic
54. Amyloidotic Polyneuropathy, Cardiac or Denmark Type
55. Amyloidotic Polyneuropathy, German-American Type
56. Andermann Syndrome
57. Andersen Cardiodysrhythmic Periodic Paralysis
58. Anderson Disease
59. Androgen Insensitivity Syndrome
60. Androgen Insensitivity Syndrome, Infertility, Male
61. Androgen Insensitivity, Complete
62. Androgen Insensitivity, Partial
63. Angelman Syndrome
64. Aniridia
65. Antenatal Bartter Syndrome, Type 2
66. Antiplasmin Alpha 2 Deficiency
67. Antithrombin III Deficiency
68. Aortic Aneurysm, Familial Thoracic
69. Aortic Aneurysm, Familial Thoracic 4
70. Aortic Aneurysm, Familial Thoracic 5
71. Apert Syndrome
72. Apolipoprotein A2 Deficiency
73. Apolipoprotein B Deficiency
74. Apolipoprotein H Deficiency
75. APRT Deficiency
76. APRT Deficiency, Japanese Type
77. Aquaporin-1 Deficiency Colton-Null
78. Argininemia
79. Argininosuccinic Aciduria
80. Arterial Aneurysms, Familial
81. Arthrogryposis, Distal, Type 2A
82. Arthrogryposis, Renal Dysfunction and Cholestasis Syndrome
83. Arylsulfatase A Pseudodeficiency
84. Ascending Spastic Paralysis, Infantile-onset
85. Leber Hereditary Optic Atrophy
86. Ataxia
87. Ataxia, Episodic / Myokymia
88. Ataxia and Retinitis Pigmentosa with Isolated Vitamin E Deficiency
89. Ataxia, Isolated Vitamin E Deficiency
90. Ataxia, Spinocerebellar
91. Ataxia, Spinocerebellar, 6
92. Ataxia Telangiectasia
93. Ataxia Telangiectasia without Immunodeficiency
94. Ataxia Telangiectasia-like Disease
95. Ataxia-ocular Apraxia 2
96. Atelosteogenesis, Type IB
97. Atelosteogenesis, Type II
98. Atrial Septal Defect
99. Atrial Septal Defect with Atrioventricular Conduction Defects
100. Atrichia with Papular Lesions
101. Atrioventricular Block, Idiopathic Second-Degree
102. Atrioventricular Septal Sefect
103. ATRX Syndrome
104. Auditory Neuropathy, Nonsyndromic
105. Auditory Neuropathy, Temperature-sensitive
106. Autoimmune and Autoinflammatory Diseases
107. Autoimmune Lymphoproliferative Syndrome, Type II
108. Autoimmune Lymphoproliferative Syndrome, Type IIA
109. Autoimmune Polyendocrinopathy-Candidiasis-Ectodermal Dystrophy
110. Azoospermia, Obstructive
111. Bardet-Biedl Syndrome 1
112. Bardet-Biedl Syndrome 4
113. Bardet-Biedl Syndrome 10
114. Bardet-Biedl Syndrome 13
115. Bardet-Biedl Syndrome 14
116. Bare Lymphocyte Syndrome, Type I
117. Bart Syndrome
118. Barth syndrome
119. Bartter Syndrome
120. Bartter Syndrome, Antenatal, Type 1
121. Bartter Syndrome, Type 3
122. Beare-Stevenson Cutis Gyrata Syndrome
123. Benign Familial Hematuria
124. Benign Hereditary Chorea
125. Berardinelli-Seip lipodystrophy
126. Bernard-Soulier Syndrome, Type A
127. Bernard-Soulier Syndrome, Type C
128. Beta Thalassemia
129. Beta-hexosaminidase A, Pseudodeficiency of
130. Bethlem Myopathy
131. Bilateral Striatal Necrosis, Infantile
132. Bile Acid Synthesis Defect, Congenital, 4
133. Biliary Atresia, Extrahepatic
134. Biotinidase Deficiency
135. Biotinidase Deficiency, Partial
136. Blau Syndrome
137. Blepharophimosis, Ptosis, and Epicanthus Inversus, Type I
138. Blindness
139. Color Blindness, Deutan
140. Night Blindness, Congenital Stationary, Type 1
141. Night Blindness, Congenital Stationary, Type 1B
142. Night Blindness, Congenital Stationary, Type 2
143. Night Blindness, Congenital Stationary, Type 2, Severe
144. Night Blindness, Congenital Stationary, Type 2B
145. Blood Group Variant: Auberger Au(a)/Au(b)
146. Blood Group Variant: Bombay Phenotype
147. Blood Group Variant: Colton
148. Blood Group Variant: Diego
149. Blood Group Variant: Dombrock
150. Blood Group Variant: Dombrock-null
151. Blood Group Variant: Froese
152. Blood Group Variant: Kidd
153. Blood Group Variant: Lewis Antigen, absence of
154. Blood Group Variant: p Phenotype
155. Blood Group Variant: P(k) Antigen
156. Blood Group Variant: Ralph
157. Blood Group Variant: Swann
158. Blood Group Variant: Waldner
159. Blood Group Variant: Wright
160. Bloom Syndrome
161. Borjeson-Forssman-Lehmann Syndrome
162. Bothnia Retinal Dystrophy Retinitis Punctata Albescens
163. Brachydactyly, Type A1
164. Brachydactyly, Type A2
165. Brachydactyly, Type B1
166. Brachydactyly, Type C
167. Brachydactyly, Type E
168. Brachydactyly, Type D
169. Butyrylcholinesterase Deficiency

170. Butyrylcholinesterase Deficiency, Fluoride-resistant, Japanese Type
171. C (Opitz Trigonocephaly) Syndrome
172. Cabezas Syndrome
173. Caffey Disease
174. Camurati-Engelmann Disease
175. Canavan Disease
176. Capillary Malformation-Arteriovenous Malformation
177. Carbohydrate-deficient Glycoprotein Syndrome, Type II
178. Carbonic Anhydrase Deficiency
179. Cardiac Conduction Disease
180. Cardioencephalomyopathy, Fatal Infantile, due to Cytochrome c Oxidase Deficiency
181. Cardiofaciocutaneous Syndrome
182. Cardiomyopathy, X-linked Infantile
183. Carney Complex
184. Carney Complex Variant
185. Carnitine Deficiency, Systemic Primary
186. Carnitine Palmitoyltransferase Deficiency, Hepatic, Type IA
187. Carnitine Palmitoyltransferase II Deficiency
188. Carnitine Palmitoyltransferase II Deficiency, Infantile
189. Carnitine Palmitoyltransferase II Deficiency, Late-onset
190. Carnitine Palmitoyltransferase II Deficiency, Lethal Neonatal
191. Carnitine-acylcarnitine Translocase Deficiency
192. Cartilage-Hair Hypoplasia
193. Caspase-8 Deficiency
194. Cataract, Central Nuclear
195. Cataract, Congenital
196. Cataract, Coppock-like
197. Cataract, Marner Type
198. Cataract, Ocular Anterior Dysgenesis and Coloboma
199. Cataract, Primary Congenital
200. Cataract, Congenital Lamellar
201. Caudal Regression Syndrome
202. CD36 Deficiency
203. Central Core Disease
204. Central Hypoventilation Syndrome
205. Centronuclear Myopathy
206. Centronuclear Myopathy, Becker Muscular Dystrophy
207. Cerebellar Ataxia, Cataracts, and Diabetes Mellitus
208. Cerebellar Ataxia, Cayman Type
209. Cerebral Arteriopathy with Subcortical Infarcts and Leukoencephalopathy
210. Cerebral Cavernous Malformations
211. Cerebroarterial Amyloidosis, Icelandic-type
212. Cerebrooculofacioskeletal Syndrome 4
213. Cerebrotendinous Xanthomatosis
214. Cerulean Cataract, Congenital
215. Cervical Artery Dissections, Spontaneous
216. Chanarin-Dorfman Syndrome
217. Charcot-Marie-Tooth Disease, Type 1A
218. Charcot-Marie-Tooth Disease, Type 1B
219. Charcot-Marie-Tooth Disease, Type 1D
220. Charcot-Marie-Tooth Disease, Axonal, Type 2A1
221. Charcot-Marie-Tooth Disease, Axonal, Type 2A2
222. Charcot-Marie-Tooth Disease, Axonal, Type 2B1
223. Charcot-Marie-Tooth Disease, Axonal, Type 2D
224. Charcot-Marie-Tooth Disease, Axonal, Type 2E
225. Charcot-Marie-Tooth Disease, Axonal, Type 2K
226. Charcot-Marie-Tooth Disease, Type 2F
227. Charcot-Marie-Tooth Disease, Type 2J
228. Charcot-Marie-Tooth Disease, Type 4C
229. Charcot-Marie-Tooth Disease, Type 4F
230. Charcot-Marie-Tooth Disease, Type 4H
231. Charcot-Marie-Tooth Disease, Type 4J
232. Charcot-Marie-Tooth Disease, Type 5
233. Charcot-Marie-Tooth Disease, Type 6
234. CHARGE Syndrome
235. Chediak-Higashi Syndrome, Childhood Type
236. Cherubism
237. Cholesterol Ester Storage Disease
238. Cholinesterasaemia
239. Chondrocalcinosis 2
240. Chondrodysplasia Punctata
241. Choreoacanthocytosis
242. Choroideremia
243. Chronic Granulomatous Disease, Cytochrome b-Negative
244. Chronic Granulomatous Disease, Cytochrome b-Positive, Chronic
245. Chronic Granulomatous Disease, Cytochrome b-Positive, Type II
246. Chronic Insomnia
247. Chronic Obstructive Pulmonary Disease, Severe Early-onset
248. Chylomicron Retention Disease
249. Ciliary Dyskinesia, Primary
250. Cleft Lip/Palate-Ectodermal Dysplasia Syndrome
251. Cleft Palate and Ankyloglossia
252. Cleidocranial Dysplasia
253. Coagulation Defect, Vitamin K-Dependent
254. Cohen Syndrome
255. Combined Deficiency of Vitamin K-dependent Clotting Factors, Type 2
256. Combined Oxidative Phosphorylation Deficiency
257. Combined SAP Deficiency
258. Complement C2 Deficiency, Type II
259. Complement C3 Deficiency
260. Complement C5 Deficiency
261. Complete Adenine Phosphoribosyltransferase Deficiency, Icelandic Type
262. Complex 1 Deficiency
263. Complex 3 Deficiency
264. Cone-rod Dystrophy
265. Cone-rod Dystrophy 3
266. Cone-rod Dystrophy 3
267. Cone-rod Dystrophy 6
268. Cone-rod Dystrophy 9
269. Congenital Adrenal Hyperplasia
270. Congenital Adrenal Hyperplasia due to Steroid-11 Beta-hydroxylase Deficiency
271. Congenital Adrenal Hyperplasia, Non-classic
272. Congenital Afibrinogenemia, Congenital Hypofibrinogenemia
273. Congenital Cataract
274. Congenital Central Hypoventilation Syndrome
275. Congenital Disorder of Glycosylation, Type Ia
276. Congenital Disorder of Glycosylation, Type Ib
277. Congenital Disorder of Glycosylation, Type Ic
278. Congenital Disorder of Glycosylation, Type Id
279. Congenital Disorder of Glycosylation, Type Ig
280. Congenital Disorder of Glycosylation, Type Ik
281. Congenital Disorder of Glycosylation, Type IIc
282. Congenital Erythropoietic Porphyria
283. Congenital Erythropoietic Porphyria, Mild, Cutaneous-only
284. Congenital Fast Channel Myasthenic Syndrome
285. Congenital Heart Defects
286. Congenital Heart Disease Heterotaxy
287. Congenital Insensitivity to Pain Syndrome
288. Congenital Lipoid Adrenal Hyperplasia
289. Congenital Merosin Deficient Muscular Dystrophy
290. Congenital Muscular Dystrophy, Type 1C, with Neurologic Abnormalities
291. Congenital Myasthenic Syndrome
292. Congenital Myasthenic Syndrome associated with Acetylcholine Receptor Deficiency
293. Congenital Nephrosis 1, Finnish Type
294. Congenital Sick Sinus Syndrome
295. Congenital Slow Channel Myasthenic Syndrome
296. Conjunctivitis, Ligneous
297. Connatal Pelizaeus-Merzbacher Disease
298. Contractural Arachnodactyly, Congenital
299. Coproporphyria
300. Corticosteroid-binding Globulin Deficiency
301. Costello Syndrome
302. Cowden Disease Bannayan-Riley-Ruvalcaba Syndrome
303. Cowden Disease Bannayan-Riley-Ruvalcaba Syndrome, Macrocephaly/Autism Syndrome
304. Craniofrontonasal Syndrome
305. Craniometaphyseal Dysplasia
306. Craniosynostosis Muenke Syndrome
307. Creutzfeldt-Jakob Syndrome
308. Crigler-Najjar Syndrome 1
309. Crigler-Najjar Syndrome 2
310. Crouzon syndrome
311. Crouzon Syndrome with Acanthosis Nigricans
312. Crouzonodermoskeletal Syndrome
313. Cutis Laxa
314. Cystathioninuria
315. Cystic Fibrosis
316. Cystic Fibrosis, Non-classic
317. Cystinuria
318. Cystinuria, Non-type I
319. Cytochrome c Oxidase Deficiency
320. Cytochrome P450 Deficiency
321. Danon Disease
322. Darier Disease
323. Deafness, Childhood-onset
324. Deafness, Aminoglycoside-induced
325. Deafness, Congenital Heart Defects, and Posterior Embryotoxon
326. Deafness, Congenital, Neurosensory
327. Deafness, Congenital, with Inner Ear Agenesis, Microtia, and Microdontia
328. Deafness, Neurosensory
329. Deafness, Neurosensory 21
330. Deafness, Neurosensory without Vestibular Involvement
331. Deafness, Nonsyndromic
332. Deafness, Nonsyndromic Sensorineural

333. Deafness, Autosomal Recessive 1
334. Deafness, Nonsyndromic Sensorineural 3
335. Deafness, Nonsyndromic Sensorineural 8
336. Deafness, Nonsyndromic Sensorineural 12
337. Deafness, Autosomal Dominant 12
338. Deafness, Nonsyndromic Sensorineural with Dentinogenesis Imperfecta, Type I
339. Deafness, Progressive
340. Deafness, Sensorineural, with Migraine
341. Deafness, Sensorineural, with Mild Renal Dysfunction
342. Decreased Androgen (DHEA) Secretion
343. Deficiency of 3-Beta-hydroxysteroid Dehydrogenase, Type II
344. Deficiency of Factor XIII, A Subunit
345. Dejerine-Sottas Neuropathy
346. Delayed Sleep Phase Syndrome
347. Demyelinating Charcot-Marie-Tooth Disease 4A Axonal Neuropathy with Vocal Cord Paresis, Axonal Charcot-Marie-Tooth Disease, Type 2K
348. Dent Disease
349. Dentinogenesis Imperfecta, Severe, with Very Mild Osteogenesis Imperfecta
350. Dentinogenesis Imperfecta, Type I
351. Dentinogenesis Imperfecta, Shields Type II
352. Dentinogenesis Imperfecta, Shields Type III
353. Denys-Drash Syndrome
354. Denys-Drash Syndrome Mesangial Sclerosis, Isolated Diffuse
355. Desmosterolosis
356. Diabetes Insipidus, Central
357. Diabetes Insipidus, Nephrogenic
358. Diabetes Mellitus, Insulin-Resistant, with Acanthosis Nigricans
359. Diabetes Mellitus, Permanent Neonatal
360. Diarrhea, Malabsorptive, Congenital
361. Diastrophic Dysplasia
362. Dihydropteridine Reductase Deficiency
363. Dihydropyrimidine Dehydrogenase Deficiency
364. Dihydropyrimidine Dehydrogenase Deficiency, Possible 5-FU Toxicity
365. Dihydropyrimidine Dehydrogenase Deficiency, Partial
366. Dilatative Myopathy
367. Disordered Steroidogenesis
368. Distal Arthrogryposis Syndrome 1
369. Distal Arthrogryposis Syndrome 2b
370. Distal Renal Tubular Acidosis
371. Distal Renal Tubular Acidosis with Late-onset Sensorineural Hearing Loss
372. Distal Renal Tubular Acidosis with Progressive Deafness
373. Dopamine Beta Hydroxylase Deficiency
374. Drug-induced Hemolysis Haemoglobin Variant
375. Dubin-Johnson Syndrome
376. Dysalbuminemic Hyperthyroxinaemia, Familial
377. Dysautonomia, Familial
378. Dysfibrinogenaemia
379. Fibrinogen Amiens 1
380. Fibrinogen Amiens 2
381. Fibrinogen Bergamo 3
382. Fibrinogen Bern 2
383. Fibrinogen Bicetre 1
384. Fibrinogen Birmingham 1
385. Fibrinogen Chapel Hill 2
386. Fibrinogen Clermont-Ferrand 1
387. Fibrinogen Giessen 1
388. Fibrinogen Leitchfield
389. Fibrinogen Long Beach 1
390. Fibrinogen Louisville 1
391. Fibrinogen Manchester 1
392. Fibrinogen Paris 6
393. Fibrinogen Petoskey 1
394. Fibrinogen Seattle 2
395. Fibrinogen Sheffield 2
396. Fibrinogen Sydney 1
397. Fibrinogen Sydney 2
398. Fibrinogen White Marsh 1
399. Dysfibrinogenemia
400. Dysfibrinogenemia, Fibrinogen Bergamo 1
401. Dysfibrinogenemia Thrombophilia, Dysfibrinogenemic
402. Fibrinogen Hershey 2
403. Fibrinogen Homburg 2
404. Fibrinogen Homburg 3
405. Fibrinogen Kawaguchi 1
406. Fibrinogen Leogan
407. Fibrinogen Metz 1
408. Fibrinogen New Albany
409. Fibrinogen Osaka 1
410. Fibrinogen Schwarzach 1
411. Fibrinogen Stony Brook 1
412. Fibrinogen Zurich 1
413. Fibrinogen Torino 1
414. Fibrinogen Ledyard
415. Fibrinogen Hershey 3
416. Fibrinogen Milano XII, Digenic
417. Dyskeratosis Congenita
418. Dystonia 12
419. Dystonia, Adult-onset
420. Dystonia, Dopa-responsive
421. Dystonia, Torsion
422. Ehlers-Danlos Syndrome
423. Ehlers-Danlos Syndrome, Cardiac Valvular Form
424. Ehlers-Danlos Syndrome, Hypermobility Type
425. Ehlers-Danlos Syndrome, Progeroid Form
426. Ehlers-Danlos Syndrome, Spondylocheiro Dysplastic Form
427. Ehlers-Danlos Syndrome, Type I
428. Ehlers-Danlos Syndrome, Type II
429. Ehlers-Danlos Syndrome, Type IV
430. Ehlers-Danlos Syndrome, Type VI-A
431. Ehlers-Danlos Syndrome, Type VI-A, Nevo Syndrome
432. Ehlers-Danlos Syndrome, Type VII
433. Ehlers-Danlos Syndrome, Type VII-A
434. Ehlers-Danlos Syndrome, Type VII-B
435. Elliptocytosis
436. Elliptocytosis, Hemolytic Anemia, Neonatal Nonimmune, Fatal and Near-fatal
437. Elliptocytosis, Rhesus-Unlinked Type
438. Elliptocytosis, Rhesus-Unlinked Type, Pyropoikilocytosis, Hereditary
439. Encephalopathy, Ethylmalonic
440. Encephalopathy, Familial, with Neuroserpin Inclusion Bodies
441. Enhanced S-cone Syndrome
442. Enhanced S-cone Syndrome, Retinitis Pigmentosa
443. Epidermolysis Bullosa Dystrophica
444. Epidermolysis Bullosa Dystrophica, Localisata Variant
445. Epidermolysis Bullosa Simplex
446. Epidermolysis Bullosa Simplex, Ogna Type
447. Epidermolysis Bullosa with Pyloric Atresia
448. Epidermolysis Bullosa without Pyloric Atresia, Generalized Atrophic Benign
449. Epidermolysis Bullosa, Dowling-Meara
450. Epidermolysis Bullosa, Generalized Atrophic Benign
451. Epidermolysis Bullosa, Herlitz
452. Epidermolysis Bullosa, Junctional
453. Epidermolysis Bullosa, Junctional, Herlitz Type
454. Epidermolysis Bullosa, Junctional, with Pyloric Atresia
455. Epidermolysis Bullosa, Koebner
456. Epidermolysis Bullosa, Lethal Acantholytic
457. Epidermolysis Bullosa, Pretibial
458. Epidermolysis Bullosa, Weber-Cockayne
459. Epidermolytic Hyperkeratosis
460. Epidermolytic Palmoplantar Keratoderma associated with Knuckle Pads
461. Epilepsy, Benign Neonatal
462. Epilepsy, Childhood Absence
463. Epilepsy, Nocturnal Frontal Lobe
464. Epilepsy, Progressive Myoclonus
465. Epilepsy, Juvenile Absence
466. Epilepsy, Juvenile Myoclonic
467. Epilepsy, Severe Myoclonic, of Infancy (Dravet Syndrome)
468. Epilepsy, Myoclonic, of Lafora
469. Epilepsy, Myoclonic, with Mental Retardation and Spasticity
470. Epiphyseal Dysplasia
471. Epoxide Hydrolase Deficiency, Susceptibility to Lymphoproliferative Disorder
472. Epstein Syndrome
473. Erythermalgia, Primary
474. Erythrocyte Lactate Transporter Defect
475. Escobar Syndrome
476. Essential Fructosuria
477. Ethylmalonic Aciduria
478. Excessive Daytime Sleepiness
479. Exostoses, Multiple, Type II
480. Extrapyramidal Movement Disorder
481. Fabry Disease
482. Faciogenital Dysplasia with Attention Deficit Hyperactivity Disorder
483. Factor H Deficiency
484. Factor V and Factor VIII Deficiency, Combined
485. Factor V Deficiency
486. Factor VII Deficiency
487. Factor X Deficiency
488. Factor XI Deficiency
489. Factor XII Deficiency
490. Familial Advanced Sleep Phase Syndrome
491. Familial Cold Autoinflammatory Syndrome
492. Familial Dysautonomia
493. Familial Mediterranean Fever
494. Fanconi Anemia, Complementation Group A

495. Fanconi Anemia, Complementation Group C
496. Fanconi Anemia, Complementation Group D1
497. Fanconi Anemia, Complementation Group E
498. Fanconi Anemia, Complementation Group J
499. Fanconi Anemia, Complementation Group N
500. Farber Lipogranulomatosis
501. Fechtner Syndrome
502. Fibromatosis, Juvenile Hyaline
503. Fish-eye Disease
504. Focal Segmental Glomerulosclerosis
505. Foveomacular Dystrophy, Adult-onset, with Choroidal Neovascularization
506. Fragile X Mental Retardation Syndrome
507. Frasier Syndrome
508. Friedreich Ataxia
509. Friedreich-Like Ataxia with Isolated Vitamin E Deficiency
510. Frontotemporal Dementia
511. Frontotemporal Dementia, with Parkinsonism
512. Frontotemporal Lobar Dementia, Ubiquitin-Positive
513. Fuchs Endothelial Corneal Dystrophy, Polymorphous Posterior
514. Fucosidosis
515. Fucosyltransferase-6 Deficiency
516. Fumarylacetoacetase Pseudodeficiency
517. Galactosemia
518. Galactosialidosis, Adult, Japanese Type
519. Galactosialidosis, Late Infantile
520. Gangliosidosis GM1
521. Gangliosidosis GM1, Late Infantile/Juvenile Type
522. Gaucher Disease, Type I
523. Gaucher Disease, Type II
524. Gaucher Disease, Type II, Perinatal Lethal Form
525. Gaucher Disease, Type III
526. Gaucher Disease, Type IIIC
527. Gelatinous Drop-like Corneal Dystrophy
528. Generalized Epilepsy and Paroxysmal Dyskinesia
529. Generalized Idiopathic Epilepsy Episodic Ataxia, Type 5
530. Gerstmann-Straeussler Syndrome
531. Giant Axonal Neuropathy
532. Gilbert Syndrome
533. Gitelman Syndrome
534. Glanzmann Thrombasthenia
535. Glaucoma, Primary Congenital
536. Glaucoma 1, Open Angle, E
537. Glaucoma 1, Open Angle, E, Glaucoma, Normal Tension
538. Glaucoma 1A, Open Angle
539. Glaucoma 1A, Primary Open Angle
540. Glaucoma 1A, Primary Open Angle, Digenic
541. Glaucoma 3A, Primary Congenital, Digenic
542. Glucocorticoid Deficiency
543. Glucose-6-phosphate Dehydrogenase Deficiency
544. Glutamate Formiminotransferase Deficiency
545. Glycine N-methyltransferase Deficiency
546. Glycogen Storage Disease, Type 0
547. Glycogen Storage Disease, Type Ia
548. Glycogen Storage Disease, Type Ib
549. Glycogen Storage Disease, Type II
550. Glycogen Storage Disease, Type II, Adult Form
551. Glycogen Storage Disease, Type II, Infantile Form
552. Glycogen Storage Disease, Type IIIa
553. Glycogen Storage Disease, Type IV, Classic Hepatic
554. Glycogen Storage Disease, Type IV, Childhood Neuromuscular
555. Glycogen Storage Disease, Type IV, Nonprogressive Hepatic
556. Glycogen Storage Disease, Type VI
557. Glycogen Storage Disease, Type VII
558. GM1-Gangliosidosis, Adult/Chronic Type
559. GM2-Gangliosidosis, Adult
560. Goiter, Familial, with Hypothyroidism
561. Goiter, Nonendemic Simple
562. Gout, HPRT-related
563. GRACILE Syndrome
564. Greater Agonists Promoted Contractility
565. Greig Cephalopolysyndactyly Syndrome
566. Griscelli Syndrome, Type 2
567. Growth Hormone Deficiency
568. Growth Hormone Deficiency, Isolated
569. Growth Hormone Deficiency, Isolated, Type 2
570. Growth Retardation due to IGF1R
571. Gyrate Atrophy
572. Gyrate Atrophy with Pyridoxine-responsive Ornithinemia
573. Haemoglobin H Disease
574. Haemorrhagic Telangiectasia 1
575. Hailey-Hailey Disease
576. Haim-Munk Syndrome
577. Harderoporphyria
578. Harlequin Ichthyosis
579. HARP Syndrome
580. Hartnup Disorder
581. Hawkinsinsuria
582. Hearing Impairment, Nonsyndromic Sensorineural
583. Hemochromatosis
584. Hemochromatosis, Type 2A
585. Hemochromatosis, Type 3
586. Hemochromatosis, Type 4
587. Hemolytic Anemia
588. Hemolytic Anemia due to Adenylate Kinase Deficiency
589. Hemolytic Anemia due to Glutathione Synthetase Deficiency of Erythrocytes
590. Hemolytic Anemia due to Triosephosphate Isomerase Deficiency
591. Hemolytic Anemia, Rh-Null, Regulator Type
592. Hemolytic Uremic Syndrome
593. Hemophagocytic Lymphohistiocytosis, Familial
594. Hemophilia A
595. Hemophilia B
596. Hemophilia B, Leyden
597. Hemophilia B, Brandenburg
598. Hemorrhagic Diathesis due to Antithrombin Pittsburgh
599. Heparin Cofactor II Deficiency
600. Hepatic Lipase Deficiency
601. Hereditary Angioedema, Type II
602. Hereditary Haemorrhagic Telangiectasia, Type 2
603. Hereditary Hypophosphatemic Rickets with Hypercalciuria
604. Hereditary Myopathy with Early Respiratory Failure
605. Hereditary Nonpolyposis Colorectal Cancer, Type 1
606. Hereditary Nonpolyposis Colorectal Cancer, Type 2
607. Hereditary Nonpolyposis Colorectal Cancer, Type 5
608. Hereditary Nonpolyposis Colorectal Cancer, Type 7
609. Hereditary Nonploy posis Colorectal Cancer, Type 9
610. Hereditary Nonploy posis Colorectal Cancer, Type 10
611. Hereditary Persistence of Fetal Hemoglobin
612. Hermansky-Pudlak Syndrome
613. Heterotaxy
614. Hidrotic Ectodermal Dysplasia
615. High Myopia
616. Hirschsprung Disease
617. Hirschsprung Disease, Congenital Hypoventilation Syndrome
618. Hirschsprung Disease, Waardenburg-Shah Syndrome
619. HLA Class I Deficiency
620. HMG-CoA Lyase Deficiency
621. HMG-CoA Synthase Deficiency
622. Holocarboxylase Synthetase Deficiency
623. Holoprosencephaly 2
624. Holoprosencephaly 3
625. Holoprosencephaly 5
626. Holoprosencephaly 7
627. Holoprosencephaly 9
628. Homocystinuria
629. Hurler Syndrome
630. Hutchinson-Gilford Progeria Syndrome, Restrictive Dermatopathy, Lethal
631. Hydatidiform Mole, Recurrent
632. Hydrocephalus, X-linked
633. Hyperbilirubinemia, Transient Familial Neonatal
634. Hypercholanemia, Familial
635. Hypercholesterolemia, Familial
636. Hyperekplexia
637. Hyperglycerolemia
638. Hyperglycinemia, Non-ketotic Glycine Encephalopathy
639. Hypergonadotrophic Hypogonadism, Female
640. Hyperhomocysteinemia due to MTHFR Deficiency, Folate Responsive
641. Hyper-IgD Syndrome
642. Hyper-IgE Syndrome
643. Hyperinsulinism-hyperammonemia Syndrome
644. Hyperkalaemic Periodic Paralysis, Paramyotonia Congenita
645. Hyperlipidemia, Familial Combined
646. Hyperornithinemia-hyperammonemia-homocitrullinemia Syndrome
647. Hyperostosis-Hyperphosphatemia Syndrome
648. Hyperphenylalaninemia
649. Hyperphenylalaninemia, Non-PKU
650. Hyperproinsulinaemia, Familial
651. Hyperprolinemia, Type 1 Schizophrenia
652. Hypertriglyceridemia, Hereditary
653. Hypocalciuric Hypercalcaemia, Familial, Hypoparathyrodism, Familial Isolated
654. Hypocholesterolaemia
655. Hypocholinesterasaemia
656. Hypochondroplasia
657. Hypogammaglobulinemia
658. Hypoglycaemia, Persistent Hyperinsulinaemic
659. Hypoglycemia

660. Hypogonadotropic Hypogonadism
661. Hypogonadotropic Hypogonadism, Fertile Eunuch Syndrome
662. Hypokalaemic Periodic Paralysis
663. Hypomagnesaemia with Secondary Hypocalcaemia
664. Hypomagnesaemia, Primary
665. Hypomagnesaemia, Renal
666. Hypoparathyroidism, Familial Isolated
667. Hypoparathyroidism, Familial Isolated, Hypocalemia with Bartter Syndrome
668. Hypoparathyroidism-retardation-dysmorphism Syndrome
669. Hypophosphatasia, Infantile
670. Hypophosphatasia, Infantile, Mild
671. Hypophosphatemia
672. Hypophosphatemic Rickets
673. Hypoplastic Left Heart Syndrome, Atrioventricular Septal Defect
674. Hypothyroidism, Thyroid Hormonogenesis, Genetic Defect in
675. Hypothyroidism, Congenital, Nongoitrous
676. Hypotrichosis
677. Hypotrichosis Simplex
678. Hypotrichosis Simplex of Scalp
679. Hypoxanthine Guanine Phosphoribosyltransferase Deficiency
680. Hystrix-Like Ichthyosis with Deafness, Keratitis-ichthyosis-deafness Syndrome
681. Ichthyosis Vulgaris
682. Ichthyosis with Hypotrichosis
683. Ichthyosis, Harlequin
684. Ichthyosis, Lamellar
685. Idiopathic Infantile Nystagmus
686. Idiopathic Pulmonary Fibrosis
687. Idiopathic Restrictive Cardiomyopathy
688. Immunodeficiency with Hyper-IgM, Type 2
689. Immunodeficiency with Hyper-IgM, Type 3
690. Immunodeficiency with Hyper-IgM, Type 5
691. Immunologically Anomalous Variant
692. Inclusion Body Myopathy
693. Infantile Nephronophthisis
694. Intrahepatic Cholestasis of Pregnancy
695. Intrahepatic Cholestasis, Familial Progressive 2
696. Intrauterine and Postnatal Growth Retardation (Short Stature)
697. Iodide Transport Defect, Thyroid Hormonogenesis, Genetic Defect in
698. IPEX syndrome
699. Iris Flocculi
700. Isolated Lissencephaly Sequence
701. Isolated Partial Atrioventricular Septal Defect
702. Isovaleric Acidemia
703. ITPase Deficiency
704. Jervell and Lange-Nielsen Syndrome
705. JK-Null Variant, Finnish Type
706. Joubert Syndrome
707. Joubert Syndrome 2
708. Joubert Syndrome 3
709. Joubert Syndrome, Leber Congenital Amaurosis, Type X
710. Juvenile Polyposis/Hereditary Hemorrhagic Telangiectasia Syndrome
711. Kallmann Syndrome
712. Kallmann Syndrome 2
713. Kartagener Syndrome
714. Hemolytic Disease of the Newborn due to Anti-K
715. Keratitis-ichthyosis-deafness Syndrome
716. Keratoderma, palmoplantar
717. Keratoderma, Palmoplantar, with Deafness, Nonsyndromic Sensorineural
718. Knuckle pads, Leukonychia, Sensorineural Deafness
719. Kowarski Syndrome
720. Larsen Syndrome
721. Late Infantile Metachromatic Leukodystrophy
722. Lateral Temporal Lobe Epilepsy
723. Lead Poisoning, Increased Susceptibility to
724. Leber Congenital Amaurosis, Type I
725. Leber Congenital Amaurosis, Type III
726. Leber Congenital Amaurosis, Type VI
727. Leber Congenital Amaurosis, Type VII
728. Leber Congenital Amaurosis, Type X
729. Leber Hereditary Optic Neuropathy
730. Leber Hereditary Optic Neuropathy, Severe
731. Leber Optic Atrophy
732. Leigh Syndrome
733. Leigh Syndrome due to Mitochondrial Complex I Deficiency
734. Leigh Syndrome, French-Canadian Type
735. Leiomyomatosis and Renal Cell Cancer
736. Leprechaunism
737. Lesch-Nyhan Syndrome
738. Lethal Arthrogryposis with Anterior Horn Cell Disease
739. Lethal Congenital Contracture Syndrome
740. Lethal Contractural Syndrome, Type 3
741. Leukocyte Adhesion Deficiency
742. Leukoencephalopathy with Vanishing White Matter
743. Leukoencephalopathy with Vanishing White Matter, Ovarioleukodystrophy
744. Leydig Cell Hypoplasia with Male Pseudohermaphroditism
745. Liddle Syndrome
746. Li-Fraumeni Syndrome
747. Lipodystrophy with Diabetes
748. Lipodystrophy, Familial Partial, Type II (Dunnigan)
749. Lipoprotein Lipase Deficiency
750. Lissencephaly, Subcortical Laminar Heterotopia
751. Liver Glycogenosis, Type I
752. Liver Glycogenosis, Type II
753. Lujan-Fryns Syndrome
754. Lymphangioleiomyomatosis
755. Lymphoedema, Hereditary
756. Lymphoedema-distichiasis Syndrome
757. Macrocephaly/Autism Syndrome
758. Macular Degeneration, Juvenile
759. Macular Dystrophy
760. Macular Dystrophy, Best
761. Macular Dystrophy, Vitelliform
762. Majeed Syndrome
763. Mal de Meleda
764. Malignant Hyperthermia
765. Malonyl-CoA Decarboxylase Deficiency
766. Mannose-binding Protein Deficiency
767. Maple Syrup Urine Disease, Type IA
768. Maple Syrup Urine Disease, Type IB
769. Maple Syrup Urine Disease, Type II, Thiamine-response
770. Maple Syrup Urine Disease, Type III
771. Marfan Syndrome
772. Marfan Syndrome, Atypical
773. Marfan Syndrome, Neonatal
774. Marfan Syndrome, Severe Classic
775. Marfan Syndrome, Type II
776. Marfanoid Skeletal Syndrome
777. MASA syndrome
778. Mast Cell Leukemia
779. Mastocytosis, Sporadic, Childhood-onset
780. Maturity-onset Diabetes of the Young, Type III
781. Maximum Parasitemia, Mild Malaria Attack
782. May-Hegglin Anomaly
783. McArdle Disease
784. McCune-Albright Syndrome
785. McKusick-Kaufman Syndrome
786. Meckel Syndrome
787. Meckel Syndrome, Type 3
788. Mediterranean Macrothrombocytopenia
789. Medium Chain Acyl CoA Dehydrogenase Deficiency
790. Meesmann Corneal Dystrophy
791. Megablastic Anemia, Norwegian
792. Megalencephalic Leukoencephalopathy with Subcortical Cysts
793. Megaloblastic Anemia, Finnish Type
794. Megaloblastic Anemia, Thiamine-Responsive
795. MELAS Syndrome
796. Melnick-Needles syndrome
797. Membranoproliferative Glomerulonephritis, Type II and Dense Deposit Disease
798. Meningioma, Li-Fraumeni Syndrome
799. Menkes Disease, Mild
800. MERRF Syndrome
801. MERRF/MELAS Overlap Syndrome
802. Metachromatic Leukodystrophy, Atypical
803. Metachromatic Leukodystrophy, Adult
804. Metachromatic Leukodystrophy, Juvenile
805. Metaphyseal Dysplasia without Hypotrichosis
806. Methionine Synthase Reductase Deficiency
807. Methylmalonic Aciduria, cblB type
808. Methylmalonic Aciduria and Homocystinuria, cblC Type
809. Methylmalonic Aciduria, mut(0) Type
810. Mevalonic Aciduria
811. Microhaematuria and Protinuria
812. Microphthalmia with Associated Anomalies
813. Microphthalmia, Posterior, with Retinitis Pigmentosa, Foveoschisis and Optic Disc Drusen
814. Migraine, Familial Hemiplegic
815. Migraine, Familial Hemiplegic with Progressive Cerebellar Ataxia
816. Migraine, Sporadic Hemiplegic with Progressive Cerebellar Ataxia
817. Mitochondrial Complex 1 Deficiency
818. Mitochondrial Cytochrome c Oxidase Deficiency
819. Mitochondrial DNA Depletion Syndrome, Hepatocerebral Form
820. Mitochondrial Myopathy and Sideroblastic Anemia

821. Mitochondrial Neurogastrointestinal Encephalopathy
822. Mitochondrial Neurogastrointestinal Encephalopathy without Leukoencephalopathy
823. Miyoshi Myopathy
824. ML / LEOPARD Syndrome
825. Monilethrix
826. Mucolipidosis, Type II
827. Mucolipidosis, Type II, Alpha/Beta
828. Mucolipidosis, Type III
829. Mucolipidosis, Type III, Alpha/Beta
830. Mucolipidosis, Type III, Gamma
831. Mucolipidosis, Type IIIC
832. Mucolipidosis, Type IV
833. Mucopolysaccharidosis, Type II, Severe Form
834. Mucopolysaccharidosis, Type IVA
835. Mucopolysaccharidosis, Type IVA, Mild
836. Mucopolysaccharidosis, Type VI
837. Multi-minicore Disease
838. Multiple Acyl-CoA Dehydrogenase Deficiency
839. Multiple Carboxylase Deficiency, Biotin-responsive
840. Multiple Cutaneous and Uterine Leiomyomata
841. Multiple Diastrophic Dysplasia
842. Multiple Endocrine Neoplasia, Type I
843. Multiple Endocrine Neoplasia, Type I, Burin Variant, Prolactinoma, Hyperparathyroid, Carcinoid Syndrome
844. Multiple Endocrine Neoplasia, Type IIA, Pheochromocytoma
845. Multiple Endocrine Neoplasia, Type IV
846. Multiple Epiphyseal Dysplasia
847. Muscle Weakness, Atrial Fibrilation, Hypertriglyceridaemia
848. Muscle-eye-brain Disease
849. Muscle-eye-brain Like Disease
850. Muscular Dystrophy, Becker
851. Muscular Dystrophy, Duchenne
852. Muscular Dystrophy, Emery-Dreifuss
853. Muscular Dystrophy, Limb Girdle, Type 1A, Myotilinopathy
854. Muscular Dystrophy, Limb Girdle, Type 1B
855. Muscular Dystrophy, Limb Girdle, Type 1C
856. Muscular Dystrophy, Limb Girdle, Type 2B
857. Muscular Dystrophy, Limb Girdle, Type 2D
858. Muscular Dystrophy, Limb Girdle, Type 2E
859. Muscular Dystrophy, Limb Girdle, Type 2I
860. Muscular Dystrophy, Limb Girdle, Type 2K
861. Muscular Dystrophy, Merosin Deficient
862. Myasthenic Syndrome
863. Myeloperoxidase Deficiency
864. Myoclonus-Dystonia Syndrome
865. Myofibrillar Myopathy, ZASP related
866. Myopathy
867. Myopathy due to Muscle Phosphoglycerate Mutase Deficiency
868. Myopathy, Desmin related
869. Myopathy, Distal, with Rimmed Vacuoles
870. Myopathy, Early-onset and Progeria
871. Myopathy, Mitochondrial, Late-onset
872. Myopathy, Mitochondrial, with Diabetes Mellitus
873. Myopathy, Variable, Inducable with Anesthesia
874. Myotilinopathy
875. Myotonia Fluctuans
876. Myotonic Dystrophy, Type 1
877. Nail-Patella Syndrome
878. Naxos Disease
879. Nemaline Myopathy
880. Neonatal Adrenoleukodystrophy
881. Neonatal Alloimmune Thrombocytopenic Purpura, Posttransfusion Purpura
882. Neonatal Death, Leigh Syndrome
883. Nephrolithiasis, Hypercalciuric
884. Nephronophthisis
885. Nephronophthisis 1
886. Nephronophthisis, Familial Juvenile
887. Nephrotic Syndrome, Steroid Resistant
888. Netherton Syndrome
889. Neural Tube Defects
890. Neuroblastoma
891. Neurofibromatosis, Type I
892. Neurofibromatosis 1–like Phenotype
893. Neurofibromatosis, Type II
894. Neuronal Ceroid Lipofuscinosis
895. Neuronal Ceroid Lipofuscinosis, Late Infantile
896. Neuropathy, Axonal, with Vocal Cord Paresis
897. Neuropathy, Axonal, Distal Hereditary Motor, Type IIB
898. Neuropathy, Distal Hereditary Motor, Type V
899. Neuropathy, Hereditary Sensory, Type I
900. Neuropathy, Hereditary Sensory, Type II
901. Neuropathy, Hereditary Sensory and Autonomic, Type V, Loss of Pain & Temperature Perception
902. Neuropathy with Liability to Pressure Palsies
903. Neutropenia, Congenital
904. Neutropenia, Cyclic
905. Neutropenia, Nonimmune Chronic Idiopathic, of Adults
906. Neutropenia, Severe Congenital
907. Nevus, Epidermal, Epidermolytic Hyerkeratotic Type
908. Niemann-Pick Disease, Type A
909. Niemann-Pick Disease, Type B
910. Niemann-Pick Disease, Type C1
911. Niemann-Pick Disease, Type C2
912. Niemann-Pick Disease, Variant Type C1
913. Nijmegen Breakage Syndrome
914. Nocturnal Frontal Lobe Epilepsy, Type 4, with Nocturnal Wandering and Ictal Fear
915. Noncompaction, Left Ventricular, associated with Congenital Heart Defects
916. Noncompaction, Left Ventricular, Isolated
917. Noncompaction, Left Ventricular Myocardium, Familial Isolated
918. Non-heterotaxy Cardiac Malformation
919. Nonsyndromic Hearing Impairment
920. Noonan Syndrome
921. Noonan Syndrome 3
922. Noonan Syndrome, LEOPARD Syndrome
923. Normal Tension Glaucoma
924. Norrie Disease
925. Nucleoside Phosphorylase Deficiency
926. Obesity, Hyperphagia, and Developmental Delay
927. Obesity, Severe
928. Obesity, Morbid, with Hypogonadism
929. Occlusive Cerebrovascular Disease
930. Oculofaciocardiodental Syndrome
931. Odontoonychodermal Dysplasia
932. Omenn Syndrome
933. Ophthalmoplegia, Progressive External with Hypogonadism
934. Opitz-Kaveggia Syndrome
935. Optic Atrophy 1
936. Optic Atrophy 1 with Deafness
937. Optic Atrophy, Deafness, Opthalmoplegia, and Myopathy
938. Optic Atrophy and Cataract
939. Ornithine Transcarbamylase Deficiency
940. Osteogenesis Imperfecta, Type I
941. Osteogenesis Imperfecta, Type II
942. Osteogenesis Imperfecta, Type III
943. Osteogenesis Imperfecta, Type IV
944. Osteogenesis Imperfecta/Ehlers-Danlos Crossover Syndrome
945. Osteopetrosis
946. Osteopetrosis, Type 2
947. Osteoporosis-pseudoglioma Syndrome
948. Otopalatodigital Syndrome 2
949. Pachyonychia Congenita, Type 1
950. Pachyonychia Congenita, Type 2
951. Palmoplantar Keratoderma, Epidermolytic
952. Pancreatitis, Chronic
953. Pantothenate Kinase-associated Neurodegeneration, Atypical Pantothenate Kinase-associated Neurodegeneration
954. PAPA syndrome
955. Papillon-Lefevre Syndrome
956. Paragangliomas, Phaeochromocytoma
957. Paramyotonia congenital
958. Parkinson Disease
959. Parkinson Disease, Early-onset
960. Parkinsonism, Juvenile, Autosomal Recessive
961. Paroxysmal Extreme Pain Disorder
962. Partial Atrioventricular Septal Defect and Heterotaxy Syndrome
963. Peeling Skin Syndrome
964. Pelizaeus-Merzbacher Disease, Mild
965. Pendred Syndrome
966. Periodic Fever, Autosomal Dominant
967. Periodontitis, Juvenile
968. Periventricular Heterotopia with Microcephaly
969. Peroxisome Biogenesis Disorder
970. Peroxisome Biogenesis Disorder, Complementation Group 3
971. Peroxisome Biogenesis Disorder, Complementation Group 8
972. Peroxisome Biogenesis Disorder, Complementation Group 9
973. Persistence of Fetal Hemoglobin
974. Peters' Anomaly
975. Peutz-Jeghers Syndrome
976. Pfeiffer Syndrome, Jackson-Weiss Syndrome
977. Pfeiffer Syndrome, Type III
978. Phenylketonuria
979. Pheochromocytoma
980. Phosphoserine Phosphatase Deficiency
981. Piebaldism with Sensorineural Deafness
982. Pigmentary Retinopathy and Sensorineural Deafness
983. Pigmented Nodular Adrenocortical Disease, Primary
984. Pigmented Paravenous Chorioretinal Atrophy

985. Pitt-Hopkins Syndrome
986. Placental Aromatase deficiency
987. Platelet Glycoprotein IV Deficiency
988. Polycystic Lipomembranous Osteodysplasia with Sclerosing Leukoencephalopathy
989. Polycythemia Vera
990. Polycythemia, Chuvash Type
991. Polymicrogyria, Bilateral Frontoparietal
992. POR Deficiency
993. Porphyria Cutanea Tarda
994. Porphyria, Acute Hepatic, Delta-aminolevulinate Dehydratase Porphyria
995. Porphyria, Acute Intermittent
996. Porphyria, Acute Intermittent, Nonerythroid Variant
997. Porphyria, Hepatoerythropoietic
998. Porphyria, Variegate
999. Postanesthetic Apnea
1000. Posterior Polymorphons Corneal Dystrophy 1
1001. Prealbumin Chicago, Euthyroid Dystransthyretinemic Hyperthyroxinemia
1002. Precocious Puberty, Male-Limited
1003. Premature Ovarian Failure
1004. Primary Congenital Glaucoma
1005. Primary Spontaneous Pneumothorax
1006. Progressive External Ophthalmoplegia with Mitochondrial DNA Deletions
1007. Progressive External Ophthalmoplegia with Mitochondrial DNA Deletions, Type 2
1008. Progressive Familial Intrahepatic Cholestasis, Benign Recurrent Intrahepatic Cholestasis
1009. Progressive Supranuclear Palsy
1010. Progressive Supranuclear Palsy, Parkinson Disease
1011. Prolonged Bleeding Time due to Plasminogen Activator Inhibitor 1 Deficiency
1012. Properdin Deficiency, Type I
1013. Properdin Deficiency, Type II
1014. Propionic Acidemia
1015. Protoporphyria, Erythropoietic
1016. Pseudoachondroplasia
1017. Pseudohermaphroditism, Leydig Cell Hypoplasia
1018. Pseudohermaphroditism, Male
1019. Pseudohypoaldosteronism, Type 1
1020. Pseudohypoparathyroidism 1a, with Testotoxicosis
1021. Pseudorheumatoid Dysplasia, Progressive
1022. Pseudovaginal Perineoscrotal Hypospadias
1023. Pseudoxanthoma Elasticum
1024. Pulmonary Arterial Hypertension, Primary, Dexfenfluramine-associated
1025. Pulmonary Hypertension, Primary
1026. Pulmonary Surfactant Metabolism Dysfunction, Type 2, Respiratory Insufficiency, Infantile-onset Progressive
1027. Pulmonary Toxicity when Exposed to Thioureas
1028. Pycnodysostosis
1029. Pyridoxine Responsive Homocystinuria
1030. Pyruvate Carboxylase Deficiency
1031. Pyruvate Dehydrogenase E1-alpha Deficiency
1032. Pyruvate Dehydrogenase E1-beta Deficiency
1033. Pyruvate Kinase Deficiency
1034. Pyruvate Kinase Deficiency, Amish Type
1035. RAPADILINO Syndrome
1036. Refsum Disease
1037. Refsum Disease, Infantile Form
1038. Reifenstein Syndrome
1039. Renal Glucosuria
1040. Renal Tubular Dysgenesis
1041. Restrictive Cardiomyopathy
1042. Retinal Degeneration in Ciliopathies
1043. Retinal Degeneration with Early Macular Involvement
1044. Retinitis Pigmentosa
1045. Retinitis Pigmentosa 1
1046. Retinitis Pigmentosa 2
1047. Retinitis Pigmentosa 4
1048. Retinitis Pigmentosa 19
1049. Retinitis Pigmentosa with Perivascular Retinal Pigment Epithelium Atrophy
1050. Retinitis Pigmentosa without Hearing Loss
1051. Retinitis Pigmentosa, Digenic
1052. Retinitis Pigmentosa, Late-onset
1053. Retinitis Punctata Albescens
1054. Retinitis Punctata Albescens, Newfoundland Rod-cone Dystrophy
1055. Retinoblastoma
1056. Retinoblastoma, Incomplete Penetrance Type
1057. Rheumatoid Arthritis, Juvenile, Systemic Onset
1058. Rhizomelic Chondrodysplasia Punctata, Type 2
1059. Rhizomelic Chondrodysplasia Punctata, Type 3
1060. Rickets, Vitamin D-dependent, Type I
1061. Rickets, Vitamin D-dependent, Type II
1062. Rickets, Vitamin D-resistant, Type I
1063. Ring Dermoid of the Cornea
1064. Rippling Muscle Disease 2
1065. Robinow Syndrome
1066. Rod Monochromacy
1067. Romano-Ward Syndrome
1068. Rothmund-Thomson Syndrome
1069. Roussy-Levy Syndrome
1070. Rubinstein-Taybi Syndrome
1071. Saddan Dysplasia
1072. Salt-Wasting Congenital Adrenal Hyperplasia
1073. Sandhoff Disease, Infantile Type
1074. Scheie Syndrome
1075. Schnyder Crystalline Corneal Dystrophy
1076. Schwannomatosis
1077. Scott Syndrome
1078. Seckel Syndrome
1079. Segawa Syndrome
1080. Senile Amyloidosis Inclusion Body Myositis
1081. Senior-Loken Syndrome
1082. Sensory Ataxic Neuropathy, Dysarthria, and Ophthalmoparesis
1083. Severe Combined Immunodeficiency (SCID), Athabascan-Type
1084. Severe Combined Immunodeficiency (SCID), B Cell-negative
1085. Severe Combined Immunodeficiency (SCID), B Cell-negative, T Cell-negative, NK Cell-negative
1086. Severe Combined Immunodeficiency (SCID), B Cell-positive T Cell-negative, NK Cell-positive
1087. Severe Metachromatic Leukodystrophy
1088. Shah-Waardenburg Syndrome
1089. Short Chain Acyl-CoA-dehydrogenase Deficiency
1090. Shwachman-Diamond Syndrome
1091. Sialidosis, Type II
1092. Sialuria
1093. Sickle Cell Disease (Sickle Cell Anemia)
1094. Sickle Cell Trait
1095. Sideroblastic Anemia and Spinocerebellar Ataxia
1096. Sideroblastic Anemia, Hereditary
1097. Simple Virilizing Congenital Adrenal Hyperplasia
1098. Simpson-Golabi-Behmel Syndrome
1099. Sinus Bradycardia Syndrome, Familial
1100. Sinus Node Disease
1101. Sitosterolemia
1102. Sjogren-Larsson Syndrome
1103. Skeleton-Skin-Brain Syndrome
1104. Skin Fragility-Woolly Hair Syndrome
1105. Small Patella Syndrome
1106. Smith-Lemli-Opitz Syndrome
1107. Spastic Paralysis, Infantile-onset
1108. Spastic Paraplegia
1109. Spastic Paraplegia 3
1110. Spastic Paraplegia 10
1111. Spastic Paraplegia, Autosomal Dominant
1112. Spherocytosis, Hereditary
1113. Spherocytosis, Hereditary, due to Protein 4.2-Notame
1114. Spherocytosis, Hereditary, Japanese Type
1115. Spinal Muscular Atrophy, Distal, Childhood-onset
1116. Spinal Muscular Atrophy, Type I
1117. Spondyloepiphyseal Dysplasia Tarda
1118. Spondyloepiphyseal Dysplasia Tarda and Arthropathy
1119. Spondyloepiphyseal Dysplasia, Omani Type
1120. Spongiform Encephalopathy with Neuropsychiatric Features
1121. Stargardt Disease
1122. Stargardt Disease, Cone-rod Dystrophy 3
1123. Stargardt Disease, Mild
1124. Steroid-5 Alpha-reductase Deficiency
1125. Stiff Skin Syndrome
1126. Subcortical Laminar Heterotopia/Pachygyria
1127. Superoxide Dismutase, Elevated Extracellular
1128. Supravalvular Aortic Stenosis
1129. Symphalangism
1130. Symphalangism, Proximal
1131. Symphalangism, Type 1
1132. Synpolydactyly 1
1133. Tangier Disease
1134. Tarsal-Carpal Coalition Syndrome
1135. Tay-Sachs Disease
1136. Tay-Sachs Disease, AB Variant
1137. Tay-Sachs Disease, B1 Variant
1138. Thalassemia Beta
1139. Thalassemia Beta-Plus
1140. Thalassemia Delta
1141. Thanatophoric Dysplasia, Type I
1142. Thanatophoric Dysplasia, Type II
1143. Thin Basement Membrane Disease

1144. Thoracic Aortic Aneurysm and Dissection
1145. Thrombocythemia, Essential
1146. Thrombocytopenia
1147. Thrombocytopenia 1
1148. Thrombocytopenia with associated Acute Myeloid Leukemia
1149. Thrombocytosis
1150. Thrombophilia due to Heparin Cofactor II Deficiency
1151. Thrombophilia due to Plasminogen Deficiency
1152. Thrombotic Thrombocytopenic Purpura, Congenital
1153. Thyroid Hormone Resistance, Generalized
1154. Thyroxine-binding Globulin Deficiency, Partial
1155. Thyroxine-binding Globulin Deficiency, Slow
1156. Tibial Muscular Dystrophy, Tardive
1157. Timothy Syndome
1158. Tolbutamide Poor Metabolizer
1159. Total Iodide Organification Defect
1160. Tourette Syndrome
1161. Tourette Syndrome Facial Tic
1162. TPMT Deficiency
1163. Transcobalamin II Deficiency
1164. Transient Bullous Dermolysis of the Newborn
1165. Transposition of the Great Arteries, Dextro-looped
1166. Treacher-Collins Syndrome
1167. Trichorhinophalangeal Syndrome, Type I
1168. Trichothiodystrophy Xeroderma Pigmentosum, Group D
1169. Trichotillomania
1170. Trigonocephaly Antley-Bixler Syndrome
1171. Trimethylaminuria
1172. Troyer Syndrome
1173. Tuberculoid Leprosy versus Lepromatous Leprosy
1174. Tuberous Sclerosis
1175. Tumoral Calcinosis, Hyperphasphatemic
1176. Tyrosinemia, Type I
1177. Tyrosinemia, Type II
1178. Tyrosinemia, Type III
1179. Unna-Thost Disease
1180. Urea Transport Defect JK-Null Variant
1181. Usher Syndrome, Type 1B
1182. Usher Syndrome, Type 1C
1183. Usher Syndrome, Type 1D
1184. Usher Syndrome, Type 1F
1185. Usher Syndrome, Type 1G
1186. Usher Syndrome, Type 2A
1187. Usher Syndrome, Type 2C
1188. Usher Syndrome, Type 3
1189. UV-sensitive Syndrome
1190. Vohwinkel Syndrome
1191. Von Hippel-Lindau syndrome
1192. Von Willebrand Disease, Type I
1193. Von Willebrand Disease, Type IIB
1194. Von Willebrand Disease, Type IIM
1195. Von Willebrand Disease, Type III
1196. Waardenburg Syndrome, Type IIA
1197. Waardenburg Syndrome, Type III
1198. Waardenburg Syndrome, Type IVA
1199. Walker-Warburg Syndrome
1200. Werner Syndrome
1201. Werner Syndrome, Atypical
1202. WHIM Syndrome
1203. Wilson Disease
1204. Wiskott-Aldrich Syndrome
1205. Wiskott-Aldrich Syndrome, Attenuated
1206. Wolff-Parkinson-White Syndrome, Hereditary
1207. Wolman Disease
1208. Xanthinuria, Type I
1209. Xeroderma Pigmentosum, Complementation Group C
1210. Xeroderma Pigmentosum, Complementation Group D
1211. Xeroderma Pigmentosum, Complementation Group E
1212. Xeroderma Pigmentosum, Complementation Group G
1213. XRCC3 Deficiency
1214. XY Sex Reversal with Adrenal Insufficiency
1215. XY Sex Reversal without Adrenal Insufficiency
1216. Zellweger Syndrome
1217. Zellweger Syndrome, Complementation Group G

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Your Genetic Testing Data

Variant ID column lists the exact position that was tested for within the gene listed in column #2. This variant ID can be thought of as the exact "GPS coordinate" within the gene.

Gene column refers to the gene that's being tested for in that row.

No Risk column refers to the letter of the genetic code that is usually not associated with having an increased or decreased risk of the disease, condition, or trait listed in column #6.

Risk column refers to the letter of the genetic code that is likely to be associated with having either an increased or decreased risk of the disease, condition, or trait listed in column #6.

Your Genetic Makeup column refers to the exact letters of YOUR genetic makeup at that position (column #1) in that gene (column #2). Single most genes come in pairs, there are usually two letters at each position. If only one letter is listed, this means you only have one copy of that gene (which is perfectly normal for some genes).

The letters of the genetic code are G, A, T, and C. You may also see an I (Insertion) or D (Deletion). Two dashed lines "--" means that variant's data did not pass quality control and therefore the data was excluded from your analysis.

Condition / Trait Assessed column lists what exactly is being analyzed at that specific position within the gene listed in column #2.

Reference(s) column refers to the scientific research studies that found that the specific position (column #1) within the gene (column #2) is associated with the specific disease, condition, or trait listed in column #6. You can find these papers by either searching pubmed.com or Google for the reference listed (one at a time) along with the Gene and the name of the condition.

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
64837	DKC1	C	T	C	Zinsser-Cole-Engman Syndrome	Knight (1999), Heiss (2001), Yaghmai (2000)
65133	PEX3	T	G	TT	Zellweger Syndrome, Complementation Group G	Muntau (2000), Ghaedi (2000)
65202	PEX10	C	T	CC	Zellweger Syndrome, Complementation Group 7	Warren (1998)
65173	PEX13	G	A	GG	Zellweger Syndrome H	Shimozawa (1999)
65174	PEX13	T	G	TT	Zellweger Syndrome H	Krause (2006)
65147	PEX5	C	T	CC	Zellweger Syndrome	Dodt (1995)
65213	PEX14	C	T	CC	Zellweger Syndrome	Shimozawa (2004)
63627	PEX26	G	A	GG	Zellweger Syndrome	Matsumoto (2003)
800963	PEX13	G	A	GG	Zellweger Syndrome	Shimozawa (1999)
30450	PEX1	C	T	CC	Zellweger Syndrome	Walter (2001)
30454	PEX1	C	G,T	CC	Zellweger Syndrome	Rosewich (2005)
33640	PXMP3	G	A,C	GG	Zellweger Syndrome	Shimozawa (1992), Shimozawa (1993), Moser (1995), Brul (1988)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
71915	PEX1	D	I	DD	Zellweger syndrome	Gould (1999)
71916	PEX1	I	D	II	Zellweger syndrome	Maxwell (2002)
30438	PEX1	C	G	CC	Zellweger syndrome	Rosewich (2005)
30439	PEX1	G	A	GG	Zellweger syndrome	Tamura (2001)
30442	PEX1	A	G	AA	Zellweger Syndrome	Tamura (1998)
30445	PEX1	G	A	GG	Zellweger syndrome	Collins (1999)
30446	PEX1	A	G	AA	Zellweger syndrome	Rosewich (2005)
102080	PEX1	C	T	CC	Zellweger Syndrome	Reuber (1997)
30449	PEX1	G	A,C,T	GG	Zellweger syndrome	Preuss (2002)
30450	PEX1	C	T	CC	Zellweger syndrome	Walter (2001)
30454	PEX1	C	G,T	CC	Zellweger syndrome	Rosewich (2005)
30456	PEX1	G	C	GG	Zellweger syndrome	Rosewich (2005)
33638	PXMP3	G	A	GG	Zellweger Syndrome	Krause (2006)
33640	PXMP3	G	A,C	GG	Zellweger Syndrome	Shimozawa (1992)
33641	PXMP3	G	A,T	GG	Zellweger Syndrome	Shimozawa (1998)
63627	PEX26	G	A	GG	Zellweger syndrome	Matsumoto (2003)
65147	PEX5	C	T	CC	Zellweger Syndrome	Dodt (1995)
65197	PEX10	G	A,C	GG	Zellweger Syndrome	Steinberg (2004)
65198	PEX10	C	A,T	CC	Zellweger Syndrome	Steinberg (2004)
65200	PEX10	G	C	GG	Zellweger Syndrome	Steinberg (2004)
65201	PEX10	C	T	CC	Zellweger Syndrome	Krause (2006)
65213	PEX14	C	T	CC	Zellweger Syndrome	Shimozawa (2004)
14075	NR5A1	C	T	CC	XY sex reversal, without adrenal failure	Lin (2007)
14074	NR5A1	D	I	DD	XY sex reversal, without adrenal failure	Achermann (1999)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
14075	NR5A1	C	T	CC	XY sex reversal, without adrenal failure	Lin (2007)
14077	NR5A1	C	T	CC	XY sex reversal, without adrenal failure	Lin (2007)
14078	NR5A1	C	T	CC	XY sex reversal, without adrenal failure	Lin (2007)
14081	NR5A1	A	T	AA	XY sex reversal, without adrenal failure	Lin (2007)
14076	NR5A1	G	A,T	GG	XY Sex Reversal without Adrenal Failure	Mallet (2004)
14079	NR5A1	C	T	CC	XY Sex Reversal with Adrenal Failure	Achermann (2002)
14398	CYP11A1	G	A	GG	XY sex reversal and adrenal insufficiency	Al Kandari (2006)
50230	XRCC3	C	T	CC	XRCC3 deficiency	Rafii (2003)
9449	ERCC5	G	A	GG	Xeroderma Pigmentosum, Group G	Emmert (2002)
9457	ERCC5	G	T	GG	Xeroderma Pigmentosum, Group G	Nouspikel (1994)
9456	ERCC5	T	C	TT	Xeroderma Pigmentosum, Group G	Cheesbrough (1978), Keijzer (1979), Constantinou , (1999), Lalle (2002)
78790	ERCC2	C	T	CC	Xeroderma Pigmentosum, Group D	Botta (1998), Botta (1998), Broughton (2001)
51400	XPC	G	A	GG	Xeroderma Pigmentosum, Complementation Group C	Chavanne (2000), Chavanne (2000), Gozukara (2001), Gozukara (2001)
43340	DDB2	G	T	GG	Xeroderma Pigmentosum (E)	Rapic-Otrin (2003)
43342	DDB2	T	C	TT	Xeroderma Pigmentosum (E)	Rapic-Otrin (2003)
9454	ERCC5	C	T	CC	Xeroderma pigmentosum	Clarkson (1994), Clarkson (1994), Nouspikel (1997)
73977	XDH	G	A	GG	Xanthinuria, Type I	Ichida (1997)
20367	SLC4A1	C	G,T	CC	Wright Blood Group Antigen	Bruce (1995)
62394	WFS1	G	A	GG	Wolfram-like Syndrome	Eiberg (2006)
62309	WFS1	T	G	TT	Wolframin variant	Torres (2001)
62311	WFS1	C	T	CC	Wolframin variant	Crawford (2002)
62315	WFS1	A	T	AA	Wolframin variant	Crawford (2002)
62317	WFS1	G	A	GG	Wolframin variant	Torres (2001)
62332	WFS1	C	T	CC	Likely harmless variant (previously associated with Wolframin variant)	Crawford (2002), GeneDx (2014), Laboratory for Molecular Medicine of Partners HealthCare Personalized Medicine (2015), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2015)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
62336	WFS1	C	T	CC	Wolframin variant	Torres (2001)
62340	WFS1	G	A	GG	Wolframin variant	Torres (2001)
62353	WFS1	A	G	AA	Wolframin variant	Crawford (2002)
62364	WFS1	C	T	CC	Wolframin variant	Crawford (2002)
2842	PRKAG2	G	C	GG	Wolff-Parkinson-White Syndrome	Gollob (2001)
60379	WAS	C	T	C	Wiskott-Aldrich Syndrome	Basile (1996), Ho (2001)
60398	WAS	T	C	T	Wiskott-Aldrich Syndrome	Kolluri (1995)
15105	ATP7B	C	A,T	CC	Wilson Disease	Kim (1998), Kusuda (2000), Wu (2001), Gu (2003)
15152	ATP7B	A	G	AA	Wilson Disease	Shah (1997), Garcia-Villarreal (2000)
15195	ATP7B	G	A	GG	Wilson Disease	Yamaguchi (1998), Takeshita (2002)
15209	ATP7B	G	A	GG	Wilson Disease	Wu (2001)
15216	ATP7B	C	T	CC	Wilson Disease	Figus (1995), Panagiotakaki (2004)
15248	ATP7B	G	A,T	GG	Wilson Disease	Tanzi (1993), Houwen (1995)
15306	ATP7B	C	T	CC	Wilson Disease	Thomas (1995), Thomas (1995)
15310	ATP7B	T	A,C	TT	Wilson Disease	Tanzi (1993), Thomas (1995), Shah , (1997), Okada , (2000), Yoo (2002)
15372	ATP7B	C	G	CC	Wilson Disease	Thomas (1995), Wilson (2000)
72331	ATP7B	A	G	AA	Wilson Disease	Davies (2008)
60955	CXCR4	G	A	GG	WHIM Syndrome	Hernandez (2003)
60956	CXCR4	C	A	CC	WHIM syndrome	Gulino (2004)
60958	CXCR4	C	A	CC	WHIM syndrome	Hernandez (2003)
800862	LMNA	G	T	GG	Werner Syndrome, Atypical	Brown , (2001), Vigouroux , (2001), Caux (2003), Chen (2003), Hegele (2003), Chen (2003), Vigouroux (2003), Caux (2003), Chen (2003), Jacob (2005), Jacob (2005)
61899	RECQL2	C	T	CC	Werner Syndrome	Oshima (1996), Muftuoglu (2008), Invitae (2015)
61955	RECQL2	C	T	CC	Werner Syndrome	Yu (1996)
61958	RECQL2	G	C	GG	Werner Syndrome	Yu (1996), Matsumoto (1997), Huang (2006)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
63282	POMT1	C	T	CC	Walker-Warburg Syndrome	Cormand (2001), Bernabe (2002)
64963	POMT2	C	A,T	CC	Walker-Warburg Syndrome	Mercuri (2006)
64965	POMT2	C	T	CC	Walker-Warburg Syndrome	Mercuri (2006)
20349	SLC4A1	C	G,T	CC	Waldner Blood Group Antigen	Bruce (1995), Jarolim (2001)
15670	EDNRB	C	A	CC	Waardenburg-Shah Syndrome	Puffenberger (1994)
15402	EDN3	C	A	CC	Waardenburg-Hirschsprung Disease	Pingault (2001)
63857	MITF	C	T	CC	Waardenburg Syndrome, Type IIA	Lalwani (1998), Nobukuni (1996)
62409	PAX3	C	G	CC	Waardenburg syndrome, Type 3	Tassabehji (1993)
18433	VWF	G	A,T	GG	Von Willebrand Disease, Type III	Zhang (1992), Zhang (1992), Zhang (1992)
48799	VHL	C	T	CC	Von Hippel-Lindau Syndrome	Crossey (1994), Kuzmin (1995), Crossey (1995), Zbar (1996), Garcia (1997), Neumann (2002), Neumann (2002)
48800	VHL	G	A	GG	Von Hippel-Lindau syndrome	Crossey (1994), Kuzmin (1995), Crossey (1995), Zbar (1996), Garcia (1997), Neumann (2002), Neumann (2002)
25922	GJB2	C	G	CC	Vohwinkel Syndrome	Vohwinkel (1929), Wigley (1929), Korge (1997), Maestrini (1999), Maestrini (1999)
53295	BEST1	A	C	AA	Vitelliform Macular Dystrophy	Petrukhin (1998), Kramer (2000)
53278	BEST1	C	T	CC	Vitelliform Macular Dystrophy	Kramer (2000), Kramer (2000)
22380	GGCX	A	C	AA	Vitamin K-Dependent Coagulation Defect	Brenner (1990), Brenner (1998), Mutucumarana (2000)
34144	CYP27B1	C	T	CC	Vitamin D-dependent Rickets, Type I	Wang (1998), Wang , (1998), Wang (2002), Wang , (2002)
34147	CYP27B1	G	A	GG	Vitamin D-dependent Rickets, Type I	Wang (1998), Wang (2002)
78406	ROBO2	G	A	GG	Vesicoureteral Reflux	Lu (2007), Lu (2007)
78405	ROBO2	T	C	TT	Vesicoureteral Reflux	Lu (2007)
20719	SERPINA10	G	T	GG	Venous Thromboembolic Disease	Water (2004)
20720	SERPINA10	A	C,G	AA	Venous Thromboembolic Disease	Water (2004)
20721	SERPINA10	C	T	CC	Venous Thromboembolic Disease	Water (2004)
100577	ERCC6	G	A	GG	UV-sensitive Syndrome	Fujiwara (1981), Horibata (2004)
62058	USH3A	A	C	AA	Usher Syndrome, Type III	Fields (2002), Adato (2002), Ness (2003)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
62071	USH3A	A	T	AA	Usher Syndrome, Type III	Joensuu (2001), Fields (2002)
62073	USH3A	A	C,G,T	AA	Usher Syndrome, Type III	Fields (2002), Joensuu (2001)
60707	USH2A	G	A,C	GG	Usher Syndrome, Type IIA	Adato (2000), Auslender (2008)
60732	USH2A	G	A	GG	Usher Syndrome, Type IIA	Kaiserman (2007), Auslender (2008)
100579	USH2A	T	C	TT	Usher Syndrome, Type IIA	Auslender (2008)
100580	USH2A	I	D	II	Usher Syndrome, Type IIA	Eudy (1998), Liu (1999), Weston (2000), Dreyer (2001), Ouyang (2004)
41356	MASS1	C	T	CC	Usher Syndrome, Type 2C	Weston (2004)
74943	USH1G	C	T	CC	Usher Syndrome, Type 1G	Ouyang (2005)
74938	PCDH15	G	A,T	GG	Usher Syndrome, Type 1F	Ben-Yosef (2003)
74940	PCDH15	I	D	II	Usher Syndrome, Type 1F	Zheng (2005)
60661	USH1C	C	A	CC	Usher Syndrome, Type 1c	Zwaenepoel (2001)
60663	USH1C	C	G,T	CC	Usher Syndrome, Type 1c	Bitner-Glindzicz (2000), Savas (2002)
100581	USH1C	D	I	DD	Usher Syndrome, Type 1c	Verpy (2000), Bitner-Glindzicz (2000), Zwaenepoel (2001)
60556	MYO7A	C	T	CC	Usher Syndrome, Type 1b	Weston (1996)
60586	MYO7A	C	T	CC	Usher Syndrome, Type 1b	Ouyang (2005)
60644	MYO7A	G	C	GG	Usher Syndrome, Type 1b	Ouyang (2005)
60566	MYO7A	G	A	GG	Likely harmless variant (previously associated with Usher syndrome 1b)	Weston (1996), Laboratory for Molecular Medicine of Partners HealthCare Personalized Medicine (2010), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2014), Illumina Clinical Services Laboratory (2016)
74962	CDH23	C	T	CC	Usher Syndrome 1	Astuto (2002)
74959	CDH23	G	A	GG	Usher Syndrome 1	Astuto (2002), Brouwer (2003), Cremers (1989)
74960	CDH23	G	C	GG	Usher Syndrome 1	Astuto (2002)
74961	CDH23	G	A	GG	Usher Syndrome 1	Astuto (2002)
74963	CDH23	G	A	GG	Usher Syndrome 1	Astuto (2002)
74965	CDH23	G	A	GG	Usher Syndrome 1	Astuto (2002)
74946	CDH23	I	D	II	Usher Syndrome 1	Zheng (2005), Astuto (2002)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
22148	SLC14A1	C	T	CC	Urea Transport Defect|JK-Null Variant	Lucien (1998)
7865	KRT1	T	A	TT	Unna-Thost Disease	Kimonis (1994), Kimonis (1994)
32750	HPD	A	C	AA	Tyrosinemia, Type III	Ruetschi (2000)
31395	TAT	G	A	GG	Tyrosinemia, Type II	Natt (1992)
31874	FAH	A	T	AA	Tyrosinemia, Type I	Rootwelt (1994)
31879	FAH	G	A	GG	Tyrosinemia, Type I	St-Louis (1994), (St-Louis (1996), St-Louis (1996)
31882	FAH	G	A	GG	Tyrosinemia, Type I	St-Louis (1995)
31892	FAH	G	A	GG	Tyrosinemia, Type I	Al-Dhalimy (1993), Grompe (1994), St-Louis (1995), Poudrier (1998)
31896	FAH	G	T	GG	Tyrosinemia, Type I	Rootwelt (1996)
13638	FGF23	A	G	AA	Tumoral Calcinosis, Hyperphasphatemic, Familial	Chefetz (2005)
13671	GALNT3	G	A	GG	Tumoral Calcinosis, Hyperphasphatemic, Familial	Engel (1961), Lyles (1985), Slavin (1993), Topaz (2004), Ichikawa (2005)
800879	GALNT3	C	T	CC	Tumoral Calcinosis, Hyperphasphatemic, Familial	Steinherz (1985), Topaz (2004), Frishberg (2005)
13678	GALNT3	C	G,T	CC	Tumoral Calcinosis, Hyperphasphatemic, Familial	Engel (1961), Lyles (1985), Slavin (1993), Topaz (2004), Ichikawa (2005)
63040	TSC2	A	C	AA	Tuberous Sclerosis	Khare (2001)
62994	TSC2	C	T	CC	Tuberous Sclerosis	Au (1998)
63068	TSC2	C	T	CC	Tuberous Sclerosis	Maheshwar (1997)
62905	TSC2	G	A	GG	Tuberous Sclerosis	Au (1998), Wilson (1996), Carsillo (2000)
68608	SPG20	I	D	II	Troyer Syndrome	Patel (2002)
32535	FMO3	C	T	CC	Trimethylaminuria	Dolphin (1997)
12671	FGFR1	A	G	AA	Trigonocephaly	Kress (2000), Hurley (2004), Huang (2005)
43899	SLITRK1	C	T	CC	Trichotillomania	Zuchner (2006)
43900	SLITRK1	T	C	TT	Trichotillomania	Zuchner (2006)
800976	ERCC2	C	T	CC	Trichothiodystrophy	Botta (1998), Botta (1998), Broughton (2001)
63533	TRPS1	C	T	CC	Trichorhinophalangeal Syndrome, Type I	Kaiser (2004)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
63534	TRPS1	G	A	GG	Trichorhinophalangeal Syndrome, Type I	Kaiser (2004)
62148	TCOF1	A	G	AA	Treacher-Collins Syndrome	Splendore (2002)
68564	TCOF1	I	D	II	Treacher-Collins Syndrome	Splendore (2000), Edwards (1997)
67887	THRAP2	C	T	CC	Transposition of the Great Arteries, Dextro-looped	Muncke (2003)
67892	THRAP2	T	C	TT	Transposition of the Great Arteries, Dextro-looped	Muncke (2003)
67902	THRAP2	T	C	TT	Transposition of the Great Arteries, Dextro-looped	Muncke (2003)
7602	COL7A1	C	G,T	CC	Transient Bullous Dermolysis of the Newborn	Hammami-Hauasli (1998)
100627	SLC44A2	A	G	GG	Transfusion-related Acute Lung Injury	X-ray and signs of hypoxemia in the absence of circulatory overload and other risk factors for acute lung injury (2004), Greinacher (2009)
21815	TCN2	T	G	TT	Transcobalamin II Deficiency	Li (1994)
21816	TCN2	T	G	TT	Transcobalamin II Deficiency	Bibi (1999), Namour (2003)
43898	SLITRK1	C	A,T	CC	Tourette Syndrome|Facial Tic	Abelson (2005)
44273	HCRTR2	C	T	CC	Tourette Syndrome, Possibly associated with increased risk of	Thompson (2004)
44279	HTR1A	C	A	CC	Possibly associated with Tourette Syndrome	Lam (1996)
13166	TPO	G	A	GG	Total Iodide Organification Defect	Bikker (1995), Pannain (1999)
72638	TPO	D	I	DD	Total Iodide Organification Defect	Niu (2002), Wu (2002)
70517	CACNA1C	G	A	GG	Timothy Syndome	Splawski (2005)
70518	CACNA1C	G	A	GG	Timothy Syndome	Splawski (2004), Splawski , (2004), Splawski (2005)
3340	TTN	A	G	AA	Tibial Muscular Dystrophy, Tardive	Hackman (2002)
73596	TTN	I	D	II	Tibial Muscular Dystrophy, Tardive	Hackman (2002), Hackman (2002)
14230	SERPINA7	G	A	G	Thyroxine-binding Globulin Deficiency, Partial	Miura , (1993), Inagaki (1996)
11019	THRB	C	T	CC	Thyroid Hormone Resistance, Generalized	Parrilla (1991), Beck-Peccoz (1994)
11020	THRB	C	A,T	CC	Thyroid Hormone Resistance, Generalized	Parrilla (1991), Beck-Peccoz (1994)
11021	THRB	C	T	CC	Thyroid Hormone Resistance, Generalized	Parrilla (1991), Beck-Peccoz (1994)
11052	THRB	C	T	CC	Thyroid Hormone Resistance, Generalized	Weinberger (1986), Parrilla (1991), Parrilla (1991), Parrilla (1991), Mixson (1992), Weiss (1993), Beck-Peccoz (1994), Pohlenz (1995), Pohlenz (1995)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
11090	THRB	C	T	CC	Thyroid Hormone Resistance, Generalized	Boothroyd (1991), Weiss (1993), Beck-Peccoz (1994)
15582	RET	T	C	TT	Thyroid Carcinoma, Sporadic Medullary|Multiple Endocrine Neoplasia, Type 2B|Pheochromocytoma, Somatic	Takahashi (1988), Hofstra (1994), Carlson (1994), Eng (1994), Carlson (1994)
15572	RET	T	G	TT	Thyroid Carcinoma, Sporadic Medullary	Hofstra (1997), Hofstra , (1997), Dang , (1999), Jimenez (2004)
15552	RET	G	T	GG	Thyroid Carcinoma, Familial Medullary	Berndt (1998)
15558	RET	G	T	GG	Thyroid Carcinoma, Familial Medullary	Farndon , (1986), Bolino , (1995), Bolino (1995), Ruiz (2001), Lombardo (2002), Patocs (2003)
15490	RET	G	A	GG	Thyroid Cancer	Blaugrund (1994), Decker (1998)
50488	MINPP1	A	G	AA	Thyroid Adenoma, Follicular	Gimm (2001)
5797	CBS	G	A	GG	Thrombosis, Hyperhomocysteinemic	Gaustadnes , (2000)
55552	PROZ	G	C	GG	Thrombosis	Souri (2005)
52756	PLG	G	A	GG	Possible increased risk of thrombophilia due to decreased plasminogen activity	Aoki (1978), Miyata (1982), Ichinose (1991), Kikuchi (1992), Murata (1997), Soonchunhyang University Bucheon Hospital of Soonchunhyang University Medical Center (2016)
4980	SERPIND1	C	T	CC	Thrombophilia due to Heparin Cofactor II Deficiency	Kanagawa (2001)
4966	SERPIND1	G	A	GG	Thrombophilia due to Heparin Cofactor II Deficiency	Blinder (1989)
46796	RUNX1	C	G	CC	Thrombocytopenia with associated Acute Myeloid Leukemia	Walker (2002)
22436	GATA1	G	T	G	Thrombocytopenia 1	Freson (2002)
22408	MASTL	G	C	GG	Thrombocytopenia	Drachman , (2000), Gandhi (2003)
72264	CYCS	C	T	CC	Thrombocytopenia	Morison (2008)
21041	THPO	C	A	CC	Thrombocythemia, Essential	Kikuchi (1995), Ghilardi (1999)
68516	ACTA2	C	T	CC	Thoracic Aortic Aneurysm and Dissection	Guo (2007)
68517	ACTA2	G	A	GG	Thoracic Aortic Aneurysm and Dissection	Guo (2007)
68518	ACTA2	T	G	TT	Thoracic Aortic Aneurysm and Dissection	Guo (2007)
68519	ACTA2	C	G,T	CC	Thoracic Aortic Aneurysm and Dissection	Guo (2007)
68520	ACTA2	A	G	AA	Thoracic Aortic Aneurysm and Dissection	Guo (2007)
68521	ACTA2	A	G	AA	Thoracic Aortic Aneurysm and Dissection	Guo (2007)
34918	FGFR3	A	G	AA	Thanatophoric Dysplasia, Type II	Tavormina (1995), Chesi (1997), Wilcox (1998), Liboi (2003)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
34952	FGFR3	C	T	CC	Thanatophoric Dysplasia, Type I	Tavormina (1995), Pokharel (1996), Wilcox (1998)
34964	FGFR3	A	G	AA	Thanatophoric Dysplasia, Type I	Rousseau (1996), Brodie (1998)
19757	HBD	G	T	GG	Thalassemia Delta	Papadakis (1997)
19758	HBD	T	C	TT	Thalassemia Delta	Papadakis (1997)
19759	HBD	A	G	AA	Thalassemia Delta	Papadakis (1997)
19760	HBD	A	G	AA	Thalassemia Delta	Papadakis (2003)
19761	HBD	T	A	TT	Thalassemia Delta	De Angioletti (2002)
19762	HBD	C	T	CC	Thalassemia Delta	Oggiano (1987)
19763	HBD	A	G	AA	Thalassemia Delta	Matsuda (1992)
20219	HBB	G	A	GG	Thalassemia Beta-Plus	Gonzalez-Redondo (1989), Ristaldi (1990), Murru (1993), Maragoudaki (1999)
6046	ZFPM2	A	G	AA	Tetralogy of Fallot	Pizzuti (2003)
2854	JAG1	C	T	CC	Tetralogy of Fallot	Eldadah (2001), Lu (2003)
8123	TYR	G	A	GG	Temperature Sensitive Oculocutaneous Albinism, Type 1	King (1989), Kwon (1989), Giebel (1991), Giebel (1991), Giebel (1991), Giebel (1991)
27505	HEXA	C	A,G,T	CC	Tay-Sachs Disease, B1 Variant	Suzuki (1988), Tanaka (1988), Goebel (1989), Santos (1991), Whitley (1992)
27358	GM2A	G	T	GG	Tay-Sachs Disease, AB variant	Chen (1999)
27360	GM2A	G	C	GG	Tay-Sachs Disease, AB variant	Schroder (1993)
27359	GM2A	T	C	TT	Tay-Sachs Disease, AB variant	Schroder (1991), Xie (1992), (Mahuran (1994)
100910	HEXA	I	D	II	Tay-Sachs Disease	Hogikyan (1986), Keats (1987), Hogikyan (1987), Hechtman (1990), Braekeleer (1992)
27487	HEXA	D	I	DD	Tay-Sachs Disease	Costigan (1988), Gravel (1990), Nishimoto (1991), McDowell (1992), Zlotogora (1993), Thurmon (1993), Frisch (2004)
27488	HEXA	C	G	CC	Tay-Sachs Disease	Ainsworth (1992)
27490	HEXA	C	T	CC	Tay-Sachs Disease	Ainsworth (1992)
27561	HEXA	C	G	CC	Tay-Sachs Disease	Arpaia (1988), Suzuki (1988), Myerowitz (1988), Strasberg (1997)
27566	HEXA	C	A,T	CC	Tay-Sachs Disease	Mules (1991)
27575	HEXA	C	G,T	CC	Tay-Sachs Disease	Hechtman (1992)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
27580	HEXA	C	A,G,T	CC	Tay-Sachs Disease	Akli (1991), McDowell (1992), Akerman (1992), Triggs-Raine (1992), Akli (1993), Landels (1993)
27487	HEXA	D	I	DD	Tay-Sachs Disease	Costigan (1988)
801001	HEXA	D	I	DD	Tay-Sachs Disease	Costigan (1988)
801002	HEXA	D	I	DD	Tay-Sachs Disease	Costigan (1988)
2358	ABCA1	T	C	TT	Tangier Disease	Bodzioch , (1999), Utech , (2001), Guo (2002)
59701	DNASE1	C	T	CC	Systemic Lupus Erythematosus	Bodano (2004)
59700	DNASE1	G	A	GG	Systemic Lupus Erythematosus	Bodano (2004)
61893	TREX1	G	A	GG	Systemic Lupus Erythematosus	Crow (2006), Lee-Kirsch (2007)
34748	HOXD13	C	T	CC	Synpolydactyly 1	Debeer (2002)
64796	BCOR	G	A,C	G	Syndromic Microphthalmia 2	Ng (2004)
64797	BCOR	G	A	G	Syndromic Microphthalmia 2	Ng (2004)
60013	GDF5	C	T	CC	Symphalangism	Wang (2006)
20363	SLC4A1	G	A,T	GG	Swann Blood Group Antigen	Zelinski (2000)
23702	TAP1	G	A	GG	Susceptibility to Pulmonary Toxoplasmosis	Dogu (2006)
79299	TLR2	C	T	CC	Susceptibility to Lepromatous Leprosy	Chae (2001), Bochud (2003)
49505	CHEK2	G	A,T	GG	Susceptibility to Cancer of Multiple Types |Breast|Colon|Stomach	Ingvarsson (2002)
2580	ELN	A	G	AA	Supravalvular Aortic Stenosis	Li (1997)
2569	ELN	C	G	CC	Supravalvular Aortic Stenosis	Metcalfe (2000)
2573	ELN	C	T	CC	Supravalvular Aortic Stenosis	Li (1997), Tassabehji (1997), Metcalfe (2000)
2575	ELN	C	T	CC	Supravalvular Aortic Stenosis	Li (1997), Metcalfe (2000)
2581	ELN	C	G	CC	Supravalvular Aortic Stenosis	Urban (1999)
71860	MTTF	G	A	?	Myopathy, Mitochondrial, Late-onset	Deschauer (2006)
71967	MTRNR1	T	G	?	Deafness, Nonsyndromic Sensorineural	Li (2004), Li (2004)
37846	PAFAH1B1	G	A	GG	Subcortical band heterotopia ?	D'Agostino (2002)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
30911	DCX	C	T	C	Subcortical band heterotopia	D'Agostino (2002)
30917	DCX	A	C	A	Subcortical band heterotopia	Matsumoto (2001)
30920	DCX	A	G	A	Subcortical band heterotopia	Matsumoto (2001)
30923	DCX	G	A	G	Subcortical band heterotopia	Kim (2005)
30924	DCX	T	A	T	Subcortical band heterotopia	Matsumoto (2001)
30926	DCX	G	A,C	G	Subcortical band heterotopia	D'Agostino (2002)
30929	DCX	G	A	G	Subcortical band heterotopia	Matsumoto (2001)
30933	DCX	T	A	T	Subcortical band heterotopia	Matsumoto (2001)
30936	DCX	T	C	T	Subcortical band heterotopia	Matsumoto (2001)
30938	DCX	C	A	C	Subcortical band heterotopia	Matsumoto (2001)
30941	DCX	C	A	C	Subcortical band heterotopia	Matsumoto (2001)
30946	DCX	G	A	G	Subcortical band heterotopia	Sakamoto (2000)
30947	DCX	C	A	C	Subcortical band heterotopia	Matsumoto (2001)
30954	DCX	C	T	C	Subcortical band heterotopia	Matsumoto (2001)
30955	DCX	A	C	A	Subcortical band heterotopia	Matsumoto (2001)
37841	PAFAH1B1	T	C	T	Subcortical band heterotopia	Pilz (1999), Leventer (2001)
100567	NAGPA	I	D	II	Stutter	Kang (2010)
71448	MTTV	C	T	?	Neonatal Death|Leigh Syndrome	McFarland (2002), McFarland (2002)
101287	FBN1	C	A,G	CC	Stiff Skin Syndrome	Loeys (2010)
101289	FBN1	A	C	AA	Stiff Skin Syndrome	Loeys (2010)
101063	SRD5A2	I	D	II	Steroid-5 Alpha-reductase Deficiency	Ko (2010)
60235	SRD5A2	G	A	GG	Steroid-5 Alpha-reductase Deficiency	Sasaki (2003), Ko (2010)
60236	SRD5A2	C	T	CC	Steroid-5 Alpha-reductase Deficiency	Imperato-McGinley (1979), Imperato-McGinley (1986), Thigpen (1992), Cai (1996), Ko (2010)
60256	SRD5A2	G	C	GG	Steroid-5 Alpha-reductase Deficiency	Sasaki (2003)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
60257	SRD5A2	C	G,T	CC	Steroid-5 alpha-reductase deficiency	Thigpen (1992)
60258	SRD5A2	A	T	AA	Steroid-5 Alpha-reductase Deficiency	Hochberg (1996)
60259	SRD5A2	T	C	TT	Steroid-5 alpha-reductase deficiency	Thigpen (1992)
60260	SRD5A2	G	T	GG	Steroid-5 alpha-reductase deficiency	Hafez (2003)
60261	SRD5A2	C	T	CC	Steroid-5 alpha-reductase deficiency	Vilchis (2000)
60262	SRD5A2	G	A,T	GG	Steroid-5 alpha-reductase deficiency	Hiort (2002)
60263	SRD5A2	A	T	AA	Steroid-5 alpha-reductase deficiency	Hiort (1996)
60266	SRD5A2	T	C	TT	Steroid-5 alpha-reductase deficiency	Thigpen (1992)
60267	SRD5A2	G	A	GG	Steroid-5 alpha-reductase deficiency	Nicoletti (2005)
60268	SRD5A2	C	T	CC	Steroid-5 alpha-reductase deficiency	Hackel (2005)
60269	SRD5A2	T	C	TT	Steroid-5 alpha-reductase deficiency	Mazen (2003)
60270	SRD5A2	T	A	TT	Steroid-5 alpha-reductase deficiency	Thigpen (1992)
60271	SRD5A2	C	G	CC	Steroid-5 alpha-reductase deficiency	Thigpen (1992)
60272	SRD5A2	G	A	GG	Steroid-5 alpha-reductase deficiency	Nicoletti (2005)
60273	SRD5A2	C	T	CC	Steroid-5 Alpha-reductase Deficiency	Imperato-McGinley (1979), Imperato-McGinley (1986), Gautier (1986), Thigpen (1992)
60274	SRD5A2	A	C	AA	Steroid-5 alpha-reductase deficiency	Zhou (1999)
60275	SRD5A2	T	C	TT	Steroid-5 alpha-reductase deficiency	Boudon (1995)
60278	SRD5A2	C	T	CC	Steroid-5 alpha-reductase deficiency	Anwar (1997)
60279	SRD5A2	G	T	GG	Steroid-5 alpha-reductase deficiency	Thigpen (1992)
60280	SRD5A2	G	C	GG	Steroid-5 Alpha-reductase Deficiency	Chavez (2000)
60281	SRD5A2	A	G	AA	Steroid-5 alpha-reductase deficiency	Thigpen (1992)
60282	SRD5A2	G	A	GG	Steroid-5 Alpha-reductase Deficiency	Vilchis (1997)
60283	SRD5A2	C	G,T	CC	Steroid-5 Alpha-reductase Deficiency	Nordenskjold (1998)
60284	SRD5A2	T	C	TT	Steroid-5 Alpha-reductase Deficiency	Nordenskjold (1998)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
60285	SRD5A2	T	A	TT	Steroid-5 alpha-reductase deficiency	Mazen (2003)
60286	SRD5A2	G	T	GG	Steroid-5 alpha-reductase deficiency	Vilchis (2000)
60287	SRD5A2	G	A	GG	Steroid-5 Alpha-reductase Deficiency	Imperato-McGinley (1979), Imperato-McGinley (1986), Gautier (1986), Thigpen (1992)
60288	SRD5A2	T	G	TT	Steroid-5 alpha-reductase deficiency	Nicoletti (2005)
60289	SRD5A2	T	A,C	TT	Steroid-5 alpha-reductase deficiency	Skordis (2005)
60290	SRD5A2	C	T	CC	Steroid-5 alpha-reductase deficiency	Hackel (2005)
60291	SRD5A2	C	A	CC	Steroid-5 alpha-reductase deficiency	Thigpen (1992)
12809	CYP17A1	G	A	GG	Steroid-17 alpha-hydroxylase deficiency ?	Adachi (2004)
12793	CYP17A1	A	G	AA	Steroid-17 alpha-hydroxylase deficiency	Costa-Santos (2004)
12794	CYP17A1	C	G	CC	Steroid-17 alpha-hydroxylase deficiency	Satoh (1998)
12798	CYP17A1	C	A,T	CC	Steroid-17 alpha-hydroxylase deficiency	Yanase (1995)
12805	CYP17A1	C	T	CC	Steroid-17 alpha-hydroxylase deficiency	Ergun-Longmire (2006)
12807	CYP17A1	C	A	CC	Steroid-17 alpha-hydroxylase deficiency	Rumsby (1993)
12811	CYP17A1	C	G	CC	Steroid-17 alpha-hydroxylase deficiency	Rosa (2007)
12813	CYP17A1	G	C	GG	Steroid-17 alpha-hydroxylase deficiency	Costa-Santos (2004)
12819	CYP17A1	T	A	TT	Steroid-17 alpha-hydroxylase deficiency	Monno (1993)
12823	CYP17A1	G	C	GG	Steroid-17 alpha-hydroxylase deficiency	Lam (2001)
12824	CYP17A1	C	T	CC	Steroid-17 alpha-hydroxylase deficiency	Ergun-Longmire (2006)
12825	CYP17A1	G	A	GG	Steroid-17 alpha-hydroxylase deficiency	Takeda (2001)
12829	CYP17A1	C	T	CC	Steroid-17 alpha-hydroxylase deficiency	Fardella (1994)
12830	CYP17A1	G	A,T	GG	Steroid-17 alpha-hydroxylase deficiency	Patocs (2005)
12832	CYP17A1	G	A	GG	Steroid-17 alpha-hydroxylase deficiency	Yanase (1992)
12834	CYP17A1	G	A	GG	Steroid-17 alpha-hydroxylase deficiency	Yanase (1992)
12835	CYP17A1	T	G	TT	Steroid-17 alpha-hydroxylase deficiency	Costa-Santos (2004)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
10630	CYP11B1	G	A,T	GG	Steroid-11 beta-hydroxylase deficiency	Merke (1998)
10633	CYP11B1	G	A,T	GG	Steroid-11 beta-hydroxylase deficiency	Grigorescu Sido (2005)
10635	CYP11B1	C	G	CC	Steroid-11 beta-hydroxylase deficiency	Krone (2005)
10641	CYP11B1	G	A	GG	Steroid-11 beta-hydroxylase deficiency	Solyom (2001)
10643	CYP11B1	T	A	TT	Steroid-11 beta-hydroxylase deficiency	Curnow (1993)
10650	CYP11B1	A	G,T	AA	Steroid-11 beta-hydroxylase deficiency	Krone (2005)
10651	CYP11B1	G	A	GG	Steroid-11 beta-hydroxylase deficiency	Lee (2005)
10652	CYP11B1	C	G	CC	Steroid-11 beta-hydroxylase deficiency	Kuribayashi (2005)
10653	CYP11B1	G	C	GG	Steroid-11 beta-hydroxylase deficiency	Merke (1998)
10655	CYP11B1	T	G	TT	Steroid-11 beta-hydroxylase deficiency	Lee (2005)
10659	CYP11B1	G	A	GG	Steroid-11 beta-hydroxylase deficiency	Curnow (1993)
10661	CYP11B1	G	A	GG	Steroid-11 beta-hydroxylase deficiency	Curnow (1993)
10662	CYP11B1	G	T	GG	Steroid-11 beta-hydroxylase deficiency	Krone (2006)
10665	CYP11B1	C	T	CC	Steroid-11 beta-hydroxylase deficiency	Curnow (1993)
10666	CYP11B1	G	C	GG	Steroid-11 beta-hydroxylase deficiency	Nakayama (1995)
10670	CYP11B1	A	C	AA	Steroid-11 beta-hydroxylase deficiency	Curnow (1993)
10671	CYP11B1	C	T	CC	Steroid-11 beta-hydroxylase deficiency	Motaghedi (2005)
10677	CYP11B1	C	T	CC	Steroid-11 beta-hydroxylase deficiency	Merke (1998)
51651	ABCA4	C	G	CC	Stargardt Disease, Mild	Allikmets (1997), Maugeri (1999), Maugeri (1999), Maugeri (1999), Maugeri (2002), Maugeri (2002), Hiller (2004), Emory Genetics Laboratory (2016), Institute of Human Genetics of the University of Regensburg (2016), GeneDx (2016), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2016), Institute of Human Genetics of the University of Regensburg (2016), GeneDx (2018)
51863	ABCA4	G	A	GG	Stargardt Disease and Cone-rod Dystrophy 3	Allikmets (1997), Allikmets (1997), Nasonkin (1998), Fishman (2003)
51945	ABCA4	C	A,T	CC	Stargardt Disease and Cone-rod Dystrophy 3	Allikmets (1997), Cremers (1998), Rozet (1998), Lewis (1999), Maugeri (1999), Fishman (1999), Lewis (1999), Maugeri (1999), Rivera (2000), Simonelli (2000), Simonelli (2000), Fishman (2003)
51642	ABCA4	A	G	AA	Stargardt Disease	Rozet (1998)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
51649	ABCA4	C	G,T	CC	Stargardt Disease	Lewis (1999)
51676	ABCA4	G	A,T	GG	Stargardt Disease	Allikmets (1997), Rozet (1998), Lewis (1999), Maugeri (1999), Fishman (1999), Lewis (1999), Maugeri (1999), Maugeri (1999), Rivera (2000), Simonelli (2000), Simonelli (2000)
51749	ABCA4	G	A	GG	Stargardt Disease	Allikmets (1997), Rozet (1998), Rozet (1998), Lewis (1999), Maugeri (1999), Fishman (1999), Lewis (1999), Maugeri (1999), Rivera (2000), Simonelli (2000), Simonelli (2000)
51778	ABCA4	G	A	GG	Stargardt Disease	Allikmets (1997), Rozet (1998), Lewis (1999), Maugeri (1999), Fishman (1999), Lewis (1999), Maugeri (1999), Maugeri (1999), Rivera (2000), Simonelli (2000), Simonelli (2000)
35179	CHST3	G	A	GG	Spondyloepiphyseal Dysplasia, Omani Type	Rajab (2004), Thiele (2004)
59410	WISP3	T	C	TT	Spondyloepiphyseal Dysplasia Tarda and Arthropathy	Liao (2004)
36461	TRAPPC2	A	G	AA	Spondyloepiphyseal Dysplasia Tarda	Grunebaum (2001), Grunebaum (2001)
36469	TRAPPC2	C	T	CC	Spondyloepiphyseal Dysplasia Tarda	Tiller (2001)
100837	TRAPPC2	I	D	II	Spondyloepiphyseal Dysplasia Tarda	Gedeon (2001)
43125	POLG	C	G	CC	Possibly associated with Sensory Ataxic Neuropathy, Dysarthria, and Ophthalmoparesis	Rantamaki (2001), Van Goethem (2004), Hakonen (2005), Winterthun (2005), Davidzon (2005), Nguyen (2005), GeneReviews (2012), Emory Genetics Laboratory of Emory University (2013), Emory Genetics Laboratory of Emory University (2016), Illumina Clinical Services Laboratory (2016), Wellcome Centre for Mitochondrial Research of Newcastle University (2017), GeneDx (2017), ARUP Laboratories (2017), Invitae (2018), SIB Swiss Institute of Bioinformatics (2018)
800849	POLG	C	G	CC	Possibly associated with Spinocerebellar Ataxia with Epilepsy	Rantamaki (2001), Van Goethem (2004), Hakonen (2005), Winterthun (2005), Davidzon (2005), Nguyen (2005), GeneReviews (2012), Emory Genetics Laboratory of Emory University (2013), Emory Genetics Laboratory of Emory University (2016), Illumina Clinical Services Laboratory (2016), Wellcome Centre for Mitochondrial Research of Newcastle University (2017), GeneDx (2017), ARUP Laboratories (2017), Invitae (2018), SIB Swiss Institute of Bioinformatics (2018)
800850	POLG	C	G	CC	Possibly associated with Alpers Syndrome	Rantamaki (2001), Van Goethem (2004), Hakonen (2005), Winterthun (2005), Davidzon (2005), Nguyen (2005), GeneReviews (2012), Emory Genetics Laboratory of Emory University (2013), Emory Genetics Laboratory of Emory University (2016), Illumina Clinical Services Laboratory (2016), Wellcome Centre for Mitochondrial Research of Newcastle University (2017), GeneDx (2017), ARUP Laboratories (2017), Invitae (2018), SIB Swiss Institute of Bioinformatics (2018)
43111	POLG	C	T	CC	Progressive External Ophthalmoplegia with Mitochondrial DNA Deletions	Rantamaki (2001), Van Goethem (2001), Van Goethem (2003), Van Broeckhoven (2004) , Van Goethem (2004), Naviaux (2004), Naviaux (2005), Nguyen (2005), Winterthun (2005), GeneReviews (2012), Courtagen Diagnostics Laboratory (2013), Laboratory for Molecular Medicine of Partners HealthCare Personalized Medicine (2014), GeneDx (2016), Emory Genetics Laboratory of Emory University (2016), Praxis fuer Humangenetik Tuebingen (2016), Illumina Clinical Services Laboratory (2016), GeneDx (2017), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2017), Wellcome Trust Centre for Mitochondrial Research of Newcastle University (2017), Invitae (2018)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
800844	POLG	C	T	CC	Sensory Ataxic Neuropathy, Dysarthria, and Ophthalmoparesis	Rantamaki (2001), Van Goethem (2001), Van Goethem (2003), Van Broeckhoven (2004) , Van Goethem (2004), Naviaux (2004), Naviaux (2005), Nguyen (2005), Winterthun (2005), GeneReviews (2012), Courtagen Diagnostics Laboratory (2013), Laboratory for Molecular Medicine of Partners HealthCare Personalized Medicine (2014), GeneDx (2016), Emory Genetics Laboratory of Emory University (2016), Praxis fuer Humangenetik Tuebingen (2016), Illumina Clinical Services Laboratory (2016), GeneDx (2017), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2017), Wellcome Trust Centre for Mitochondrial Research of Newcastle University (2017), Invitae (2018)
800845	POLG	C	T	CC	Spinocerebellar Ataxia with Epilepsy	Rantamaki (2001), Van Goethem (2001), Van Goethem (2003), Van Broeckhoven (2004) , Van Goethem (2004), Naviaux (2004), Naviaux (2005), Nguyen (2005), Winterthun (2005), GeneReviews (2012), Courtagen Diagnostics Laboratory (2013), Laboratory for Molecular Medicine of Partners HealthCare Personalized Medicine (2014), GeneDx (2016), Emory Genetics Laboratory of Emory University (2016), Praxis fuer Humangenetik Tuebingen (2016), Illumina Clinical Services Laboratory (2016), GeneDx (2017), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2017), Wellcome Trust Centre for Mitochondrial Research of Newcastle University (2017), Invitae (2018)
800846	POLG	C	T	CC	Alpers Syndrome	Rantamaki (2001), Van Goethem (2001), Van Goethem (2003), Van Broeckhoven (2004) , Van Goethem (2004), Naviaux (2004), Naviaux (2005), Nguyen (2005), Winterthun (2005), GeneReviews (2012), Courtagen Diagnostics Laboratory (2013), Laboratory for Molecular Medicine of Partners HealthCare Personalized Medicine (2014), GeneDx (2016), Emory Genetics Laboratory of Emory University (2016), Praxis fuer Humangenetik Tuebingen (2016), Illumina Clinical Services Laboratory (2016), GeneDx (2017), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2017), Wellcome Trust Centre for Mitochondrial Research of Newcastle University (2017), Invitae (2018)
38953	CACNA1A	C	T	CC	Spinocerebellar Ataxia 6	Yue (1997), Yue (1997), Yue (1997), Wan (2005)
71765	TTBK2	I	D	II	Spinocerebellar Ataxia	Houlden (2007)
100570	AFG3L2	C	T	CC	Spinocerebellar Ataxia	Di Bella (2010)
100572	AFG3L2	C	T	CC	Spinocerebellar Ataxia	Di Bella (2010)
101085	AFG3L2	T	C,G	TT	Spinocerebellar Ataxia	Di Bella (2010) wrote that autosomal dominant spinocerebellar ataxias (SCAs) are genetically heterogeneous neurological disorders characterized by cerebellar dysfunction mostly due to Purkinje cell degeneration. Here we show that AFG3L2 mutations cause SC
100924	SMN1	I	D	II	Spinal Muscular Atrophy, Type I	Bussaglia (1995), Cusco (2003)
79095	PLEKHG5	A	G	AA	Spinal Muscular Atrophy, Distal, Childhood-onset	Maystadt (2006), Maystadt (2007), Maystady (2007)
19720	EPB42	C	T	CC	Spherocytosis, Hereditary, Japanese Type	Bouhassira (1991), Bouhassira (1992), Iwamoto (1993), Perrotta (1999)
19731	EPB42	C	T	CC	Spherocytosis, Hereditary, due to Protein 4.2-Notame	Matsuda , (1995)
19965	ANK1	G	A	GG	Spherocytosis, Hereditary	Hayette (1998)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
20390	SLC4A1	C	T	CC	Spherocytosis	Alloisio (1996)
21496	SPTB	A	G	AA	Spherocytosis	Goodman (1982), Wolfe (1982), Becker (1993), Becker (1993)
38070	SPAST	A	G	AA	Spastic paraplegia, autosomal dominant	Orlacchio (2004)
38143	SPAST	A	G	AA	Spastic paraplegia, autosomal dominant	Hazan (1999)
38337	SPG3A	C	T	CC	Spastic Paraplegia 3	Hazan (1993), Zhao (2001), Abel (2004), Abel (2004), Abel (2004), Namekawa (2006)
38284	KIF5A	A	G	AA	Spastic Paraplegia 10	Reid (2002)
38285	KIF5A	C	T	CC	Spastic Paraplegia 10	Fichera (2004)
38286	KIF5A	C	T	CC	Spastic Paraplegia 10	Giudice (2006)
38705	PLP1	T	C	T	Spastic paraplegia	McKusick (1962), McKusick (1962), Kobayashi (1994), Naidu (1997), Edgar (2004)
43620	ALS2	C	A	CC	Spastic paralysis, infantile-onset	Eymard-Pierre (2002)
63668	DHCR7	G	A	GG	Smith-Lemli-Opitz Syndrome	Fitzky (1998), Brasi (1999), Witsch-Baumgartner (2001), Scalco (2005)
63743	DHCR7	C	A,T	CC	Smith-Lemli-Opitz Syndrome	Matsumoto (2005)
63754	DHCR7	G	A,C,T	GG	Smith-Lemli-Opitz Syndrome	Witsch-Baumgartner (2001), Fitzky (1998)
63680	DHCR7	C	G,T	CC	Smith-Lemli-Opitz Syndrome	Fitzky (1998), Nowaczyk (1998), Löffler (2000), Witsch-Baumgartner (2000), Witsch-Baumgartner (2001), Ciara (2004), GeneReviews (2007), Witsch-Baumgartner (2008), Centre for Mendelian Genomics of the University Medical Centre Ljubljana (2013), Emory Genetics Laboratory of Emory University (2013), Courtagen Diagnostics Laboratory of Courtagen Life Sciences (2014), Laboratory for Molecular Medicine of Partners HealthCare Personalized Medicine (2014), Emory Genetics Laboratory of Emory University (2015), Praxis fuer Humangenetik Tuebingen (2016), GeneDx (2016), Illumina Clinical Services Laboratory (2016),
63681	DHCR7	C	T	CC	Smith-Lemli-Opitz Syndrome	Witsch-Baumgartner (2000), Löffler (2000), Witsch-Baumgartner (2001), Ciara (2004), Witsch-Baumgartner (2008)
801989	DHCR7	C	G	CC	Smith-Lemli-Opitz Syndrome	Wassif (1998), Waterham (1998), Yu (2000), Witsch-Baumgartner (2000), Krakowiak (2000), Witsch-Baumgartner (2001), Nowaczyk (2001), Wright (2003), Yu (2005), Scalco (2005), Witsch-Baumgartner (2008)
63736	DHCR7	C	A	CC	Smith-Lemli-Opitz Syndrome	Fitzky (1998), Yu (2000), Witsch-Baumgartner (2001), Ciara (2004), Yu (2005)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
63777	DHCR7	C	G	CC	Smith-Lemli-Opitz Syndrome	Wassif (1998), Waterham (1998), Witsch-Baumgartner (2000), Yu (2000), Witsch-Baumgartner (2001), Krakowiak (2000), Nowaczyk (2001), Wright (2003), Scalco (2005), Witsch-Baumgartner (2008), Koo (2010), Lazarin (2013), Emory Genetics Laboratory of Emory University (2013), Emory Genetics Laboratory of Emory University (2015), Genetic Services Laboratory of the University of Chicago (2015), Center for Pediatric Genomic Medicine of Children's Mercy Hospital and Clinics (2015), GeneDx (2016), Illumina Clinical Services Laboratory (2016), Knight Diagnostic Laboratories of the Oregon Health and Sciences University (2016)
34738	TBX4	A	G	AA	Small Patella Syndrome	Bongers (2004)
34744	TBX4	C	T	CC	Small Patella Syndrome	Bongers (2001), Bongers (2004)
34737	TBX4	G	T	GG	Small Patella Syndrome	Bongers (2001), Bongers (2004)
42138	ARHGEF10	C	T	CC	Slowed Motor and Sensory Nerve Conduction Velocities	Nelis (1998), Verhoeven (2003)
6743	DSP	C	T	CC	Skin Fragility-Woolly Hair Syndrome	Watt, (1997), Fujiwara , (2001), Whittock (2002)
75048	ALDH3A2	C	T	CC	Sjogren-Larsson Syndrome	Sillen (1997), Laurenzi (1997), IJIst (1999)
75049	ALDH3A2	I	D	II	Sjogren-Larsson Syndrome	Tsukamoto (1997), IJIst (1999)
5245	ABCG5	G	A	GG	Sitosterolemia	Berge (2000), Lee (2001)
5246	ABCG5	C	G,T	CC	Sitosterolemia	Lee (2001)
5281	ABCG8	G	A	GG	Sitosterolemia	Connor, (1974), Berge (2000), Lu (2001), GeneDx (2016), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2018), Illumina Clinical Services Laboratory (2018)
2548	HCN4	C	T	CC	Sinus Node Disease	Ueda (2004)
65393	GPC3	G	A	G	Simpson-Golabi-Behmel Syndrome, Type 1	Veugelers (2000)
18982	ALAS2	C	T	C	Sideroblastic Anemia, Hereditary	Prades (1995)
19494	ABCB7	A	C,T	A	Sideroblastic Anemia and Spinocerebellar Ataxia	Pagon (1985), Allikmets (1999)
19495	ABCB7	C	T	C	Sideroblastic Anemia and Spinocerebellar Ataxia	Bekri (2000)
40623	GNE	C	T	CC	Sialuria	Weiss (1989), Seppala (1999), Leroy (2001)
27743	NEU1	C	T	CC	Sialidosis, Type II	Pattison (2004)
11172	GH1	T	C	TT	Short stature	Millar (2003)
34987	ACADS	C	T	CC	Short Chain Acyl-CoA-dehydrogenase Deficiency	Corydon (2001)
34992	ACADS	C	T	CC	Short Chain Acyl-CoA-dehydrogenase Deficiency	Corydon (2001)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
34994	ACADS	C	T	CC	Short Chain Acyl-CoA-dehydrogenase Deficiency	Corydon (2001)
34995	ACADS	C	T	CC	Short Chain Acyl-CoA-dehydrogenase Deficiency	Gregersen (1998), Corydon (2001)
35009	ACADS	C	T	CC	Short Chain Acyl-CoA-dehydrogenase Deficiency	Naito (1989), Naito (1989)
35015	ACADS	C	T	CC	Short Chain Acyl-CoA-dehydrogenase Deficiency	Corydon (2001), Corydon (2001), SCAD deficiency (2014)
35011	ACADS	G	T	GG	Short Chain Acyl-CoA-dehydrogenase Deficiency	Gregersen (1998)
35016	ACADS	G	T	GG	Short Chain Acyl-CoA-dehydrogenase Deficiency	Seidel (2003)
35014	ACADS	T	C	TT	Short Chain Acyl-CoA-dehydrogenase Deficiency	Gregersen (1998)
35010	ACADS	G	A	GG	Short Chain Acyl-CoA-dehydrogenase Deficiency	Corydon (2001)
15400	EDN3	G	T	GG	Shah-Waardenburg Syndrome	Hofstra (1996)
40496	ARSA	G	A	GG	Severe Metachromatic Leukodystrophy	Harvey (1993)
40542	ARSA	C	T	CC	Severe Metachromatic Leukodystrophy	Pastor-Soler (1994)
24995	JAK3	T	C	TT	Severe Combined Immunodeficiency, T-negative/B-positive Type	Macchi (1995), Zhou (2001)
24998	JAK3	G	A	GG	Severe Combined Immunodeficiency, T-negative/B-positive Type	Schumacher (2000), Schumacher (2000)
24103	IL7R	C	T	CC	Severe Combined Immunodeficiency, T Cell-negative, B Cell-positive, NK Cell-positive	Roifman (2000)
42935	PTPRC	G	A	GG	Severe Combined Immunodeficiency, T Cell-negative, B Cell-positive, NK Cell-positive	Kung (2000)
24267	CD3D	G	A,T	GG	Severe Combined Immunodeficiency, T Cell-negative, B Cell-positive, NK Cell-positive	Dadi (2003), Basile (2004)
24268	CD3D	G	T	GG	Severe Combined Immunodeficiency, T Cell-negative, B Cell-positive, NK Cell-positive	Basile (2004)
24269	CD3D	T	C	TT	Severe Combined Immunodeficiency, T Cell-negative, B Cell-positive, NK Cell-positive	Takada (2005)
24753	DCLRE1C	G	A,T	GG	Severe Combined Immunodeficiency, Athabascan-Type	Li (2002), Li (2002)
22816	ADA	G	A	GG	Severe Combined Immunodeficiency (SCID), T Cell-negative, B Cell-negative, NK Cell-negative	Akeson (1987), Markert (1989), Hirschhorn (1992)
24297	RAG2	C	A,T	CC	Severe Combined Immunodeficiency (SCID), B Cell-negative|Omenn Syndrome	Schwarz (1996), Corneo (2001)
24305	RAG2	C	T	CC	Severe Combined Immunodeficiency (SCID), B Cell-negative	Schwarz (1996)
24456	IL2RG	A	G	A	Severe Combined Immunodeficiency	Stephan (1996), Stephan (1996), Stephan (1996)
24513	IL2RG	C	T	C	Severe Combined Immunodeficiency	Clark (1995), Jones (1997), Jones (1997), Jones (1997)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
24515	IL2RG	A	T	A	Severe Combined Immunodeficiency	Schmalstieg (1995)
40522	ARSA	G	A,T	GG	Severe Arylsulfatase A Pseudodeficiency	Zlotogora (1994)
100904	BSND	T	C	TT	Sensorineural Deafness with Mild Renal Dysfunction	Riazuddin (2009)
71857	MTTL1	A	G	?	Melas Syndrome	Goto (1990), Kobayashi (1990), Kobayashi (1991), Enter (1991), Reardon (1992), Yoneda (1992), Ouweland , (1992), Ciafaloni (1992), Moraes (1992), Lertrit (1992), Mosewich (1993), Schulz (1993), Matthews (1994), Vries (1994), Matthews (1995), Chuang (1995), Yang (1995), Manouvrier (1995), Matthews (1995), Odawara (1995), Morten (1995), Velho , (1996), Yorifuji (1996), Yorifuji (1996), Vilarinho (1997)
63231	NPHP4	G	A	GG	Senior-Loken Syndrome 4	Otto (2002), Fillastre (1976)
39794	TH	C	T	CC	Segawa Syndrome	Swaans (2000)
39795	TH	T	G	TT	Segawa Syndrome	Swaans (2000)
39817	TH	C	T	CC	Segawa Syndrome	Heuvel (1998), Heuvel (1998)
39822	TH	G	A,T	GG	Segawa Syndrome	Ludecke (1995), Ludecke (1995), Knappskog (1995)
100881	TH	C	T	CC	Segawa Syndrome	Tinti , (1997), Verbeek (2007)
68619	PCNT	G	T	GG	Seckel Syndrome 1	Griffith (2008)
2466	ABCA1	C	A,T	CC	Possibly Associated with Scott Syndrome	Albrecht (2005)
78878	SMARCB1	C	T	CC	Schwannomatosis	Hulsebos (2007)
100850	UBIAD1	A	G	AA	Schnyder Crystalline Corneal Dystrophy	Yellore (2007), Orr (2007), Orr (2007)
70751	UBIAD1	C	G	CC	Schnyder Crystalline Corneal Dystrophy	Yellore (2007)
70750	UBIAD1	A	G	AA	Schnyder Crystalline Corneal Dystrophy	Yellore (2007)
43193	NR4A2	T	C	TT	Schizophrenia	Buervenich (2000)
27905	IDUA	C	T	CC	Scheie Syndrome	Beesley (2001)
47478	TP53	G	A	GG	Variant of Unknown Significance (Possibly associated with Sarcoma)	Hwang (2003), Ambry Genetics (2015)
72587	CYP21A2	I	D	II	Salt-Wasting Congenital Adrenal Hyperplasia	White (1988), Lee (2003)
34975	FGFR3	A	T	AA	Saddan Dysplasia|Skeleton-Skin-Brain Syndrome|Thanatophoric Dysplasia, Type I	II mutation, (1948), The (1949), M mutation due to a (1988), Francomano (1996), Chesi (1997), Francomano, (1999), Tavormina (1999)
49892	EP300	C	T	CC	Rubinstein-Taybi Syndrome	Roelfsema (2005)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
43525	MPZ	G	T	GG	Roussy-Levy syndrome	Levy (1926), Plante-Bordeneuve (1999)
61857	RECQL4	G	A	GG	Rothmund-Thomson syndrome	Kitao (1999)
100859	RECQL4	I	D	II	Rothmund-Thomson syndrome	Siitonen (2003)
100860	RECQL4	I	D	II	Rothmund-Thomson syndrome	Lindor (2000)
54142	CNGA3	C	T	CC	Rod Monochromacy	Kohl (1998), Wissinger (2001)
54166	CNGA3	C	A	CC	Rod Monochromacy	Kohl (1998), Wissinger (2001)
34876	ROR2	G	A	GG	Robinow Syndrome	Afzal (2000), Tufan (2005)
34881	ROR2	G	A	GG	Robinow Syndrome	Afzal (2000)
40553	CAV3	G	A	GG	Rippling Muscle Disease 2	Kubisch (2003), Kubisch (2003), Kubisch (2003), Kubisch (2005)
64473	PITX2	C	T	CC	Ring Dermoid of the Cornea	Xia (2004)
59559	VDR	C	T	CC	Rickets, Vitamin D-dependent, Type II	Sone (1990)
59550	VDR	G	A,C	GG	Rickets, Vitamin D Resistant	Mechica (1997), Mechica , (1997), Zhu (1998)
22192	RHAG	C	T	CC	Rh-Null Hemolytic Anemia, Regulator Type	Hyland (1998), Hyland , (1998), Huang (1998)
22197	RHAG	C	T	CC	Rh-Null Hemolytic Anemia, Regulator Type	Cherif-Zahar (1998)
30429	GNPAT	C	T	CC	Rhizomelic Chondrodysplasia Punctata, Type 3	Ofman (1998)
30428	GNPAT	G	A	GG	Rhizomelic Chondrodysplasia Punctata, Type 2	Ofman (1998), Clayton (1994)
60773	CDKL5	C	T	C	Rett Syndrome, Atypical	Archer (2006)
60777	CDKL5	C	T	C	Rett Syndrome, Atypical	Archer (2006)
60778	CDKL5	C	T	C	Rett Syndrome, Atypical	Nectoux (2006)
60775	CDKL5	G	T	G	Rett Syndrome, Atypical	Tao (2004)
60786	MECP2	G	A	G	Rett syndrome	Bourdon (2001), Heilstedt (2002), Schanen (2004)
60787	MECP2	G	A	G	Rett syndrome	Amir (1999), Villard (2000)
60789	MECP2	G	A,T	G	Rett syndrome	Orrico (2000), Couvert (2001), Klauck (2002), Winnepenninckx (2002)
60833	MECP2	C	A,T	C	Rett syndrome	Bona (2000)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
60861	MECP2	G	A,T	G	Rett syndrome	Wan (1999)
60883	MECP2	G	A	G	Rett syndrome	Amir (1999), Cheadle (2000), Bienvenu (2000), Huppke (2000)
60887	MECP2	G	A,C	G	Rett syndrome	Huppke (2000), Bienvenu (2000), Bona (2000), Topcu (2002), Jian (2005)
60893	MECP2	G	A,C	G	Rett syndrome	Bona (2000), Carney (2003)
48269	RB1	T	C	TT	Retinoblastoma, Incomplete Penetrance Type	Cowell (1998), Otterson (1999)
48234	RB1	C	T	CC	Retinoblastoma	Hogg (1993), Cowell (1994), Cowell (1994), Liu (1995), Onadim (1997)
48255	RB1	C	T	CC	Retinoblastoma	Onadim (1992), Lohmann (1994), Bia (1998), Bia (1998), Otterson (1999)
48306	RB1	G	A	GG	Retinoblastoma	Dunn (1989)
48381	RB1	G	T	GG	Retinoblastoma	Klutz (2002)
52218	RLBP1	C	G,T	CC	Retinitis Punctata Albescens|Retinitis Pigmentosa|Fundus Albipunctatus	Maw (1997), Katsanis (2001)
52234	RLBP1	C	T	CC	Retinitis Punctata Albescens|Newfoundland Rod-cone Dystrophy	Morimura (1999), Eichers (2002)
52235	RLBP1	A	G	AA	Retinitis Punctata Albescens|Newfoundland Rod-cone Dystrophy	Morimura (1999), Eichers (2002)
52822	CRX	G	A	GG	Retinitis Pigmentosa, Late-onset	Swain (1997), Swain (1997), Sohocki (1998), Sohocki (1998)
70748	TOPORS	D	I	DD	Retinitis Pigmentosa with Perivascular Retinal Pigment Epithelium Atrophy	Chakarova (2007)
52147	RHO	C	T	CC	Retinitis Pigmentosa 4|Retinitis Punctata Albescens	Sung (1991), Jacobson (1991), Andreasson (1992), Pannarale (1996), Pannarale (1996), Souied (1996), Souied (1996), Ponjavic (1997), Ponjavic (1997)
52131	RHO	G	A	GG	Retinitis Pigmentosa 4|Nightblindness, Congenital Stationary	Sieving (1992), Maghtheh (1993), Rao (1994)
52114	RHO	C	A	CC	Retinitis Pigmentosa 4	Dryja (1990), Dryja (1990), Dryja , (1990), Farrar (1990), Berson (1991), Heckenlively (1991), Heckenlively (1991), Naash (1993)
52255	RP2	C	T	C	Retinitis Pigmentosa 2	Hardcastle (1999), Vorster (2004)
52238	RP2	G	T	G	Retinitis Pigmentosa 2	Schwahn , (1998), Sharon , (2000), Miano (2001)
52536	RP1	C	T	CC	Retinitis Pigmentosa 1	Blanton (1991), Iannaccone (1996), Pierce (1999), Sullivan (1999), Pierce (1999), Pierce (1999), Guillonneau (1999)
52356	NR2E3	G	A	GG	Retinitis Pigmentosa	Gerber (2000), Haider (2000)
70703	NR2E3	G	A	GG	Retinitis Pigmentosa	Coppieters (2007), Coppieters (2007)
60735	USH2A	C	A	CC	Retinitis Pigmentosa	Rivolta (2000), Rivolta (2002), Zlotogora (2004)
70754	PRPH2	A	G	AA	Retinitis Pigmentosa	Kajiwara (1991), Kajiwara (1994), Loewen (2001)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
70756	PRPH2	T	C,G	TT	Retinitis Pigmentosa	Passerini (2007)
70757	CERKL	G	A	GG	Retinitis Pigmentosa	Tuson (2004)
70758	CERKL	C	T	CC	Retinal Degeneration with Early Macular Involvement	Auslender (2007)
800864	LMNA	C	T	CC	Restrictive Dermatopathy, Lethal	HGPS patients carrying the (1824), HGPS due to the (1824), Eriksson (2003), De Sandre-Giovannoli (2003), Hegele (2003), Eriksson (2003), D'Apice (2004), Goldman (2004), Goldman (2004), Goldman (2004), Navarro (2004), Wuyts (2005), Shumaker (2006), They referred to this mutation as (2036)
57878	SFTPC	T	C	TT	Respiratory Insufficiency, Infantile-onset Progressive	Tredano (2004), Brasch (2004), Percopo (2004)
20003	HBB	C	G,T	CC	Resistance to Malaria	Neel (1950), Neel (1953), Ranney (1953), Allison, (1954), Ingram (1959), Krevans (1959), Ingram (1961), River (1961), Heller (1977), Fabry (1981), Ballas , (1982), Pearson , (1985), Boehm (1985), Fischel-Ghodsian (1990), Trabuchet (1991), Lane (1994), Lane (1994), Agarwal (2000), Agarwal , (2000), Modiano (2001), Modiano , (2001), Rihet (2004), Hedrick, (2004), Rihet , (2004), Wood (2005), Fairhurst (2005), Wood (2005), Fairhurst (2005)
25124	CCR5	G	T	GG	Resistance to HIV Infection	Carrington (1997), Tamasauskas (2001)
25122	CCR5	T	A	TT	Resistance to HIV Infection	Carrington (1997), Quillent (1998), Quillent (1998), Ometto (1999)
831	AGT	G	A	GG	Renal Tubular Dysgenesis	Uematsu (2006)
67102	SLC5A2	G	A	GG	Renal Glucosuria	Heuvel (2002), Heuvel (2002)
46854	MET	G	T	GG	Renal Carcinoma, Papillary	Schmidt (1997)
46856	MET	G	A	GG	Renal Carcinoma, Papillary	Schmidt (1997)
13257	AR	G	A	G	Reifenstein syndrome	Klocker (1992)
63624	PEX26	T	C	TT	Refsum Disease, Infantile Form	Matsumoto (2003)
30440	PEX1	C	T	CC	Refsum Disease, Infantile Form	Walter (2001)
30448	PEX1	C	T	CC	Refsum Disease, Infantile Form	Reuber (1997), Portsteffen (1997), Gartner (1999)
33639	PXMP3	C	T	CC	Refsum Disease, Infantile Form	Shimozawa (1999)
62050	PHYH	T	C	TT	Refsum Disease, Adult	Mihalik (1997), Jansen (1997)
61865	RECQL4	G	A	GG	RAPADILINO syndrome	Siitonen (2003)
22538	CD151	G	A	GG	Ralph Blood Group Variation	Crew (2003)
4271	KCNH2	C	T	CC	QT Prolongation and Torsade de Pointes, Drug-induced, Amiodarone	Yang (2002)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
32907	PKLR	C	T	CC	Pyruvate Kinase Deficiency of Red Cells	Procopio (1963), Bowman (1965), Kanno (1994), Valentini (2002)
32910	PKLR	G	A	GG	Pyruvate Kinase Deficiency of Red Cells	Zarza (1998), Zanella (2001)
32922	PKLR	C	T	CC	Pyruvate Kinase Deficiency of Red Cells	Beutler (1995), Lenzner (1997), Wang (2001)
32563	PDHB	T	C	TT	Pyruvate Dehydrogenase E1-beta Deficiency	Brown (2004)
32564	PDHB	G	A	GG	Pyruvate Dehydrogenase E1-beta Deficiency	Brown (2004)
30723	PC	G	A	GG	Pyruvate Carboxylase Deficiency	Wexler (1998)
30724	PC	C	T	CC	Pyruvate Carboxylase Deficiency	Carbone (1998)
30725	PC	C	A	CC	Pyruvate Carboxylase Deficiency	Carbone (1998)
19431	SPTA1	A	G	AA	Pyropoikilocytosis, Hereditary	G-to-A transition (1828), Zarkowsky (1975), Zarkowsky (1975), Gallagher (1992), Venezia (1993), Venezia (1993), Costa (2005)
801968	SPTA1	A	G	AA	Pyropoikilocytosis, Hereditary	G-to-A transition (1828), Zarkowsky (1975), Zarkowsky (1975), Gallagher (1992), Venezia (1993), Venezia (1993), Costa (2005)
5795	CBS	C	T	CC	Pyridoxine Responsive Homocystinuria	Kluijtmans (1996), Kluijtmans (1996)
5800	CBS	A	G	AA	Pyridoxine Responsive Homocystinuria	Kozich (1992), Kraus (1992), Hu (1993), Hu (1993), Shih (1995), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2013), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2014), Laboratory for Molecular Medicine of Partners HealthCare Personalized Medicine (2014), Ambry Genetics (2016), Invitae (2016), Illumina Clinical Services Laboratory (2016), GeneDx (2017), Invitae (2017), Ambry Genetics (2017)
5814	CBS	C	T	CC	Pyridoxine Responsive Homocystinuria	Sebastio , (1995), Sperandeo , (1995), Tsai , (1996), Sperandeo , (1996), Tsai (1996), Kim (1997), Kim (1997), Kim (1997), Kim (1997), Kim (1997)
5848	CBS	C	A,T	CC	Pyridoxine Responsive Homocystinuria	Shan (2001)
5898	CBS	T	C	TT	Pyridoxine Responsive Homocystinuria	Aral (1997)
34782	CTSK	A	G	AA	Pycnodysostosis	Haagerup (2000)
34789	CTSK	G	A,T	GG	Pycnodysostosis	Polymeropoulos (1995), Gelb (1996), Johnson (1996), Johnson (1996)
34794	CTSK	T	C	TT	Pycnodysostosis	Gelb (1996)
800959	SFTPC	T	C	TT	Pulmonary Surfactant Metabolism Dysfunction, Type 2	Tredano (2004), Brasch (2004), Percopo (2004)
9032	ABCC6	G	A	GG	Pseudoxanthoma Elasticum	Saux (2000), Bergen (2000), Ringpfeil (2000), Saux (2001), Trip (2002), Hu (2003), Centre for Mendelian Genomics of the University Medical Centre Ljubljana (2014), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2016), Genetic Services Laboratory of the University of Chicago (2016), GeneDx (2017), Fulgent Genetics (2018), GeneDx (2019)
8926	ABCC6	G	A	GG	Pseudoxanthoma Elasticum	Saux (2002)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
60276	SRD5A2	C	G,T	CC	Pseudovaginal Perineoscrotal Hypospadias	Thigpen , (1992), Forti , (1996), Ivarsson (1998)
59408	WISP3	G	A	GG	Pseudorheumatoid Dysplasia, Progressive	Hurvitz (1999)
61574	GNAS	G	T	GG	Pseudohypoparathyroidism, Type 1A	Iiri (1994)
5578	SCNN1G	G	A	GG	Pseudohypoaldosteronism, Type I	Strautnieks (1996), Strautnieks (1996), Strautnieks (1996), Strautnieks (1996)
100883	SCNN1A	I	D	II	Pseudohypoaldosteronism, Type 1	Schaedel (1999)
100884	SCNN1A	I	D	II	Pseudohypoaldosteronism, Type 1	Schaedel (1999)
13354	AR	G	A	G	Pseudohermaphroditism, male	Cabral (1998)
14365	LHCGR	A	G	AA	Pseudohermaphroditism|Leydig Cell Hypoplasia	Themmen (2000), Martens (2002)
68184	COMP	I	D	II	Pseudoachondroplasia	Hecht (1995), Briggs (1995)
9798	EPP	A	C	AA	Protoporphyria, Erythropoietic	Bloomer (1998)
49861	EPHB2	G	A	GG	Prostate Cancer, Somatic|Prostate Cancer, Hereditary	Huusko (2004), Kokko (2006)
13453	AR	G	A	G	Prostate Cancer	Taplin (1999)
45914	CDH1	T	A	TT	Prostate Cancer	Jonsson (2002)
801250	ELAC2	C	G,T	CC	Prostate Cancer	Wang (2001)
801251	ELAC2	C	A,T	CC	Prostate Cancer	Wang (2001)
801252	ELAC2	C	A,T	CC	Possibly associated with Prostate Cancer	Tavtigian (2001), Mayo Clinic Genetic Testing Laboratories (2016), Invitae (2017), GeneDx (2017), Institute of Human Genetics of Klinikum rechts der Isar (2017), Baylor Miraca Genetics Laboratories (2017)
801253	CHEK2	C	T	CC	Prostate Cancer	Dong (2003)
801255	CHEK2	C	A,G,T	CC	Prostate Cancer	Dong (2003)
801256	CHEK2	C	A,G,T	CC	Prostate Cancer	Dong (2003)
801257	CHEK2	T	A,C	TT	Prostate Cancer	Dong (2003)
30259	PCCB	A	G	AA	Propionic Acidemia	Yorifuji (2002)
30257	PCCB	C	T	CC	Propionic Acidemia	Tahara (1993), Tahara (1990), Ugarte (1999)
30258	PCCB	C	T	CC	Propionic Acidemia	Ohura (1993)
33595	PCCA	C	T	CC	Propionic Acidemia	Campeau (1999)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
30249	PCCB	G	A	GG	Propionic Acidemia	Rodriguez-Pombo (1998)
71914	PCCB	D	I	DD	Propionic Acidemia	Rodriguez-Pombo (1998)
24243	PFC	G	A	G	Properdin Deficiency, Type II	Sjoholm , (1988), Westberg (1995), Westberg (1995)
24245	PFC	G	A	G	Properdin Deficiency, Type I	Sjoholm (1982), Westberg (1995)
71610	MAPT	I	D	II	Progressive Supranuclear Palsy|Parkinson Disease	Antonarakis, (2000), Pastor (2001), Pastor (2001), Pastor (2001), Yoshida , (2002), Rossi (2004), Pastor (2004), Rossi (2004), Pastor (2004), Pastor (2004)
16086	ATP8B1	A	G	AA	Progressive Familial Intrahepatic Cholestasis	Jensen (1969), Bull (1998), Tygstrup (1999), Klomp (2004)
801957	ATP8B1	A	G	AA	Benign Recurrent Intrahepatic Cholestasis	Jensen (1969), Bull (1998), Tygstrup (1999), Klomp (2004)
41238	SLC25A4	A	G	AA	Progressive External Ophthalmoplegia with Mitochondrial DNA Deletions, Type 2	Komaki (2002)
41254	SLC25A4	G	C	GG	Progressive External Ophthalmoplegia with Mitochondrial DNA Deletions, Type 2	Kaukonen (1996), Kaukonen (2000)
53424	PEO1	C	A	CC	Progressive External Ophthalmoplegia with Mitochondrial DNA Deletions	Lewis (2002)
43103	POLG	A	C	AA	Progressive External Ophthalmoplegia with Mitochondrial DNA Deletions	Van Goethem (2001), Van Goethem (2001), Rantamaki (2001), Van Goethem (2003), Van Goethem (2003), Van Goethem (2003), Van Broeckhoven (2004), Van Goethem (2004), Nguyen (2004), Nguyen (2005), Nguyen (2005), Winterthun (2005), Alpers syndrome (2037)
43118	POLG	G	A	GG	Likely associated with Progressive External Ophthalmoplegia with Mitochondrial DNA Deletions	Van Goethem (2003), Filosto (2003), Lamantea (2004), GeneReviews (2012), University of Chicago (2014), Centre for Mendelian Genomics of the University Medical Centre Ljubljana (2014), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2016), Athena Diagnostics Inc (2016), Children's Hospital of Philadelphia (2016), Children's Mercy Hospital and Clinics (2016), Invitae (2017), Wellcome Trust Centre for Mitochondrial Research of Newcastle University (2017)
800847	POLG	G	A	GG	Likely associated with Mitochondrial Neurogastrointestinal Encephalopathy Syndrome without Leukoencephalopathy	Van Goethem (2003), Filosto (2003), Lamantea (2004), GeneReviews (2012), University of Chicago (2014), Centre for Mendelian Genomics of the University Medical Centre Ljubljana (2014), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2016), Athena Diagnostics Inc (2016), Children's Hospital of Philadelphia (2016), Children's Mercy Hospital and Clinics (2016), Invitae (2017), Wellcome Trust Centre for Mitochondrial Research of Newcastle University (2017)
43101	POLG	G	A	GG	Likely associated with Progressive External Ophthalmoplegia with Mitochondrial DNA Deletions	Vissing (2002), Van Goethem (2003), Zeviani (2004), GeneReviews (2012), Courtagen Life Sciences (2014), Centre for Mendelian Genomics of the University Medical Centre Ljubljana (2014), University of Chicago (2014), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2016), Illumina Clinical Services Laboratory (2016), Children's Hospital of Philadelphia (2016), GeneDx (2017), Children's Mercy Hospital and Clinics (2017), Invitae (2017)
800843	POLG	G	A	GG	Likely associated with Mitochondrial Neurogastrointestinal Encephalopathy Syndrome without Leukoencephalopathy	Vissing (2002), Van Goethem (2003), Zeviani (2004), GeneReviews (2012), Courtagen Life Sciences (2014), Centre for Mendelian Genomics of the University Medical Centre Ljubljana (2014), University of Chicago (2014), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2016), Illumina Clinical Services Laboratory (2016), Children's Hospital of Philadelphia (2016), GeneDx (2017), Children's Mercy Hospital and Clinics (2017), Invitae (2017)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
100899	FLCN	D	I	DD	Primary Spontaneous Pneumothorax	Nickerson (2002), Khoo (2002), Gunji (2007), Ren (2008)
100901	FLCN	I	D	II	Primary Spontaneous Pneumothorax	Ren (2008)
100902	FLCN	I	D	II	Primary Spontaneous Pneumothorax	Ren (2008)
53852	CYP1B1	C	T	CC	Primary Congenital Glaucoma	Bejjani (1998), Stoilov (1998), Belmouden (2002), Chitsazian (2007)
53853	CYP1B1	G	A	GG	Primary Congenital Glaucoma	Bejjani (1998), Chitsazian (2007)
53854	CYP1B1	C	T	CC	Primary Congenital Glaucoma	Bejjani (1998)
53855	CYP1B1	C	T	CC	Primary Congenital Glaucoma	Bejjani (2000), Vincent (2002), Vincent (2002), Chitsazian (2007), Mookherjee (2012), Emory Genetics Laboratory of Emory University (2013), Laboratory for Molecular Medicine of Partners HealthCare Personalized Medicine (2014), Emory Genetics Laboratory of Emory University (2015)
8654	FOXL2	A	T	AA	Premature Ovarian Failure	Harris (2002)
78905	NOBOX	C	G,T	C	Premature Ovarian Failure	Qin (2007)
14364	LHCGR	T	G	TT	Precocious Puberty, Male-Limited	Laue (1995), Kremer (1999)
14370	LHCGR	G	A,T	GG	Precocious Puberty, Male-Limited	Latronico , (1995), Latronico (2000)
14376	LHCGR	T	C,G	TT	Precocious Puberty, Male-Limited	Shenker (1993), Kremer (1993), Yano (1994), Kosugi (1995), Laue (1995), Kawate (1995), Rosenthal (1996)
37038	TTR	C	T	CC	Prealbumin Chicago	Harrison (1991), Ii (1992), Ii (1992)
801977	TTR	C	T	CC	Variant of Unknown Significance (Possibly associated with Euthyroid Dystransthyretinemic Hyperthyroxinemia)	Harrison (1991), Ii (1992), Ii (1992)
53141	VSX1	C	A,T	CC	Posterior Polymorphons Corneal Dystrophy 1	Heon (2002)
801949	GJB2	A	C	AA	Nonsyndromic Sensorineural Deafness	Verbov (1987), Kelsell (1997), White (1998), Kelley (1998), Scott (1998), Griffith (2000), Kelsell (2000), Houseman (2001), D'Andrea (2002), Oshima (2003), Feldmann (2004), Tang (2006)
26333	PAX9	A	G	AA	Possibly associated with Nonsyndromic Cleft Lip with or without Cleft Palate	Ichikawa (2006)
100745	MTTL1	T	C	?	MELAS Syndrome	Goto (1991), Hayashi (1993), Stenqvist (2005), Stenqvist (2005)
102507	ANKRD1	G	C	GG	Possible Risk of Total Anomalous Pulmonary Venous Return	Cinquetti (2008)
55551	PROZ	G	A	GG	Possible Protein Z Deficiency	Iwata (2005)
14804	CARD15	G	A	GG	Possible Association with Crohn's Disease|Sarcoidosis, Early-onset	Lesage (2002), Kanazawa (2005)
8702	PPOX	C	T	CC	Porphyria, Variegate	Dean, (1972), Hift , (1993), Meissner (1996), Meissner (1996), Warnich (1996), De Villiers (1999)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
31314	HMBS	G	A	GG	Porphyria, Acute Intermittent, Nonerythroid Variant	Grandchamp (1989), Petrides (1998)
31317	HMBS	G	T	GG	Porphyria, Acute Intermittent, Nonerythroid Variant	Grandchamp (1989)
31239	HMBS	C	T	CC	Porphyria, Acute Intermittent	Lee (1990), Gu (1993)
31249	HMBS	C	T	CC	Porphyria, Acute Intermittent	Llewellyn (1992), Gu (1992), Kauppinen (1992), Solis (2004)
31251	HMBS	G	A	GG	Porphyria, Acute Intermittent	Delfau (1990), Kauppinen (1992)
31256	HMBS	G	A	GG	Porphyria, Acute Intermittent	Anvret (1991)
31300	HMBS	G	A	GG	Porphyria, Acute Intermittent	Chen (1994), Schneider-Yin (2002)
31370	HMBS	G	C	GG	Porphyria, Acute Intermittent	Lundin (1997)
31252	HMBS	T	G	TT	Porphyria, Acute Intermittent	Mgone (1992)
31235	HMBS	G	A	GG	Porphyria, Acute Intermittent	Gu (1994), Siervi (1999)
71918	HMBS	D	I	DD	Porphyria, Acute Intermittent	Flachsová (2007)
801005	HMBS	D	I	DD	Porphyria, Acute Intermittent	Flachsová (2007)
55392	ALAD	G	A	GG	Porphyria, Acute Hepatic|Delta-aminolevulinate Dehydratase Porphyria	Doss (1986), Fujita (1987), Sassa (1991), Ishida (1992)
800958	ALAD	C	T	CC	Porphyria, Acute Hepatic|Delta-aminolevulinate Dehydratase Porphyria	Doss (1986), Fujita (1987), Sassa (1991), Ishida (1992)
32689	UROD	C	T	CC	Porphyria Cutanea Tarda	Cappellini (2001)
32698	UROD	G	A	GG	Porphyria Cutanea Tarda	Verneuil (1986), Verneuil (1988), Garey (1989), Roberts (1995), Moran-Jimenez (1996)
32665	UROD	T	C	TT	Porphyria Cutanea Tarda	Christiansen (1999)
32666	UROD	G	A	GG	Porphyria Cutanea Tarda	Phillips (2001)
32667	UROD	G	A	GG	Porphyria Cutanea Tarda	Martinez (2001)
32668	UROD	A	G	AA	Porphyria Cutanea Tarda	Christiansen (2000)
32669	UROD	T	A	TT	Porphyria Cutanea Tarda	Ged (2002)
32670	UROD	C	T	CC	Porphyria Cutanea Tarda	Moran-Jimenez (1996)
32671	UROD	C	G	CC	Porphyria Cutanea Tarda	McManus (1996)
32672	UROD	G	T	GG	Porphyria Cutanea Tarda	Brady (2000)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
32673	UROD	C	T	CC	Porphyria Cutanea Tarda	di Montemuro (2002)
32674	UROD	C	T	CC	Porphyria Cutanea Tarda	McManus (1999)
32675	UROD	G	A	GG	Porphyria Cutanea Tarda	Martinez (1999)
32676	UROD	G	C	GG	Porphyria Cutanea Tarda	Brady (2000)
32677	UROD	C	A	CC	Porphyria Cutanea Tarda	Poblete-Gutierrez (2004)
32678	UROD	C	T	CC	Porphyria Cutanea Tarda	di Montemuro (2002)
32679	UROD	G	A	GG	Porphyria Cutanea Tarda	Phillips (2001)
32680	UROD	G	A	GG	Porphyria Cutanea Tarda	Mendez (2000)
32681	UROD	C	T	CC	Porphyria Cutanea Tarda	Poblete-Gutierrez (2004)
32682	UROD	T	A	TT	Porphyria Cutanea Tarda	Martinez (2001)
32683	UROD	T	G	TT	Porphyria Cutanea Tarda	Mendez (1998)
32685	UROD	T	A	TT	Porphyria Cutanea Tarda	Poblete-Gutierrez (2004)
32686	UROD	T	C	TT	Porphyria Cutanea Tarda	di Montemuro (2002)
32687	UROD	G	C	GG	Porphyria Cutanea Tarda	Phillips (2001)
32688	UROD	C	T	CC	Porphyria Cutanea Tarda	Mendez (1998)
32689	UROD	C	T	CC	Porphyria Cutanea Tarda	Cappellini (2001)
32690	UROD	T	A	TT	Porphyria Cutanea Tarda	Brady (2000)
32691	UROD	G	A	GG	Porphyria Cutanea Tarda	Brady (2000)
32692	UROD	C	T	CC	Porphyria Cutanea Tarda	Martinez (1999)
32693	UROD	A	C	AA	Porphyria Cutanea Tarda	Meguro (1994)
32694	UROD	C	A	CC	Porphyria Cutanea Tarda	Christiansen (1999)
32695	UROD	T	C	TT	Porphyria Cutanea Tarda	Phillips (2001)
32696	UROD	C	T	CC	Porphyria Cutanea Tarda	Martinez (1999)
32697	UROD	T	C	TT	Porphyria Cutanea Tarda	Phillips (2001)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
32699	UROD	G	T	GG	Porphyria Cutanea Tarda	Garey (1989)
32700	UROD	T	G	TT	Porphyria Cutanea Tarda	Brady (2000)
32701	UROD	C	G	CC	Porphyria Cutanea Tarda	Verneuil (1992)
32702	UROD	G	A	GG	Porphyria Cutanea Tarda	Brady (2000)
32703	UROD	C	A	CC	Porphyria Cutanea Tarda	Mendez (1998)
32704	UROD	A	G	AA	Porphyria Cutanea Tarda	Moran-Jimenez (1996)
32705	UROD	G	A	GG	Porphyria Cutanea Tarda	McManus (1996)
32706	UROD	T	C	TT	Porphyria Cutanea Tarda	Christiansen (1999)
32707	UROD	G	A	GG	Porphyria Cutanea Tarda	Mendez (1998)
32708	UROD	T	C	TT	Porphyria Cutanea Tarda	McManus (1996)
32709	UROD	G	T	GG	Porphyria Cutanea Tarda	Martinez (2003)
32710	UROD	A	G	AA	Porphyria Cutanea Tarda	Poblete-Gutierrez (2004)
32711	UROD	A	T	AA	Porphyria Cutanea Tarda	Christiansen (1999)
32712	UROD	G	A	GG	Porphyria Cutanea Tarda	McManus (1999)
32713	UROD	A	G	AA	Porphyria Cutanea Tarda	Christiansen (1999)
32714	UROD	G	C	GG	Porphyria Cutanea Tarda	Garey (1990)
32715	UROD	C	G	CC	Porphyria Cutanea Tarda	Martinez (2002)
32716	UROD	G	C	GG	Porphyria Cutanea Tarda	Martinez (1999)
32717	UROD	G	C	GG	Porphyria Cutanea Tarda	Mendez (2000)
32718	UROD	G	A	GG	Porphyria Cutanea Tarda	Mendez (1998)
54600	CYP2C8	G	C	GG	Poor Paclitaxel Metabolism	Soyama (2001)
44131	GPR56	T	A	TT	Polymicrogyria, Bilateral Frontoparietal	Piao (2004)
48773	VHL	C	T	CC	Polycythemia, Chuvash Type	Ang (2002), Ang (2002), Percy (2002), Percy (2002), Pastore (2003), Liu (2004), Gordeuk (2004), Gordeuk (2004), Cario (2005), Perrotta (2006), Perrotta (2006)
21056	JAK2	G	T	GG	Polycythemia Vera	Baxter (2005), Kralovics (2005), James (2005), Patel (2006), Sozer (2009), Cheng (2013), Karaköse (2015)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
801985	JAK2	G	A	GG	Thrombocythemia 3	Mead (2015)
39828	TREM2	C	A	CC	Polycystic Lipomembranous Osteodysplasia with Sclerosing Leukoencephalopathy	Paloneva (2002)
39841	TREM2	T	C	TT	Polycystic Lipomembranous Osteodysplasia with Sclerosing Leukoencephalopathy	Bird (1983), Paloneva (2002)
39845	TREM2	C	T	CC	Polycystic Lipomembranous Osteodysplasia with Sclerosing Leukoencephalopathy	Paloneva (2002)
10939	PKD1	G	A	GG	Polycystic kidney disease 1	Rossetti (2003)
65756	PKHD1	G	A	GG	Polycystic Kidney Disease	Ward (2002), Furu (2003)
65786	PKHD1	G	A,C	GG	Polycystic Kidney Disease	Bergmann (2003), Bergmann (2003)
65937	PKHD1	A	C	AA	Polycystic Kidney Disease	Bergmann (2003), Bergmann (2003)
75062	PKHD1	T	C	TT	Polycystic Kidney Disease	Michel-Calemard (2009)
19803	CD36	C	T	CC	Platelet Glycoprotein IV Deficiency	Kashiwagi (1993), Kashiwagi (1995), Yanai (2000)
44334	CYP19A1	G	A	GG	Placental Aromatase deficiency	Smith (1994), Morishima (1995), Morishima (1995), Morishima (1995), Bilezikian (1998)
68648	TCF4	G	A	GG	Pitt-Hopkins Syndrome	Amiel (2007), Peippo (2006), Zweier (2007)
68649	TCF4	C	T	CC	Pitt-Hopkins Syndrome	Amiel (2007)
52930	CRB1	G	A	GG	Pigmented Paravenous Chorioretinal Atrophy	McKay (2005)
14269	PDE11A	G	A	GG	Pigmented Nodular Adrenocortical Disease, Primary	Horvath (2006)
45956	KIT	G	A	GG	Piebaldism with Sensorineural Deafness	Giebel (1991), Spritz (1991)
32357	PSPH	C	T	CC	Phosphoserine Phosphatase Deficiency	Jaeken (1997), Veiga-da-Cunha (2004)
32358	PSPH	A	G	AA	Phosphoserine Phosphatase Deficiency	Jaeken (1997), Veiga-da-Cunha (2004)
800941	VHL	C	T	CC	Pheochromocytoma	Crossey (1994), Kuzmin (1995), Crossey (1995), Zbar (1996), Garcia (1997), Neumann (2002), Neumann (2002)
48806	VHL	G	T	GG	Pheochromocytoma	Kuzmin (1995), Crossey (1995), Abbott (2006)
15520	RET	T	C	TT	Pheochromocytoma	Takahashi (1988), Mendonca (1988), Mulligan (1994), Mulligan (1994), Gardner (1994), Hofstra (1996), Tessitore (1999), Nunes (2002), Neumann (2002), Benazzouz (2008)
29023	PAH	C	A,G,T	CC	Phenylketonuria	Abadie (1989), Okano (1990), Superti-Furga (1991), Kleiman (1993), Counsyl (2015), Emory Genetics (2016), GeneDx (2016)
801970	PAH	C	G	CC	Phenylketonuria	Abadie (1989), Okano (1990), Superti-Furga (1991), Kleiman (1993), Cousyl (2016)
29058	PAH	A	G	AA	Phenylketonuria	Konecki (1991), Stojiljkovic (2006)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
29097	PAH	G	A	GG	Phenylketonuria	Wang (1989), Huang (1990)
29120	PAH	C	G,T	CC	Phenylketonuria	Dworniczak (1989)
29230	PAH	G	A,C,T	GG	Phenylketonuria	Abadie (1989), Okano (1991), Kalanin (1994)
29233	PAH	A	G	AA	Phenylketonuria	Hofman (1991)
29255	PAH	C	A	CC	Phenylketonuria	Apold (1990), Melle (1991), Apold (1993)
29272	PAH	C	G,T	CC	Phenylketonuria	Lyonnet (1989), Okano (1990), Byck (1997)
29278	PAH	G	A	GG	Phenylketonuria	Dworniczak (1991), Baric (1994)
29293	PAH	A	C	AA	Phenylketonuria	Eiken (1992), Okano (1989)
29383	PAH	C	G,T	CC	Phenylketonuria	Takahashi (1992), Desviat (1995), Leandro (1995)
29401	PAH	C	T	CC	Phenylketonuria	Eiken (1992), Lin (1992)
29402	PAH	G	A	GG	Phenylketonuria	DiLella (1987), Woo (1988), John (1990), Kalaydjieva (1991), Tsai (1990), Jaruzelska (1991), Zygulska (1991), Baranov (1993), Tighe (2003), Stojiljkovic (2006)
29405	PAH	C	G	CC	Phenylketonuria	Wang (1991)
29408	PAH	T	C	TT	Phenylketonuria	Okano (1991), Genome Diagnostics Laboratory of the University Medical Center Utrecht (2014), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2015), GeneDx (2016), Counsyl (2016), Laboratory Corporation of America (2016), Laboratory for Molecular Medicine of Partners HealthCare Personalized Medicine (2016), Mayo Clinic Genetic Testing Laboratories (2016), Quest Diagnostics (2017), Center for Pediatric Genomic Medicine of Children's Mercy Hospital and Clinics (2017), Fulgent Genetics (2017), Invitae (2017),
29431	PAH	C	T	CC	Phenylketonuria	Dworniczak (1991), Perez (1992), Perez (1993), Desviat (1997), Kalanin (1994)
29450	PAH	C	A,T	CC	Phenylketonuria	Marvit (1987)
29467	PAH	C	T	CC	Phenylketonuria	Wang (1991), Zuo (1986)
29044	PAH	C	A,G,T	CC	Phenylketonuria	Eiken (1992)
46896	SDHD	C	A	CC	Phaeochromocytoma	Neumann (2002)
12666	FGFR1	G	C	GG	Pfeiffer Syndrome|Jackson-Weiss Syndrome	Muenke (1994), Roscioli (2000), Ibrahimi (2004), Ibrahimi (2004)
65087	FGFR2	G	C	GG	Pfeiffer Syndrome	Cohen (1993), Gripp (1998), Kerr (1996), Okajima (1999), Chun (1998), Gorlin (1999), Gripp (1999), Chun (1999), Reardon (2000), Gonzales (2005)
46054	STK11	C	A	CC	Peutz-Jeghers syndrome	Jenne (1998)
46077	STK11	G	A	GG	Peutz-Jeghers syndrome	Jenne (1998), Jenne (1998)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
53014	PAX6	G	C	GG	Peters' anomaly	Stone (1976), Beauchamp (1978), Holmstrom , (1991), Ton , (1991), Hanson (1994), Hanson (1994), Hanson (1994)
63633	PEX26	C	T	CC	Peroxisome Biogenesis Disorder, Complementation Group 8	Steinberg (2004)
30125	PEX12	G	A	GG	Peroxisome Biogenesis Disorder, Complementation Group 3	Gootjes (2004), Preuss (2002)
30126	PEX12	C	A,T	CC	Peroxisome Biogenesis Disorder, Complementation Group 3	Gould (1998)
30444	PEX1	G	A,C	GG	Peroxisome Biogenesis Disorder	Maxwell (2002)
42158	ARFGEF2	G	A	GG	Periventricular Heterotopia with Microcephaly	Sheen (2004)
2566	ELN	G	A	GG	Peripheral pulmonary artery stenosis	Metcalfe (2000)
64071	ECGF1	A	G	AA	Peripheral neuropathy with intestinal pseudo-occlusion	Said (2005)
26165	CTSC	T	C	TT	Periodontitis, Juvenile	Hewitt (2004)
26198	CTSC	C	G,T	CC	Periodontitis, juvenile	Hewitt (2004)
24889	TNFRSF1A	A	C,G	AA	Periodic fever, autosomal dominant	D'Osualdo (2006)
24890	TNFRSF1A	A	C,G	AA	Periodic fever, autosomal dominant	Dode (2002)
24891	TNFRSF1A	A	C,G	AA	Periodic fever, autosomal dominant	Stojanov (2004)
24893	TNFRSF1A	G	A	GG	Periodic fever, autosomal dominant	Aksentijevich (2001)
24894	TNFRSF1A	C	A,T	CC	Periodic fever, autosomal dominant	Simon (2001)
24895	TNFRSF1A	A	G	AA	Periodic fever, autosomal dominant	McDermott (1999)
24897	TNFRSF1A	C	G	CC	Periodic fever, autosomal dominant	Dode (2000), Aksentijevich (2001)
24898	TNFRSF1A	A	C	AA	Periodic fever, autosomal dominant	Aksentijevich (2001)
24899	TNFRSF1A	C	T	CC	Periodic fever, autosomal dominant	Williamson (1982), McDermott (1999)
24900	TNFRSF1A	C	T	CC	Periodic fever, autosomal dominant	Aganna (2003)
24901	TNFRSF1A	G	A	GG	Periodic fever, autosomal dominant	Aganna (2003)
24902	TNFRSF1A	T	C,G	TT	Periodic fever, autosomal dominant	Simon (2001)
24903	TNFRSF1A	C	G	CC	Periodic fever, autosomal dominant	Federici (2006)
24904	TNFRSF1A	A	G	AA	Periodic fever, autosomal dominant	Obici (2002)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
24905	TNFRSF1A	C	G,T	CC	Periodic fever, autosomal dominant	Tchernitchko (2004)
24906	TNFRSF1A	C	G	CC	Periodic fever, autosomal dominant	Siebert (2005)
24908	TNFRSF1A	G	A,T	GG	Periodic fever, autosomal dominant	Aganna (2003)
24909	TNFRSF1A	G	A,T	GG	Periodic fever, autosomal dominant	McDermott (1998), McDermott (1999)
24910	TNFRSF1A	A	G	AA	Periodic fever, autosomal dominant	Aganna (2003)
24911	TNFRSF1A	C	A,T	CC	Periodic fever, autosomal dominant	D'Osualdo (2006)
24912	TNFRSF1A	C	A,T	CC	Periodic fever, autosomal dominant	McDermott (1999)
24913	TNFRSF1A	A	G	AA	Periodic fever, autosomal dominant	Rudofsky (2006)
24914	TNFRSF1A	C	G,T	CC	Periodic fever, autosomal dominant	D'Osualdo (2006)
24916	TNFRSF1A	A	C,G	AA	Periodic fever, autosomal dominant	Haas (2006)
24917	TNFRSF1A	G	C	GG	Periodic fever, autosomal dominant	Aganna (2003)
24918	TNFRSF1A	G	A,T	GG	Periodic fever, autosomal dominant	Ida (2004)
24919	TNFRSF1A	T	A	TT	Periodic fever, autosomal dominant	Aganna (2003)
24920	TNFRSF1A	A	G	AA	Periodic fever, autosomal dominant	Dode (2002)
24921	TNFRSF1A	A	T	AA	Periodic fever, autosomal dominant	Aganna (2003)
24922	TNFRSF1A	A	C,G,T	AA	Periodic fever, autosomal dominant	Aganna (2002)
24923	TNFRSF1A	A	C,G,T	AA	Periodic fever, autosomal dominant	Horiuchi (2004)
24924	TNFRSF1A	C	T	CC	Periodic fever, autosomal dominant	Simon (2001)
24925	TNFRSF1A	A	G	AA	Periodic fever, autosomal dominant	Stjernberg-Salmela (2004)
24926	TNFRSF1A	G	C	GG	Periodic fever, autosomal dominant	Federici (2006)
24927	TNFRSF1A	A	G	AA	Periodic fever, autosomal dominant	Aksentijevich (2001)
24928	TNFRSF1A	C	T	CC	Periodic fever, autosomal dominant	Karenko (1992), McDermott (1999)
24929	TNFRSF1A	A	G	AA	Periodic fever, autosomal dominant	Mulley (1998), McDermott (1999)
24931	TNFRSF1A	C	G	CC	Periodic fever, autosomal dominant	Aganna (2001)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
24933	TNFRSF1A	C	T	CC	Periodic fever, autosomal dominant	Dode (2002)
24934	TNFRSF1A	C	T	CC	Periodic fever, autosomal dominant	Weyhreter (2003)
24936	TNFRSF1A	A	T	AA	Periodic fever, autosomal dominant	Nevala (2002)
24937	TNFRSF1A	A	T	AA	Periodic fever, autosomal dominant	Kriegel (2003)
24932	TNFRSF1A	C	T	CC	Periodic fever, atypical	Rack (2006)
24896	TNFRSF1A	C	A,G,T	CC	Periodic fever with amyloidosis	Broeders (2004)
24915	TNFRSF1A	C	G	CC	Periodic fever with AA amyloidosis	Jadoul (2001)
25662	SLC26A4	A	C	AA	Pendred Syndrome	Van Hauwe (1998), Coyle (1998), Alasti (1998), Campbell (2001), Napiontek (2004), Yoon (2008), GeneReviews (2011), Laboratory for Molecular Medicine of Partners HealthCare Personalized Medicine (2015), GeneDx (2016)
25710	SLC26A4	A	G	AA	Pendred syndrome	Scott (2000), Rotman-Pikielny (2002), Borck (2003), Shears (2004), Blons (2004), Prasad (2004), Pryor (2005), Hutchin (2005), Yoon (2008), Dai (2009), Choi (2009), Jonard (2010), Laboratory for Molecular Medicine of Partners HealthCare Personalized Medicine (2011), GeneReviews (2014)
25779	SLC26A4	A	G	AA	Pendred syndrome	Coucke (1999)
25692	SLC26A4	G	T	GG	Pendred syndrome	Borck (2003), Borck (2003)
25784	SLC26A4	G	A	GG	Pendred syndrome	England, the region in which the family described in (1896), Coyle (1998), Coyle (1998)
25701	SLC26A4	T	C	TT	Pendred syndrome	Van Hauwe (1998), Coyle (1998)
25721	SLC26A4	T	G	TT	Pendred syndrome	Masmoudi (2000)
9237	TGM5	C	A	CC	Peeling Skin Syndrome	Cassidy (2005)
30649	PDHA1	C	A	C	PDH deficiency	Brown (1994), Naito (2002)
30668	PDHA1	C	T	C	PDH deficiency	Dahl (1992), Otero (1998)
30675	PDHA1	G	A	G	PDH deficiency	Hansen (1991), Matthews (1994)
6021	CRELD1	G	A	GG	Partial Atrioventricular Septal Defect and Heterotaxy Syndrome	Robinson (2003)
40244	SCN9A	G	A,C	GG	Paroxysmal extreme pain disorder	Fertleman (2006)
39883	PARK2	C	T	CC	Parkinsonism, juvenile, autosomal recessive	Pineda-Trujillo (2001), Hoenicka (2002)
38201	PINK1	C	T	CC	Parkinson disease, early-onset	Hatano (2004)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
38223	PINK1	G	A	GG	Parkinson disease, early-onset	Valente (2004), Silvestri (2005), Piccoli (2008), Piccoli (2008)
802007	PINK1	G	C	GG	PINK1-Related Parkinsonism	GeneDx (2015)
802008	PINK1	I	D	II	Parkinson Disease 6, Early-onset	Laboratory for Molecular Medicine of Partners HealthCare Personalized Medicine (2014)
802009	PINK1	C	A	CC	Parkinson Disease 6, Early-onset	Leutenegger (2006)
802010	PINK1	C	T	CC	Parkinson Disease 6, Early-onset	Hatano (2004)
38177	PINK1	C	A	CC	Parkinson Disease 6, Early-onset	Hatano (2004)
38181	PINK1	T	C	TT	Parkinson Disease 6, Early-onset	Hatano (2004), Rogaeva (2004), Beilina (2005)
42313	LRRK2	A	G	AA	Parkinson Disease	Wszolek (1997), Paisan-Ruiz (2004), Zimprich (2004), Gasser (2005), GeneReviews (2012)
42258	LRRK2	C	G	CC	Parkinson Disease	Paisan-Ruiz (2004), Gasser (2005), GeneReviews (2012)
42259	LRRK2	C	T	CC	Parkinson Disease	Wszolek (1995), Zimprich (2004), West (2005), GeneReviews (2012), Bioscientia Institut fuer Medizinische Diagnostik GmbH of Sonic Healthcare (2017), Invitae (2017)
802006	LRRK2	C	A	CC	Parkinson Disease	Zabetian UW Neurogenetics Lab of the University of Washington VAPSHCS - Parkinson's Genetic Research Study PaGeR (2016)
42260	LRRK2	G	A	GG	Parkinson Disease	Mata (2005), Zabetian (2005), GeneReviews (2012)
42249	LRRK2	G	T	GG	Likely Harmless Variant (Previously associated with Parkinson Disease)	Berg (2005), Illumina Clinical Services Laboratory (2016)
43205	UCHL1	C	G	CC	Increased risk of Parkinson Disease	Leroy (1998), Healy (2006)
42202	SNCAIP	C	T	CC	Likely Harmless Variant (Previously associated with Parkinson Disease)	Marx (2003), Marx (2003), Pankratz (2007), Illumina Clinical Services Laboratory (2016)
41377	SNCA	C	T	CC	Parkinson Disease	Zarranz (2004), Choi (2004), Greenbaum (2005)
43190	NR4A2	A	C	AA	Possibly associated with Parkinson Disease	Le (2003), Le (2003), Hering (2004)
43192	NR4A2	I	D	TT	Possibly associated with Parkinson Disease	Le (2003), Hering (2004), Ibanez (2004)
47278	CDC73	C	T	CC	Parathyroid Cancer	Shattuck (2003)
47279	CDC73	C	T	CC	Parathyroid Cancer	Cetani (2004)
44173	SCN4A	C	T	CC	Paramyotonia congenita	Koch (1995)
44186	SCN4A	C	A,G,T	CC	Paramyotonia congenita	Meyer-Kleine (1994)
46909	SDHD	G	T	GG	Paragangliomas|Phaeochromocytoma	Baysal (2000), Neumann (2002)
26201	CTSC	G	A	GG	Papillon-Lefevre Syndrome	Hart (1999), Hart (2000)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
46863	MET	G	A	GG	Papillary renal carcinoma	Olivero (1999)
63791	PSTPIP1	G	A	GG	PAPA syndrome	Wise (2002)
63792	PSTPIP1	G	C	GG	PAPA syndrome	Wise (2002)
40745	PANK2	C	T	CC	Pantothenate Kinase-associated Neurodegeneration	Zhou (2001), Hayflick (2003)
40744	PANK2	G	A	GG	Pantothenate Kinase-associated Neurodegeneration	Zhou (2001)
15919	PRSS1	G	A	GG	Pancreatitis, hereditary	Whitcomb (1996), Ferec (1999), Ferec, (2000), Chen (2000), Audrezet (2002), Simon (2002), Simon (2002), Teich (2006), Teich (2006)
16162	SPINK1	C	T	CC	Pancreatitis, Chronic	Kaneko (2001)
801956	SPINK1	C	A	CC	Pancreatitis, Chronic	Chandak (2002)
16164	SPINK1	G	A	GG	Pancreatitis, Chronic	Witt (2000)
8602	KRT9	G	A	GG	Palmoplantar Keratoderma, Epidermolytic	Thost (1880), Hutchinson (1989), Becker, (1992), Hennies (1993), Reis (1994), Bonifas (1994), Reis (1994), Bonifas (1994)
8603	KRT9	A	G	AA	Palmoplantar Keratoderma, Epidermolytic	Rothnagel (1995)
34245	SQSTM1	A	G	AA	Paget Disease of Bone	Hocking (2004)
34242	SQSTM1	C	T	CC	Paget Disease of Bone	Laurin (2002)
9482	KRT17	T	C	TT	Pachyonychia Congenita, Type 2	Todd , (1990), Munro , (1994), Smith (1997), Covello (1998), Covello (1998)
9732	KRT6A	A	C,G	AA	Pachyonychia Congenita, Type 1	Terrinoni (2001)
9733	KRT6A	C	T	CC	Pachyonychia Congenita, Type 1	Terrinoni (2001)
19331	A4GALT	G	A,T	GG	P Blood Group System, p Phenotype	Furukawa (2002)
50020	FH	C	T	CC	Ovarian mucinous cystadenoma	Ylisaukko-Oja (2006)
64761	CREBBP	T	C	TT	Increased Risk of Ovarian Cancer	Ward (2005)
45780	BARD1	C	G	CC	Increased Risk of Ovarian Cancer	Thai (1998)
64761	CREBBP	T	C	TT	Increased Risk of Ovarian Cancer	Ward (2005)
31518	OTC	C	T	C	OTC Deficiency	Maddalena (1988), Hata (1989)
31440	OTC	G	A	G	OTC Deficiency	Ploechl (2001), Tuchman (1994), Leibundgut (1995), Matsuda (1996), Mavinakere (2001)
31503	OTC	G	A	G	OTC Deficiency	Garcia-Perez (1995)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
31515	OTC	G	A	G	OTC Deficiency	Maddalena (1988), Nussbaum (1989), Strautnieks (1991), Suess (1992)
47386	TP53	T	C	TT	Osteosarcoma	McIntyre (1994)
100836	CLCN7	I	D	AA	Osteopetrosis, Type 2	Cleiren (2001)
71837	TCIRG1	G	A	GG	Osteopetrosis	Frattini (2000), Sobacchi (2001)
69947	TNFSF11	T	A	TT	Osteopetrosis	Sobacchi (2007)
36045	OSTM1	C	T	CC	Osteopetrosis	Chalhoub (2003), Chalhoub (2003)
34116	COL1A2	G	A	GG	Osteogenesis Imperfecta/Ehlers-Danlos Crossover Syndrome	Nathanson (1997), Nathanson (1997)
34107	COL1A2	T	C	TT	Osteogenesis Imperfecta/Ehlers-Danlos Crossover Syndrome	Nicholls (2001)
33982	COL1A2	G	T	GG	Osteogenesis Imperfecta, Type IV|Osteogenesis Imperfecta, Type III	Bateman (1991), Forlino (1994), Cole (1996), Lund (1997)
34018	COL1A2	G	A	GG	Osteogenesis Imperfecta, Type III	De Paepe (1997)
34091	COL1A2	A	G	AA	Osteogenesis imperfecta IV	Zolezzi (1997)
35658	COL1A1	C	A,T	CC	Osteogenesis Imperfecta IV	Bateman (1992), Korkko (1997), Korkko (1997)
35889	COL1A1	C	G,T	CC	Osteogenesis Imperfecta IV	Schwarze (1999), Schwarze (1999), Schwarze (1999), Schwarze (1999), Schwarze (1999)
34033	COL1A2	G	A	GG	Osteogenesis imperfecta III	Versfeld, (1985), Wallis , (1993), Rose (1994), Rose (1994)
35603	COL1A1	C	T	CC	Osteogenesis Imperfecta III	Pruchno (1991), Zhuang (1996)
35793	COL1A1	C	T	CC	Osteogenesis Imperfecta III	Aitchison (1988), Pruchno (1991), Wallis (1993), Zhuang (1996), Zhuang (1996)
33965	COL1A2	G	A	GG	Osteogenesis imperfecta II	Rose (1994), Steinmann (1995)
35779	COL1A1	G	A,T	GG	Osteogenesis Imperfecta I	Willing (1994), Korkko (1997)
35847	COL1A1	C	A,T	CC	Osteogenesis Imperfecta I	Genovese (1989), Stover (1993), Stover (1993)
59443	FRZB	C	T	CC	Osteoarthritis of the Hip	Loughlin (2004)
37022	SLC6A2	G	C	GG	Orthostatic Intolerance and Tachycardia	Shannon (2000), Paczkowski (2002)
53436	OPA3	G	C	GG	Optic Atrophy and Cataract	Reynier (2004)
53437	OPA3	C	T	CC	Optic Atrophy and Cataract	Garcin (1961), Reynier (2004)
52613	OPA1	G	A	GG	Optic Atrophy 1 with Deafness|Optic Atrophy, Deafness, Opthalmoplegia, and Myopathy	Treft (1984), Meire (1985), Shimizu (2003), Amati-Bonneau (2003), Payne (2004), Li (2005), Li (2005), Amati-Bonneau (2005)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
70704	OPA1	I	D	II	Optic Atrophy 1	Thiselton (2001)
100971	OPA1	I	D	II	Optic Atrophy 1	Delettre (2000), Votruba (1998), Johnston (1999), Toomes (2001)
78407	MED12	C	T	C	Opitz-Kaveggia Syndrome	Risheg (2007)
63919	MID1	G	A	G	Opitz GBBB Syndrome	Chen (2007)
64526	FLNA	C	T	C	OPD Syndrome 2	Robertson (2003)
24154	RAG1	C	T	CC	Omenn Syndrome|Severe Combined Immunodeficiency, B Cell-negative	Villa (1998), Corneo (2001)
24165	RAG1	G	A	GG	Omenn syndrome	Villa (1998)
100874	RAG1	I	D	II	Omenn Syndrome	Corneo (2001), De Villartay (2005)
100875	RAG1	I	D	II	Omenn Syndrome	Villa (1998), Santagata (2000), De Villartay (2005)
101055	WNT10A	C	A	CC	Odontoonychodermal Dysplasia	Bohring (2009)
78500	WNT10A	G	T	GG	Odontoonychodermal Dysplasia	Adaimy (2007)
7963	TYR	C	T	CC	Oculocutaneous Albinism, Type 1A	Giebel (1990), Giebel (1990), Giebel (1991)
7999	TYR	G	A	GG	Oculocutaneous Albinism, Type 1A	Tripathi (1992), Oetting (1993), IA oculocutaneous albinism (2031)
7958	TYR	A	G	AA	Ocular Albinism	Fukai (1995)
31037	ATP7A	C	T	C	Occipital Horn Syndrome	Ronce (1997), Tumer (1997)
10253	MC4R	G	A	??	Obesity, Severe, Early-onset	Lubrano-Berthelier (2003), Lubrano-Berthelier (2003), Hinney (2003), Valli-Jaakola (2004), Valli-Jaakola (2004)
10176	UCP3	G	A	GG	Obesity, Severe|Diabetes Mellitus, Type 2	Brown (1999), Brown (1999)
10187	UCP3	G	A	GG	Obesity, Severe|Diabetes Mellitus, Type 2	Argyropoulos (1998), Argyropoulos (1998)
14047	PPARG	C	A	CC	Obesity, Severe	Ristow (1998), Wang , (1999)
10469	LEP	C	T	CC	Obesity, Morbid, with Hypogonadism	Strobel (1998)
10444	LEPR	G	A	GG	Obesity, Morbid, with Hypogonadism	Clement (1998)
78600	NTRK2	A	G	AA	Obesity, Hyperphagia, and Developmental Delay	Yeo (2004), Xu (2003)
11972	POMC	G	C	GG	Obesity, Early-onset, Increased risk	Challis (2002), GeneDx (2017)
10289	MC4R	C	T	??	Obesity, Early-onset	Yeo (2003), Farooqi (2003)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
10443	LEPR	A	C	AA	Obesity, early-onset	Farooqi (2007)
10439	LEPR	C	A	CC	Obesity, early-onset	Farooqi (2007)
10108	GHRL	C	T	CC	Metabolic Syndrome and Obesity, Delayed-onset, Increased risk	Ukkola (2001), Hinney (2002), Steinle (2005)
10269	MC4R	G	A	??	Obesity, autosomal dominant	Giudice (2002)
10275	MC4R	T	C	??	Obesity, autosomal dominant	Lubrano-Berthelier (2006)
10278	MC4R	A	C,G	??	Obesity, autosomal dominant	Lubrano-Berthelier (2006)
10284	MC4R	G	A,T	??	Obesity, autosomal dominant	Lubrano-Berthelier (2006)
10295	MC4R	C	T	??	Obesity, autosomal dominant	Farooqi (2000)
10298	MC4R	T	C	??	Obesity, autosomal dominant	Mergen (2001)
10303	MC4R	A	G	??	Obesity, autosomal dominant	Vaisse (2000)
10307	MC4R	A	C	??	Obesity, autosomal dominant	Farooqi (2003), Yeo (2003)
12001	POMC	C	A	CC	Obesity ?	Hinney (1998)
10088	NR0B2	G	A	GG	Obesity	Nishigori (2001)
10091	NR0B2	C	T	CC	Obesity	Nishigori (2001)
10246	MC4R	G	T	??	Obesity	Hinney (1999)
10248	MC4R	T	A	??	Obesity	Hinney (1999)
10257	MC4R	G	T	??	Obesity	Lubrano-Berthelier (2003)
10259	MC4R	A	G	??	Obesity	Hinney (2003)
11996	POMC	C	G	CC	Obesity	Giudice (2001)
11997	POMC	G	A	GG	Obesity	Giudice (2001)
12003	POMC	G	A	GG	Obesity	Buono (2005)
12004	POMC	C	A	CC	Obesity	Buono (2005)
53043	PAX6	G	C	GG	Nystagmus	Sonoda (2000)
24206	NP	G	C	GG	Nucleoside Phosphorylase Deficiency	Aust (1992), Markert (1997)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
27611	NDP	T	C	T	Norrie Disease	Isashiki (1995)
27613	NDP	A	C	A	Norrie Disease	Fuchs (1994)
33553	SOS1	A	C,G	AA	Noonan Syndrome 4	Roberts (2007), Tartaglia (2007)
65433	KRAS2	C	T	CC	Noonan Syndrome 3	Schubbert (2006)
65433	KRAS2	C	T	CC	Noonan Syndrome 3	Schubbert (2006)
65440	KRAS2	G	A	GG	Noonan Syndrome 3	Schubbert (2006)
65442	KRAS2	A	C	AA	Noonan Syndrome 3	Carta (2006)
65443	KRAS2	T	A	TT	Noonan Syndrome 3	Niihori (2006), Schubbert (2006), Carta (2006)
61813	PTPN11	A	G	AA	Noonan Syndrome 1	Tartaglia (2001), Tartaglia (2002), Kosaki (2002), Jongmans (2005)
33551	SOS1	C	T	CC	Noonan Syndrome	Tartaglia (2007)
33552	SOS1	G	T	GG	Noonan Syndrome	Roberts (2007)
800895	SOS1	A	C,G	AA	Noonan Syndrome	Roberts (2007)
33554	SOS1	C	A	CC	Noonan Syndrome	Roberts (2007)
33555	SOS1	T	C	TT	Noonan Syndrome	Roberts (2007)
33556	SOS1	A	G	AA	Noonan Syndrome	Tartaglia (2007)
33557	SOS1	C	T	CC	Noonan Syndrome	Tartaglia (2007)
33558	SOS1	C	G	CC	Noonan Syndrome	Roberts (2007)
33559	SOS1	C	T	CC	Noonan Syndrome	Tartaglia (2007)
33560	SOS1	T	G	TT	Noonan Syndrome	Tartaglia (2007)
33561	SOS1	A	C,G	AA	Noonan Syndrome	Tartaglia (2007)
33562	SOS1	T	A,C	TT	Noonan Syndrome	Roberts (2007)
33563	SOS1	C	A,G,T	CC	Noonan Syndrome	Tartaglia (2007)
33564	SOS1	C	G	CC	Noonan Syndrome	Tartaglia (2007)
33566	SOS1	A	G	AA	Noonan Syndrome	Tartaglia (2007)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
33567	SOS1	C	A	CC	Noonan Syndrome	Tartaglia (2007)
33568	SOS1	T	A	TT	Noonan Syndrome	Tartaglia (2007)
33569	SOS1	C	T	CC	Noonan Syndrome	Tartaglia (2007)
78851	RAF1	G	A,C	GG	Noonan Syndrome	Razzaque (2007), Pandit (2007)
800836	RAF1	G	A,C	GG	LEOPARD Syndrome	Pandit (2007), Razzaque (2007)
78852	RAF1	G	A	GG	Noonan Syndrome	Pandit (2007)
78853	RAF1	G	C	GG	Noonan Syndrome	Pandit (2007)
78856	RAF1	G	A	GG	Noonan Syndrome	Razzaque (2007)
78858	RAF1	T	C	TT	Noonan Syndrome	Razzaque (2007)
78861	RAF1	G	A	GG	Noonan Syndrome	Pandit (2007)
78862	RAF1	G	C	GG	Noonan Syndrome	Pandit (2007)
61770	PTPN11	C	T	CC	Noonan syndrome	Sarkozy (2003)
61771	PTPN11	A	G	AA	Noonan syndrome	Tartaglia (2002)
61773	PTPN11	C	G	CC	Noonan syndrome	Musante (2003)
61774	PTPN11	A	C	AA	Noonan syndrome	Limal (2006)
61775	PTPN11	A	G	AA	Noonan syndrome	Zenker (2004)
61776	PTPN11	G	C	GG	Noonan syndrome	Tartaglia (2002)
61777	PTPN11	G	T	GG	Noonan syndrome	Limal (2006)
61778	PTPN11	A	G	AA	Noonan syndrome	Tartaglia (2001)
61779	PTPN11	G	A	GG	Noonan syndrome	Tartaglia (2002)
61780	PTPN11	T	A	TT	Noonan syndrome	Tartaglia (2006)
61781	PTPN11	T	G	TT	Noonan syndrome	Tartaglia (2002)
61782	PTPN11	A	G	AA	Noonan syndrome	Tartaglia (2001)
61784	PTPN11	G	C	GG	Noonan syndrome	Musante (2003)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
61785	PTPN11	T	A	TT	Noonan syndrome	Niihori (2005)
61786	PTPN11	T	C	TT	Noonan syndrome	Musante (2003)
61788	PTPN11	C	G	CC	Noonan syndrome	Tartaglia (2001)
61790	PTPN11	G	T	GG	Noonan syndrome	Tartaglia (2001)
61791	PTPN11	C	T	CC	Noonan syndrome	Tartaglia (2002)
61793	PTPN11	G	C	GG	Noonan syndrome	Tartaglia (2001)
61794	PTPN11	G	T	GG	Noonan syndrome	Tartaglia (2006)
61797	PTPN11	A	C	AA	Noonan syndrome	Sarkozy (2003)
61798	PTPN11	A	G	AA	Noonan syndrome	Tartaglia (2001)
61799	PTPN11	A	C	AA	Noonan syndrome	Tartaglia (2002)
61800	PTPN11	A	C	AA	Noonan syndrome	Zenker (2004)
61801	PTPN11	G	C	GG	Noonan syndrome	Tartaglia (2002)
61802	PTPN11	G	T	GG	Noonan syndrome	Tartaglia (2002)
61803	PTPN11	C	A	CC	Noonan syndrome	Binder (2005)
61804	PTPN11	A	G	AA	Noonan syndrome	Musante (2003)
61805	PTPN11	G	A	GG	Noonan syndrome	Tartaglia (2006)
61806	PTPN11	G	T	GG	Noonan syndrome	Tartaglia (2006)
61808	PTPN11	A	G	AA	Noonan syndrome	Tartaglia (2002)
61809	PTPN11	A	G	AA	Noonan syndrome	Tartaglia (2001)
61810	PTPN11	T	C	TT	Noonan syndrome	Tartaglia (2002)
61811	PTPN11	T	C	TT	Noonan syndrome	Tartaglia (2002)
61812	PTPN11	T	G	TT	Noonan syndrome	Tartaglia (2006)
800961	PTPN11	A	G	AA	Noonan Syndrome	Tartaglia (2001), Tartaglia (2002), Tartaglia (2002), Tartaglia (2002), Kosaki (2002), Jongmans (2005)
61814	PTPN11	A	C	AA	Noonan syndrome	Zenker (2004)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
61815	PTPN11	A	G	AA	Noonan syndrome	Tartaglia (2002)
61816	PTPN11	A	G	AA	Harmless variant (previously associated with Noonan syndrome)	Tartaglia (2002), Laboratory for Molecular Medicine of Partners HealthCare Personalized Medicine (2014), Ambry Genetics (2014), GeneDx (2016), ClinGen RASopathy Expert Panel (2017), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2016), Invitae (2016)
61817	PTPN11	G	C	GG	Noonan syndrome	Zenker (2007)
61818	PTPN11	C	T	CC	Noonan syndrome	Bertola (2004)
61822	PTPN11	C	T	CC	Noonan syndrome	Binder (2005)
61823	PTPN11	C	T	CC	Noonan syndrome	Zenker (2004)
61826	PTPN11	G	A	GG	Noonan syndrome	Tartaglia (2002)
61827	PTPN11	T	A	TT	Noonan syndrome	Maheshwari (2002)
61828	PTPN11	T	G	TT	Noonan syndrome	Kratz (2005)
61830	PTPN11	G	A	GG	Noonan syndrome	Tartaglia (2006)
61831	PTPN11	G	C	GG	Noonan syndrome	Sarkozy (2003)
61832	PTPN11	A	G	AA	Noonan syndrome	Tartaglia (2001)
61834	PTPN11	C	G	CC	Noonan syndrome	Takahashi (2005)
61836	PTPN11	A	G	AA	Noonan syndrome	Bertola (2005)
61837	PTPN11	C	T	CC	Noonan syndrome	Sarkozy (2003)
65435	KRAS2	T	A,C	TT	Noonan syndrome	Zenker (2007)
65436	KRAS2	G	T	GG	Noonan syndrome	Zenker (2007)
65438	KRAS2	G	A,C	GG	Noonan syndrome	Zenker (2007)
65439	KRAS2	T	C	TT	Noonan syndrome	Zenker (2007)
65444	KRAS2	A	T	AA	Noonan syndrome	Zenker (2007)
65445	KRAS2	G	C	GG	Noonan syndrome	Zenker (2007), Zenker (2007)
26517	EYA4	C	T	CC	Nonsyndromic Sensorineural Deafness	Wayne (2001)
71974	ESRRB	G	T	GG	Nonsyndromic Hearing Impairment	Collin (2008)
100805	SCN1B	G	C	GG	Nonspecific Cardiac Conduction Defect	Watanabe (2008)

SEQUENCING

Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
100807	SCN1B	G	A	GG	Nonspecific Cardiac Conduction Defect	Watanabe (2008)
48432	EGFR	C	T	CC	Nonsmall Cell Lung Cancer, Bronchoalveolar Subtype, Resistant to Tyrosine Kinase Inhibitors	Bell (2005)
42648	CHRNA2	A	T	AA	Nocturnal Frontal Lobe Epilepsy, Type 4, with Nocturnal Wandering and Ictal Fear	Aridon (2006), Aridon (2006)
75034	NBN	G	A,T	GG	Nijmegen Breakage Syndrome	Nakanishi (2002)
52133	RHO	C	T	CC	Nightblindness, Congenital Stationary	Jandal (1999)
53981	CABP4	C	T	CC	Night Blindness, Congenital Stationary, Type 2B	Zeitz (2006)
54023	CACNA1F	A	G	A	Night Blindness, Congenital Stationary, Type 2, Severe	Hemara-Wahanui (2005), Hemara-Wahanui (2005), Hemara-Wahanui (2005)
54015	CACNA1F	C	T	C	Night Blindness, Congenital Stationary, Type 2	Strom (1998)
54027	CACNA1F	G	A	G	Night Blindness, Congenital Stationary, Type 2	Strom (1998)
100849	CACNA1F	D	I	D	Night Blindness, Congenital Stationary, Type 2	Boycott (1998), Bech-Hansen (1998)
54110	GRM6	G	A	GG	Night Blindness, Congenital Stationary, Type 1B	Dryja (2005)
52796	NYX	G	A	G	Night Blindness, Congenital Stationary, Type 1	Bech-Hansen (2000)
52780	NYX	T	C	T	Night Blindness, Congenital Stationary, Type 1	Xiao (2006)
26771	NPC1	C	A,T	CC	Niemann-Pick type II disease	Greer (1999)
26773	NPC1	G	A	GG	Niemann-Pick type II disease	Greer (1999)
26778	NPC1	C	T	CC	Niemann-Pick type II disease	Greer (1999)
26781	NPC1	G	A	GG	Niemann-Pick type II disease	Greer (1999)
26783	NPC1	C	T	CC	Niemann-Pick Disease, Variant Type C1	Sun (2001)
26799	NPC1	G	C	GG	Niemann-Pick Disease, Variant Type C1	Sun (2001)
26796	NPC1	C	A,G,T	CC	Niemann-Pick Disease, Type D	Greer (1997)
26867	NPC2	C	A,T	CC	Niemann-Pick Disease, Type C2	Naureckiene (2000), Millat (2001)
26867	NPC2	C	A,T	CC	Niemann-Pick Disease, Type C2	Naureckiene (2000), Millat (2001)
26868	NPC2	C	T	CC	Likely Harmless Variant (Previously associated with Niemann-Pick Disease, Type C2)	Park (2003), Emory Genetics Laboratory of Emory University (2015)
26869	NPC2	C	T	CC	Niemann-Pick Disease, Type C2	Klunemann (2002)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
26870	NPC2	G	A	GG	Niemann-Pick Disease, Type C2	Chikh (2005)
26871	NPC2	C	A	CC	Niemann-Pick Disease, Type C2	Park (2003)
26872	NPC2	G	T	GG	Niemann-Pick Disease, Type C2	Chikh (2005)
26873	NPC2	A	G	AA	Niemann-Pick Disease, Type C2	Millat (2001)
26874	NPC2	C	A	CC	Niemann-Pick Disease, Type C2	Park (2003)
26875	NPC2	A	G	AA	Niemann-Pick Disease, Type C2	Chikh (2005)
26876	NPC2	C	A,T	CC	Niemann-Pick Disease, Type C2	Millat (2001)
26877	NPC2	G	A	GG	Niemann-Pick Disease, Type C2	Millat (2005)
26878	NPC2	G	A	GG	Niemann-Pick Disease, Type C2	Millat (2005)
26879	NPC2	A	G	AA	Niemann-Pick Disease, Type C2	Park (2003)
26880	NPC2	C	T	CC	Niemann-Pick Disease, Type C2	Millat (2001)
26816	NPC1	T	C	TT	Niemann-Pick Disease, Type C1, Juvenile Form	Yamamoto (1999)
26838	NPC1	C	G	CC	Niemann-Pick Disease, Type C1, Juvenile Form	Yamamoto (1999)
26759	NPC1	C	T	CC	Niemann-Pick Disease, Type C1, Adult Form	Yamamoto (1999)
800891	NPC1	G	C	GG	Niemann-Pick Disease, Type C1	Sun (2001), Millat (2001)
26811	NPC1	A	G	AA	Niemann-Pick Disease, Type C1	Millat (1999)
26826	NPC1	T	A,C	TT	Niemann-Pick Disease, Type C1	Carstea (1997)
26858	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Yamamoto (1999)
26672	NPC1	A	G	AA	Niemann-Pick Disease, Type C1	Meiner (2001)
26673	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Park (2003)
26674	NPC1	T	C	TT	Niemann-Pick Disease, Type C1	Ribeiro (2001)
26675	NPC1	A	G	AA	Niemann-Pick Disease, Type C1	Blom (2003)
26676	NPC1	G	A,T	GG	Niemann-Pick Disease, Type C1	Kaminski (2002)
26677	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Sun (2001)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
26678	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Millat (2005)
26679	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Park (2003)
26680	NPC1	A	C	AA	Niemann-Pick Disease, Type C1	Yamamoto (1999)
26681	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Ribeiro (2001)
26682	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Millat (2005)
26683	NPC1	G	T	GG	Niemann-Pick Disease, Type C1	Yamamoto (1999)
26685	NPC1	A	C	AA	Niemann-Pick Disease, Type C1	Kaminski (2002)
26687	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Sun (2001)
26688	NPC1	C	G	CC	Niemann-Pick Disease, Type C1	Millat (2001)
26689	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Park (2003)
26690	NPC1	C	A,T	CC	Niemann-Pick Disease, Type C1	Sun (2001)
26691	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Fernandez-Valero (2005)
26692	NPC1	A	C	AA	Niemann-Pick Disease, Type C1	Millat (2001)
26693	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Fernandez-Valero (2005)
26694	NPC1	A	G	AA	Niemann-Pick Disease, Type C1	Millat (2001)
26695	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Park (2003)
26696	NPC1	C	G	CC	Niemann-Pick Disease, Type C1	Park (2003)
26697	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Park (2003)
26698	NPC1	G	A,T	GG	Niemann-Pick Disease, Type C1	Sun (2001)
26699	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Park (2003)
26700	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Millat (2001)
26701	NPC1	C	G	CC	Niemann-Pick Disease, Type C1	Millat (2005)
26702	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Park (2003)
26703	NPC1	G	T	GG	Niemann-Pick Disease, Type C1	Fernandez-Valero (2005)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
26704	NPC1	G	A,C	GG	Niemann-Pick Disease, Type C1	Park (2003)
26705	NPC1	G	A,C	GG	Niemann-Pick Disease, Type C1	Fernandez-Valero (2005)
26706	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Tarugi (2002)
26707	NPC1	A	G	AA	Niemann-Pick Disease, Type C1	Yamamoto (1999)
26708	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Tarugi (2002)
26709	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Fernandez-Valero (2005)
26710	NPC1	T	G	TT	Niemann-Pick Disease, Type C1	Park (2003)
26711	NPC1	T	G	TT	Niemann-Pick Disease, Type C1	Yamamoto (1999)
26712	NPC1	T	C	TT	Niemann-Pick Disease, Type C1	Bauer (2002)
26713	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Ribeiro (2001)
26714	NPC1	C	A,T	CC	Niemann-Pick Disease, Type C1	Park (2003)
26715	NPC1	A	T	AA	Niemann-Pick Disease, Type C1	Millat (2005)
26716	NPC1	C	G	CC	Niemann-Pick Disease, Type C1	Sevin (2007)
26717	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Park (2003)
26718	NPC1	T	C	TT	Niemann-Pick Disease, Type C1	Fernandez-Valero (2005)
26719	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Millat (2001)
26720	NPC1	G	A,T	GG	Niemann-Pick Disease, Type C1	Bauer (2002)
26721	NPC1	T	G	TT	Niemann-Pick Disease, Type C1	Sun (2001)
26722	NPC1	C	A,T	CC	Niemann-Pick Disease, Type C1	Millat (2005)
26723	NPC1	G	A,T	GG	Niemann-Pick Disease, Type C1	Park (2003)
26724	NPC1	A	C	AA	Niemann-Pick Disease, Type C1	Millat (2001)
26725	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Park (2003)
26726	NPC1	A	T	AA	Niemann-Pick Disease, Type C1	Fernandez-Valero (2005)
26727	NPC1	G	C	GG	Niemann-Pick Disease, Type C1	Sun (2001)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
26728	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Park (2003)
26729	NPC1	C	G,T	CC	Niemann-Pick Disease, Type C1	Park (2003)
26730	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Tamura (2006)
26731	NPC1	G	C	GG	Niemann-Pick Disease, Type C1	Bauer (2002)
26732	NPC1	C	A	CC	Niemann-Pick Disease, Type C1	Park (2003)
26733	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Park (2003)
26734	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Park (2003)
26735	NPC1	G	C	GG	Niemann-Pick Disease, Type C1	Park (2003)
26736	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Park (2003)
26737	NPC1	A	G	AA	Niemann-Pick Disease, Type C1	Yamamoto (2000)
26738	NPC1	A	G	AA	Niemann-Pick Disease, Type C1	Millat (2001)
26739	NPC1	C	A	CC	Niemann-Pick Disease, Type C1	Fernandez-Valero (2005)
26740	NPC1	C	A	CC	Niemann-Pick Disease, Type C1	Park (2003)
26741	NPC1	G	A,T	GG	Niemann-Pick Disease, Type C1	Park (2003)
26742	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Park (2003)
26743	NPC1	G	T	GG	Niemann-Pick Disease, Type C1	Park (2003)
26744	NPC1	A	T	AA	Niemann-Pick Disease, Type C1	Fernandez-Valero (2005)
26745	NPC1	A	G	AA	Niemann-Pick Disease, Type C1	Millat (2005)
26746	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Park (2003)
26747	NPC1	T	G	TT	Niemann-Pick Disease, Type C1	Millat (2001)
26748	NPC1	G	C	GG	Niemann-Pick Disease, Type C1	Park (2003)
26749	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Sun (2001)
26750	NPC1	G	C	GG	Niemann-Pick Disease, Type C1	Yamamoto (2000)
26751	NPC1	T	C	TT	Niemann-Pick Disease, Type C1	Millat (2001)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
26752	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Park (2003)
26753	NPC1	C	A	CC	Niemann-Pick Disease, Type C1	Bauer (2002)
26754	NPC1	T	A	TT	Niemann-Pick Disease, Type C1	Millat (2005)
26755	NPC1	G	A,T	GG	Niemann-Pick Disease, Type C1	Fernandez-Valero (2005)
26756	NPC1	T	C	TT	Niemann-Pick Disease, Type C1	Millat (2005)
26757	NPC1	T	A	TT	Niemann-Pick Disease, Type C1	Millat (2001)
26758	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Sun (2001)
26760	NPC1	T	C	TT	Niemann-Pick Disease, Type C1	Tarugi (2002)
26761	NPC1	G	C	GG	Niemann-Pick Disease, Type C1	Sun (2001)
26762	NPC1	A	C	AA	Niemann-Pick Disease, Type C1	Tarugi (2002)
26763	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Tarugi (2002)
26764	NPC1	C	A	CC	Niemann-Pick Disease, Type C1	Millat (2005)
26765	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Sun (2001)
26766	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Tarugi (2002)
26767	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Fernandez-Valero (2005)
26768	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Meiner (2001)
26769	NPC1	T	G	TT	Niemann-Pick Disease, Type C1	Carstea (1997)
26770	NPC1	A	G	AA	Niemann-Pick Disease, Type C1	Sun (2001)
26772	NPC1	G	T	GG	Niemann-Pick Disease, Type C1	Millat (2001)
26774	NPC1	C	A	CC	Niemann-Pick Disease, Type C1	Ribeiro (2001)
26775	NPC1	G	C	GG	Niemann-Pick Disease, Type C1	Millat (2001)
26776	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Millat (2001)
26777	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Park (2003)
26779	NPC1	C	G	CC	Niemann-Pick Disease, Type C1	Fernandez-Valero (2005)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
26780	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Millat (2001)
26782	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Yamamoto (1999)
26784	NPC1	C	A	CC	Niemann-Pick Disease, Type C1	Bauer (2002)
26785	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Sun (2001)
26786	NPC1	A	T	AA	Niemann-Pick Disease, Type C1	Fernandez-Valero (2005)
26787	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Park (2003)
26788	NPC1	T	C	TT	Niemann-Pick Disease, Type C1	Yang (2005)
26789	NPC1	A	G	AA	Niemann-Pick Disease, Type C1	Sun (2001)
26790	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Ribeiro (2001)
26791	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Millat (2001)
26792	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Millat (2005)
26793	NPC1	C	G	CC	Niemann-Pick Disease, Type C1	Sevin (2007)
26794	NPC1	C	A,G,T	CC	Niemann-Pick Disease, Type C1	Millat (2001)
26795	NPC1	C	G	CC	Niemann-Pick Disease, Type C1	Millat (2001)
26797	NPC1	A	C	AA	Niemann-Pick Disease, Type C1	Yamamoto (2000)
26798	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Sun (2001)
26800	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Meiner (2001)
26801	NPC1	C	A	CC	Niemann-Pick Disease, Type C1	Yang (2005)
26802	NPC1	T	C	TT	Niemann-Pick Disease, Type C1	Park (2003)
26803	NPC1	A	C	AA	Niemann-Pick Disease, Type C1	Sun (2001)
26804	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Yang (2005)
26805	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Ribeiro (2001)
26807	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Carstea (1997)
26808	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Millat (2001)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
26809	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Fernandez-Valero (2005)
26810	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Park (2003)
26811	NPC1	A	G	AA	Niemann-Pick Disease, Type C1	Millat (1999)
26812	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Millat (2005)
26813	NPC1	A	C	AA	Niemann-Pick Disease, Type C1	Yamamoto (2004)
26814	NPC1	G	T	GG	Niemann-Pick Disease, Type C1	Fernandez-Valero (2005)
26815	NPC1	A	G	AA	Niemann-Pick Disease, Type C1	Park (2003)
26817	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Sun (2001)
26818	NPC1	A	G	AA	Niemann-Pick Disease, Type C1	Kaminski (2002)
26819	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Millat (2005)
26820	NPC1	T	A	TT	Niemann-Pick Disease, Type C1	Park (2003)
26821	NPC1	C	A	CC	Niemann-Pick Disease, Type C1	Park (2003)
26822	NPC1	A	G	AA	Niemann-Pick Disease, Type C1	Millat (2001)
26823	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Millat (2001)
26824	NPC1	G	A,T	GG	Niemann-Pick Disease, Type C1	Sun (2001)
26825	NPC1	A	T	AA	Niemann-Pick Disease, Type C1	Millat (2005)
26826	NPC1	T	A,C	TT	Niemann-Pick Disease, Type C1	Carstea (1997)
26827	NPC1	T	A	TT	Niemann-Pick Disease, Type C1	Fernandez-Valero (2005)
26828	NPC1	C	A,T	CC	Niemann-Pick Disease, Type C1	Sun (2001)
26829	NPC1	A	G	AA	Niemann-Pick Disease, Type C1	Carstea (1997)
26830	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Millat (2001)
26831	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Millat (2005)
26832	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Carstea (1997)
26833	NPC1	T	C	TT	Niemann-Pick Disease, Type C1	Sun (2001)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
26834	NPC1	G	C,T	GG	Niemann-Pick Disease, Type C1	Park (2003)
26835	NPC1	G	C	GG	Niemann-Pick Disease, Type C1	Yamamoto (2000)
26836	NPC1	C	A	CC	Niemann-Pick Disease, Type C1	Yang (2005)
26837	NPC1	A	C	AA	Niemann-Pick Disease, Type C1	Greer (1999)
26839	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Millat (2005)
26840	NPC1	G	C	GG	Niemann-Pick Disease, Type C1	Fernandez-Valero (2005)
26841	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Yamamoto (2000)
26842	NPC1	C	G	CC	Niemann-Pick Disease, Type C1	Millat (2005)
26843	NPC1	T	C	TT	Niemann-Pick Disease, Type C1	Park (2003)
26844	NPC1	C	A	CC	Niemann-Pick Disease, Type C1	Millat (2005)
26845	NPC1	A	C,T	AA	Niemann-Pick Disease, Type C1	Park (2003)
26846	NPC1	T	C	TT	Niemann-Pick Disease, Type C1	Park (2003)
26847	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Park (2003)
26849	NPC1	G	A	GG	Niemann-Pick Disease, Type C1	Park (2003)
26850	NPC1	C	G	CC	Niemann-Pick Disease, Type C1	Park (2003)
26851	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Millat (2005)
26852	NPC1	A	C	AA	Niemann-Pick Disease, Type C1	Tarugi (2002)
26853	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Ribeiro (2001)
26854	NPC1	C	G	CC	Niemann-Pick Disease, Type C1	Sun (2001)
26855	NPC1	C	G	CC	Niemann-Pick Disease, Type C1	Millat (2001)
26856	NPC1	T	C	TT	Niemann-Pick Disease, Type C1	Di Leo (2004)
26857	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Fernandez-Valero (2005)
26858	NPC1	C	T	CC	Niemann-Pick Disease, Type C1	Yamamoto (1999)
26859	NPC1	C	G	CC	Niemann-Pick Disease, Type C1	Sun (2001)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
26806	NPC1	G	T	GG	Niemann-Pick Disease, Type C	Fernandez-Valero (2005)
26605	SMPD1	C	T	CC	Niemann-Pick Disease, Type B	Simonaro (2002)
26574	SMPD1	G	A	GG	Niemann-Pick Disease, Type B	Takahashi (1992)
26600	SMPD1	A	G	AA	Niemann-Pick Disease, Type B	Takahashi (1992)
26609	SMPD1	C	A	CC	Niemann-Pick Disease, Type B	Takahashi (1992)
26610	SMPD1	C	T	CC	Niemann-Pick Disease, Type B	Lee (2003)
26624	SMPD1	G	T	GG	Niemann-Pick Disease, Type A	Levran (1990, 1991)
26582	SMPD1	T	A	TT	Niemann-Pick Disease, Type A	Takahashi (1992)
26599	SMPD1	G	A	GG	Niemann-Pick Disease, Type A	Takahashi (1992)
26634	SMPD1	G	A	GG	Niemann-Pick Disease, Type A	Ferlinz (1991)
26602	SMPD1	T	G	TT	Niemann-Pick Disease, Intermediate Form, with Macular Halo	Sperl (1994)
26588	SMPD1	T	C	TT	Niemann-Pick Disease	Levran (1992)
100908	SMPD1	I	D	II	Niemann-Pick Disease	Levran (1991), Vanier (1993), Fernandez-Burriel (2003), Rodriguez-Pascau (2009)
100909	SMPD1	I	D	II	Niemann-Pick Disease	Levran (1993)
26553	SMPD1	T	G	TT	Niemann-Pick Disease	Pittis (2004)
26554	SMPD1	C	T	CC	Niemann-Pick Disease	Simonaro (2002)
26555	SMPD1	G	A	GG	Niemann-Pick Disease	Pittis (2004)
26556	SMPD1	A	T	AA	Niemann-Pick Disease	Simonaro (2002)
26557	SMPD1	C	G	CC	Niemann-Pick Disease	Simonaro (2002)
26558	SMPD1	T	C	TT	Niemann-Pick Disease	Ricci (2004)
26559	SMPD1	T	C	TT	Niemann-Pick Disease	Dardis (2005)
26560	SMPD1	T	C	TT	Niemann-Pick Disease	Simonaro (2002)
26561	SMPD1	T	C	TT	Niemann-Pick Disease	Simonaro (2002)
26562	SMPD1	G	A	GG	Niemann-Pick Disease	Pavlu-Pereira (2005)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
26563	SMPD1	G	A	GG	Niemann-Pick Disease	Simonaro (2002)
26564	SMPD1	T	A	TT	Niemann-Pick Disease	Mussig (2006)
26565	SMPD1	C	T	CC	Niemann-Pick Disease	Pavlu-Pereira (2005)
26566	SMPD1	G	C	GG	Niemann-Pick Disease	Simonaro (2002)
26567	SMPD1	C	T	CC	Niemann-Pick Disease	Simonaro (2002)
26568	SMPD1	C	A	CC	Niemann-Pick Disease	Simonaro (2002)
26569	SMPD1	T	C	TT	Niemann-Pick Disease	Pittis (2004)
26570	SMPD1	C	T	CC	Niemann-Pick Disease	Simonaro (2002)
26571	SMPD1	G	A	GG	Niemann-Pick Disease	Pavlu-Pereira (2005)
26572	SMPD1	G	A	GG	Niemann-Pick Disease	Simonaro (2002)
26573	SMPD1	C	T	CC	Niemann-Pick Disease	Pavlu-Pereira (2005)
26575	SMPD1	G	C	GG	Niemann-Pick Disease	Pittis (2004)
26576	SMPD1	G	A	GG	Niemann-Pick Disease	Simonaro (2002)
26577	SMPD1	G	A	GG	Niemann-Pick Disease	Ricci (2004)
26578	SMPD1	C	A	CC	Niemann-Pick Disease	Simonaro (2002)
26579	SMPD1	A	C	AA	Niemann-Pick Disease	Sikora (2003)
26580	SMPD1	C	A	CC	Niemann-Pick Disease	Pavlu-Pereira (2005)
26581	SMPD1	G	T	GG	Niemann-Pick Disease	Ida (1996)
26583	SMPD1	A	C	AA	Niemann-Pick Disease	Pavlu-Pereira (2005)
26584	SMPD1	G	A	GG	Niemann-Pick Disease	Pittis (2004)
26585	SMPD1	G	A	GG	Niemann-Pick Disease	Simonaro (2002)
26586	SMPD1	C	A	CC	Niemann-Pick Disease	Pavlu (1997)
26589	SMPD1	T	C	TT	Niemann-Pick Disease	Ricci (2004)
26590	SMPD1	C	T	CC	Niemann-Pick Disease	Sikora (2003)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
26591	SMPD1	C	G	CC	Niemann-Pick Disease	Simonaro (2002)
26592	SMPD1	C	G	CC	Niemann-Pick Disease	Simonaro (2002)
26593	SMPD1	T	C	TT	Niemann-Pick Disease	Pavlu (1997)
26594	SMPD1	C	A	CC	Niemann-Pick Disease	Simonaro (2002)
26595	SMPD1	C	T	CC	Niemann-Pick Disease	Sikora (2003)
26596	SMPD1	G	A	GG	Niemann-Pick Disease	Simonaro (2002)
26597	SMPD1	G	T	GG	Niemann-Pick Disease	Simonaro (2002)
26598	SMPD1	T	C	TT	Niemann-Pick Disease	Simonaro (2002)
26601	SMPD1	A	C	AA	Niemann-Pick Disease	Schuchman (1995)
26603	SMPD1	C	T	CC	Niemann-Pick Disease	Simonaro (2002)
26604	SMPD1	C	T	CC	Niemann-Pick Disease	Ricci (2004)
26606	SMPD1	T	C	TT	Niemann-Pick Disease	Simonaro (2002)
26607	SMPD1	T	C	TT	Niemann-Pick Disease	Simonaro (2002)
26608	SMPD1	G	C	GG	Niemann-Pick Disease	Simonaro (2002)
26611	SMPD1	A	G	AA	Niemann-Pick Disease	Takahashi (1995)
26612	SMPD1	T	C	TT	Niemann-Pick Disease	Ricci (2004)
26613	SMPD1	C	T	CC	Niemann-Pick Disease	Simonaro (2002)
26614	SMPD1	G	A	GG	Niemann-Pick Disease	Simonaro (2002)
26615	SMPD1	T	C	TT	Niemann-Pick Disease	Sikora (2003)
26616	SMPD1	C	T	CC	Niemann-Pick Disease	Simonaro (2002)
26617	SMPD1	C	T	CC	Niemann-Pick Disease	Simonaro (2002)
26618	SMPD1	C	A	CC	Niemann-Pick Disease	Simonaro (2002)
26620	SMPD1	T	A	TT	Niemann-Pick Disease	Simonaro (2002)
26621	SMPD1	G	A	GG	Niemann-Pick Disease	Simonaro (2002)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
26622	SMPD1	C	T	CC	Niemann-Pick Disease	Simonaro (2002)
26623	SMPD1	G	A	GG	Niemann-Pick Disease	Ricci (2004)
26625	SMPD1	T	G	TT	Niemann-Pick Disease	Simonaro (2002)
26626	SMPD1	A	T	AA	Niemann-Pick Disease	Simonaro (2002)
26627	SMPD1	A	G	AA	Niemann-Pick Disease	Ricci (2004)
26628	SMPD1	T	C	TT	Niemann-Pick Disease	Simonaro (2002)
26629	SMPD1	T	C	TT	Niemann-Pick Disease	Sikora (2003)
26630	SMPD1	T	C	TT	Niemann-Pick Disease	Simonaro (2002)
26631	SMPD1	G	T	GG	Niemann-Pick Disease	Dardis (2005)
26632	SMPD1	G	A	GG	Niemann-Pick Disease	Ricci (2004)
26633	SMPD1	G	C	GG	Niemann-Pick Disease	Simonaro (2002)
26635	SMPD1	G	A	GG	Niemann-Pick Disease	Simonaro (2002)
26636	SMPD1	G	C	GG	Niemann-Pick Disease	Simonaro (2002)
26637	SMPD1	G	C	GG	Niemann-Pick Disease	Levran (1993)
31022	HAX1	C	T	CC	Neutropenia, Severe Congenital, Autosomal Recessive 3	Klein (2007)
31023	HAX1	C	T	CC	Neutropenia, Severe Congenital, Autosomal Recessive 3	Kostmann (1956), Horwitz (1999), Klein (2007)
19018	ELA2	G	A	GG	Neutropenia, Severe Congenital	Dale (2000)
23576	GFI1	T	C	TT	Neutropenia, Nonimmune Chronic Idiopathic, of Adults	Papadaki , (2002), Person (2003)
19077	ELA2	G	A	GG	Neutropenia, Cyclic	Horwitz (1999)
25652	TMPRSS3	G	A	GG	Neurosensory deafness	Ben-Yosef (2001)
25654	TMPRSS3	C	A	CC	Neurosensory deafness	Ben-Yosef (2001)
25658	TMPRSS3	A	G	AA	Neurosensory deafness	Ben-Yosef (2001)
43422	HSN2	C	T	CC	Neuropathy, Hereditary Sensory, Type II	Cho (2006)
43424	HSN2	C	T	CC	Neuropathy, Hereditary Sensory, Type II	Roddier (2005)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
43425	HSN2	C	T	CC	Neuropathy, Hereditary Sensory, Type II	Lafreniere (2004), Roddier (2005)
40265	SPTLC1	A	C	AA	Neuropathy, Hereditary Sensory, Type I	England before (1800), Hicks (1922), Hoffman (1964), Dawkins (2001), Bejaoui (2001), Nicholson (2001)
40266	SPTLC1	A	T	AA	Neuropathy, Hereditary Sensory, Type I	Dawkins (2001)
44441	NGFB	G	A	GG	Neuropathy, Hereditary Sensory and Autonomic, Type V|Loss of Pain & Temperature Perception	Einarsdottir (2004)
40363	HSPB1	C	T	CC	Neuropathy, Distal Hereditary Motor, Type IIB|Axonal Charcot-Marie-Tooth Disease, Type 2F	Evgrafov (2004), Tang (2005)
41386	CLN8	C	T	CC	Neuronal ceroid lipofuscinosis, late infantile	Mitchell (2001), Topcu (2004), Ranta (2004), Topcu (2004), Ranta (2004)
42165	CLN5	G	A	GG	Neuronal Ceroid Lipofuscinosis, Late Infantile	Savukoski (1998)
71921	CLN3	I	D	II	Neuronal Ceroid Lipofuscinosis, Juvenile	The International Batten Disease Consortium (1995)
41382	CLN8	G	C	GG	Neuronal Ceroid Lipofuscinosis	Topcu (2004), Ranta (2004), Topcu (2004), Ranta (2004)
30498	CLN6	C	A,G,T	CC	Neuronal Ceroid Lipofuscinoses 6	Gao (2002), Wheeler (2002)
55805	RTN4R	G	A,C	GG	Neuroleptic Resistant Schizophrenia	Sinibaldi (2004)
55806	RTN4R	C	T	CC	Neuroleptic Resistant Schizophrenia	Sinibaldi (2004)
50731	NF2	T	C	TT	Neurofibromatosis, Type II	Bourn (1994), Bourn (1994), Scoles (1996), Fernandez-Valle (2002)
50773	NF2	T	C	TT	Neurofibromatosis, Type II	Myers (1985), Rouleau (1993)
79192	SPRED1	A	T	AA	Neurofibromatosis 1–like Phenotype	Brems (2007)
79187	SPRED1	C	T	CC	Neurofibromatosis 1–like Phenotype	Brems (2007)
79188	SPRED1	C	T	CC	Neurofibromatosis 1–like Phenotype	Brems (2007)
79190	SPRED1	G	A	GG	Neurofibromatosis 1–like Phenotype	Brems (2007)
79194	SPRED1	G	A	GG	Neurofibromatosis 1–like Phenotype	Brems (2007)
36779	NF1	C	T	CC	Neurofibromatosis 1	Cawthon , (1990), Marchuk (1991), Estivill , (1991), Ainsworth , (1993), Horiuchi (1994), Horiuchi (1994), Heim (1994), Lazaro (1995), Park (2000)
51013	NME1	A	G	AA	Neuroblastoma, Somatic	Chang (1994)
47958	PHOX2B	C	A	CC	Neuroblastoma	Trochet (2004)
78048	VANGL1	G	A	GG	Neural Tube Defects|Caudal Regression Syndrome	Kibar (2007)
78049	VANGL1	G	A	GG	Neural Tube Defects	Kibar (2007)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
78050	VANGL1	T	C	TT	Neural Tube Defects	Kibar (2007)
72139	SPINK5	D	I	DD	Netherton Syndrome	Chavanas (2000)
66242	NPHS2	C	T	CC	Nephrotic Syndrome, Steroid Resistant	Fuchshuber (1995), Boute (2000)
63204	NPHP4	A	G	AA	Nephronophthisis 4	Mollet (2002)
66787	NPHP1	A	G	AA	Nephronophthisis 1	Maire (2005)
78400	GLIS2	G	T	GG	Nephronophthisis	Attanasio (2007)
66753	CLCN5	G	C	G	Nephrolithiasis, hypercalciuric	Lloyd (1997)
72591	INS	G	T	GG	Neonatal Diabetes, Permanent	Stoy (2007)
72592	INS	C	T	CC	Neonatal Diabetes, Permanent	Stoy (2007)
101044	INS	G	C	GG	Neonatal Diabetes	Garin (2010)
36119	TPM2	C	T	CC	Nemaline Myopathy	Donner (2002), Donner (2002)
36120	TPM2	T	G	TT	Nemaline Myopathy	Donner (2002), Donner (2002)
100814	JUP	I	D	II	Naxos Disease	McKoy (2000)
41826	HCRT	A	C	AA	Narcolepsy, Early-onset	Peyron (2000)
62495	LMX1B	G	A	GG	Nail-Patella Syndrome	McIntosh (1998)
55151	RYR1	C	T	CC	Myotonic Dystrophy, Type 1	Gambelli (2007)
44175	SCN4A	C	G	CC	Myotonia Fluctuans	Ricker (1994)
38841	TTID	C	G	CC	Myotilinopathy	Engel (2004)
70728	MTND1	G	A	?	Leber Hereditary Optic Neuropathy, Severe	Howell , (1991), Huoponen , (1991), Majander , (1991), Brown , (1992), Howell , (1992), Johns, (1992), Johns , (1992), Paulus , (1993), Wong (2002)
70734	MTND1	G	A	?	Leber Hereditary Optic Neuropathy	Valentino (2004), Valentino (2004)
2274	LMNA	G	T	GG	Myopathy, early-onset and progeria	D'Amico (2005)
40604	GNE	C	G	CC	Myopathy, distal, with rimmed vacuoles	Saito (2004), Kim (2006)
40641	GNE	C	G	CC	Myopathy, distal, with rimmed vacuoles	Kayashima (2002), Tomimitsu (2002), Arai (2002), Tomimitsu (2004), Kim (2006)
39171	DES	T	C	TT	Myopathy, Desmin related	Horowitz (1994), Sjoberg (1999)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
70733	MTND1	A	G	?	Dystonia, Adult-onset	Herrnstadt (2002), Simon (2003), Simon (2003)
1547	LDB3	C	T	CC	Myofibrillar Myopathy, ZASP related	Engel (2005)
1548	LDB3	C	T	CC	Myofibrillar Myopathy, ZASP related	Engel (2005)
1546	LDB3	G	A	GG	Myofibrillar Myopathy, ZASP related	Engel (2005)
43221	SGCE	G	A,T	GG	Myoclonus-Dystonia Syndrome	Zimprich (2001), Valente (2005)
43223	SGCE	G	A	GG	Myoclonus-Dystonia Syndrome	Zimprich (2001), Han (2003)
43225	SGCE	A	C	AA	Myoclonus-Dystonia Syndrome	Leung (2001), Klein (2002), Doheny (2002), Doheny (2002), Rajput, (2002)
43229	SGCE	G	A	GG	Myoclonus-Dystonia Syndrome	Valente (2005)
42567	JRK	G	A	GG	Myoclonic Epilepsy, Juvenile	Moore (2001)
38813	ARX	G	A	G	Myoclonic Epilepsy with Mental Retardation and Spasticity	Scheffer (2002), Stromme (2002)
42070	NHLRC1	A	T	AA	Myoclonic Epilepsy of Lafora	Chan (2003)
39669	MPO	T	G	TT	Myeloperoxidase Deficiency	Marchetti (2004), Laboratory for Molecular Medicine of Partners HealthCare Personalized Medicine (2014), GeneDx (2017)
39675	MPO	G	A	GG	Myeloperoxidase Deficiency	Nauseef (1994), Kizaki (1994), Nauseef (1996), GeneDx (2016)
44183	SCN4A	A	T	AA	Myasthenic Syndrome	Tsujino (2003), Tsujino (2003), Tsujino (2003)
100885	LAMA2	I	D	II	Muscular Dystrophy, Merosin Deficient	Oliveira (2008)
63280	POMT1	G	C	GG	Muscular Dystrophy, Limb Girdle, Type 2K	Dincer (2003), Balci (2005)
40567	CAV3	G	A	GG	Muscular Dystrophy, Limb Girdle, Type 1C|Rippling Muscle Disease 2|Elevated Serum Creatine Phosphokinase |Distal Myopathy with Decreased Caveolin 3	Vorgerd (1999), Carbone (2000), Carbone (2000), Betz (2001), Vorgerd (2001), Vorgerd (2001), Tateyama (2002), Tateyama (2002), Figarella-Branger (2003), Figarella-Branger (2003)
40573	CAV3	G	A	GG	Muscular Dystrophy, Limb Girdle, Type 1C|Rippling Muscle Disease 2|Elevated Serum Creatine Phosphokinase	Torbergsen (1975), Vorgerd (1999), Herrmann (2000), Betz (2001), Fulizio (2005)
2252	LMNA	G	A	GG	Muscular Dystrophy, Limb Girdle, Type 1B	Muchir (2000), Charniot (2003), Sebillon (2003)
38840	TTID	C	T	CC	Muscular Dystrophy, Limb Girdle, Type 1A|Myotilinopathy	Hauser (2002), Engel (2004)
2275	LMNA	T	C	TT	Muscular dystrophy, limb girdle	Pegoraro (2005)
2221	LMNA	A	C	AA	Muscular dystrophy, Emery-Dreifuss	Bonne (2000)
2193	LMNA	C	A	CC	Muscular Dystrophy, Emery-Dreifuss	Brown (2001)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
36236	EMD	C	A	C	Muscular Dystrophy, Emery-Dreifuss	Ellis (1999), Yates (1999)
2226	LMNA	G	A	GG	Muscular dystrophy, Emery-Dreifuss	di Barletta (2000)
1182	DMD	G	A,C	G	Muscular dystrophy, Duchenne	Lenk (1994), Barbieri (1995), Muntoni (1995)
1292	DMD	G	A,C	G	Muscular dystrophy, Duchenne	Deburgrave (2007), Hwa (2008)
1303	DMD	G	A	G	Muscular dystrophy, Duchenne	Buzin (2005)
1332	DMD	G	A	G	Muscular dystrophy, Duchenne	Roberts (1992), Hwa (2008)
1434	DMD	C	A,T	C	Muscular dystrophy, Duchenne	Hodgson (1992), Emery (1993), Lenk (1993), Tuffery (1995)
1358	DMD	T	A,G	T	Muscular dystrophy, Becker	Laing (1992), Sharp (1992)
1417	DMD	T	C	T	Muscular dystrophy, Becker	Tuffery-Giraud (2003)
41032	POMGNT1	C	T	CC	Muscle-eye-brain Disease|Walker-Warburg Syndrome, Mild	Yoshida (2001), Taniguchi (2003)
41004	POMGNT1	G	A	GG	Muscle-eye-brain Disease	Vervoort (2004)
41016	POMGNT1	G	C	GG	Muscle-eye-brain Disease	Yoshida (2001)
41023	POMGNT1	C	A,G,T	CC	Muscle-eye-brain Disease	Vervoort (2004), Biancheri (2006)
41029	POMGNT1	C	G,T	CC	Muscle-eye-brain Disease	Lehesjoki (2004)
800896	FGFR3	A	G	AA	Multiple Myeloma, Somatic	Tavormina (1995), Chesi (1997), Wilcox (1998), Liboi (2003)
59479	MATN3	G	A	GG	Multiple Epiphyseal Dysplasia	Chapman (2001), Jackson (2004)
78828	CDKN1B	G	A	GG	Multiple Endocrine Neoplasia, Type IV	Pellegata (2006)
800880	RET	G	A	GG	Multiple Endocrine Neoplasia, Type 2A, with Hirschsprung Disease	Blaugrund (1994), Decker (1998)
800882	RET	G	T	GG	Multiple Endocrine Neoplasia, Type 2A	Berndt (1998)
800881	RET	T	C	TT	Multiple Endocrine Neoplasia, Type 2A	Takahashi (1988), Mendonca (1988), Mulligan (1994), Mulligan (1994), Gardner (1994), Hofstra (1996), Tessitore (1999), Nunes (2002), Neumann (2002), Benazzouz (2008)
800883	RET	T	G	TT	Multiple Endocrine Neoplasia, Type 2A	Hofstra (1997), Hofstra , (1997), Dang , (1999), Jimenez (2004)
47904	MEN1	G	A,T	GG	Multiple Endocrine Neoplasia, Type 1, Burin Variant|Prolactinoma, Hyperparathyroid, Carcinoid Syndrome	Farid (1980), Bear (1985), Agarwal (1997), Olufemi (1998)
47934	MEN1	C	T	CC	Multiple Endocrine Neoplasia, Type 1	Gortz (1999), Turner (2002)
70858	MEN1	I	D	II	Multiple Endocrine Neoplasia, Type 1	Teh (1998), Ebeling (2004)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
70860	MEN1	I	D	II	Multiple Endocrine Neoplasia, Type 1	Bassett (1998), Turner (2002), Turner (2002)
70861	MEN1	I	D	II	Multiple Endocrine Neoplasia, Type 1	Turner (2002)
70862	MEN1	I	D	II	Multiple Endocrine Neoplasia, Type 1	Turner (2002)
47889	MEN1	C	A	CC	Multiple endocrine neoplasia 1	Bassett (1998), Turner (2002)
49999	FH	T	C	TT	Multiple Cutaneous and Uterine Leiomyomata|Leiomyomatosis and Renal Cell Cancer	Tomlinson (2002), Wei (2006)
49996	FH	C	A,T	CC	Multiple Cutaneous and Uterine Leiomyomata|Leiomyomatosis and Renal Cell Cancer	Tomlinson (2002), Toro (2003), Toro (2003), Wei (2006)
50000	FH	T	G	TT	Multiple Cutaneous and Uterine Leiomyomata	Tomlinson (2002)
30088	HLCS	G	A	GG	Multiple Carboxylase Deficiency, Biotin-responsive	Yang (2001)
30092	HLCS	C	T	CC	Multiple Carboxylase Deficiency, Biotin-responsive	Yang (2001), Aoki (1997)
30102	HLCS	C	T	CC	Multiple Carboxylase Deficiency, Biotin-responsive	Yang (2001)
46681	PTEN	G	A	GG	Multiple cancers	Vivo (2000)
46701	PTEN	G	A	GG	Multiple cancers	Vivo (2000)
55183	RYR1	A	G	AA	Multi-minicore Disease	Monnier (2003)
28958	IDS	C	A,G,T	C	Mucopolysaccharidosi, Type II	Whitley (1993), Sukegawa (1997)
27427	MCOLN1	A	G	AA	Mucolipidosis IV	Slaugenhaupt (1999), Bargal (2000), Bassi (2000), Edelmann (2002), Bach (2005)
71899	GNPTG	D	I	DD	Mucolipidosis III, Gamma	Raas-Rothschild (2000)
100714	GNPTAB	I	D	II	Mucolipidosis III, Alpha/Beta	Kudo (2006), Plante (2008),GeneDx (2015), Emory Genetics (2015), Illumina (2016)
100716	GNPTAB	A	G	AA	Mucolipidosis III, Alpha/Beta	Otomo (2009)
27387	GNPTAB	G	A	GG	Mucolipidosis II, Alpha/Beta	Paik (2005), Otomo (2009)
27391	GNPTAB	C	T	CC	Mucolipidosis II, Alpha/Beta	Paik (2005)
78483	BRWD3	C	A	C	MRX93	Field (2007)
78484	BRWD3	T	C,G	T	MRX93	Field (2007)
28143	ARSB	A	C	AA	MPS 6	Petry (2005), Garrido (2007)
27945	GALNS	C	T	CC	Morquio Syndrome A	Montano (2003)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
27962	GALNS	T	A	TT	Morquio Syndrome A	Tomatsu (1995)
27978	GALNS	G	A	GG	Morquio Syndrome A	Kato (1997)
28006	GALNS	C	A	CC	Morquio Syndrome A	Kato (1997)
28010	GALNS	G	C	GG	Morquio Syndrome A	Yamada (1998)
28030	GALNS	G	A	GG	Morquio Syndrome A	Tomatsu (1992), Tomatsu (2004)
9535	KRTHB6	G	A	GG	Monilethrix	Winter (1997), Korge (1997), Bowden (1998), Winter (1998), Korge (1998), Winter (1999), Khandpur (2004)
64047	ECGF1	C	T	CC	MNGIE Syndrome	Gamez (2002)
38634	DYSF	C	T	CC	Miyoshi Myopathy|Limb Girdle Muscular Dystrophy, Type 2B|Distal Myopathy with Anterior Tibial Onset	Vilchez (2005)
38573	DYSF	C	G	CC	Miyoshi Myopathy|Limb Girdle Muscular Dystrophy, Type 2B	Weiler (1996), Weiler (1999)
38526	DYSF	G	A	GG	Miyoshi Myopathy	Aoki (2001), Takahashi (2003)
50229	XRCC3	G	A	GG	Mitomycin-C resistance	Rafii (2003)
19458	PUS1	C	T	CC	Mitochondrial Myopathy and Sideroblastic Anemia	Bykhovskaya (2004)
32169	HMGCS2	C	T	CC	Mitochondrial HMG-CoA Synthase Deficiency	Aledo (2001)
32171	HMGCS2	C	T	CC	Mitochondrial HMG-CoA Synthase Deficiency	Aledo (2001)
41882	DGUOK	G	T	GG	Mitochondrial DNA Depletion Syndrome, Hepatocerebral Form|Cystathioninuria	Tadiboyina (2005)
41869	DGUOK	G	A	GG	Mitochondrial DNA Depletion Syndrome, Hepatocerebral Form	Salviati (2002)
61602	NDUFV1	C	T	CC	Mitochondrial Complex 1 Deficiency	Schuelke (1999)
8626	UROS	G	T	GG	Mild, Cutaneous-only Congenital Erythropoietic Porphyria	Xu (1995), Solis (2001)
38663	PLP1	G	A	G	Mild Pelizaeus-Merzbacher Disease	Sistermans (1996), Sistermans (1996)
40003	ATP1A2	G	A	GG	Migraine	Todt (2005)
14381	LHCGR	G	T	GG	Micropenis	Latronico (1996)
60254	SRD5A2	C	T	CC	Micropenis	Makridakis (2000), Sasaki (2003)
66895	COL4A3	C	T	CC	Microhaematuria and protinuria	Longo (2002)
33070	MUT	G	A	GG	Methylmalonic Aciduria, mut(0) Type	Worgan (2006)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
33073	MUT	C	A	CC	Methylmalonic Aciduria, mut(0) Type	Ogasawara (1994)
33091	MUT	C	A,T	CC	Methylmalonic Aciduria, mut(0) Type	Cavicchi (2006)
33094	MUT	T	A	TT	Methylmalonic Aciduria, mut(0) Type	Acquaviva (2001), Acquaviva (2005), Berger (2001)
33179	MUT	C	A	CC	Methylmalonic Aciduria, mut(0) Type	Ledley (1990), Crane (1992), Worgan (2006)
32587	MMAB	G	A	GG	Methylmalonic Aciduria, cblB type	Dobson (2002)
33266	MMACHC	C	T	CC	Methylmalonic Aciduria and Homocystinuria, cblC Type	Lerner-Ellis (2006)
33262	MMACHC	T	C	TT	Methylmalonic Aciduria and Homocystinuria, cblC Type	Lerner-Ellis (2006)
44594	MTRR	C	T	CC	Methionine synthase reductase deficiency	Zavadakova (2005)
73809	PPARA	C	G	CC	Increased risk of Metabolic Syndrome and Hyperapobetalipoproteinemia	Vohl (2000), Tai (2002), Robitaille (2004)
19851	ATRX	G	A	G	Mental Retardation-hypotonic Facies Syndrome	Chudley (1988), Guerrini (2000), Howard (2004), Howard (2004), Abidi (2005), Abidi (2005)
64101	HADH2	G	T	G	Mental Retardation, X-linked, Syndromic 10	Reyniers (1999), Lenski (2007)
63971	RPS6KA3	G	A	G	Mental Retardation, X-linked 19	Merienne (1999)
78614	UPF3B	A	C	A	Mental Retardation, Nonsyndromic	Tarpey (2007)
70531	CC2D2A	G	C	GG	Mental Retardation with Retinitis Pigmentosa	Noor (2008)
56248	ZNF81	G	A	G	Mental Retardation	Hamel (1999), Kleefstra (2004)
38818	ARX	A	G	A	Mental Retardation	Bienvenu (2002)
56329	ZNF41	G	A	G	Mental Retardation	Shoichet (2003)
70556	FACL4	G	T	G	Mental Retardation	Meloni (2002)
70557	FACL4	T	C	T	Mental Retardation	Meloni (2002)
70558	FACL4	G	A	G	Mental Retardation	Longo (2003)
22001	FGG	G	T	GG	Menorrhagia	Brennan (2000)
31096	ATP7A	A	T	A	Menkes Disease	Kaler (1994)
47409	TP53	T	G	TT	Meningioma|Li-Fraumeni Syndrome	Joachim (2001), Capponcelli (2005)
64515	FLNA	G	A	G	Melnick-Needles syndrome	Robertson (2003), Robertson (2006)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
64516	FLNA	C	T	C	Melnick-Needles syndrome	Robertson (2003)
100746	MTTQ	A	G	?	Sensorineural Deafness and Migraine	Finnila (2001)
100726	MTTQ	D	I	?	Myopathy	Dey (2000), Graham (1997)
18744	SLC19A2	C	A,T	CC	Megaloblastic Anemia, Thiamine-Responsive	Labay (1999)
18762	SLC19A2	C	T	CC	Megaloblastic Anemia, Thiamine-Responsive	Scharfe (2000)
20633	CUBN	G	A	GG	Megaloblastic Anemia, Finnish Type	Aminoff (1999), Kristiansen (2000)
56378	MLC1	C	T	CC	Megalencephalic Leukoencephalopathy with Subcortical Cysts	Ben-Zeev (2002), Ben-Zeev (2002)
56389	MLC1	G	A	GG	Megalencephalic Leukoencephalopathy with Subcortical Cysts	Leegwater (2001), Leegwater (2001), Tsujino (2003)
21827	AMN	C	T	CC	Megablastic Anemia, Norwegian	Tanner (2003)
53498	KRT12	A	G	AA	Meesmann Corneal Dystrophy	Corden (2000)
53514	KRT12	C	G	CC	Meesmann Corneal Dystrophy	Irvine (1997), Corden (2000)
33305	ACADM	T	C	TT	ACADM Deficiency	Andresen (2001), Zschocke (2001)
33313	ACADM	C	T	CC	ACADM Deficiency	Andresen (2001), Nielsen (2007)
33343	ACADM	A	G	AA	Medium Chain Acyl CoA Dehydrogenase Deficiency	Matsubara (1990), Yokota (1990), Duran (1986), Kelly (1990), Blakemore (1991), Gregersen (1991), Kolvraa (1991), Curtis (1991), Yokota (1991), Ding (1991), Miller (1992), Opdal (1995), Leung (1992), Matsubara (1991), Matsubara (1992), Yokota (1992), Courtagen Life Sciences (2014), Laboratory for Molecular Medicine of Partners HealthCare Personalized Medicine (2014), Center for Pediatric Genomic Medicine of Children's Mercy Hospital and Clinics (2015), ARUP Laboratories (2015), Knight Diagnostic Laboratories of Oregon Health and Sciences University (2015), Counsyl (2015), GeneDx (2016), Illumina Clinical Services Laboratory (2016), Baylor Miraca Genetics Laboratories (2016)
801983	ACADM	A	C	AA	Medium Chain Acyl CoA Dehydrogenase Deficiency	ARUP Laboratories (2015)
21861	GP1BA	C	T	CC	Mediterranean Macrothrombocytopenia |Bernard-Soulier Syndrome, Type A	Ware (1993), Savoia (2001)
68634	CEP290	I	D	II	Meckel Syndrome, Type 4	Baala (2007), Perrault (2007)
63871	TMEM67	A	C	AA	Meckel Syndrome, Type 3	Smith (2006)
68624	MKS1	C	T	CC	Meckel Syndrome, Type 1	Consugar (2007)
68625	MKS1	I	D	II	Meckel Syndrome, Type 1	Kyttala (2006)
61996	MKKS	G	A	GG	McKusick-Kaufman Syndrome	Stone (2000)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
61998	MKKS	C	A	CC	Likely Harmless Variant (Previously associated with McKusick-Kaufman Syndrome)	Stone (2000), Invitae (2017), Center for Pediatric Genomic Medicine of Children's Mercy Hospital and Clinics (2017), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2018)
61554	GNAS	G	A	GG	McCune-Albright Syndrome	Weinstein (1991), Schwindinger (1992), Happle (1986), Nerlich (1991), Falconer (1942), Wrong (1992), Premawardhana (1992), Cremonini (1992), Tinschert (1999), Malchoff (1994), Candeliere (1995), Coutant (2001), Fragoso (2003), Farfel (1996), Iiri (1997),
30293	MCCC2	G	C	GG	MCC2 Deficiency	Bannwart (1992), Baumgartner (2001), Gibson (1998)
31955	MCCC1	T	G	TT	MCC1 Deficiency	Steen (1999), Baumgartner (2001), Gallardo (2001), Baumgartner (2004)
28686	PYGM	C	T	CC	McArdle Disease	Fernandez-Cadenas (2003)
28689	PYGM	G	A	GG	McArdle Disease	Tsujino (1993), Munsat (1976), Schmidt (1987), Papadimitriou (1990), Bartram (1993), Vorgerd (1998), Martin (2001), Martin (2003)
28700	PYGM	C	T	CC	McArdle Disease	Tsujino (1993), Munsat (1976), Schmidt (1987), Papadimitriou (1990), Martin (2001)
28718	PYGM	T	G	TT	McArdle Disease	Tsujino (1993), Munsat (1976), Schmidt (1987), Papadimitriou (1990)
28736	PYGM	A	G,T	AA	McArdle Disease	Martin (2001), Fernandez (2000)
68363	MYH9	G	A	GG	May-Hegglin Anomaly	Consortium (2000), Kelley (2000)
68364	MYH9	C	T	CC	May-Hegglin Anomaly	Consortium (2000), Kelley (2000)
12045	TCF1	C	T	CC	Maturity-onset Diabetes of the Young, Type III	Bjorkhaug (2003)
70809	KIT	G	A	GG	Mastocytosis, Sporadic, Childhood-onset	Longley (1999)
45962	KIT	A	T	AA	Mast Cell Leukemia	Furitsu (1993), Furitsu (1993)
62603	L1CAM	G	A,C,T	G	MASA syndrome	Jouet (1994)
67734	TGFBR2	T	C	TT	Marfan Syndrome, Type II	Mizuguchi (2004)
33987	COL1A2	G	A	GG	Marfan Syndrome, Atypical	Byers (1981), Byers (1981), Henke (1985), Dalgleish (1986), Phillips (1990)
61054	FBN1	G	A,C	GG	Marfan Syndrome	Liu (1997)
61061	FBN1	G	A	GG	Marfan Syndrome	Stahl-Hallengren (1994), Stevenson (1982)
61079	FBN1	G	A	GG	Marfan Syndrome	Edwards (1994), Ades (2004)
61167	FBN1	C	T	CC	Marfan Syndrome	Collod-Beroud (1999)
61189	FBN1	C	T	CC	Marfan Syndrome	Reinhardt (2000), Ades (2006)
61207	FBN1	C	G	CC	Marfan Syndrome	Dietz (1991)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
61222	FBN1	C	A,T	CC	Marfan Syndrome	Hewett (1994), Dietz (1995), Sood (1996)
61480	FBN1	C	A,T	CC	Marfan Syndrome	Nijbroek (1995), Liu (1996), Collod-Beroud (2003)
27342	DLD	G	T	GG	Maple Syrup Urine Disease, Type III	Shaag (1999)
100575	DLD	D	I	DD	Maple Syrup Urine Disease, Type III	Hong (1996), Elpeleg (1997)
801010	DLD	D	I	DD	Maple Syrup Urine Disease, Type III	Hong (1996), Elpeleg (1997)
27290	BCKDHB	G	C	GG	Maple Syrup Urine Disease, Type IB	Edelmann (2001)
27266	BCKDHA	G	A	GG	Maple Syrup Urine Disease, Type IA	Chuang (1995)
27276	BCKDHA	T	G	TT	Maple Syrup Urine Disease, Type IA	Chuang (1995)
27279	BCKDHA	T	A	TT	Maple Syrup Urine Disease, Type IA	Zhang (1989), Chuang (1994), Matsuda (1990), Dariush (1991), Mitsubuchi (1992), Puffenberger (2003)
27317	DBT	A	C	AA	Maple Syrup Urine Disease, Thiamine-response, Type II	Fisher (1991)
47115	ATM	A	G	AA	Mantle cell lymphoma	Fang (2003)
47139	ATM	A	G	AA	Mantle cell lymphoma	Fang (2003)
32012	MLYCD	C	G	CC	Malonyl-CoA Decarboxylase Deficiency	MacPhee (1993), FitzPatrick (1999)
55057	RYR1	A	G	AA	Variant of Unknown Significance but Likely Harmless (previously associated with Malignant Hyperthermia)	Marchant (2004), Biesecker Lab/Human Development Section of the National Institutes of Health (2013), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2014), University of Washington Medical Center (2014), Praxis fuer Humangenetik Tuebingen (2016), Illumina Clinical Services Laboratory (2016), Invitae (2017), Children's Mercy Hospital and Clinics (2017), GeneDx (2017), Laboratory for Molecular Medicine of Partners HealthCare Personalized Medicine (2017)
68660	TNP1	I	D	II	Male Infertility	Miyagawa (2005), Nishimune (2006)
78550	SPATA16	C	T	CC	Male Infertility	Dam (2007)
9408	SLURP1	G	A,C	GG	Mal de Meleda	Fischer (2001)
9410	SLURP1	C	A,T	CC	Mal de Meleda	Charfeddine (2003), Mokni (2004)
9411	SLURP1	C	T	CC	Mal de Meleda	Fischer (2001)
78171	LPIN2	G	A	GG	Majeed Syndrome	Majeed (1989), Ferguson (2005)
101250	ETFDH	G	A	GG	MADD	Liang (2009)
101252	ETFDH	G	A	GG	MADD	

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
101247	ETFDH	T	C	TT	MADD	Beard (1993)
800964	PRPH2	T	C,G	TT	Macular Dystrophy, Vitelliform, Adult-onset	Passerini (2007)
70753	PRPH2	G	A	GG	Macular Dystrophy	Wells (1993), Wroblewski (1994), Reig (1995), Payne (1998)
51979	CNGB3	A	C,T	AA	Macular Degeneration, Juvenile and Stargardt Disease	Nishiguchi (2005)
46673	PTEN	A	G	AA	Macrocephaly/Autism Syndrome	Butler (2005)
46722	PTEN	A	G	AA	Macrocephaly/Autism Syndrome	Butler (2005)
46719	PTEN	T	C	TT	Macrocephaly/Autism Syndrome	Butler (2005)
67858	FLT4	C	T	CC	Lymphoedema, Hereditary	Ghalamkarpour (2006)
67869	FLT4	C	T	CC	Lymphoedema, Hereditary	Spiegel (2006)
43788	TSC1	G	T	GG	Lymphangioleiomyomatosis	Knudson, (1971), Sato (2002)
9458	ERCC5	A	G	AA	Lung cancer, susceptibility to	Matakidou (2006)
9455	ERCC5	C	T	CC	Lung cancer, susceptibility to	Matakidou (2006)
9453	ERCC5	G	C	GG	Lung cancer, susceptibility to	Matakidou (2006)
78613	UPF3B	G	A,T	G	Lujan-Fryns Syndrome	Tarpey (2007)
54653	CYP2J2	A	T	AA	Lower Arachidonic & Linoleic Acid Metabolism	King (2002)
54655	CYP2J2	T	A	TT	Lower Arachidonic & Linoleic Acid Metabolism	King (2002)
68636	OCRL	C	T	C	Lowe Syndrome	Leahey (1992), Leahey (1993)
68637	OCRL	C	T	C	Lowe Syndrome	Monnier (2000), Sethi (2008)
68638	OCRL	I	D	I	Lowe Syndrome	Lin (1997), Sethi (2008)
4748	PCSK9	C	G	CC	Low LDL Cholesterol Plasma Levels	Cohen (2005), Cohen (2006), Zhao (2006), Mbikay (2007)
28158	GYS2	T	C	TT	Liver Glycogen Storage Disease 0	Orho (1998)
28160	GYS2	G	A	GG	Liver Glycogen Storage Disease 0	Orho (1998)
30913	DCX	C	A,T	C	Lissencephaly	Pilz (1999)
30931	DCX	C	A,T	C	Lissencephaly	Demelas (2001)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
5658	LPL	C	T	CC	Lipoprotein lipase deficiency	Ma (1991), Normand , (1992), Normand (1992), Wood (1993)
5649	LPL	G	A	GG	Lipoprotein lipase deficiency	Wilson (1983), Gagne , (1989), Emi (1990), Monsalve (1990), Wilson (1990), Paulweber (1991), Henderson (1992), Gilbert (2001)
2180	LMNA	C	T	CC	Lipodystrophy with Diabetes, Hepatic Steatosis, Hypertrophic Cardiomyopathy, Leukomelanodermic Papules	Novelli (2003)
800863	LMNA	G	T	GG	Lipodystrophy with Diabetes, Hepatic Steatosis, Hypertrophic Cardiomyopathy, Leukomelanodermic Papules	Brown (2001), Vigouroux (2001), Caux (2003), Hegele (2003), Vigouroux (2003), Chen (2003), Jacob (2005)
2205	LMNA	G	T	GG	Werner Syndrome, Atypical	Brown (2001), Vigouroux (2001), Caux (2003), Hegele (2003), Vigouroux (2003), Chen (2003), Jacob (2005)
38364	FKRP	C	A	CC	Limb-girdle Muscular Dystrophy, Type 2I	Brockington (2001), Mercuri (2003), de Paula (2003), Frosk (2005), Frosk (2005), Sveen (2006), Chong (2012), Athena Diagnostics Inc (2015), Genetic Services Laboratory of the University of Chicago (2016), Counsyl (2016), GeneDx (2017), Laboratory for Molecular Medicine of Partners HealthCare Personalized Medicine (2017), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2017), Fulgent Genetics (2017), Centre for Mendelian Genomics of the University Medical Centre Ljubljana (2017), Undiagnosed Diseases Network of the NIH (2017), Center for Pediatric Genomic Medicine of Children's Mercy Hospital and Clinics (2017), Mayo Clinic Genetic Testing Laboratories (2017), Invitae (2018)
40091	SGCB	C	G	CC	Limb-girdle Muscular Dystrophy, Type 2E	Bönnemann (1998)
802231	SGCB	C	T	CC	Limb-girdle Muscular Dystrophy, Type 2E	Bönnemann (1998), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2017)
39082	SGCA	C	T	CC	Limb Girdle Muscular Dystrophy, Type 2D	Piccolo (1995), Piccolo (1995), Bueno (1995), Kawai (1995)
47347	TP53	A	G	AA	Li-Fraumeni Syndrome	Law (1991), Hung (1999)
47423	TP53	C	A,G,T	CC	Li-Fraumeni Syndrome	Metzger (1991)
5528	SCNN1B	C	T	CC	Liddle syndrome	Liddle (1963), Shimkets (1994)
5572	SCNN1G	G	A	GG	Liddle syndrome	Hansson (1995), Hansson (1995), Hansson (1995), Hansson (1995)
21926	FUT3	C	T	CC	Possible Absence of Lewis Antigen	Koda (1993)
42485	EIF2B2	A	G	AA	Leukoencephalopathy with Vanishing White Matter|Ovarioleukodystrophy	Leegwater (2001), Fogli (2003)
41412	EIF2B5	G	A	GG	Leukoencephalopathy with Vanishing White Matter|Ovarioleukodystrophy	Leegwater (2001), Leegwater , (2001), Biancheri (2003), Fogli (2003), Fogli (2003), Fogli (2003), Biancheri (2004), der Knaap (2004)
41402	EIF2B5	A	G	AA	Leukoencephalopathy with Vanishing White Matter	Leegwater (2001)
42439	EIF2B1	C	A	CC	Leukoencephalopathy with Vanishing White Matter	Williams , (2001), Ohlenbusch (2005)
42440	EIF2B1	T	A	TT	Leukoencephalopathy with Vanishing White Matter	der Knaap (2002)
42441	EIF2B1	C	A,G,T	CC	Leukoencephalopathy with Vanishing White Matter	der Knaap (2002)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
100892	EIF2B3	A	G	AA	Leukoencephalopathy with Vanishing White Matter	Wu (2009)
24831	ITGB2	A	G	AA	Leukocyte Adhesion Deficiency	Hogg (1999), Hogg (1999)
24839	ITGB2	C	T	CC	Leukocyte Adhesion Deficiency	Hogg (1999), Hogg (1999)
71450	GLE1L	A	G	AA	Lethal Congenital Contracture Syndrome	Nousiainen (2008)
78460	PIP5K1C	C	T	CC	Lethal Congenital Contractural Syndrome 3	Narkis (2007)
78062	ERBB3	A	G	AA	Lethal Congenital Contractural Syndrome	Narkis (2007)
71452	GLE1L	G	A	GG	Lethal Arthrogryposis with Anterior Horn Cell Disease	Nousiainen (2008)
71453	GLE1L	T	C	TT	Lethal Arthrogryposis with Anterior Horn Cell Disease	Nousiainen (2008), Nousiainen (2008)
59005	HPRT1	C	T	C	Lesch-Nyhan syndrome	Gibbs (1989), Gibbs (1989), Tarle (1991), Marcus (1992), Marcus (1992), Marcus (1992), Marcus (1992), De Gregorio (2000)
11509	INSR	T	C	TT	Leprechaunism	Kadowaki (1988)
61821	PTPN11	C	T	CC	Leopard Syndrome 1	Digilio (2002)
50031	FH	G	A	GG	Leiomyomatosis and Renal Cell Cancer	Tomlinson (2002)
75052	LRPPRC	G	A	GG	Leigh Syndrome, French-Canadian Type	Mootha (2003)
75053	LRPPRC	I	D	II	Leigh Syndrome, French-Canadian Type	Mootha (2003)
70746	MTCO1	T	C	?	Acquired Idiopathic Sideroblastic Anemia	Gattermann (1997)
800825	MTCO1	T	C	?	Acquired Idiopathic Sideroblastic Anemia	Gattermann (1997)
70744	COX1	G	A	?	Leber Optic Atrophy	Brown , (1992), Brown , (1992), Neufield, (1993), Wallace, (1994), Guan , (1998), Pandya (1999), Pandya (1999), Yuan (2005), Jin (2007)
801991	COX1	G	A	?	Leber Hereditary Optic Neuropathy	Brown , (1992), Brown , (1992), Neufield, (1993), Wallace, (1994), Guan , (1998), Pandya (1999), Pandya (1999), Yuan (2005), Jin (2007)
801992	COX1	G	A	?	Deafness, Aminogycloside-induced	Brown , (1992), Brown , (1992), Neufield, (1993), Wallace, (1994), Guan , (1998), Pandya (1999), Pandya (1999), Yuan (2005), Jin (2007)
801993	COX1	G	A	?	Deafness, Nonsyndromic Sensorineural	Brown , (1992), Brown , (1992), Neufield, (1993), Wallace, (1994), Guan , (1998), Pandya (1999), Pandya (1999), Yuan (2005), Jin (2007)
100754	MTTS1	D	I	?	Deafness, Aminoglycoside-induced and Mitochondrial Cytochrome C Oxidase Deficiency	Tiranti (1995), Jaksch (1998), Verhoeven (1999), Verhoeven (1999), Verhoeven (1999), Friedman (1999), Friedman (1999), Toompuu (1999), Hutchin (2001)
68616	MTTK	A	G	?	MERRF Syndrome	Shoffner (1990), Yoneda (1990), Berkovic (1991), Seibel (1991), Shih (1991), Tanno (1991), Zeviani (1991), Noer (1991), Boulet (1992), Suomalainen (1993), Lertrit (1992), Wallace (1992), Penisson-Besnier (1992), Silvestri (1993), Hammans (1993), Fang (199
42366	NDUFS8	C	T	CC	Leigh syndrome	Procaccio and Wallace (2004)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
42368	NDUFS8	G	A	GG	Leigh syndrome	Procaccio and Wallace (2004)
42347	NDUFS8	C	T	CC	Leigh Syndrome	Loeffen (1998)
42367	NDUFS8	G	A	GG	Leigh syndrome	Loeffen (1998)
801986	COX15	G	A	GG	Congenital Myasthenic Syndrome, Acetazolamide-responsive	Kennaway (1990), Antonicka (2003), Oquendo (2004), Alfadhel (2011)
801987	COX15	G	A	GG	Cardioencephalomyopathy, Fatal Infantile, due to Cytochrome C Oxidase Deficiency 2	Kennaway (1990), Antonicka (2003), Oquendo (2004), Alfadhel (2011)
102073	DLD	G	A	GG	Leigh Syndrome	Grafakou (2003)
30659	PDHA1	A	C	A	Leigh Syndrome	Matthews (1993)
30661	PDHA1	C	G	C	Leigh Syndrome	Naito (1997)
30678	PDHA1	A	G	A	Leigh Syndrome	De Meirleir (1994)
38290	SURF1	C	T	CC	Leigh Syndrome	Poyau (2000)
38297	SURF1	C	T	CC	Leigh syndrome	Tiranti (1999)
38298	SURF1	G	A	GG	Leigh syndrome	Santoro (2000)
38299	SURF1	C	T	CC	Leigh syndrome	Poyau (2000)
38300	SURF1	A	C	AA	Leigh syndrome	Yuksel (2006)
38303	SURF1	G	A	GG	Leigh syndrome	Capkova (2002)
38305	SURF1	A	G	AA	Leigh syndrome	Sacconi (2003)
38306	SURF1	G	A	GG	Leigh syndrome	Ogawa (2002)
38307	SURF1	G	A	GG	Leigh syndrome	Coenen (1999)
38308	SURF1	A	G	AA	Leigh syndrome	Pecina (2004)
38309	SURF1	A	G	AA	Leigh syndrome	Poyau (2000)
38310	SURF1	G	A	GG	Leigh syndrome	Tiranti (1998), Tiranti (1999)
38311	SURF1	C	A	CC	Leigh syndrome	Ogawa (2002)
38312	SURF1	A	C	AA	Leigh syndrome	Teraoka (1999)
38313	SURF1	C	T	CC	Leigh syndrome	Tay (2005)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
1504	TAZ	T	C	T	Left Ventricular Noncompaction, Isolated	Ichida (2001)
6563	DTNA	C	T	CC	Left Ventricular Noncompaction associated with Congenital Heart Defects	Ichida (2001)
800804	MTATP6	T	C	?	Leigh Syndrome	van Erven (1987), de Vries (1993), Rahman (1996), Chakrapani (1998), Fujii (1998), Vilarinho (2001), Rantamaki (2005), Debray (2007), Craig (2007)
800823	MTATP6	T	C	?	Ataxia and Polyneuropathy, Adult-onset	van Erven (1987), de Vries (1993), Rahman (1996), Chakrapani (1998), Fujii (1998), Vilarinho (2001), Rantamaki (2005), Debray (2007), Craig (2007)
68615	MTATP6	T	C	?	Striatonigral Degeneration, Infantile, Mitochondrial	Thyagarajan (1995), Dionisi-Vici (1998), Makino (1998)
70735	MTND4	G	A	?	Leber Hereditary Optic Neuropathy	Wallace , (1988), Wallace , (1988), Vilkki , (1989), Hotta , (1989), Holt , (1989), Yoneda , (1989), Singh , (1989), Singh , (1989), Johns, (1990), Stone , (1990), Wallace, (1990), Huoponen , (1990), Bolhuis , (1990), Lott , (1990), Newman , (1991), Majander , (1991), Cortelli , (1991), Larsson , (1991), Berman, (1991), Poulton , (1991), Nakagawa, (1991), Carducci , (1991), Hiida , (1991), Kormann , (1991), Johns , (1992), Mashima , (1992), Howell , (1992), Sudoyo , (1992), Zhu , (1992), Weiner , (1993), Newman, (1993), Norby, (1993), Nakamura , (1993), Moorman , (1993), Castora, (1993), Cullom , (1993), Cavelier , (1993), Smith , (1993), Torroni (1997), Chinnery (2001), Guy (2002), Guy (2002), Wong (2002), Wong (2002), Guy (2002), Mimaki (2003), Qu (2006), Carelli (2006), Phasukkijwatana (2006), Qu (2006)
100741	MTTH	G	A	?	MERRF Syndrome	Melone (2004), Taylor (2004)
100743	MTTH	G	A	?	Pigmentary Retinopathy and Sensorineural Deafness	Crimi (2003), Crimi (2003)
52928	CRB1	T	G	TT	Leber Congenital Amaurosis, Type VIII	Lotery (2001)
52934	CRB1	C	T	CC	Leber Congenital Amaurosis, Type VIII	Lotery (2001)
52935	CRB1	C	T	CC	Leber Congenital Amaurosis, Type VIII	Yzer (2006)
52936	CRB1	G	A	GG	Leber Congenital Amaurosis, Type VIII	Lotery (2001)
52939	CRB1	C	T	CC	Leber Congenital Amaurosis, Type VIII	Yzer (2006)
52940	CRB1	G	C	GG	Leber Congenital Amaurosis, Type VIII	Yzer (2006)
52941	CRB1	T	C	TT	Leber Congenital Amaurosis, Type VIII	Lotery (2001)
52942	CRB1	T	G	TT	Leber Congenital Amaurosis, Type VIII	Lotery (2001)
52943	CRB1	T	C	TT	Leber Congenital Amaurosis, Type VIII	Galvin (2005)
52944	CRB1	T	C	TT	Leber Congenital Amaurosis, Type VIII	Vallespin (2007)
52947	CRB1	G	T	GG	Leber Congenital Amaurosis, Type VIII	Hanein (2004)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
52949	CRB1	G	A	GG	Leber Congenital Amaurosis, Type VIII	Lotery (2001)
52950	CRB1	G	C	GG	Leber Congenital Amaurosis, Type VIII	Hanein (2004)
52951	CRB1	T	C	TT	Leber Congenital Amaurosis, Type VIII	Hanein (2004)
52952	CRB1	T	C	TT	Leber Congenital Amaurosis, Type VIII	Galvin (2005)
52955	CRB1	G	T	GG	Leber Congenital Amaurosis, Type VIII	Hanein (2004)
52960	CRB1	T	C	TT	Leber Congenital Amaurosis, Type VIII	Hanein (2004)
52963	CRB1	T	A	TT	Leber Congenital Amaurosis, Type VIII	Hanein (2004)
52967	CRB1	T	C	TT	Leber Congenital Amaurosis, Type VIII	Khaliq (2003)
52969	CRB1	T	A	TT	Leber Congenital Amaurosis, Type VIII	Vallespin (2006)
52970	CRB1	G	T	GG	Leber Congenital Amaurosis, Type VIII	Hanein (2004)
52974	CRB1	T	G	TT	Leber Congenital Amaurosis, Type VIII	Hollander (2001)
52975	CRB1	G	A	GG	Leber Congenital Amaurosis, Type VIII	Hanein (2004)
52976	CRB1	T	C	TT	Leber Congenital Amaurosis, Type VIII	Hanein (2004)
52979	CRB1	G	A	GG	Leber Congenital Amaurosis, Type VIII	Lotery (2001)
52981	CRB1	C	T	CC	Leber Congenital Amaurosis, Type VIII	Yzer (2006)
52982	CRB1	G	A	GG	Leber Congenital Amaurosis, Type VIII	Hanein (2004)
52983	CRB1	A	C	AA	Leber Congenital Amaurosis, Type VIII	Lotery (2001)
52984	CRB1	T	A	TT	Leber Congenital Amaurosis, Type VIII	Hanein (2004)
52985	CRB1	G	T	GG	Leber Congenital Amaurosis, Type VIII	Vallespin (2006)
52987	CRB1	C	A	CC	Leber Congenital Amaurosis, Type VIII	Lotery (2001)
52988	CRB1	G	T	GG	Leber Congenital Amaurosis, Type VIII	Hollander (2001)
52826	CRX	C	T	CC	Leber Congenital Amaurosis, Type VII	Freund (1998), Swaroop (1999), Swaroop (1999)
52570	RPGRIP1	G	A	GG	Leber Congenital Amaurosis, Type VI	Dryja (2001)
52571	RPGRIP1	C	T	CC	Leber Congenital Amaurosis, Type VI	Gerber (2001)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
52572	RPGRIP1	G	A	GG	Leber Congenital Amaurosis, Type VI	Gerber (2001)
52573	RPGRIP1	C	T	CC	Leber Congenital Amaurosis, Type VI	Hanein (2004)
52574	RPGRIP1	C	T	CC	Leber Congenital Amaurosis, Type VI	Gerber (2001)
53488	RDH12	A	G	AA	Leber Congenital Amaurosis, Type III	Janecke (2004), Perrault (2004)
53476	RDH12	C	T	CC	Leber Congenital Amaurosis, Type III	Janecke (2004), Perrault (2004)
70708	RDH12	I	D	II	Leber Congenital Amaurosis, Type III	Janecke (2004), Perrault (2004)
53473	RDH12	C	T	CC	Leber Congenital Amaurosis, Type III	Janecke (2004)
53474	RDH12	T	A	TT	Leber Congenital Amaurosis, Type III	Perrault (2004)
53479	RDH12	G	T	GG	Leber Congenital Amaurosis, Type III	Perrault (2004)
53481	RDH12	C	A	CC	Leber Congenital Amaurosis, Type III	Perrault (2004)
53482	RDH12	C	G	CC	Leber Congenital Amaurosis, Type III	Perrault (2004)
53484	RDH12	T	C	TT	Leber Congenital Amaurosis, Type III	Perrault (2004)
53485	RDH12	C	T	CC	Leber Congenital Amaurosis, Type III	Janecke (2004)
53488	RDH12	A	G	AA	Leber Congenital Amaurosis, Type III	Janecke (2004)
53489	RDH12	C	G	CC	Leber Congenital Amaurosis, Type III	Perrault (2004)
52325	GUCY2D	T	C	TT	Leber Congenital Amaurosis, Type I	Perrault (1996), Duda (1999), Duda (1999)
52310	GUCY2D	T	A	TT	Leber Congenital Amaurosis, Type I	Hanein (2004)
52311	GUCY2D	G	A	GG	Leber Congenital Amaurosis, Type I	Perrault (2000)
52312	GUCY2D	G	C	GG	Leber Congenital Amaurosis, Type I	Perrault (2000)
52313	GUCY2D	C	T	CC	Leber Congenital Amaurosis, Type I	Perrault (2000)
52314	GUCY2D	G	A	GG	Leber Congenital Amaurosis, Type I	Dharmaraj (2000)
52315	GUCY2D	C	A	CC	Leber Congenital Amaurosis, Type I	Perrault (2000)
52316	GUCY2D	C	T	CC	Leber Congenital Amaurosis, Type I	Galvin (2005)
52317	GUCY2D	C	T	CC	Leber Congenital Amaurosis, Type I	Perrault (2000)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
52318	GUCY2D	T	C	TT	Leber Congenital Amaurosis, Type I	Dharmaraj (2000)
52319	GUCY2D	A	G	AA	Leber Congenital Amaurosis, Type I	Hanein (2004)
52320	GUCY2D	C	A	CC	Leber Congenital Amaurosis, Type I	Perrault (2000)
52322	GUCY2D	C	T	CC	Leber Congenital Amaurosis, Type I	Perrault (2000)
52323	GUCY2D	T	G	TT	Leber Congenital Amaurosis, Type I	Dharmaraj (1999)
52324	GUCY2D	T	C	TT	Leber Congenital Amaurosis, Type I	Perrault (2000)
52326	GUCY2D	G	A	GG	Leber Congenital Amaurosis, Type I	Lotery (2000)
52327	GUCY2D	C	T	CC	Leber Congenital Amaurosis, Type I	Lotery (2000)
52329	GUCY2D	A	G	AA	Leber Congenital Amaurosis, Type I	Lotery (2000)
52330	GUCY2D	C	T	CC	Leber Congenital Amaurosis, Type I	Lotery (2000)
52331	GUCY2D	A	T	AA	Leber Congenital Amaurosis, Type I	Lotery (2000)
52332	GUCY2D	G	A	GG	Leber Congenital Amaurosis, Type I	Weigell-Weber (2000)
52333	GUCY2D	C	T	CC	Leber Congenital Amaurosis, Type I	El-Shanti (1999)
52334	GUCY2D	C	T	CC	Leber Congenital Amaurosis, Type I	Dharmaraj (2000)
52335	GUCY2D	G	C	GG	Leber Congenital Amaurosis, Type I	Hanein (2004)
52336	GUCY2D	C	T	CC	Leber Congenital Amaurosis, Type I	Yzer (2006)
52337	GUCY2D	T	C	TT	Leber Congenital Amaurosis, Type I	Koenekoop (2002)
52338	GUCY2D	G	T	GG	Leber Congenital Amaurosis, Type I	Perrault (2000)
52339	GUCY2D	G	A	GG	Leber Congenital Amaurosis, Type I	Galvin (2005)
52340	GUCY2D	C	T	CC	Leber Congenital Amaurosis, Type I	Perrault (2000)
52341	GUCY2D	A	C	AA	Leber Congenital Amaurosis, Type I	Perrault (2000)
52342	GUCY2D	A	C	AA	Leber Congenital Amaurosis, Type I	Perrault (2000)
52343	GUCY2D	C	T	CC	Leber Congenital Amaurosis, Type I	Perrault (2000)
52344	GUCY2D	C	G	CC	Leber Congenital Amaurosis, Type I	Lotery (2000)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
52345	GUCY2D	C	T	CC	Leber Congenital Amaurosis, Type I	Hanein (2004)
52346	GUCY2D	A	T	AA	Leber Congenital Amaurosis, Type I	Hanein (2004)
52347	GUCY2D	T	C	TT	Leber Congenital Amaurosis, Type I	Hanein (2004)
52348	GUCY2D	G	A	GG	Leber Congenital Amaurosis, Type I	Perrault (2000)
52349	GUCY2D	G	T	GG	Leber Congenital Amaurosis, Type I	Perrault (2000)
52350	GUCY2D	T	A	TT	Leber Congenital Amaurosis, Type I	Perrault (2000)
68632	CEP290	T	C	TT	Leber Congenital Amaurosis 10	Hollander (2006), Brancati (2007)
52827	CRX	C	T	CC	Leber congenital amaurosis ?	Zhang (2001)
52304	GUCY2D	C	A	CC	Leber congenital amaurosis	Payne (2001)
52824	CRX	G	A	GG	Leber congenital amaurosis	Lotery (2000)
52977	CRB1	G	T	GG	Leber congenital amaurosis	Hollander (2001)
53352	BEST1	G	T	GG	Leber congenital amaurosis	Lotery (2000)
53462	RDH12	C	A	CC	Leber congenital amaurosis	Perrault (2004)
52867	LCAT	G	A,T	GG	LCAT Deficiency	Funke (1993)
52869	LCAT	G	A	GG	LCAT Deficiency	Funke (1993)
52870	LCAT	C	T	CC	LCAT Deficiency	Funke (1993)
52876	LCAT	T	A	TT	LCAT Deficiency	Okubo (1996)
52879	LCAT	G	A	GG	LCAT Deficiency	Argyropoulos (1998)
52880	LCAT	C	T	CC	LCAT Deficiency	Owen (1996)
52882	LCAT	C	G	CC	LCAT Deficiency	Wiebusch (1995)
52883	LCAT	A	T	AA	LCAT Deficiency	Calabresi (2005)
52884	LCAT	G	T	GG	LCAT Deficiency	Funke (1993)
52889	LCAT	C	T	CC	LCAT Deficiency	Steyrer (1995)
52890	LCAT	G	A	GG	LCAT Deficiency	Maruyama (2004)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
52891	LCAT	G	C	GG	LCAT Deficiency	Gotoda (1991)
52894	LCAT	G	A	GG	LCAT Deficiency	Vergani (1983), Taramelli (1990)
52895	LCAT	A	T	AA	LCAT Deficiency	Klein (1993)
52896	LCAT	G	A	GG	LCAT Deficiency	Funke (1993)
52897	LCAT	A	C	AA	LCAT Deficiency	Guerin (1997)
52899	LCAT	C	T	CC	LCAT Deficiency	Frasca (2004)
52900	LCAT	C	T	CC	LCAT Deficiency	Guerin (1997)
52902	LCAT	G	T	GG	LCAT Deficiency	Gotoda (1991)
52903	LCAT	C	G	CC	LCAT Deficiency	Miettinen (1998)
52904	LCAT	C	T	CC	LCAT Deficiency	Calabresi (2005)
52905	LCAT	G	C	GG	LCAT Deficiency	Skretting (1992)
52906	LCAT	G	C	GG	LCAT Deficiency	Maruyama (2004)
52907	LCAT	A	T	AA	LCAT Deficiency	Skretting (1992)
52908	LCAT	G	A	GG	LCAT Deficiency	Calabresi (2005)
52909	LCAT	T	C	TT	LCAT Deficiency	Sessa (2001)
52910	LCAT	C	T	CC	LCAT Deficiency	Maeda (1991)
52911	LCAT	G	A	GG	LCAT Deficiency	Argyropoulos (1998)
52913	LCAT	C	T	CC	LCAT Deficiency	Moriyama (1995)
52915	LCAT	A	C	AA	LCAT Deficiency	Nanjee (2003)
52917	LCAT	G	A,T	GG	LCAT Deficiency	Miettinen (1995)
41229	LGI1	C	T	CC	Lateral Temporal Lobe Epilepsy	Morante-Redolat (2002), Bisulli (2004)
41207	LGI1	T	G	TT	Lateral Temporal Lobe Epilepsy	Fertig (2003)
41220	LGI1	T	C	TT	Lateral Temporal Lobe Epilepsy	Gu (2002), Pizzuti (2003)
40510	ARSA	C	A,T	CC	Late Infantile Metachromatic Leukodystrophy	Kreysing (1993)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
22563	PROCR	T	G	TT	Late Fetal Loss	Franchi (2001)
64154	FLNB	A	C	AA	Larsen syndrome	Zhang (2006)
64155	FLNB	T	G	TT	Larsen syndrome	Krakow (2004)
64167	FLNB	G	A	GG	Larsen syndrome	Krakow (2004)
64168	FLNB	C	G	CC	Larsen syndrome	Bicknell (2007)
64170	FLNB	G	A	GG	Larsen syndrome	Bicknell (2007)
64171	FLNB	G	A	GG	Larsen syndrome	Bicknell (2007)
64175	FLNB	T	G	TT	Larsen syndrome	Bicknell (2007)
64176	FLNB	G	A	GG	Larsen syndrome	Zhang (2006)
64177	FLNB	C	A	CC	Larsen syndrome	Zhang (2006)
64178	FLNB	G	A	GG	Larsen syndrome	Krakow (2004)
64179	FLNB	T	A	TT	Larsen syndrome	Bicknell (2007)
64181	FLNB	C	T	CC	Larsen syndrome	Bicknell (2007)
64185	FLNB	G	T	GG	Larsen syndrome	Zhang (2006)
64186	FLNB	G	A	GG	Larsen Syndrome	Bicknell (2007)
64187	FLNB	G	A	GG	Larsen syndrome	Bicknell (2007)
11183	GH1	T	C	TT	Kowarski Syndrome	Takahashi (1997)
25908	GJB2	G	T	GG	Knuckle pads, Leukonychia, Sensorineural Deafness	Richard (2004)
22146	SLC14A1	C	G	CC	Kidd blood group variant	Lucien (2002)
25846	GJB2	C	T	CC	Keratoderma, Palmoplantar, with Deafness|Deafness, Nonsyndromic Sensorineural	Uyguner (2002), Feldmann (2005), Feldmann (2005)
7893	KRT1	C	T	CC	Keratoderma, palmoplantar	Hatsell (2001)
25900	GJB2	C	T	CC	Keratitis-ichthyosis-deafness Syndrome	Ohtsuka , (2003), Janecke (2005), Janecke (2005), Janecke (2005), Abe (2007)
58435	DNAI1	G	A	GG	Kartagener Syndrome	Guichard (2001)
12662	FGFR1	C	A	CC	Kallmann Syndrome 2	Dode (2003)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
12655	FGFR1	C	T	CC	Kallmann Syndrome 2	Trarbach (2006)
12656	FGFR1	C	T	CC	Kallmann Syndrome 2	Dode (2003)
12657	FGFR1	T	C	TT	Kallmann Syndrome 2	Dode (2003)
12658	FGFR1	C	A	CC	Kallmann Syndrome 2	Dode (2007)
12659	FGFR1	C	T	CC	Kallmann Syndrome 2	Albuisson (2005)
12661	FGFR1	T	C,G	TT	Kallmann Syndrome 2	Albuisson (2005)
12663	FGFR1	C	G	CC	Kallmann Syndrome 2	Zenaty (2006)
12664	FGFR1	A	G	AA	Kallmann Syndrome 2	Trarbach (2006)
12665	FGFR1	G	A	GG	Kallmann Syndrome 2	Trarbach (2006)
12667	FGFR1	C	T	CC	Kallmann Syndrome 2	Dode (2007)
12668	FGFR1	C	T	CC	Kallmann Syndrome 2	Albuisson (2005)
12669	FGFR1	C	T	CC	Kallmann Syndrome 2	Dode (2003)
12670	FGFR1	G	C	GG	Kallmann Syndrome 2	Dode (2007)
12672	FGFR1	C	A	CC	Kallmann Syndrome 2	Dode (2007)
12674	FGFR1	G	C	GG	Kallmann Syndrome 2	Dode (2007)
12675	FGFR1	A	G	AA	Kallmann Syndrome 2	
12676	FGFR1	G	A	GG	Kallmann Syndrome 2	Trarbach (2006)
12677	FGFR1	G	A	GG	Kallmann Syndrome 2	Trarbach (2006)
12681	FGFR1	C	T	CC	Kallmann Syndrome 2	Albuisson (2005)
12682	FGFR1	C	T	CC	Kallmann Syndrome 2	Dode (2003)
12683	FGFR1	T	C	TT	Kallmann Syndrome 2	Dode (2007)
12684	FGFR1	G	C	GG	Kallmann Syndrome 2	Zenaty (2006)
12685	FGFR1	G	A,C	GG	Kallmann Syndrome 2	Dode (2003)
12686	FGFR1	G	A	GG	Kallmann Syndrome 2	Dode (2007)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
12687	FGFR1	A	T	AA	Kallmann Syndrome 2	Dode (2003)
12688	FGFR1	G	A	GG	Kallmann Syndrome 2	Dode (2007)
12689	FGFR1	C	G,T	CC	Kallmann Syndrome 2	Sato (2005)
12690	FGFR1	T	A	TT	Kallmann Syndrome 2	Dode (2007)
12691	FGFR1	A	C	AA	Kallmann Syndrome 2	Dode (2003)
12692	FGFR1	G	A	GG	Kallmann Syndrome 2	Pitteloud (2006)
12693	FGFR1	G	C	GG	Kallmann Syndrome 2	Albuisson (2005)
12694	FGFR1	G	A	GG	Kallmann Syndrome 2	Sato (2004)
12696	FGFR1	C	T	CC	Kallmann Syndrome 2	Trarbach (2006)
12547	KAL1	C	T	C	Kallmann Syndrome	Maya-Nunez (1998)
12548	KAL1	C	T	C	Kallmann Syndrome	Albuisson (2005)
12549	KAL1	G	A	G	Kallmann Syndrome	Izumi (1999)
12550	KAL1	C	T	C	Kallmann Syndrome	Sato (2004)
12551	KAL1	A	G	A	Kallmann Syndrome	Oliveira (2001)
12552	KAL1	G	A	G	Kallmann Syndrome	Oliveira (2001)
12553	KAL1	C	T	C	Kallmann Syndrome	Hardelin (1992)
12554	KAL1	G	A	G	Kallmann Syndrome	Hardelin (1992)
12555	KAL1	C	G,T	C	Kallmann Syndrome	Hardelin (1992)
12556	KAL1	C	G	C	Kallmann Syndrome	Albuisson (2005)
12557	KAL1	G	A	G	Kallmann Syndrome	Soderlund (2002)
12558	KAL1	A	C	A	Kallmann Syndrome	Loidi (2005)
12559	KAL1	A	T	A	Kallmann Syndrome	Hardelin (1993)
12561	KAL1	C	A	C	Kallmann Syndrome	Albuisson (2005)
12562	KAL1	G	C	G	Kallmann Syndrome	Georgopoulos (1997)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
12563	KAL1	G	A	G	Kallmann Syndrome	Hardelin (1993)
12564	KAL1	G	A	G	Kallmann Syndrome	Hardelin (1993)
12565	KAL1	G	A	G	Kallmann Syndrome	Sato (2004)
12566	KAL1	G	A	G	Kallmann Syndrome	Oliveira (2001)
12569	KAL1	G	C	G	Kallmann Syndrome	Georgopoulos (1997)
12570	KAL1	A	T	A	Kallmann Syndrome	Albuisson (2005)
12571	KAL1	G	A,T	G	Kallmann Syndrome	Jansen (2000)
12572	KAL1	C	A	C	Kallmann Syndrome	Albuisson (2005)
12573	KAL1	C	T	C	Kallmann Syndrome	Hardelin (1993)
12574	KAL1	C	A	C	Kallmann Syndrome	Sato (2004)
12575	KAL1	C	T	C	Kallmann Syndrome	Neill (1998)
12576	KAL1	C	T	C	Kallmann Syndrome	Izumi (2001)
12577	KAL1	C	A	C	Kallmann Syndrome	Neill (1997)
12578	KAL1	C	A	C	Kallmann syndrome	Trarbach (2006)
48104	SMAD4	G	A	GG	Juvenile Polyposis/Hereditary Hemorrhagic Telangiectasia Syndrome	Baert (1983), Burger (2002), Burger (2002), Gallione (2004), Gallione (2004)
39856	PARK2	G	A,T	GG	Juvenile Parkinson Disease 2	Abbas (1999), Cookson (2003), Klein (2005), Invitae (2016), Illumina Clinical Services Laboratory (2016), Children's Mercy Hospital and Clinics (2017)
39881	PARK2	T	A	TT	Juvenile Parkinson Disease 2	Klein (2005)
40401	EFHC1	T	C	TT	Harmless variant (previously associated with Juvenile Myoclonic Epilepsy, Type 1)	Bai (2002), Suzuki (2004), University of Chicago (2014), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2015), GeneDx (2015), Invitae (2017), Children's Mercy Hospital and Clinics (2017), Athena Diagnostics (2017)
42619	CACNB4	G	A	GG	Juvenile Myoclonic Epilepsy	Escayg (2000), Escayg (2000)
61795	PTPN11	G	A	GG	Juvenile Myelomonocytic Leukemia	Tartaglia (2003)
40538	ARSA	C	T	CC	Juvenile Metachromatic Leukodystrophy|Adult Metachromatic Leukodystrophy|Arylsulfatase A, Allele I	Polten (1991), Polten (1991), Polten , (1991), Barth , (1993), Gieselmann , (1994), Heinisch (1995), Draghia (1997), Comabella (2001)
39134	CLCN2	C	T	CC	Juvenile Absence Epilepsy	Haug (2003)
63870	TMEM67	C	T	CC	Joubert syndrome 6	Baala (2007)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
68631	CEP290	C	A	CC	Joubert Syndrome 5	Sayer (2006), Valente (2006), Brancati (2007)
101925	AHI1	G	A	GG	Joubert Syndrome 3	Valente (2006)
101926	AHI1	A	T	AA	Joubert Syndrome 3	Ferland (2004)
101913	TMEM216	G	T	GG	Joubert Syndrome 2	Edvardson (2010), Valente (2010), GeneReviews (2012), Ambry Genetics (2014), GeneDx (2016), Counsyl (2016), Invitae (2016), Integrated Genetics of Laboratory Corporation of America (2016)
802229	TMEM216	G	A	GG	Joubert Syndrome 2	Valente (2010)
802230	TMEM216	G	A	GG	Meckel Syndrome Type 2	Valente (2010)
22147	SLC14A1	T	C	TT	JK-Null Variant, Finnish Type	Sidoux-Walter (2000)
2808	KCNQ1	G	A	GG	Jervell and Lange-Nielsen Syndrome	Tyson (2000)
32224	IVD	T	C	TT	Isovaleric Acidemia, Type I	Vockley (1991)
32234	IVD	G	T	GG	Isovaleric Acidemia, Type I	Vockley (1991)
32213	IVD	C	G	CC	Isovaleric Acidemia	Vockley (2000)
800805	IVD	C	T	CC	Isovaleric Acidemia	Vockley (2000)
32225	IVD	G	A	GG	Isovaleric Acidemia	Ensenauer (2004)
32226	IVD	G	C	GG	Isovaleric Acidemia	Mohsen (1998)
32227	IVD	G	A	GG	Isovaleric Acidemia	Mohsen (1998)
32228	IVD	G	T	GG	Isovaleric Acidemia	Ensenauer (2004)
32229	IVD	C	T	CC	Isovaleric Acidemia	Lin (2007)
32230	IVD	G	A	GG	Isovaleric Acidemia	Ensenauer (2004)
32231	IVD	G	A	GG	Isovaleric Acidemia	Ensenauer (2004)
32232	IVD	A	G	AA	Isovaleric Acidemia	Lin (2007)
32233	IVD	A	G	AA	Isovaleric Acidemia	Ensenauer (2004)
32235	IVD	G	A	GG	Isovaleric Acidemia	Ensenauer (2004)
32236	IVD	G	A	GG	Isovaleric Acidemia	Ensenauer (2004)
32237	IVD	C	T	CC	Isovaleric Acidemia	Ensenauer (2004)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
32238	IVD	A	C	AA	Isovaleric Acidemia	Ensenauer (2004)
32239	IVD	G	A	GG	Isovaleric Acidemia	Lin (2007)
32240	IVD	T	C	TT	Isovaleric Acidemia	Mohsen (1998)
32241	IVD	T	C	TT	Isovaleric Acidemia	Mohsen (1998)
32242	IVD	T	C	TT	Isovaleric Acidemia	Ensenauer (2004)
32243	IVD	C	T	CC	Isovaleric Acidemia	Mohsen (1998)
32244	IVD	A	G	AA	Isovaleric Acidemia	Lin (2007)
32245	IVD	G	T	GG	Isovaleric Acidemia	Mohsen (1998)
32246	IVD	C	T	CC	Isovaleric Acidemia	Vockley (2000)
32247	IVD	G	C	GG	Isovaleric Acidemia	Ensenauer (2004)
32248	IVD	G	T	GG	Isovaleric Acidemia	Ensenauer (2004)
32249	IVD	A	G	AA	Isovaleric Acidemia	Vockley (2000)
32250	IVD	G	A	GG	Isovaleric Acidemia	Vockley (2000)
32251	IVD	T	C	TT	Isovaleric Acidemia	Vockley (2000)
32252	IVD	G	A	GG	Isovaleric Acidemia	Vockley (2000)
32253	IVD	A	G	AA	Isovaleric Acidemia	Ensenauer (2004)
6011	CRELD1	C	T	CC	Isolated Partial Atrioventricular Septal Defect	Robinson (2003)
6012	CRELD1	C	T	CC	Isolated Partial Atrioventricular Septal Defect	Pierpont , (2000), Robinson (2003), Maslen (2006)
37828	PAFAH1B1	G	A	GG	Isolated Lissencephaly Sequence	Leventer (2001)
37836	PAFAH1B1	C	T	CC	Subcortical Band Heterotopia	Sicca (2003)
801972	PAFAH1B1	C	T	CC	Isolated Lissencephaly Sequence	Sicca (2003), Genetic Services Laboratory of the University of Chicago (2014)
11190	GH1	T	A	TT	Isolated Growth Hormone Deficiency, Type 2	Fofanova (2003)
11193	GH1	C	G,T	CC	Isolated Growth Hormone Deficiency, Type 2	Cogan (1995), Hayashi (1999), Hayashi (1999), Saitoh (1999)
11195	GH1	C	T	CC	Isolated Growth Hormone Deficiency, Type 2	Sirand-Pugnet (1995), Cogan (1997), Cogan (1997)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
11887	HESX1	C	T	CC	Isolated growth hormone deficiency	McNay (2007)
72253	TMPRSS6	C	T	CC	Iron Deficiency Anemia, Iron-refractory	Finberg (2008)
72255	TMPRSS6	C	T	CC	Iron Deficiency Anemia, Iron-refractory	Finberg (2008)
72257	TMPRSS6	I	D	II	Iron Deficiency Anemia, Iron-refractory	Finberg (2008)
72261	TMPRSS6	G	A	GG	Iron Deficiency Anemia, Iron-refractory	Finberg (2008)
68515	ACTA2	G	A	GG	Iris Flocculi	Guo (2007)
11118	FOXP3	G	A	G	IPEX syndrome	Wildin (2001), Wildin (2001)
11129	FOXP3	C	T	C	IPEX syndrome	Ferguson (2000), Wildin (2001), Bennett (2001)
12949	SLC5A5	A	C	AA	Iodide Transport Defect|Thyroid Hormonogenesis, Genetic Defect in	Fujiwara (1997), Kosugi (1997), Fujiwara (1998), Kosugi (1998), Levy (1998)
78121	IGF1R	A	C	AA	Intrauterine and Postnatal Growth Retardation (Short Stature)	Abuzzahab (2003)
78122	IGF1R	G	A	GG	Intrauterine and Postnatal Growth Retardation (Short Stature)	Kawashima (2005)
15704	ABCB11	T	C	TT	Intrahepatic cholestasis, familial progressive 2	Strautnieks (1998), Jansen (1999), Van Mil (2004), Noe (2005)
15711	ABCB11	G	A	GG	Intrahepatic cholestasis, familial progressive 2	Strautnieks (1998)
16064	**ATP8B1**	**C**	**A,T**	**CT**	**Likely harmless variant (previously associated with Intrahepatic Cholestasis of Pregnancy)**	**Klomp (2004), Mullenbach (2005), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2017), GeneDx (2017), Genomic Research Center of Shahid Beheshti University of Medical Sciences (2018)**
100756	MTTS2	G	A	?	MERRF/MELAS Overlap Syndrome	Wong (2006), Wong (2006)
14124	LHB	C	T	CC	Infertility, Male|Infertility, Female	Roy (1996), Liao , (1998), Liao (1998), Ramanujam , (1999), Ramanujam (2000), Liao (2002), Lio (2002)
13278	AR	A	G	A	Infertility, Male	January (1996), January (2005), Ferlin (2006)
13359	AR	A	G	A	Infertility, Male	Hiort (1996), Giwercman (2001)
13210	AR	C	A	C	Infertility, Male	Giwercman (2001)
13213	AR	C	T	C	Infertility, Male	January (1996), January (2005), Ferlin (2006)
66704	INVS	C	T	CC	Infantile Nephronophthisis	Otto (2003)
66703	INVS	T	C	TT	Infantile Nephronophthisis	Gagnadoux , (1989), Otto (2003)
22576	F13A1	T	A	TT	Increased risk of Stroke with Oral Contraceptives	Anwar (1999), Pruissen (2008)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
73033	IL12RB1	T	A	TT	Increased Risk of Disseminated Infection with BCG and Salmonella enteritidis	Altare (1998)
73034	IL12RB1	C	G	CC	Increased Risk of Disseminated Infection with BCG	Altare (1998)
5457	ACE	C	T	CC	Increased ACE Serum Levels, Benign	Kramers (2001), Kramers (2001), Eyries (2001), Linnebank (2003), Linnebank (2003)
40591	GNE	A	G	AA	Inclusion Body Myopathy	Eisenberg (2001), Broccolini (2002), Argov (2003), Argov (2003), Tomimitsu (2004)
23767	CD40	T	C	TT	Immunodeficiency with Hyper-IgM, Type III	Ferrari (2001)
24657	UNG	T	C	TT	Immunodeficiency with Hyper-IgM, Type 5	Imai (2003), Kavli (2005)
24614	AICDA	C	T	CC	Immunodeficiency with Hyper-IgM, Type 2	Revy (2000)
24630	AICDA	G	T	GG	Immunodeficiency with Hyper-IgM, Type 2	Revy (2000)
45992	PDGFRA	C	T	CC	Imatinib resistance in hypereosinophilic syndrome	Cools (2003)
100827	TERT	T	G	TT	Idiopathic Pulmonary Fibrosis	Armanios (2007)
70759	FRMD7	A	C	A	Idiopathic Infantile Nystagmus	Tarpey (2006)
8825	TGM1	G	A	GG	Ichthyosis, Lamellar	Huber (1995), Laiho (1997)
8873	TGM1	T	C	TT	Ichthyosis, Lamellar	Huber (1995), Pigg (1998)
8912	ABCA12	C	T	CC	Ichthyosis, Harlequin	Thomas (2006)
78175	ST14	G	A	GG	Ichthyosis with Hypotrichosis	Basel-Vanagaite (2007)
25904	GJB2	C	A,T	CC	Hystrix-Like Ichthyosis with Deafness|Keratitis-ichthyosis-deafness Syndrome	Richard (2002), Richard (2002), Van Geel (2002), Alvarez (2003), Janecke (2005), Janecke (2005)
58985	HPRT1	T	G	T	Hypoxanthine guanine phosphoribosyltransferase deficiency	Fujimori (1988)
78176	CDSN	G	A	GG	Hypotrichosis Simplex of Scalp	Metzker (1987), Betz (2000), Levy-Nissenbaum (2003)
73084	P2RY5	G	A	GG	Hypotrichosis Simplex	Pasternack (2008)
73085	P2RY5	I	D	II	Hypotrichosis Simplex	Pasternack (2008)
73021	LIPH	I	D	II	Hypotrichosis	Jelani (2008)
11275	TSHR	G	A	GG	Hypothyroidism, Congenital, Nongoitrous	Clifton-Bligh (1997), Jordan (2003), Jordan (2003), Jordan (2003), Jordan (2003)
11321	PAX8	C	T	CC	Hypothyroidism, Congenital, Nongoitrous	Vilain (2001)
13070	DUOX2	G	A,T	GG	Hypothyroidism|Thyroid Hormonogenesis, Genetic Defect in	Vigone (2005)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
13071	DUOX2	G	A,C	GG	Hypothyroidism|Thyroid Hormonogenesis, Genetic Defect in	Moreno (2002)
101074	ENPP1	A	C	AA	Hypophosphatemic Rickets	Levy-Litan (2010)
13640	FGF23	C	T	CC	Hypophosphatemic Rickets	Bianchine (1971), McEnery (1997), Consortium (2000)
13560	PHEX	C	T	C	Hypophosphatemia	Goji (2006), Goji (2006)
34418	ALPL	A	T	AA	Hypophosphatasia, Infantile|Hypophosphatasia, Mild	Henthorn (1992), Moore (1999), Moore (1999)
34375	ALPL	G	A	GG	Hypophosphatasia, Infantile	Henthorn (1992), Henthorn (1992), Herasse (2002), Herasse (2002)
64596	TBCE	T	A	TT	Hypoparathyroidism-Retardation-Dysmorphism Syndrome	Parvari (2002), Tian (2006)
11482	CASR	C	A	CC	Hypoparathyroidism, Familial Isolated	MacLeod, (2001), Watanabe (2002), Watanabe (2002), Sato (2002)
13711	PTH	A	G	AA	Hypoparathyroidism, Familial Isolated	Ahn (1986), Arnold (1990), Arnold (1990)
13712	PTH	A	G	AA	Hypoparathyroidism, Familial Isolated	Sunthornthepvarakul (1999)
13785	GCM2	C	A,T	CC	Hypoparathyroidism, Familial Isolated	Baumber (2005)
13786	GCM2	C	T	CC	Hypoparathyroidism, Familial Isolated	Thomee (2005)
12423	FXYD2	C	T	CC	Hypomagnesaemia, Renal	Meij (2000)
12494	CLDN16	G	A	GG	Hypomagnesaemia, Primary	Simon (1999)
12364	TRPM6	G	A	GG	Hypomagnesaemia with Secondary Hypocalcaemia	Chubanov (2004)
12376	TRPM6	C	T	CC	Hypomagnesaemia with Secondary Hypocalcaemia	Walder (1997), Walder (2002)
36562	CACNA1S	C	A,T	CC	Hypokalaemic periodic paralysis	Jurkat-Rott (1994), Elbaz (1995), Boerman (1995), Tricarico (1999), Tricarico (1999)
12326	GNRHR	T	C	TT	Hypogonadotropic Hypogonadism|Fertile Eunuch Syndrome	Roux (1997), De Roux (1997), De Roux (1999), Seminara (2000), Pitteloud (2001), Costa (2001), Pitteloud (2007), Pitteloud (2007)
12390	KISS1R	T	C	TT	Hypogonadotropic Hypogonadism	Bo-Abbas (2003), Seminara (2003)
12335	GNRHR	C	T	CC	Hypogonadotropic Hypogonadism	Caron (1999), De Roux (1999), Seminara (2000), Pitteloud (2007), Pitteloud (2007)
12853	NELF	T	C	TT	Hypogonadotropic Hypogonadism	Miura (2004), Miura (2004)
13928	ABCC8	C	G,T	CC	Hypoglycaemia, persistent hyperinsulinaemic	Thomas (1995), Nestorowicz (1996), Glaser (1999)
23198	BTK	T	C	T	Hypogammaglobulinemia	Rowlands, (1973), Puck (1988), Saffran (1994), Buckley (1994), Conley (1994)
23955	IGHM	C	T	CC	Hypogammaglobulinemia	Granados (2002)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
23956	IGHM	A	C	AA	Hypogammaglobulinemia	Yel (1996)
34916	FGFR3	C	G	CC	Hypochondroplasia	Prinos (1995), Bellus (1995), Fofanova (1998), Tsai (1999), GeneReviews (2013), Programa de Pós-Graduação em Ciências Genômicas e Biotecnologia de Universidade Católica de Brasília (2015), Centre for Mendelian Genomics of the University Medical Centre Ljubljana (2015), Emory Genetics (2016), GeneDx (2016), GeneDx (2017), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2017), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2018), Invitae (2018)
801961	FGFR3	C	A	CC	Hypochondroplasia	Bellus (1995), Angle (1998), Bober (1999), GeneReviews (2013), Emory Genetics (2015), GeneDx (2016)
34973	FGFR3	C	A	CC	Hypochondroplasia	McKusick , (1973), Bellus (1995), Prinos (1995), Bellus (1995), Prinos (1995), Prinster (1998), Ramaswami (1998), Angle (1998), Huggins (1999), Chitayat (1999)
800876	CASR	C	A	CC	Hypocalemia with Bartter Syndrome	MacLeod, (2001), Watanabe (2002), Watanabe (2002), Sato (2002)
11413	CASR	C	T	CC	Hypocalciuric Hypercalcaemia, Familial	Lovlie (1996), Lovlie (1996), Pearce (1996)
801960	CASR	C	T	CC	Hypoparathyrodism, Familial Isolated	Lovlie (1996), Lovlie (1996), Pearce (1996)
100624	MTTS2	C	A	?	Cerebellar Ataxia, Cataracts, and Diabetes Mellitus	Lynn (1998), Lynn (1998)
801998	MTND5	T	C	?	Possibly associated with Leigh Syndrome	Taylor (2002), Thorburn (2003), Lebon (2003), GeneReviews (2014)
800820	MTND5	G	A	?	Leigh Syndrome due to Mitochondrial Complex I Deficiency	Naini (2005), Blok (2007)
800821	MTND5	G	A	?	Leigh Syndrome due to Mitochondrial Complex I Deficiency	Naini (2005), Blok (2007)
100730	MTND5	G	A	?	Leigh Syndrome due to Mitochondrial Complex I Deficiency	Naini (2005), Blok (2007)
68611	MTND5	G	A	?	MELAS Syndrome	Santorelli (1997), Kirby (2003), Chol (2003), Sudo (2004), Dickerson (2005), Blok (2007), Shanske (2008), GeneReviews (2014), Children's Mercy Hospital and Clinics (2015)
5691	LPL	G	A	GG	Hypertriglyceridemia in Pregnancy|Elevated Total Cholesterol in Pregnancy	Kobayashi (1993), Kobayashi (1993)
3147	APOA5	G	A	GG	Hypertriglyceridemia	Marcais (2005)
11277	TSHR	T	C	TT	Hyperthyroidism	Tonacchera (2000)
989	PTGIS	A	G	AA	Hypertension	Nakayama (2002)
11620	INS	C	T	CC	Hyperproinsulinaemia, Familial	Robbins (1981), Robbins (1981), Shibasaki (1985), Barbetti (1990), Roder (1996), Collinet (1998)
33011	PTS	A	G	AA	Hyperphenylalaninemia	Liu (1998)
33018	PTS	C	T	CC	Hyperphenylalaninemia	Liu (1998)
29055	PAH	C	G,T	CC	Hyperphenylalaninemia	Eiken (1996)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
29215	PAH	A	G,T	AA	Hyperphenylalaninemia	Guldberg (1994)
29378	PAH	G	A	GG	Hyperphenylalaninemia	Zschocke (1994)
29386	PAH	T	C	TT	Hyperphenylalaninemia	Abadie (1993), Zschocke (1999)
29409	PAH	C	T	CC	Hyperphenylalaninemia	Economou-Petersen (1992)
13677	GALNT3	C	T	CC	Hyperostosis-Hyperphosphatemia Syndrome	Steinherz (1985), Topaz (2004), Frishberg (2005)
100509	SLC25A15	C	T	CC	Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome	Tsujino (2000), Miyamoto (2001), Nakajima (1988), Tessa (2009)
100510	SLC25A15	T	A	TT	Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome	Tessa (2009)
100508	SLC25A15	I	D	II	Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome	Camacho (1999)
44164	SCN4A	G	A	GG	Hyperkalaemic Periodic Paralysis|Paramyotonia Congenita	The same family had been described in the medical literature since (1934), Ptacek (1991), Sillen (1996), Kim (2001), Brancati (2003), Miller (2004), Hisama (2005), Hisama (2005)
44193	SCN4A	T	C	TT	Hyperkalaemic Periodic Paralysis|Paramyotonia Congenita	Rojas (1991), Rojas (1992), Heine (1993), Kelly (1997)
14111	GLUD1	G	A,C	GG	Hyperinsulinism-Hyperammonemia Syndrome	Stanley (1997), Stanley (1997), Miki (2000)
14114	GLUD1	C	A,T	CC	Hyperinsulinism-Hyperammonemia Syndrome	Stanley (1998)
79305	STAT3	G	A	GG	Hyper-IgE Syndrome	Minegishi (2007), Holland (2007)
79307	STAT3	C	T	CC	Hyper-IgE Syndrome	Holland (2007)
31841	MVK	G	A	GG	Hyper-IgD Syndrome	Houten (1999), Drenth (1999), Houten (2001), Cuisset (2001), D'Osualdo (2005)
31828	MVK	T	C	TT	Hyper-IgD Syndrome	Houten (1999), Drenth (1999), Cuisset (2001)
41760	GLDC	C	G	CC	Hyperglycinemia, Non-ketotic|Glycine Encephalopathy	Toone (2000), Toone (2001)
41763	GLDC	C	A	CC	Hyperglycinemia, Non-ketotic|Glycine Encephalopathy	Wendt , (1979), Kure (1992)
41778	GLDC	C	T	CC	Hyperglycinemia, Non-ketotic|Glycine Encephalopathy	Kure (1999), Toone (2001), Toone (2002)
30825	GK	A	G	AA	Hyperglycerolemia	Gaudet (2000)
41931	GLRA1	C	A,T	CC	Hyperekplexia	Shiang (1993), Rees (1994), Elmslie (1996)
3652	LDLR	C	G	CC	Hypercholesterolemia	Komuro (1987), Kotze (1990), Kotze (1990), Hobbs (1990), Kotze (1991), Vergotine (2001)
4026	LDLR	C	A	CC	Hypercholesterolemia	Khachadurian (1964), Lehrman (1987), Lehrman (1987), Lehrman (1987), Oppenheim (1991)
3605	LDLR	G	A	GG	Hypercholesterolemia	van Roggen (1991)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
3618	LDLR	G	A	GG	Hypercholesterolemia	Lee (1998)
4075	LDLR	G	A	GG	Hypercholesterolemia	Koivisto (1995), Koivisto (1995), Koivisto (2001)
4101	LDLR	G	A	GG	Hypercholesterolemia	Takada (2002), Takada (2002), Takada (2002), Takada (2002), Takada (2003), Sato (2004)
4126	LDLR	G	A	GG	Hypercholesterolemia	Leren (1994), Sun , (1995), Feussner , (1996)
3818	LDLR	T	A	TT	Hypercholesterolemia	Koivisto (1995), Koivisto (1995)
4135	LDLR	T	C	TT	Hypercholesterolemia	Gudnason (1997), Gudnason (1997)
16221	TJP2	T	C	TT	Hypercholanaemia	Carlton (2003)
62626	L1CAM	G	A	G	Hydrocephalus, X-linked	Gu (1996)
44726	NALP7	T	C	TT	Hydatidiform Mole, Recurrent	Murdoch (2006)
44728	NALP7	C	T	CC	Hydatidiform Mole, Recurrent	Murdoch (2006)
2303	LMNA	C	T	CC	Hutchinson-Gilford Progeria Syndrome	Eriksson (2003), De Sandre-Giovannoli (2003), Cao (2003), Hegele (2003), Goldman (2004), Navarro (2004), Wuyts (2005), Shumaker (2006)
800833	LMNA	C	T	CC	Restrictive Dermatopathy, Lethal	Eriksson (2003), De Sandre-Giovannoli (2003), Cao (2003), Hegele (2003), Goldman (2004), Navarro (2004), Wuyts (2005), Shumaker (2006)
27841	IDUA	C	T	CC	Hurler Syndrome	Scott (1992), Beesley (2001)
27894	IDUA	C	G	CC	Hurler Syndrome	Scott (1992), Alif (1999)
27836	IDUA	G	A	GG	Hurler Syndrome	Bunge (1994)
27847	IDUA	G	A	GG	Hurler Syndrome	Yamagishi (1996), Scott (1993)
27871	IDUA	G	C	GG	Hurler Syndrome	Bunge (1995)
27885	IDUA	G	A	GG	Hurler Syndrome	Scott (1992), McKusick (1965), Beesley (2001)
64100	HADH2	G	A	G	HSD10 Deficiency	Ofman (2003)
5853	CBS	G	A,T	GG	Homocystinuria	Urreizti (2006)
5877	CBS	C	T	CC	Homocystinuria	Ireland since (1971), Gu (1991), Hu (1993), Hu (1993), Gallagher (1995), Gallagher (1995)
5887	CBS	G	A	GG	Homocystinuria	Kruger (2003)
44100	GLI2	G	A	GG	Holoprosencephaly 9	Roessler (2003)
47601	PTCH	A	C	AA	Holoprosencephaly 7	Ribeiro (2006)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
44073	ZIC2	C	T	CC	Holoprosencephaly 5	Dubourg (2004)
44063	SIX3	T	C	TT	Holoprosencephaly 2	Wallis (1999), Laflamme (2004)
30081	HLCS	A	G	AA	Holocarboxylase Synthetase Deficiency	Narisawa (1982), Suzuki (1994), Yang (2000)
23912	NFKBIA	G	A,T	GG	Hodgkin Lymphoma	Osborne (2005)
32375	HMGCL	C	T	CC	HMG-CoA Lyase Deficiency	Ozand (1992), Mitchell (1998)
32385	HMGCL	C	T	CC	HMG-CoA Lyase Deficiency	Muroi (2000)
23734	TAP2	G	A,C,T	GG	HLA class I deficiency	Matamoros (2001)
800884	EDNRB	C	A	CC	Hirschsprung Disease	Puffenberger (1994)
7946	GJB6	G	A	GG	Hidrotic Ectodermal Dysplasia	Lamartine (2000)
7954	GJB6	C	G,T	CC	Hidrotic Ectodermal Dysplasia	Lamartine (2000)
6573	ZIC3	C	T	C	Heterotaxy	Ware (2004)
70766	TLR3	C	T	CC	Herpes Simplex Encephalitis	In 2 unrelated French children with herpes simplex encephalitis, Zhang (2007) identified heterozygosity for a 1660C-T transition in the TLR3 gene, resulting in a pro554-to-ser (P554S) substitution. Pro554 is highly conserved in a leucine-rich region of TL
55678	DTNBP1	G	A	GG	Hermansky-Pudlak Syndrome	Li (2003), Li (2003)
21272	HBG2	C	G	GG	Hereditary Persistence of Fetal Hemoglobin	Collins (1984)
70867	MLH1	I	D	II	Hereditary Nonpolyposis Colorectal Cancer, Type 2	Nystrom-Lahti (1995), Nystrom-Lahti (1995), Moisio (1996), Moisio (1996), Moisio (1996)
3338	TTN	G	A	GG	Hereditary Myopathy with Early Respiratory Failure	Nicolao (1999), Lange (2005)
13697	SLC34A3	C	A	CC	Hereditary Hypophosphatemic Rickets with Hypercalciuria	Lorenz-Depiereux (2006)
13696	SLC34A3	G	T	GG	Hereditary Hypophosphatemic Rickets with Hypercalciuria	Lorenz-Depiereux (2006)
71455	ACVRL1	D	I	DD	Hereditary Haemorrhagic Telangiectasia, Type 2	Abdalla (2003), Abdalla (2003), Lesca (2004), Lesca (2004)
58379	C1NH	G	A	GG	Hereditary Angioedema, Type II	Levy (1990), Davis (1992)
5997	LIPC	C	T	CC	Hepatic Lipase Deficiency	Hegele (1991), Hegele (1991), Ruel (2003)
4979	SERPIND1	G	A	GG	Heparin Cofactor II Deficiency	Corral (2004)
17211	F9	T	A	T	Hemophilia B (Leyden)	Veltkamp (1970), Briet (1982), Reitsma (1988), Reijnen (1992)
17221	F9	G	C	G	Hemophilia B (Brandenburg)	Reijnen (1992), Crossley (1992)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
17008	F9	C	T	C	Hemophilia B	Koeberl (1989), Ketterling (1991)
16762	F9	G	A	G	Hemophilia B	Denton (1988), Chen (1989), Poort (1989)
17208	F9	G	A	G	Hemophilia B	Fahner (1988), Hirosawa (1990), Crossley (1990)
16656	F9	T	C	T	Hemophilia B	Green (1993)
17188	F9	T	C	T	Hemophilia B	Ware (1988), Geddes (1989), Bottema (1990), Sarkar (1991), Chen (1991)
17307	F8	C	A,T	CC	Hemophilia A	Youssoufian (1988), Millar (1991)
17318	F8	G	A	GG	Hemophilia A	Higuchi (1991), Antonarakis (1995)
17336	F8	C	T	CC	Hemophilia A	Higuchi (1991), Antonarakis (1995)
17344	F8	G	A,T	GG	Hemophilia A	Gitschier (1988), Arai (1990), Aly (1992), Hoyer (1992)
17354	F8	G	A,C	GG	Hemophilia A	Higuchi (1991)
17358	F8	C	A,T	CC	Hemophilia A	Naylor (1993)
17708	F8	G	A	GG	Hemophilia A	Higuchi (1991)
18037	F8	G	T	GG	Hemophilia A	Antonarakis (1995)
101733	F8	G	A	GG	Hemophilia A	Khayat (2008)
49173	PRF1	G	A,C	GG	Hemophagocytic Lymphohistiocytosis, Familial	Stepp (1999), Ericson (2001)
21892	CD46	T	C	TT	Hemolytic Uremic Syndrome	Richards (2003), Richards (2003)
32299	GSS	T	C,G	TT	Hemolytic Anemia due to Glutathione Synthetase Deficiency of Erythrocytes	Shi (1996), Corrons (2001)
21302	AK1	C	T	CC	Hemolytic Anemia due to Adenylate Kinase Deficiency	Corrons (2003)
21305	AK1	T	C	TT	Hemolytic Anemia due to Adenylate Kinase Deficiency	Qualtieri (1997)
22232	PGK1	A	T	A	Hemolytic Anemia	Valentine , (1969), Cohen-Solal , (1994), Turner , (1995), Flanagan (2006), Flanagan (2006)
19317	HBA2	T	C	TT	Hemoglobin Variant	Clegg , (1971), Clegg (1971), Dayhoff (1972), Kosasih (1988), Hsia (1989), Hsia (1989), Laig (1990), Wen (1992)
800888	HBB	C	G,T	CC	Hemoglobin Variant	Neel (1950), Neel (1953), Ranney (1953), Allison, (1954), Ingram (1959), Krevans (1959), Ingram (1961), River (1961), Heller (1977), Fabry (1981), Ballas , (1982), Pearson , (1985), Boehm (1985), Fischel-Ghodsian (1990), Trabuchet (1991), Lane (1994), Lane (1994), Agarwal (2000), Agarwal , (2000), Modiano (2001), Modiano , (2001), Rihet (2004), Hedrick, (2004), Rihet , (2004), Wood (2005), Fairhurst (2005), Wood (2005), Fairhurst (2005)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
21413	SLC40A1	G	T	GG	Hemochromatosis, Type 4	Montosi (2001), Agarwal (2006), Agarwal (2006)
21392	TFR2	T	C,G	TT	Hemochromatosis, Type 3	Mattman (2002), Mattman (2002)
25770	SLC26A4	G	A	GG	Hearing loss	Yang (2005)
25833	TECTA	C	T	CC	Hearing Impairment, Nonsyndromic Sensorineural	Plantinga (2006)
32782	SLC6A19	G	A	GG	Hartnup Disorder	Shih (1971), Auray-Blais (2003), Seow (2004), Azmanov (2007), Bröer (2011), Laboratory for Molecular Medicine of Partners HealthCare Personalized Medicine (2014), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2015), GeneDx (2015), Genetic Services Laboratory of the University of Chicago (2016)
32783	SLC6A19	C	T	CC	Hartnup Disorder	Seow (2004)
40736	PANK2	C	T	CC	HARP syndrome	Higgins (1992), Ching (2002)
40690	PANK2	T	C	TT	HARP Syndrome	Orrell (1995), Houlden (2003)
26200	CTSC	T	C	TT	Haim-Munk Syndrome	Hart (2000), Hart (2000)
9651	ATP2C1	C	T	CC	Hailey-Hailey Disease	Sudbrak (2000), Li (2007)
67414	ENG	G	A,T	GG	Haemorrhagic Telangiectasia 1	Brusgaard (2004), Brusgaard (2004)
67451	ENG	C	A,T	CC	Haemorrhagic Telangiectasia 1	Gallione (2000)
67477	ENG	C	T	CC	Haemorrhagic telangiectasia 1	Gallione (2000)
20169	HBB	C	A,G,T	CC	Haemoglobin variant (Hemoglobin O also referred to as Hemoglobin O Arab and Hemoglobin Egypt)	Kamel (1967), Little (1980), GeneReviews (2014), Laboratory Corporation of America (2015), Laboratory Corporation of America (2016), ARUP Laboratories (2017), Quest Diagnostics of Nichols Institute San Juan Capistrano (2017)
20170	HBB	C	G	CC	Haemoglobin variant (Hemoglobin D also referred to as Hemoglobin D Punjab, Hemoglobin D Los Angeles, Hemoglobin D Chicago, Hemoglobin D North Carolina, Hemoglobin D Portugal and Hemoglobin Oak Ridge)	Benzer (1958), Ingram (1961), Stout (1964), Schneider (1968), Carrell (1969), Ozsoylu (1970), Riggs (1972), Bunn (1978), Trent (1982), Lehmann (1985), Husquinet (1986), Li , (1986), Harano (1987), Zeng (1989), Schnee (1990), GeneReviews (2014), Laboratory Corporation of America (2015), Counsyl (2016), Knight Diagnostic Laboratories of Oregon Health and Sciences University (2016), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2016), ARUP Laboratories (2017), Quest Diagnostics of Nichols Institute San Juan Capistrano (2016),
802011	HBB	C	A	CC	Beta Thalassemia, Dominant Inclusion Body Type	Weatherall (1973), Thein (1990), Quest Diagnostics of Nichols Institute San Juan Capistrano (2017)
19295	HBA2	G	A	GG	Haemoglobin H Disease	Morle (1995), Khan (2000)
52429	OAT	G	A	GG	Gyrate Atrophy with Pyridoxine-responsive Ornithinemia	Michaud (1995), Michaud (1995)
52443	OAT	C	T	CC	Gyrate Atrophy with Pyridoxine-responsive Ornithinemia	Ramesh (1988), Dougherty (1993), Dougherty (1993)
52451	OAT	A	C,G	AA	Gyrate Atrophy	Mitchell (1989), Martin (1989)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
78123	IGF1R	G	A	GG	Growth Retardation	Raile (2006)
11878	HESX1	T	C	TT	Growth Hormone Deficiency	Thomas (2001)
63843	RAB27A	C	G	CC	Griscelli Syndrome, Type 2	Menasche (2000), Bizario (2004)
63396	GLI3	G	A	GG	Greig Cephalopolysyndactyly Syndrome	Johnston (2005), Fujioka (2005), Johnston (2005), Ariga (2005), Baraitser (1983)
64191	BCS1L	A	G	AA	Gracile Syndrome	Visapaa (2002), Lonlay (2001)
58974	HPRT1	C	A	C	Gout, HPRT-related	Kelley (1984), Cariello (1988), Cariello (1988), Cariello (1988), Palella (1990)
12964	TG	G	T	GG	Goiter, Nonendemic Simple	Corral (1993), Vono-Toniolo (2005)
13015	TG	C	T	CC	Goiter, Familial, with Hypothyroidism	Targovnik (1989), Graaf (1999), Gutnisky (2004), Children's Hospital of Philadelphia (2014), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2015), Illumina Clinical Services Laboratory (2016), GeneDx (2017)
27527	HEXA	C	G,T	CC	GM2-Gangliosidosis, Adult	Proia (1989), Paw (1989), Navon (1990), Kappler (1990), Proia (1990), Mahuran (1993)
28654	GBE1	A	G	AA	Glycogen Storage Disease, Type IV	Bao (1996)
28664	GBE1	G	A,C	GG	Glycogen Storage Disease, Type IV	Bao (1996), Bruno (2004)
28418	GAA	C	A	CC	Glycogen Storage Disease, Type II	Shieh (1996), Lin (1998)
28435	GAA	T	G	TT	Glycogen Storage Disease, Type II	Huie (1994), Boerkoel (1995), Kroos (1995), Montalvo (2006)
71905	GAA	I	D	II	Glycogen Storage Disease, Type II	Hermans (1994), Kroos (1995)
28195	G6PC	C	T	CC	Glycogen Storage Disease, Type IA	Lei (1993), Lei (1994), Waddell (1990), Lei (1995), Qu (1996), Parvari (1997), Ekstein (2004)
28239	G6PC	C	T	CC	Glycogen Storage Disease, Type IA	Lei (1994), Chevalier-Porst (1996)
28218	G6PC	G	C	GG	Glycogen Storage Disease, Type IA	Chevalier-Porst (1996), Weston (2000)
28244	G6PC	G	T	GG	Glycogen Storage Disease, Type IA	Kajihara (1995), Sakamoto (1994), Akanuma (2000)
100590	G6PC	I	D	II	Glycogen Storage Disease, Type IA	Chevalier-Porst (1996)
801012	G6PC	I	D	II	Glycogen Storage Disease, Type IA	Chevalier-Porst (1996)
28631	PFKM	G	T	GG	Glycogen Storage Disease VII	Tarui (1965), Nakajima (1990), Nakajima (1997), Yamasaki (1991)
28634	PFKM	G	A	GG	Glycogen Storage Disease VII	Raben (1993), Sherman (1994), Ristow (1997), Vorgerd (1996)
28561	PYGL	T	C	TT	Glycogen Storage Disease VI	Burwinkel (1998)
28563	PYGL	C	A,G,T	CC	Glycogen Storage Disease VI	Chang (1998)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
28583	PHKA2	G	A	G	Glycogen Storage Disease Type IXa2	Burwinkel (1996)
28584	PHKA2	C	T	C	Glycogen Storage Disease Type IXa2	Burwinkel (1996), Hendrickx (1998)
28601	PHKA2	G	A	G	Glycogen Storage Disease Type IXa1	Fernandez (1969), Willems (1990), Berg (1995)
28529	AGL	A	G	AA	Glycogen Storage Disease Type 3A	Okubo (1998), Shaiu (2000)
28506	AGL	C	T	CC	Glycogen Storage Disease Type 3A	Santer (2001)
71907	AGL	I	D	II	Glycogen Storage Disease Type 3A	Shaiu (2000)
28263	SLC37A4	C	T	CC	Glycogen Storage Disease Ib	Hiraiwa (1999)
28274	SLC37A4	A	G	AA	Glycogen Storage Disease Ib	Kure (1998)
28297	SLC37A4	C	A	CC	Glycogen Storage Disease Ib	Gerin (1997)
32267	GNMT	A	G	AA	Glycine N-methyltransferase Deficiency	Augoustides-Savvopoul (2003)
32268	GNMT	C	A	CC	Glycine N-methyltransferase Deficiency	Mudd (2001), Luka (2002)
30062	FTCD	G	A	GG	Glutamate Formiminotransferase Deficiency	Hilton (2003)
30063	FTCD	C	G	CC	Glutamate Formiminotransferase Deficiency	Hilton (2003)
20403	G6PD	A	G	A	Glucose-6-phosphate Dehydrogenase Variant	Beutler (1989)
20407	G6PD	C	A,T	C	Glucose-6-phosphate Dehydrogenase Variant	Beutler (1989)
20422	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Weimer (1993)
20423	G6PD	T	A	T	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1996)
20424	G6PD	G	A	G	Glucose-6-phosphate Dehydrogenase Deficiency	Kirkman (1964), Ben-Ishay (1969), Lenzerini (1969), Testa (1980), Morelli (1984), Vita (1989), Kuhl (1990), Kurdi-Haidar (1990), Matsuoka (2003), Corcoran (1992), Kaplan (1997), Kaplan (2001)
20425	G6PD	C	A,T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Mesbah-Namin (2002)
20426	G6PD	C	A,T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Galanello (1997)
20427	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Weimer (1998)
20428	G6PD	T	C	T	Glucose-6-phosphate Dehydrogenase Deficiency	Du (1985), Chiu (1993)
20429	G6PD	T	C	T	Glucose-6-phosphate Dehydrogenase Deficiency	Hirono (1995)
20430	G6PD	A	G	A	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1997)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
20431	G6PD	G	C	G	Glucose-6-phosphate Dehydrogenase Deficiency	Kaeda (1995), Ranney (1990)
20432	G6PD	A	G	A	Glucose-6-phosphate Dehydrogenase Deficiency	Nafa (1993), Niazi (1996)
20433	G6PD	G	A	G	Glucose-6-phosphate Dehydrogenase Deficiency	Drousiotou (2004)
20434	G6PD	C	G	C	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (2002)
20435	G6PD	G	A	G	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (2002)
20436	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1988)
20437	G6PD	G	A	G	Glucose-6-phosphate Dehydrogenase Deficiency	Hirono (1997)
20438	G6PD	A	T	A	Glucose-6-phosphate Dehydrogenase Deficiency	Laosombat (2005)
20439	G6PD	A	G	A	Glucose-6-phosphate Dehydrogenase Deficiency	Ganczakowski (1995)
20440	G6PD	T	C	T	Glucose-6-phosphate Dehydrogenase Deficiency	Rovira (1995)
20441	G6PD	A	G	A	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1993)
20442	G6PD	G	A	G	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1993)
20443	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1993)
20444	G6PD	G	A	G	Glucose-6-phosphate Dehydrogenase Deficiency	Jiang (2006)
20445	G6PD	G	C	G	Glucose-6-phosphate Dehydrogenase Deficiency	Maeda (1992)
20447	G6PD	G	A	G	Glucose-6-phosphate Dehydrogenase Deficiency	Maeda (1992)
20448	G6PD	G	A	G	Glucose-6-phosphate Dehydrogenase Deficiency	Corcoran (1992)
20449	G6PD	A	T	A	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1997)
20450	G6PD	T	C	T	Glucose-6-phosphate Dehydrogenase Deficiency	Chen (1998)
20451	G6PD	A	C,G	A	Glucose-6-phosphate Dehydrogenase Deficiency	Ganczakowski (1995)
20452	G6PD	A	C,G	A	Glucose-6-phosphate Dehydrogenase Deficiency	Martinez (2001)
20453	G6PD	C	A,T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1993)
20454	G6PD	T	G	T	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (2002)
20455	G6PD	G	A	G	Glucose-6-phosphate Dehydrogenase Deficiency	Zarza (1997)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
20456	G6PD	G	T	G	Glucose-6-phosphate Dehydrogenase Deficiency	Menounos (2000)
20457	G6PD	G	C	G	Glucose-6-phosphate Dehydrogenase Deficiency	Drousiotou (2004)
20458	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Usanga (1977), Luzzatto (1979), Vulliamy (1988)
20459	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Panich (1972), Tang (1992), Matsuoka (2004)
20460	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1993)
20461	G6PD	T	C	T	Glucose-6-phosphate Dehydrogenase Deficiency	Tang (1992)
20462	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Ganczakowski (1995)
20463	G6PD	G	A	G	Glucose-6-phosphate Dehydrogenase Deficiency	Roos (1999)
20464	G6PD	A	G	A	Glucose-6-phosphate Dehydrogenase Deficiency	Chen (1996)
20465	G6PD	G	C	G	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1997)
20466	G6PD	T	C	T	Glucose-6-phosphate Dehydrogenase Deficiency	Hirono (1994)
20467	G6PD	T	A	T	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (2002)
20469	G6PD	C	G	C	Glucose-6-phosphate Dehydrogenase Deficiency|Anemia, Nonspherocytic Hemolytic, Due to G6PD Deficiency	Beutler (1992)
20470	G6PD	T	C	T	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1993)
20471	G6PD	C	A,G	C	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (1991)
20472	G6PD	A	C,G	A	Glucose-6-phosphate Dehydrogenase Deficiency	Poggi (1989), Town (1990)
20473	G6PD	G	A	G	Glucose-6-phosphate Dehydrogenase Deficiency	Jablonska-Skwiecinska (1999)
20474	G6PD	C	A,T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (1992)
20476	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Hirono (1997)
20477	G6PD	T	C	T	Glucose-6-phosphate Dehydrogenase Deficiency	Zimmerman (1997)
20478	G6PD	G	C	G	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (1991)
20479	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Costa (2000)
20480	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Xu (1995)
20481	G6PD	G	A	G	Glucose-6-phosphate Dehydrogenase Deficiency	Taki (2001)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
20482	G6PD	A	G	A	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (2002)
20483	G6PD	G	A	G	Glucose-6-phosphate Dehydrogenase Deficiency	Mason (1995)
20484	G6PD	T	C	T	Glucose-6-phosphate Dehydrogenase Deficiency	Cai (2001)
20485	G6PD	T	A	T	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (1992)
20486	G6PD	C	G	C	Glucose-6-phosphate Dehydrogenase Deficiency	Lenzerini (1969), Rattazzi (1969), De Vita (1989)
20487	G6PD	C	A,G,T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Kawamoto (2006)
20488	G6PD	G	T	G	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1997)
20489	G6PD	G	A	G	Glucose-6-phosphate Dehydrogenase Deficiency	Okano (2001)
20491	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Poon (1988), Beutler (1991), Nuchprayoon (2005), Matsuoka (2005), Illumina Clinical Services Laboratory (2013), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2015), Counsyl (2017)
20492	G6PD	C	A	C	Glucose-6-phosphate Dehydrogenase Deficiency	Xu (1995)
20493	G6PD	C	A	C	Glucose-6-phosphate Dehydrogenase Deficiency	Xu (1995)
20494	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (2002)
20495	G6PD	C	G	C	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1997)
20496	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1997)
20497	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Ahluwalia (1992)
20498	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1997)
20499	G6PD	A	G	A	Glucose-6-phosphate Dehydrogenase Deficiency	Iancovici-Kidon (2000)
20500	G6PD	G	A,T	G	Glucose-6-phosphate Dehydrogenase Deficiency	Weimer (1998)
20501	G6PD	G	T	G	Glucose-6-phosphate Dehydrogenase Deficiency	Xu (1995)
20502	G6PD	T	C	T	Glucose-6-phosphate Dehydrogenase Deficiency	Jablonska-Skwiecinska (1999)
20503	G6PD	G	A	G	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1993)
20504	G6PD	C	G	C	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1997)
20505	G6PD	C	A	C	Glucose-6-phosphate Dehydrogenase Deficiency	Cappellini (1996)
20506	G6PD	C	A	C	Glucose-6-phosphate Dehydrogenase Deficiency	Miwa (1996)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
20507	G6PD	G	A	G	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (1992)
20508	G6PD	A	G	A	Glucose-6-phosphate Dehydrogenase Deficiency	Patrinos (2005)
20509	G6PD	G	A	G	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1997)
20510	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1997)
20511	G6PD	G	T	G	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (1991)
20512	G6PD	G	C	G	Glucose-6-phosphate Dehydrogenase Deficiency	Efferth (2000)
20513	G6PD	T	C	T	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (2002)
20514	G6PD	A	G	A	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (1995)
20515	G6PD	T	C	T	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (1995)
20516	G6PD	A	T	A	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (2002)
20517	G6PD	A	G	A	Glucose-6-phosphate Dehydrogenase Deficiency	Xu (1995)
20518	G6PD	A	G	A	Glucose-6-phosphate Dehydrogenase Deficiency	Hirono (1989)
20519	G6PD	C	A	C	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1997)
20520	G6PD	G	C	G	Glucose-6-phosphate Dehydrogenase Deficiency	Zarza (1997)
20521	G6PD	T	C	T	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (1986), Hirono (1989)
20522	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Hirono (1989)
20523	G6PD	G	A	G	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (1992)
20524	G6PD	T	C	T	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1997)
20525	G6PD	T	C	T	Glucose-6-phosphate Dehydrogenase Deficiency	Xu (1995)
20526	G6PD	G	C	G	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (1995)
20527	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Filosa, (1989)
20528	G6PD	C	G	C	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (1992)
20529	G6PD	G	A	G	Glucose-6-phosphate Dehydrogenase Deficiency	Filosa (1994)
20530	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (1991)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
20531	G6PD	T	C	T	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (2002)
20532	G6PD	G	T	G	Glucose-6-phosphate Dehydrogenase Deficiency	Manco (2005)
20533	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Rovira (1995)
20534	G6PD	T	G	T	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1997)
20535	G6PD	G	C	G	Glucose-6-phosphate Dehydrogenase Deficiency	Grabowska (2004)
20536	G6PD	G	A	G	Glucose-6-phosphate Dehydrogenase Deficiency	van Wijk (2004)
20537	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (1992)
20538	G6PD	C	G	C	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1997)
20539	G6PD	C	A	C	Glucose-6-phosphate Dehydrogenase Deficiency	Hirono (1989)
20540	G6PD	T	C	T	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (2002)
20541	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Hirono (1992)
20542	G6PD	G	T	G	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1993)
20543	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Iwai (2001)
20544	G6PD	A	C	A	Glucose-6-phosphate Dehydrogenase Deficiency	Saad (1997)
20546	G6PD	C	G	C	Glucose-6-phosphate Dehydrogenase Deficiency|Anemia, Nonspherocytic Hemolytic, Due to G6PD Deficiency	Prchal (1985), Beutler (1992)
20547	G6PD	G	A	G	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1993)
20548	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1988)
20549	G6PD	T	C	T	Glucose-6-phosphate Dehydrogenase Deficiency	Miwa (1996)
20550	G6PD	C	G	C	Glucose-6-phosphate Dehydrogenase Deficiency	Calabro (1993)
20551	G6PD	A	T	A	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1997)
20552	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Vives-Corrons (1990)
20553	G6PD	G	A	G	Glucose-6-phosphate Dehydrogenase Deficiency	Perng (1992), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2013), Center for Pediatric Genomic Medicine of Children's Mercy Hospital and Clinics (2014), ARUP Laboratories (2016), Invitae (2017)
20554	G6PD	C	G	C	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (2002)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
20555	G6PD	C	G	C	Glucose-6-phosphate Dehydrogenase Deficiency	Calabro (1993)
20556	G6PD	C	A,G,T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Stevens (1990), Tang (1992)
20557	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Ren (2001)
20558	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Panich (1973), Panich (1980), Shatskaya (1980), Fujii (1981), Du (1988), Zuo (1990), Chiu (1991), Bean (2013), Emory Genetics Laboratory of Emory University (2013)
20559	G6PD	G	T	G	Glucose-6-phosphate Dehydrogenase Deficiency	Rodrigues (2002)
20560	G6PD	G	A	G	Glucose-6-phosphate Dehydrogenase Deficiency	Hirono (1997)
20561	G6PD	G	C	G	Glucose-6-phosphate Dehydrogenase Deficiency	Alfinito (1997)
20562	G6PD	G	C	G	Glucose-6-phosphate Dehydrogenase Deficiency	Barisic (2005)
20563	G6PD	C	T	C	Glucose-6-phosphate Dehydrogenase Deficiency	Beutler (2002)
20564	G6PD	C	A	C	Glucose-6-phosphate Dehydrogenase Deficiency	Baronciani (1993)
20565	G6PD	G	A	G	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1997)
20566	G6PD	A	C	A	Glucose-6-phosphate Dehydrogenase Deficiency	Vulliamy (1997)
20567	G6PD	T	C	T	Glucose-6-phosphate Dehydrogenase Deficiency	Efferth (2004)
12215	GCCR	A	T	AA	Glucocorticoid Resistance, Generalized	Karl (1996)
12214	GCCR	C	T	CC	Glucocorticoid Resistance, Familial	Ruiz (2001)
12217	GCCR	T	A	TT	Glucocorticoid Resistance, Familial	Vingerhoeds (1976), Chrousos (1982), Lipsett (1985), Hurley (1991)
12218	GCCR	C	T	CC	Glucocorticoid Resistance, Familial	Ruiz (2001)
12219	GCCR	C	T	CC	Glucocorticoid Resistance, Familial	Malchoff (1993)
12220	GCCR	A	C,G	AA	Glucocorticoid Resistance, Familial	Vottero (2002)
12221	GCCR	A	G	AA	Glucocorticoid Resistance, Familial	Charmandari (2005)
11327	MC2R	T	C	TT	Glucocorticoid Deficiency	Fluck (2002)
11348	MC2R	G	C	GG	Glucocorticoid deficiency	Tsigos (1993)
11327	MC2R	T	C	TT	Glucocorticoid Deficiency	Fluck (2002)
11344	MC2R	G	C	GG	Glucocorticoid deficiency	Weber (1995)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
11345	MC2R	C	A	CC	Glucocorticoid deficiency	Clark (1993)
11346	MC2R	C	G,T	CC	Glucocorticoid deficiency	Clark (1998)
11347	MC2R	C	T	CC	Glucocorticoid deficiency	Naville (1996)
11348	MC2R	G	C	GG	Glucocorticoid deficiency	Tsigos (1993)
11349	MC2R	G	A	GG	Glucocorticoid deficiency	Weber (1994)
11350	MC2R	A	T	AA	Glucocorticoid deficiency	Naville (2000)
11351	MC2R	G	A,T	GG	Glucocorticoid deficiency	Fluck (2002)
11352	MC2R	G	A	GG	Glucocorticoid deficiency	Naville (2000)
11353	MC2R	C	A,T	CC	Glucocorticoid deficiency	Penhoat (2002)
11354	MC2R	C	A,T	CC	Glucocorticoid deficiency	Weber (1995)
11355	MC2R	G	A,C,T	GG	Glucocorticoid deficiency	Clark (1998)
11356	MC2R	A	G,T	AA	Glucocorticoid deficiency	Naville (2000)
11357	MC2R	G	A,T	GG	Glucocorticoid deficiency	Tsigos (1993)
11358	MC2R	C	G	CC	Glucocorticoid deficiency	Penhoat (2002)
11360	MC2R	C	A,G,T	CC	Glucocorticoid deficiency	Naville (1996)
11361	MC2R	G	T	GG	Glucocorticoid deficiency	Wu (1998)
53926	CYP1B1	C	T	CC	Glaucoma, primary congenital	Stoilov (1998), Chitsazian (2007)
52000	MYOC	C	A	CC	Glaucoma 1A, Primary Open Angle, Digenic	Vincent (2002)
52049	MYOC	G	A	GG	Glaucoma 1A, Primary Open Angle	Australia in the early (1800), Stone (1997), Stone (1997), Stone, (1999), Craig (2001), Baird (2001), Baird (2003)
52051	MYOC	G	A	GG	Glaucoma 1A, Primary Open Angle	Adam (1997), Suzuki (1997)
52064	MYOC	A	G	AA	Glaucoma 1A, Primary Open Angle	Vasconcellos (2000)
52066	MYOC	A	G	AA	Glaucoma 1A, Primary Open Angle	Sheffield (1993), Stone (1997), Stone (1997), Alward (1998), Alward (1998)
52077	MYOC	G	T	GG	Glaucoma 1A, Primary Open Angle	Adam (1997), Brezin (1998), Brezin (1998)
52021	MYOC	C	A	CC	Glaucoma 1A, Open Angle|Glaucoma 3A, Primary Congenital, Digenic	Sripriya (2004), Kaur (2005)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
52836	OPTN	G	A	GG	Glaucoma 1, Open Angle, E	Rezaie (2002)
4672	ITGB3	C	T	CC	Glanzmann thrombasthenia 2	French (1997)
4680	ITGB3	T	C	TT	Glanzmann thrombasthenia 2	French (1997)
4693	ITGB3	G	T	GG	Glanzmann thrombasthenia 2	Ambo (1998)
4696	ITGB3	G	A	GG	Glanzmann thrombasthenia 2	Ambo (1998)
4688	ITGB3	T	C	TT	Glanzmann thrombasthenia 1	Jallu (2005)
4689	ITGB3	G	A	GG	Glanzmann thrombasthenia 1	Ruan (1999)
4691	ITGB3	T	C	TT	Glanzmann thrombasthenia 1	Ruan (1999)
4666	ITGB3	T	G	TT	Glanzmann Thrombasthenia	Basani , (1997), Peretz (2006)
4644	ITGB3	A	C	AA	Glanzmann Thrombasthenia	Ambo (1998), Corinaldesi (2009), Kannan (2009), Favaloro (2010)
4660	ITGB3	C	T	CC	Glanzmann Thrombasthenia	Negrier (1998)
4661	ITGB3	T	G	TT	Glanzmann Thrombasthenia	Peretz (2006)
4662	ITGB3	C	T	CC	Glanzmann Thrombasthenia	Vinciguerra (1995)
4663	ITGB3	C	T	CC	Glanzmann Thrombasthenia	D'Andrea (2002)
4664	ITGB3	G	A	GG	Glanzmann Thrombasthenia	Peretz (2006)
4665	ITGB3	A	G	AA	Glanzmann Thrombasthenia	Nair (2005)
4666	ITGB3	T	G	TT	Glanzmann Thrombasthenia	Basani (1997), Peretz (2006)
4667	ITGB3	G	A	GG	Glanzmann Thrombasthenia	French (1997)
4668	ITGB3	G	T	GG	Glanzmann Thrombasthenia	Ginsberg (1986), Loftus (1990)
4669	ITGB3	T	C	TT	Glanzmann Thrombasthenia	Nair (2004)
4670	ITGB3	A	G	AA	Glanzmann Thrombasthenia	Gonzalez-Manchon (2004)
4673	ITGB3	C	T	CC	Glanzmann Thrombasthenia	Peretz (2006)
4674	ITGB3	G	A	GG	Glanzmann Thrombasthenia	Fullard (2001)
4675	ITGB3	T	C	TT	Glanzmann Thrombasthenia	Nurden (2002)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
4676	ITGB3	C	T	CC	Glanzmann Thrombasthenia	Lanza (1992)
4677	ITGB3	G	A	GG	Glanzmann Thrombasthenia	Bajt (1992), Bajt (1992)
4678	ITGB3	G	A	GG	Glanzmann Thrombasthenia	French (1997)
4679	ITGB3	A	T	AA	Glanzmann Thrombasthenia	D'Andrea (2002)
4681	ITGB3	C	A	CC	Glanzmann Thrombasthenia	Peretz (2006)
4682	ITGB3	A	C	AA	Glanzmann Thrombasthenia	Nair (2005)
4683	ITGB3	T	A	TT	Glanzmann Thrombasthenia	Tanaka (2005)
4684	ITGB3	G	A	GG	Glanzmann Thrombasthenia	Chen (1993)
4685	ITGB3	G	A	GG	Glanzmann Thrombasthenia	Peretz (2006)
4687	ITGB3	G	T	GG	Glanzmann Thrombasthenia	Peretz (2006)
4690	ITGB3	G	A	GG	Glanzmann Thrombasthenia	Nair (2002)
4692	ITGB3	T	C	TT	Glanzmann Thrombasthenia	Ruiz (2001)
4694	ITGB3	T	C	TT	Glanzmann Thrombasthenia	D'Andrea (2002)
4695	ITGB3	T	G	TT	Glanzmann thrombasthenia	Nelson (2006)
4697	ITGB3	G	T	GG	Glanzmann Thrombasthenia	The patient was found to be homozygous for a (1846), Ferrer (1998)
4698	ITGB3	A	T	AA	Glanzmann Thrombasthenia	Vinciguerra (2001)
4700	ITGB3	C	T	CC	Glanzmann Thrombasthenia	Wang (1997), Wang (1997)
4701	ITGB3	T	C	TT	Glanzmann Thrombasthenia	Chen (1992)
4702	ITGB3	C	A	CC	Glanzmann Thrombasthenia	Srivastava (2003)
4703	ITGB3	G	T	GG	Glanzmann Thrombasthenia	Tanaka (2003)
4705	ITGB3	G	T	GG	Glanzmann Thrombasthenia	French (1997)
4706	ITGB3	G	A	GG	Glanzmann Thrombasthenia	French (1997)
4707	ITGB3	G	A	GG	Glanzmann Thrombasthenia	Jin (1996)
64326	SLC12A3	C	T	CC	Gitelman Syndrome	Simon (1996)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
64306	SLC12A3	G	T	GG	Gitelman Syndrome	Cruz (2001)
64269	SLC12A3	C	T	CC	Gitelman Syndrome	Simon (1996)
64274	SLC12A3	T	C	TT	Gitelman Syndrome	Simon (1996)
64289	SLC12A3	G	A	GG	Variant of Unknown Significance (questionable association with Gitelman Syndrome)	De Jong (2002)
64302	SLC12A3	A	T	AA	Gitelman Syndrome	Maki (2004), Akoi (2008)
64303	SLC12A3	C	T	CC	Gitelman Syndrome	Maki (2004)
64304	SLC12A3	G	A	GG	Gitelman Syndrome	Mastroianni (1996)
64305	SLC12A3	C	T	CC	Gitelman Syndrome	Lin (2005)
64307	SLC12A3	A	T	AA	Gitelman Syndrome	Cruz (2001)
64308	SLC12A3	G	A	GG	Gitelman Syndrome	Syren (2002)
64309	SLC12A3	C	T	CC	Gitelman Syndrome	Syren (2002)
64310	SLC12A3	T	C	TT	Gitelman Syndrome	Syren (2002)
64311	SLC12A3	C	T	CC	Gitelman Syndrome	Cruz (2001)
64312	SLC12A3	C	A	CC	Gitelman Syndrome	Monkawa (2000)
64313	SLC12A3	G	A	GG	Gitelman Syndrome	Mastroianni (1996)
64314	SLC12A3	G	A	GG	Gitelman Syndrome	Cruz (2001)
64315	SLC12A3	T	C	TT	Gitelman Syndrome	Lemmink (1998)
64316	SLC12A3	G	A	GG	Gitelman Syndrome	Lemmink (1998)
64317	SLC12A3	G	A	GG	Gitelman Syndrome	Lemmink (1998)
64318	SLC12A3	C	A	CC	Gitelman Syndrome	Lin (2005)
64319	SLC12A3	A	G	AA	Gitelman Syndrome	Cruz (2001)
64320	SLC12A3	A	C	AA	Gitelman Syndrome	Melander (2000)
64321	SLC12A3	C	T	CC	Gitelman Syndrome	Cruz (2001)
64322	SLC12A3	C	T	CC	Gitelman Syndrome	Reissinger (2002)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
64323	SLC12A3	G	T	GG	Gitelman Syndrome	Syren (2002)
64324	SLC12A3	C	T	CC	Gitelman Syndrome	Cruz (2001)
64325	SLC12A3	C	T	CC	Gitelman Syndrome	Cruz (2001)
64326	SLC12A3	C	T	CC	Gitelman Syndrome	Simon (1996)
64327	SLC12A3	G	T	GG	Gitelman Syndrome	Syren (2002)
64328	SLC12A3	C	T	CC	Gitelman Syndrome	Cruz (2001)
64329	SLC12A3	C	T	CC	Gitelman Syndrome	Reissinger (2002)
64330	SLC12A3	A	C	AA	Gitelman Syndrome	Yoo (2003)
64331	SLC12A3	C	G	CC	Gitelman Syndrome	Lin (2005)
64332	SLC12A3	G	A	GG	Gitelman Syndrome	Mastroianni (1996)
64333	SLC12A3	G	A	GG	Gitelman Syndrome	Syren (2002)
64334	SLC12A3	G	A	GG	Gitelman Syndrome	Syren (2002)
64335	SLC12A3	A	G	AA	Gitelman Syndrome	Mastroianni (1996)
64336	SLC12A3	G	A	GG	Gitelman Syndrome	Simon (1996)
64337	SLC12A3	G	T	GG	Gitelman Syndrome	Simon (1996)
64338	SLC12A3	A	T	AA	Gitelman Syndrome	Cruz (2001)
64339	SLC12A3	T	C	TT	Gitelman Syndrome	De Jong (2002)
64340	SLC12A3	T	C	TT	Gitelman Syndrome	Mastroianni (1996)
64341	SLC12A3	C	T	CC	Gitelman Syndrome	Cruz (2001)
64342	SLC12A3	C	A	CC	Gitelman Syndrome	Lemmink (1998)
64343	SLC12A3	C	T	CC	Gitelman Syndrome	Monkawa (2000)
64344	SLC12A3	C	T	CC	Gitelman Syndrome	Yoo (2003)
64345	SLC12A3	G	A	GG	Gitelman Syndrome	Monkawa (2000)
64346	SLC12A3	C	T	CC	Gitelman Syndrome	Simon (1996)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
64347	SLC12A3	G	A	GG	Gitelman Syndrome	Lemmink (1998)
64348	SLC12A3	C	G	CC	Gitelman Syndrome	Syren (2002)
64349	SLC12A3	C	T	CC	Gitelman Syndrome	Cruz (2001)
64350	SLC12A3	T	C	TT	Gitelman Syndrome	Takeuchi (1996)
64351	SLC12A3	G	T	GG	Gitelman Syndrome	Simon (1996)
64352	SLC12A3	C	G	CC	Gitelman Syndrome	Cruz (2001)
64353	SLC12A3	C	T	CC	Gitelman Syndrome	Yahata (1999)
64354	SLC12A3	C	T	CC	Gitelman Syndrome	Cruz (2001)
64355	SLC12A3	C	G	CC	Gitelman Syndrome	Lemmink (1998)
64356	SLC12A3	C	T	CC	Gitelman Syndrome	Lemmink (1998)
64357	SLC12A3	G	A	GG	Gitelman Syndrome	Simon (1996)
64358	SLC12A3	G	T	GG	Gitelman Syndrome	Simon (1996)
64359	SLC12A3	G	A	GG	Gitelman Syndrome	Terui (2006)
64360	SLC12A3	G	A	GG	Gitelman Syndrome	Syren (2002)
64361	SLC12A3	C	A	CC	Gitelman Syndrome	Lin (2004)
64362	SLC12A3	G	T	GG	Gitelman Syndrome	Cruz (2001)
64363	SLC12A3	G	C	GG	Gitelman Syndrome	Mastroianni (1996)
64364	SLC12A3	T	G	TT	Gitelman Syndrome	Lemmink (1998)
64365	SLC12A3	G	A	GG	Gitelman Syndrome	Simon (1996)
64366	SLC12A3	G	A	GG	Gitelman Syndrome	Lin (2005)
64367	SLC12A3	T	A	TT	Gitelman Syndrome	Monkawa (2000)
64368	SLC12A3	T	C	TT	Gitelman Syndrome	Simon (1996)
64369	SLC12A3	C	A	CC	Gitelman Syndrome	Syren (2002)
64370	SLC12A3	C	T	CC	Gitelman Syndrome	Lemmink (1998)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
64371	SLC12A3	G	A	GG	Gitelman Syndrome	Lin (2005)
64372	SLC12A3	G	A	GG	Gitelman Syndrome	Kurschat (2003)
64373	SLC12A3	G	A	GG	Gitelman Syndrome	Simon (1996)
64374	SLC12A3	A	G	AA	Gitelman Syndrome	Syren (2002)
64375	SLC12A3	C	T	CC	Gitelman Syndrome	Simon (1996)
64376	SLC12A3	G	A	GG	Gitelman Syndrome	Syren (2002)
64377	SLC12A3	C	T	CC	Gitelman Syndrome	Fukuyama (2003)
64380	SLC12A3	G	A	GG	Gitelman Syndrome	Lemmink (1998)
43721	GAN	G	A	GG	Giant Axonal Neuropathy	Bomont (2000)
37180	PRNP	C	T	CC	Gerstmann-Straeussler syndrome	Hsiao (1989), Goldgaber (1989), Doh-ura (1989), Goldfarb (1990), Doh-ura (1990), Doh-ura (1990), Speer (1991), Goldhammer (1993), Mishra (2002), Webb (2008)
42622	CACNB4	C	A	CC	Generalized Idiopathic Epilepsy|Episodic Ataxia, Type 5	Escayg (2000), Escayg (2000)
42622	CACNB4	C	A	CC	Generalized Idiopathic Epilepsy|Episodic Ataxia, Type 5	Escayg (2000), Escayg (2000)
42230	GABRG2	A	T	AA	Generalized Epilepsy with Febrile Seizures Plus, Type 3	Baulac (2001)
42231	GABRG2	C	T	CC	Generalized Epilepsy with Febrile Seizures Plus, Type 3	Harkin (2002)
40877	SCN1A	G	A	GG	Generalized Epilepsy with Febrile Seizures Plus, Type 2|Intractable Childhood Epilepsy with Generalized Tonic-clonic Seizures	Fujiwara (2003)
40854	SCN1A	A	G	AA	Generalized Epilepsy with Febrile Seizures Plus, Type 2	Sugawara (2001)
40862	SCN1A	C	A	CC	Generalized Epilepsy with Febrile Seizures Plus, Type 2	Fujiwara (2003)
800903	SCN1A	G	A	GG	Generalized Epilepsy with Febrile Seizures Plus, Type 2	Fujiwara (2003)
40811	SCN1A	T	A,C	TT	Generalized epilepsy with febrile seizures plus	Annesi (2003)
40813	SCN1A	G	A	GG	Generalized epilepsy with febrile seizures plus	Barela (2006)
40836	SCN1A	A	G	AA	Generalized epilepsy with febrile seizures plus	Escayg (2001)
40845	SCN1A	T	G	TT	Generalized epilepsy with febrile seizures plus	Abou-Khalil (2001)
40849	SCN1A	C	G	CC	Generalized epilepsy with febrile seizures plus	Wallace (2001)
40868	SCN1A	G	C	GG	Generalized epilepsy with febrile seizures plus	Wallace (2001)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
40869	SCN1A	G	A	GG	Generalized epilepsy with febrile seizures plus	Lossin (2003)
40878	SCN1A	T	C	TT	Generalized epilepsy with febrile seizures plus	Pineda-Trujillo (2005)
40885	SCN1A	A	G	AA	Generalized epilepsy with febrile seizures plus	Annesi (2003)
40887	SCN1A	C	A	CC	Generalized epilepsy with febrile seizures plus	Spampanato (2004)
40892	SCN1A	T	C	TT	Generalized epilepsy with febrile seizures plus	Combi (2007)
43881	SCN1B	C	G	CC	Generalized Epilepsy With Febrile Seizures	Wallace (1998), Wallace (1998), Tammaro (2002), Wallace (2002)
43883	SCN1B	G	A	GG	Generalized Epilepsy With Febrile Seizures	Scheffer (2007)
43884	SCN1B	C	T	CC	Generalized Epilepsy With Febrile Seizures	Scheffer (2007)
43885	SCN1B	A	C	AA	Generalized Epilepsy With Febrile Seizures	Audenaert (2003), Audenaert (2003)
40823	SCN1A	G	A,T	GG	Generalized epilepsy of infancy	Ebach (2005)
42605	KCNMA1	T	C	TT	Generalized Epilepsy and Paroxysmal Dyskinesia	Du (2005), Du (2005)
42605	KCNMA1	T	C	TT	Generalized Epilepsy and Paroxysmal Dyskinesia	Du (2005), Du (2005)
10374	ENPP1	G	C	GG	Generalized Arterial Calcification of Infancy	Rutsch (2003)
10383	ENPP1	G	T	GG	Generalized Arterial Calcification of Infancy	Rutsch (2001)
40774	SCN1A	T	A	TT	Generalised Epilepsy with Febrile Seizures Plus, Type 2	Wallace (2001)
40815	SCN1A	G	A,T	GG	Generalised epilepsy with febrile seizures plus 2	Moulard (1999), Escayg (2000)
40867	SCN1A	C	A,T	CC	Generalised epilepsy with febrile seizures plus 2	Baulac (1999), Escayg (2000)
40886	SCN1A	C	A	CC	Generalised epilepsy with febrile seizures plus 2	Nagao (2005)
78784	TACSTD2	C	T	CC	Gelatinous Drop-like Corneal Dystrophy	Alavi (2007)
78785	TACSTD2	G	A	GG	Gelatinous Drop-like Corneal Dystrophy	Tsujikawa (1999)
42461	ROBO3	T	C	TT	Gaze Palsy, Horizontal, with Progressive Scoliosis	Jen (2004)
42462	ROBO3	A	C	AA	Gaze Palsy, Horizontal, with Progressive Scoliosis	Jen (2004)
42463	ROBO3	C	T	CC	Gaze Palsy, Horizontal, with Progressive Scoliosis	Chan (2006)
42464	ROBO3	G	A	GG	Gaze Palsy, Horizontal, with Progressive Scoliosis	Jen (2004)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
42465	ROBO3	G	A	GG	Gaze Palsy, Horizontal, with Progressive Scoliosis	Jen (2004)
42466	ROBO3	G	T	GG	Gaze Palsy, Horizontal, with Progressive Scoliosis	Jen (2004)
42467	ROBO3	G	C	GG	Gaze Palsy, Horizontal, with Progressive Scoliosis	Jen (2004)
42468	ROBO3	T	C	TT	Gaze Palsy, Horizontal, with Progressive Scoliosis	Jen (2004)
42469	ROBO3	C	T	CC	Gaze Palsy, Horizontal, with Progressive Scoliosis	Chan (2006)
42470	ROBO3	G	A	GG	Gaze Palsy, Horizontal, with Progressive Scoliosis	Jen (2004)
102057	GBA	A	G	AA	Gaucher Disease, Type III	Zimran (1989), Latham (1990)
27154	GBA	G	C	GG	Gaucher Disease, Type III	Parenti (1998)
102070	GBA	C	T	CC	Gaucher Disease, Type III	Amaral (1999)
27203	GBA	T	A	TT	Gaucher Disease, Type III	Theophilus (1989)
102072	GBA	G	A	GG	Gaucher Disease, Type III	Hong (1990), Mistry (1992)
27016	GBA	A	C	AA	Gaucher Disease, Type II	Stone (2000)
102066	GBA	C	G	CC	Gaucher Disease, Perinatal Lethal	Theophilus (1989), Cormand (1995), Chabas (1995), Uyama (1997), Uyama (1992), Bohlega (2000), Inui (2001), Mignot (2003)
27018	GBA	C	G	CC	Gaucher Disease, Type I	Theophilus (1989), Cormand (1995), Chabas (1995), Uyama (1997), Uyama (1992), Bohlega (2000), Inui (2001), Mignot (2003)
102063	GBA	C	G	CC	Gaucher Disease, Type II	Theophilus (1989), Cormand (1995), Chabas (1995), Uyama (1997), Uyama (1992), Bohlega (2000), Inui (2001), Mignot (2003)
102064	GBA	C	G	CC	Gaucher Disease, Type III	Theophilus (1989), Beutler (1992), Uyama (1992), Beutler (1995), Cormand (1995), Chabas (1995), Abrahamov (1995), Uyama (1997), Bohlega (2000), Inui (2001), Mignot (2003)
102065	GBA	C	G	CC	Gaucher Disease, Type IIIC	Theophilus (1989), Beutler (1992), Uyama (1992), Beutler (1995), Cormand (1995), Chabas (1995), Abrahamov (1995), Uyama (1997), Bohlega (2000), Inui (2001), Mignot (2003)
102056	GBA	A	G	AA	Gaucher Disease, Type II	Zimran (1989), Latham (1990)
27143	GBA	C	T	CC	Gaucher Disease, Type II	Eyal (1990)
27149	GBA	A	C	AA	Gaucher Disease, Type II	Eyal (1990)
27209	GBA	G	C	GG	Gaucher Disease, Type II	Wigderson (1989)
102071	GBA	G	A	GG	Gaucher Disease, Type II	Hong (1990), Mistry (1992)
27011	GBA	A	G	AA	Gaucher Disease, Type I	Tsuji (1987), Wigderson (1989), Firon (1990), Dahl (1990), Hong (1990), Koprivica (2000), Lwin (2004), GeneReviews (2011), GeneDx (2016), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2016), Counsyl (2016)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
801958	GBA	A	G	AA	Possibly associated with an increased risk of Parkinson disease, late-onset	Sidransky (2009), Alcalay (2014)
801959	GBA	A	G	AA	Possibly associated with an increased risk of Dementia, Lewy body	Sidransky (2009), Alcalay (2014)
27022	GBA	A	T	AA	Gaucher Disease, Type I	Kawame (1991), Eto (1991), He (1992), Pastores (2000), GeneReviews (2011), Bean (2013), Emory Genetics Laboratory of Emory University (2014)
27036	GBA	C	G	CC	Gaucher Disease, Type I	Kim (1996)
27043	GBA	C	T	CC	Gaucher Disease, Type I	Kim (1996)
27049	GBA	C	G	CC	Gaucher Disease, Type I	Zhao (2003)
27059	GBA	C	G,T	CC	Gaucher Disease, Type I	Graves (1988), Felderhoff-Mueser (2004)
27061	GBA	G	A	GG	Gaucher Disease, Type I	Beutler (1993)
27076	GBA	T	G	TT	Gaucher Disease, Type I	Eyal (1991)
27108	GBA	A	G	AA	Gaucher Disease, Type I	Beutler (1993)
27110	GBA	A	C	AA	Gaucher Disease, Type I	Zimran (1994)
27111	GBA	A	T	AA	Gaucher Disease, Type I	Gelbart (1990)
27129	GBA	G	A	GG	Gaucher Disease, Type I	He (1992)
27134	GBA	G	A,C	GG	Gaucher Disease, Type I	Latham (1991)
27136	GBA	C	A	CC	Gaucher Disease, Type I	Latham (1991)
27141	GBA	G	A	GG	Gaucher Disease, Type I	He (1992)
27163	GBA	C	G	CC	Gaucher Disease, Type I	Latham (1991)
27169	GBA	T	C,G	TT	Gaucher Disease, Type I	Tsuji (1988), Zimran (1989), Firon (1990), Zimran (1990), Zimran (1991), Beutler (1993), Mistry (1992), Weely (1993), Walley (1993), Ida (1995), Cormand (1998), Koprivica (2000), Lwin (2004)
27171	GBA	G	C	GG	Gaucher Disease, Type I	Shamseddine (2004)
27173	GBA	C	T	CC	Gaucher Disease, Type I	Amaral (1999), Zimran (1989), Park (2003)
27189	GBA	C	A	CC	Gaucher Disease, Type I	Theophilus (1989), Latham (1990)
27199	GBA	G	A	GG	Gaucher Disease, Type I	Wasserstein (1999)
27223	GBA	G	A,T	GG	Gaucher Disease, Type I	Hong (1990), Mistry (1992), Park (2003)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
27226	GBA	C	T	CC	Gaucher Disease, Type I	Beutler (1993)
27230	GBA	C	T	CC	Gaucher Disease, Type I	Beutler (1993)
27235	GBA	C	A,T	CC	Gaucher Disease, Type I	Beutler (1992), GeneReviews (2011), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2005), Counsyl (2016), Laboratory Corporation of America (2016), Fulgent Genetics (2018)
102068	GBA	C	T	CC	Gaucher Disease, Perinatal Lethal	Graves (1988), Felderhoff-Mueser (2004)
27067	GBA	C	A,T	CC	Gaucher Disease, Perinatal Lethal	Stone (2000)
27117	GBA	G	T	GG	Gaucher Disease, Perinatal Lethal	Zhao (2003)
27135	GBA	T	A,C	TT	Gaucher Disease, Perinatal Lethal	Stone (1999)
27160	GBA	G	A	GG	Gaucher Disease, Perinatal Lethal	Stone (1999)
27196	GBA	C	A,G	CC	Gaucher Disease, Perinatal Lethal	Stone (1999)
32987	PSAP	C	A	CC	Gaucher Disease, Atypical	Schnabel (1991)
32988	PSAP	G	A	GG	Gaucher Disease, Atypical	Diaz-Font (2005)
27095	GBA	C	T	CC	Gaucher disease 3	Koprivica (2000)
27132	GBA	A	C	AA	Gaucher disease 3	Koprivica (2000)
27195	GBA	C	G	CC	Gaucher disease 3	Seeman (1996)
27201	GBA	T	A	TT	Gaucher disease 3	Orvisky (2002)
27229	GBA	G	A	GG	Gaucher disease 3	Seeman (1996)
27233	GBA	A	T	AA	Gaucher disease 3	Orvisky (2002)
800892	GBA	A	C	AA	Gaucher disease 2	Stone (2000)
27041	GBA	C	T	CC	Gaucher disease 2	Sinclair (1998)
27062	GBA	G	A	GG	Gaucher disease 2	Tang (2005)
27066	GBA	G	A	GG	Gaucher disease 2	Sinclair (1998)
27091	GBA	G	A	GG	Gaucher disease 2	Montfort (2004)
27092	GBA	A	G	AA	Gaucher disease 2	Choy (2000)
27096	GBA	G	A,T	GG	Gaucher disease 2	Stone (2000)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
27103	GBA	T	C	TT	Gaucher disease 2	Orvisky (2002)
27114	GBA	C	A	CC	Gaucher disease 2	Grace (1999)
27120	GBA	G	T	GG	Gaucher disease 2	Stone (2000)
27124	GBA	G	A	GG	Gaucher disease 2	Cooper (2000)
27128	GBA	C	T	CC	Gaucher disease 2	Stone (2000)
27131	GBA	T	C	TT	Gaucher disease 2	Stone (2000)
27162	GBA	T	C	TT	Gaucher disease 2	Tang (2005)
27179	GBA	G	T	GG	Gaucher disease 2	Tang (2005)
27180	GBA	A	C	AA	Gaucher disease 2	Tang (2005)
27181	GBA	A	G	AA	Gaucher disease 2	Tang (2005)
27184	GBA	C	T	CC	Gaucher disease 2	Cormand (1998)
27187	GBA	T	A	TT	Gaucher disease 2	Cormand (1998)
27210	GBA	T	C	TT	Gaucher disease 2	Tang (2005)
27225	GBA	C	A	CC	Gaucher disease 2	Choy (1998)
27227	GBA	A	G	AA	Gaucher disease 2	Filocamo (2000)
27033	GBA	C	T	CC	Gaucher disease 1	Cormand (1998)
27038	GBA	C	T	CC	Gaucher disease 1	Miocic (2005)
27040	GBA	A	C	AA	Gaucher disease 1	Choy (1997)
27050	GBA	C	T	CC	Gaucher disease 1	Koprivica (2000)
27051	GBA	A	C	AA	Gaucher disease 1	Choy (2002)
27053	GBA	A	C	AA	Gaucher disease 1	Amaral (2000)
27055	GBA	C	T	CC	Gaucher disease 1	Cormand (1998)
27056	GBA	T	C	TT	Gaucher disease 1	Koprivica (2000)
27058	GBA	A	C,G	AA	Gaucher disease 1	Miocic (2005)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
27068	GBA	G	A	GG	Gaucher disease 1	Koprivica (2000)
27069	GBA	T	G	TT	Gaucher disease 1	Cormand (1998)
27070	GBA	A	C	AA	Gaucher disease 1	Koprivica (2000)
27072	GBA	C	G	CC	Gaucher disease 1	Eyal (1991)
27080	GBA	T	G	TT	Gaucher disease 1	Orvisky (2002)
27082	GBA	G	A	GG	Gaucher disease 1	Cormand (1998)
27084	GBA	G	A	GG	Gaucher disease 1	Koprivica (2000)
27097	GBA	A	T	AA	Gaucher disease 1	Miocic (2005)
27106	GBA	T	C	TT	Gaucher disease 1	Choy (1999)
27119	GBA	G	A	GG	Gaucher disease 1	Cormand (1998)
27148	GBA	A	G	AA	Gaucher disease 1	Choy (2002)
27152	GBA	G	A	GG	Gaucher disease 1	Orvisky (2002)
27157	GBA	G	A	GG	Gaucher disease 1	Cooper (2001)
27186	GBA	G	A	GG	Gaucher disease 1	Cormand (1998)
27190	GBA	C	G	CC	Gaucher disease 1	Amaral (2000)
27191	GBA	G	A,C	GG	Gaucher disease 1	Rozenberg (2006)
27194	GBA	C	T	CC	Gaucher disease 1	Rozenberg (2006)
27198	GBA	C	A	CC	Gaucher disease 1	Koprivica (2000)
27205	GBA	A	G	AA	Gaucher disease 1	Cormand (1998)
27211	GBA	A	C	AA	Gaucher disease 1	Choy (1994)
27213	GBA	C	T	CC	Gaucher disease 1	Miocic (2005)
27219	GBA	C	G,T	CC	Gaucher disease 1	Cooper (2001)
27238	GBA	C	T	CC	Gaucher disease 1	Rozenberg (2006)
27239	GBA	G	C	GG	Gaucher disease 1	Dominissini (2006)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
27240	GBA	C	A	CC	Gaucher disease 1	Cormand (1998)
27241	GBA	T	C	TT	Gaucher disease 1	Dominissini (2006)
27023	GBA	A	G	AA	Gaucher disease	Hodanova (1999)
27025	GBA	A	C	AA	Gaucher disease	Germain (1998)
27026	GBA	T	C	TT	Gaucher disease	Kim (1996)
27034	GBA	C	A	CC	Gaucher disease	Beutler (2005)
27035	GBA	C	T	CC	Gaucher disease	Alfonso (2004)
27037	GBA	C	G	CC	Gaucher disease	Church (2000)
27042	GBA	G	A	GG	Gaucher disease	Beutler (1995)
27044	GBA	G	A	GG	Gaucher disease	Alfonso (2001)
27045	GBA	G	A	GG	Gaucher disease	Beutler (1995)
27046	GBA	A	G	AA	Gaucher disease	Beutler (2005)
27047	GBA	G	A	GG	Gaucher disease	Beutler (2005)
27048	GBA	T	A	TT	Gaucher disease	Grace (1997)
27052	GBA	G	A,T	GG	Gaucher disease	Beutler (1998)
27057	GBA	A	C,G	AA	Gaucher disease	Germain (1998)
27063	GBA	A	G	AA	Gaucher disease	Alfonso (2004)
27064	GBA	T	C	TT	Gaucher disease	Beutler (2005)
27065	GBA	T	A	TT	Gaucher disease	Beutler (2005)
27071	GBA	G	T	GG	Gaucher disease	Orvisky (2002)
27077	GBA	G	T	GG	Gaucher disease	Filocamo (2002)
27078	GBA	G	A	GG	Gaucher disease	Beutler (1998)
27079	GBA	A	C,T	AA	Gaucher disease	Beutler (1998)
27081	GBA	G	A	GG	Gaucher disease	Beutler (1998)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
27083	GBA	C	G	CC	Gaucher disease	Germain (1998)
27085	GBA	G	C	GG	Gaucher disease	Beutler (1995)
27087	GBA	G	T	GG	Gaucher disease	Beutler (1994)
27088	GBA	G	A	GG	Gaucher disease	Choy (1995)
27089	GBA	C	T	CC	Gaucher disease	Grace (1997)
27090	GBA	G	A,T	GG	Gaucher disease	Beutler (1994)
27094	GBA	C	A	CC	Gaucher disease	Ida (1997)
27098	GBA	C	T	CC	Gaucher disease	Grace (1997)
27099	GBA	C	A	CC	Gaucher disease	Sarria (1999)
27100	GBA	G	A	GG	Gaucher disease	Beutler (2005)
27104	GBA	C	T	CC	Gaucher disease	Beutler (1994)
27105	GBA	C	T	CC	Gaucher disease	Hodanova (1999)
27109	GBA	A	C	AA	Gaucher disease	Cooper (2001)
27113	GBA	G	C	GG	Gaucher disease	Filocamo (2002)
27115	GBA	A	G	AA	Gaucher disease	Germain (1998)
27118	GBA	C	T	CC	Gaucher disease	Beutler (1994)
27121	GBA	A	G	AA	Gaucher disease	Fernandez (2005)
27122	GBA	C	T	CC	Gaucher disease	Beutler (1998)
27123	GBA	G	C	GG	Gaucher disease	Walley (1995)
27125	GBA	G	C	GG	Gaucher disease	Beutler (2005)
27126	GBA	C	T	CC	Gaucher disease	Grace (1997)
27127	GBA	G	A	GG	Gaucher disease	Beutler (1994)
27130	GBA	T	A	TT	Gaucher disease	Germain (1998)
27137	GBA	A	G	AA	Gaucher disease	Cormand (1996)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
27138	GBA	C	G	CC	Gaucher disease	Walley (1995)
27139	GBA	G	T	GG	Gaucher disease	Walley (1995)
27142	GBA	A	G	AA	Gaucher disease	Germain (1998)
27144	GBA	C	A	CC	Gaucher disease	Torralba (2001)
27146	GBA	A	G	AA	Gaucher disease	Alfonso (2004)
27147	GBA	C	T	CC	Gaucher disease	Beutler (1998)
27150	GBA	C	T	CC	Gaucher disease	Beutler (2005)
27156	GBA	C	T	CC	Gaucher disease	Cooper (2001)
27158	GBA	C	T	CC	Gaucher disease	Durr (2004)
27159	GBA	C	T	CC	Gaucher disease	Kawame (1992)
27161	GBA	C	T	CC	Gaucher disease	Filocamo (2002)
27164	GBA	T	G	TT	Gaucher disease	Alfonso (2004)
27165	GBA	C	T	CC	Gaucher disease	Beutler (1998)
27166	GBA	C	G	CC	Gaucher disease	Cooper (2001)
27167	GBA	T	C	TT	Gaucher disease	Ida (1997)
27172	GBA	C	A,G	CC	Gaucher disease	Cormand (1997)
27174	GBA	A	C	AA	Gaucher disease	Beutler (1994)
27175	GBA	C	T	CC	Gaucher disease	Beutler (2005)
27176	GBA	C	T	CC	Gaucher disease	Beutler (1994)
27177	GBA	C	G	CC	Gaucher disease	Pomponio (2005)
27178	GBA	T	G	TT	Gaucher disease	Walley (1993)
27182	GBA	G	A	GG	Gaucher disease	Morar (1996)
27183	GBA	C	A,T	CC	Gaucher disease	Beutler (2005)
27185	GBA	C	T	CC	Gaucher disease	Beutler (2005)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
27188	GBA	A	T	AA	Gaucher disease	Beutler (1998)
27192	GBA	T	G	TT	Gaucher disease	Amaral (1996)
27193	GBA	A	G	AA	Gaucher disease	Beutler (2005)
27197	GBA	C	T	CC	Gaucher disease	Beutler (1994)
27200	GBA	A	G	AA	Gaucher disease	Cormand (1997)
27202	GBA	T	C	TT	Gaucher disease	Beutler (2005)
27204	GBA	A	T	AA	Gaucher disease	Beutler (1998)
27206	GBA	T	G	TT	Gaucher disease	Ida (1997)
27207	GBA	G	A	GG	Gaucher disease	Beutler (1996)
27208	GBA	T	C	TT	Gaucher disease	Beutler (2005)
27212	GBA	T	C	TT	Gaucher disease	Tuteja (1994)
27214	GBA	T	C	TT	Gaucher disease	Kawame (1992)
27215	GBA	T	C	TT	Gaucher disease	Ida (1997)
27217	GBA	C	G	CC	Gaucher disease	Germain (1998)
27218	GBA	T	C,G	TT	Gaucher disease	Filocamo (2002)
27222	GBA	G	C	GG	Gaucher disease	Hatton (1997)
27231	GBA	G	A	GG	Gaucher disease	Kawame (1992)
27232	GBA	C	T	CC	Gaucher disease	Felderhoff-Mueser (2004)
27234	GBA	A	C	AA	Gaucher disease	Beutler (1995)
27236	GBA	C	A,T	CC	Gaucher disease	Germain (1998)
27237	GBA	T	C	TT	Gaucher disease	Alfonso (2001)
45993	PDGFRA	G	T	GG	Gastrointestinal stromal tumour	Chompret (2004)
45896	CDH1	C	T	CC	Gastric Cancer, Familial Diffuse	Gayther (1998), Huntsman (2001)
45899	CDH1	C	T	CC	Gastric Cancer, Familial Diffuse	Guilford (1998)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
45876	CDH1	G	T	GG	Gastric Cancer, Familial Diffuse	Guilford (1999), Lynch (2000)
45916	CDH1	G	T	GG	Gastric Cancer, Familial Diffuse	Jones (1964), Lauren, (1965), Zanghieri (1990), Vecchia (1992), Guilford (1998), Guilford (1998)
45918	CDH1	G	A	GG	Gastric Cancer|Cleft Lip with or without Cleft Palate, with Gastric Cancer, Familial Diffuse	Frebourg (2006)
45890	CDH1	A	G	AA	Gastric cancer	Oliveira (2002), Suriano (2003)
45898	CDH1	C	T	CC	Gastric Cancer	Suriano (2003)
46861	MET	C	T	CC	Gastric cancer	Lee, (2000), Kim (2003)
47458	TP53	G	A	GG	Gastric cancer	Keller (2004)
33704	GLB1	A	G,T	AA	Gangliosidosis GM1, Adult/Chronic Type	Nishimoto (1991), Yoshida (1991), Yoshida (1992)
33726	GLB1	G	A	GG	Gangliosidosis GM1	Nishimoto (1991), Yoshida (1991)
33727	GLB1	G	A	GG	Gangliosidosis GM1	Boustany (1993), Chiu (1996)
33702	GLB1	C	T	CC	Gangliosidosis GM1	Roze (2005)
33703	GLB1	G	A	GG	Gangliosidosis GM1	Nishimoto (1991)
33705	GLB1	G	C	GG	Gangliosidosis GM1	Georgiou (2004)
33706	GLB1	G	A	GG	Gangliosidosis GM1	Caciotti (2005)
33707	GLB1	C	T	CC	Gangliosidosis GM1	Silva (1999)
33708	GLB1	G	A	GG	Gangliosidosis GM1	Caciotti (2003)
33709	GLB1	T	C	TT	Gangliosidosis GM1	Roze (2005)
33713	GLB1	C	A	CC	Gangliosidosis GM1	Silva (1999)
33714	GLB1	C	T	CC	Gangliosidosis GM1	Yoshida (1991)
33715	GLB1	C	A	CC	Gangliosidosis GM1	Santamaria (2007)
33716	GLB1	G	A	GG	Gangliosidosis GM1	Santamaria (2006)
33717	GLB1	G	A,T	GG	Gangliosidosis GM1	Morrone (2000)
33718	GLB1	G	A,T	GG	Gangliosidosis GM1	Roze (2005)
33719	GLB1	C	A	CC	Gangliosidosis GM1	Georgiou (2004)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
33720	GLB1	T	A	TT	Gangliosidosis GM1	Santamaria (2006)
33721	GLB1	A	C	AA	Gangliosidosis GM1	Santamaria (2007)
33722	GLB1	A	G	AA	Gangliosidosis GM1	Santamaria (2007)
33723	GLB1	A	G	AA	Gangliosidosis GM1	Santamaria (2006)
33724	GLB1	T	C	TT	Gangliosidosis GM1	Santamaria (2006)
33725	GLB1	C	T	CC	Gangliosidosis GM1	Kaye (1997)
33728	GLB1	C	A	CC	Gangliosidosis GM1	Morrone (2000)
33729	GLB1	A	G	AA	Gangliosidosis GM1	Morrone (2000)
33730	GLB1	G	A	GG	Gangliosidosis GM1	Caciotti (2005)
33731	GLB1	C	T	CC	Gangliosidosis GM1	Silva (1999)
33732	GLB1	G	A	GG	Gangliosidosis GM1	Morrone (2000)
33733	GLB1	A	G	AA	Gangliosidosis GM1	Santamaria (2006)
33734	GLB1	T	C	TT	Gangliosidosis GM1	Kaye (1997)
33736	GLB1	C	T	CC	Gangliosidosis GM1	Santamaria (2006)
33739	GLB1	T	C	TT	Gangliosidosis GM1	Yoshida (1991)
33741	GLB1	C	T	CC	Gangliosidosis GM1	Zhang (2000)
33742	GLB1	C	G	CC	Gangliosidosis GM1	Santamaria (2006)
33743	GLB1	T	C	TT	Gangliosidosis GM1	Santamaria (2006)
33744	GLB1	G	A	GG	Gangliosidosis GM1	Hinek (2000)
33749	GLB1	G	A	GG	Gangliosidosis GM1	Santamaria (2007)
33765	GLB1	C	A	CC	Gangliosidosis GM1	Santamaria (2006)
6512	GCLC	G	A	GG	Gamma-glutamylcysteine synthetase deficiency	Hamilton (2003)
6513	GCLC	G	A	GG	Gamma-glutamylcysteine synthetase deficiency	Ristoff (2000)
22378	GGCX	C	T	CC	Gamma-glutamyl carboxylase deficiency	Darghouth (2006)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
22379	GGCX	A	C,G	AA	Gamma-glutamyl carboxylase deficiency	Darghouth (2006)
22382	GGCX	G	T	GG	Gamma-glutamyl carboxylase deficiency	Darghouth (2006)
27801	PPGB	T	A	TT	Galactosialidosis, Late Infantile	Zhou (1996)
27803	PPGB	T	C	TT	Galactosialidosis, Late Infantile	Zhou (1996)
27805	PPGB	T	G	TT	Galactosialidosis, Late Infantile	Zhou (1991)
27806	PPGB	A	G	AA	Galactosialidosis, Late Infantile	Takiguchi (2000)
27799	PPGB	G	A	GG	Galactosialidosis, Early Infantile	Zhou (1996)
27800	PPGB	T	C	TT	Galactosialidosis, Early Infantile	Zhou (1996)
27804	PPGB	G	A	GG	Galactosialidosis, Early Infantile	Zhou (1996)
27808	PPGB	A	G	AA	Galactosialidosis	Shimmoto (1990)
800893	PPGB	T	A	TT	Galactosialidosis	Zhou (1996)
800894	PPGB	T	G	TT	Galactosialidosis	Zhou (1991), Chitayat (1988), Strisciuglio (1990), Zhou (1996)
27794	PPGB	T	C	TT	Galactosialidosis	Shimmoto (1993)
27795	PPGB	A	G	AA	Galactosialidosis	Shimmoto (1993)
27796	PPGB	C	A	CC	Galactosialidosis	Zhou (1996)
27797	PPGB	G	A	GG	Galactosialidosis	Groener (2003)
27798	PPGB	C	T	CC	Galactosialidosis	Rudenko (1998)
27802	PPGB	A	G	AA	Galactosialidosis	Fukuhara (1992), Shimmoto (1993), Shimmoto (1993)
27807	PPGB	G	T	GG	Galactosialidosis	Malvagia (2004)
29594	GALT	C	T	CC	Galactosemia	Baker (1966), Lai (1996)
29679	GALT	C	G	CC	Galactosemia	Murphy (1999)
29657	GALT	G	T	GG	Galactosemia	Greber-Platzer (1997)
29549	GALT	G	C	GG	Galactosemia	Item (2002)
29550	GALT	A	G	AA	Galactosemia	Yang (2001)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
29551	GALT	G	A	GG	Galactosemia	Tang (1999)
29552	GALT	G	C	GG	Galactosemia	Gort (2006)
29553	GALT	G	T	GG	Galactosemia	Greber-Platzer (1997)
29554	GALT	T	A	TT	Galactosemia	Greber-Platzer (1997)
29555	GALT	G	A	GG	Galactosemia	Gort (2006)
29556	GALT	T	C	TT	Galactosemia	Emory (2001)
29557	GALT	A	C	AA	Galactosemia	Greber-Platzer (1997)
29558	GALT	G	A	GG	Galactosemia	Woo (1991)
29559	GALT	G	T	GG	Galactosemia	Tyfield (1999)
29560	GALT	C	T	CC	Galactosemia	Tyfield (1999)
29561	GALT	G	A	GG	Galactosemia	Bosch (2005)
29562	GALT	G	T	GG	Galactosemia	Tyfield (1999)
29563	GALT	T	A	TT	Galactosemia	Emory (2001)
29564	GALT	C	T	CC	Galactosemia	Langley (1997)
29565	GALT	G	T	GG	Galactosemia	Tyfield (1999)
29567	GALT	C	A	CC	Galactosemia	Shin (2004)
29568	GALT	C	T	CC	Galactosemia	Zekanowski (1999)
29569	GALT	C	T	CC	Galactosemia	Sommer (1995)
29570	GALT	G	A	GG	Galactosemia	Emory (2001)
29571	GALT	A	C	AA	Galactosemia	Emory (2001)
29572	GALT	C	T	CC	Galactosemia	Emory (2001)
29573	GALT	T	C	TT	Galactosemia	Reichardt (1992)
29574	GALT	C	T	CC	Galactosemia	Tyfield (1999)
29575	GALT	G	A	GG	Galactosemia	Leslie (1992)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
29576	GALT	A	G	AA	Galactosemia	Emory (2001)
29577	GALT	T	G	TT	Galactosemia	Zekanowski (1999)
29578	GALT	A	G	AA	Galactosemia	Hirokawa (1999)
29579	GALT	G	A	GG	Galactosemia	Tyfield (1999)
29580	GALT	G	A	GG	Galactosemia	Tyfield (1999)
29581	GALT	A	C	AA	Galactosemia	Elsas (1998)
29582	GALT	A	T	AA	Galactosemia	Emory (2001)
29583	GALT	C	G	CC	Galactosemia	Emory (2001)
29584	GALT	T	C	TT	Galactosemia	Tyfield (1999)
29585	GALT	A	C	AA	Galactosemia	Tyfield (1999)
29586	GALT	G	A	GG	Galactosemia	Tyfield (1999)
29587	GALT	C	G	CC	Galactosemia	Greber-Platzer (1997)
29588	GALT	T	C	TT	Galactosemia	Tyfield (1999)
29589	GALT	A	G	AA	Galactosemia	Tyfield (1999)
29590	GALT	T	C	TT	Galactosemia	Tyfield (1999)
29591	GALT	G	A	GG	Galactosemia	Tyfield (1999)
29592	GALT	C	T	CC	Galactosemia	Tyfield (1999)
29593	GALT	C	G	CC	Galactosemia	Bosch (2005)
29594	GALT	C	T	CC	Galactosemia	Baker (1966), Cuatrecasas, (1968)
29595	GALT	C	T	CC	Galactosemia	Elsas (1995)
29596	GALT	T	C	TT	Galactosemia	Leslie (1992)
29597	GALT	T	A	TT	Galactosemia	Woo (1990)
29598	GALT	T	C	TT	Galactosemia	Zaffanello (2005)
29599	GALT	A	G	AA	Galactosemia	Tyfield (1999)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
29600	GALT	C	T	CC	Galactosemia	Greber-Platzer (1997)
29601	GALT	C	G	CC	Galactosemia	Tyfield (1999)
29602	GALT	C	T	CC	Galactosemia	Reichardt (1992)
29603	GALT	G	A	GG	Galactosemia	Elsas (1995)
29604	GALT	G	C	GG	Galactosemia	Maceratesi (1996)
29605	GALT	T	C	TT	Galactosemia	Elsas (1995)
29606	GALT	T	G	TT	Galactosemia	Tyfield (1999)
29607	GALT	G	A	GG	Galactosemia	Tyfield (1999)
29608	GALT	T	C	TT	Galactosemia	Tyfield (1999)
29609	GALT	C	T	CC	Galactosemia	Tyfield (1999)
29610	GALT	T	C	TT	Galactosemia	Tyfield (1999)
29611	GALT	C	A	CC	Galactosemia	Kozak (1999)
29612	GALT	T	C	TT	Galactosemia	Reichardt (1992)
29613	GALT	G	A	GG	Galactosemia	Gort (2006)
29614	GALT	C	T	CC	Galactosemia	Emory (2001)
29615	GALT	G	A	GG	Galactosemia	Tyfield (1999)
29616	GALT	T	G	TT	Galactosemia	Gort (2006)
29617	GALT	C	A	CC	Galactosemia	Ninfali (1996)
29618	GALT	C	A	CC	Galactosemia	Tyfield (1999)
29619	GALT	C	G	CC	Galactosemia	Tang (1999)
29620	GALT	C	T	CC	Galactosemia	Gort (2006)
29621	GALT	C	T	CC	Galactosemia	Tyfield (1999)
29622	GALT	A	G	AA	Galactosemia	Reichardt (1991), Elsas (1994), Ashino (1995), Fridovich-Keil (1996), LabCorp (2011). Center for Pediatric Genomic Medicine of Children's Mercy Hospital and Clinics (2014), Emory Genetics Laboratory of Emory University (2014), Counsyl (2015), GeneDx (2016), Knight Diagnostic Laboratories of Oregon Health and Sciences University (2016)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
29623	GALT	G	A	GG	Galactosemia	Elsas (1995)
29624	GALT	A	G	AA	Galactosemia	Gort (2006)
29625	GALT	T	C	TT	Galactosemia	
29626	GALT	T	C	TT	Galactosemia	Reichardt (1992)
29627	GALT	T	C	TT	Galactosemia	Tyfield (1999)
29628	GALT	T	G	TT	Galactosemia	Tyfield (1999)
29629	GALT	G	A	GG	Galactosemia	Tyfield (1999)
29630	GALT	G	A	GG	Galactosemia	Elsas (1995)
29631	GALT	G	A	GG	Galactosemia	Elsas (1995)
29632	GALT	G	C	GG	Galactosemia	Tyfield (1999)
29633	GALT	C	T	CC	Galactosemia	Tyfield (1999)
29634	GALT	C	T	CC	Galactosemia	Yang (2001)
29635	GALT	A	C	AA	Galactosemia	Kozak (1999)
29636	GALT	A	G	AA	Galactosemia	Tyfield (1999)
29637	GALT	C	T	CC	Galactosemia	Gathof (1995)
29638	GALT	T	C	TT	Galactosemia	Tyfield (1999)
29639	GALT	T	C	TT	Galactosemia	Tyfield (1999)
29640	GALT	G	T	GG	Galactosemia	Bosch (2005)
29641	GALT	C	T	CC	Galactosemia	Tang (1999)
29642	GALT	G	A	GG	Galactosemia	Ashino (1995)
29643	GALT	G	T	GG	Galactosemia	Emory (2001)
29644	GALT	T	C	TT	Galactosemia	Tyfield (1999)
29645	GALT	G	A	GG	Galactosemia	Tyfield (1999)
29646	GALT	A	C	AA	Galactosemia	Tyfield (1999)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
29647	GALT	A	G	AA	Galactosemia	Tyfield (1999)
29648	GALT	G	T	GG	Galactosemia	Bosch (2005)
29649	GALT	C	T	CC	Galactosemia	Tyfield (1999)
29650	GALT	C	T	CC	Galactosemia	Shin (1996)
29651	GALT	G	C	GG	Galactosemia	Tyfield (1999)
29652	GALT	G	A	GG	Galactosemia	Emory (2001)
29653	GALT	C	G	CC	Galactosemia	Gort (2006)
29654	GALT	C	G	CC	Galactosemia	Tyfield (1999)
29655	GALT	C	G	CC	Galactosemia	Emory (2001)
29656	GALT	C	G	CC	Galactosemia	Tyfield (1999)
29657	GALT	G	T	GG	Galactosemia	Greber-Platzer (1997)
29658	GALT	T	G	TT	Galactosemia	Tyfield (1999)
29659	GALT	G	A	GG	Galactosemia	Elsas (1995)
29660	GALT	T	A	TT	Galactosemia	Seyrantepe (1999)
29661	GALT	C	A	CC	Galactosemia	Gort (2006)
29662	GALT	G	T	GG	Galactosemia	Emory (2001)
29663	GALT	G	A	GG	Galactosemia	Elsas (1995)
29664	GALT	G	A	GG	Galactosemia	Sommer (1995)
29665	GALT	A	G	AA	Galactosemia	Tyfield (1999)
29666	GALT	G	T	GG	Galactosemia	Tyfield (1999)
29667	GALT	C	G	CC	Galactosemia	Flach (1990), Reichardt (1993)
29668	GALT	G	A	GG	Galactosemia	Elsas (1995)
29669	GALT	G	C	GG	Galactosemia	Emory (2001)
29670	GALT	C	T	CC	Galactosemia	Emory (2001)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
29671	GALT	T	C	TT	Galactosemia	Tyfield (1999)
29672	GALT	T	G	TT	Galactosemia	Elsas (1995)
29673	GALT	C	T	CC	Galactosemia	Tyfield (1999)
29674	GALT	C	T	CC	Galactosemia	Greber-Platzer (1997)
29675	GALT	C	T	CC	Galactosemia	Emory (2001)
29676	GALT	G	A	GG	Galactosemia	Tyfield (1999)
29677	GALT	C	T	CC	Galactosemia	Tyfield (1999)
29678	GALT	C	T	CC	Galactosemia	Sommer (1995)
29679	GALT	C	G	CC	Galactosemia	
29680	GALT	C	T	CC	Galactosemia	Woo (1990), Reichardt (1991)
29681	GALT	G	A	GG	Galactosemia	Tyfield (1999)
29682	GALT	A	G	AA	Galactosemia	Leslie (1992)
29683	GALT	A	T	AA	Galactosemia	Tyfield (1999)
29684	GALT	G	A	GG	Galactosemia	Kozak (1999)
29685	GALT	G	T	GG	Galactosemia	Gathof (1995)
29686	GALT	C	A	CC	Galactosemia	Item (2002)
29687	GALT	C	A	CC	Galactosemia	Tyfield (1999)
29688	GALT	C	A	CC	Galactosemia	Emory (2001)
29689	GALT	A	G	AA	Galactosemia	Shin (1996)
29690	GALT	C	T	CC	Galactosemia	Gathof (1995)
29691	GALT	A	T	AA	Galactosemia	Shin (2004)
29692	GALT	C	A	CC	Galactosemia	Greber-Platzer (1997)
29693	GALT	C	T	CC	Galactosemia	Reichardt (1993)
29694	GALT	T	C	TT	Galactosemia	Tyfield (1999)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
29695	GALT	A	C	AA	Galactosemia	Bosch (2005)
29696	GALT	C	T	CC	Galactosemia	Wadelius (1993)
29697	GALT	A	G	AA	Galactosemia	Yang (2001)
29698	GALT	A	G	AA	Galactosemia	Ashino (1995)
29699	GALT	A	C	AA	Galactosemia	Elsas (1995)
29700	GALT	G	A	GG	Galactosemia	Hirokawa (1997)
29701	GALT	G	T	GG	Galactosemia	Tyfield (1999)
29702	GALT	T	C	TT	Galactosemia	Tyfield (1999)
29703	GALT	A	G	AA	Galactosemia	Elsas (1995)
29704	GALT	G	A	GG	Galactosemia	Tyfield (1999)
29705	GALT	T	C	TT	Galactosemia	Tyfield (1999)
29706	GALT	A	G	AA	Galactosemia	Bosch (2005)
28772	GALE	C	T	CC	Galactose Epimerase Deficiency, Severe	Wohlers (1999)
28767	GALE	G	A	GG	Galactose Epimerase Deficiency	Park (2005)
28768	GALE	T	C	TT	Galactose Epimerase Deficiency	Quimby (1997)
28769	GALE	G	A,T	GG	Galactose Epimerase Deficiency	Park (2005)
28770	GALE	G	T	GG	Galactose Epimerase Deficiency	Park (2005)
28771	GALE	C	T	CC	Galactose Epimerase Deficiency	Maceratesi (1998)
28773	GALE	T	C	TT	Galactose Epimerase Deficiency	Maceratesi (1998)
28774	GALE	G	A	GG	Galactose Epimerase Deficiency	Openo (2006)
28775	GALE	C	A	CC	Galactose Epimerase Deficiency	Openo (2006)
28776	GALE	C	T	CC	Galactose Epimerase Deficiency	Park (2005)
28777	GALE	G	A,T	GG	Galactose Epimerase Deficiency	Park (2005)
28778	GALE	A	G	AA	Galactose Epimerase Deficiency	Quimby (1997)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
28779	GALE	G	A	GG	Galactose Epimerase Deficiency	Park (2005)
28781	GALE	G	A	GG	Galactose Epimerase Deficiency	Openo (2006)
28782	GALE	C	T	CC	Galactose Epimerase Deficiency	Park (2005)
28784	GALE	C	T	CC	Galactose Epimerase Deficiency	Maceratesi (1998)
28785	GALE	C	G,T	CC	Galactose Epimerase Deficiency	Park (2005)
28786	GALE	C	T	CC	Galactose Epimerase Deficiency	Park (2005)
31883	FAH	C	T	CC	Variant of Unknown Significance (questionable association with Fumarylacetoacetase Pseudodeficiency)	Kvittingen (1985), Rootwelt (1994), Bergeron (2001)
29534	FUCA1	G	A	GG	Fucosidosis	Kretz (1989)
70760	COL8A2	G	T	GG	Fuchs Endothelial Corneal Dystrophy	Biswas (2001)
70761	COL8A2	A	C,G	AA	Fuchs Endothelial Corneal Dystrophy	Magovern (1979), Gottsch (2005) PMID 16303941, Gottsch (2005) PMID 15914606
33209	KHK	G	A	GG	Fructosuria	Bonthron (1994)
33210	KHK	G	A	GG	Fructosuria	Bonthron (1994)
100888	GRN	I	D	II	Frontotemporal Lobar Dementia, Ubiquitin-Positive	Baker (2006)
100889	GRN	I	D	II	Frontotemporal Lobar Dementia, Ubiquitin-Positive	Borroni (2008)
100891	GRN	D	I	DD	Possibly associated with Frontotemporal Lobar Dementia, Ubiquitin-Positive	Mackenzie (2006), Baker (2006), Mackenzie (2006), Rohrer (2008)
43291	MAPT	C	T	CC	Frontotemporal dementia, with parkinsonism	Heutink (1997), Hutton (1998), Poorkaj (1998), Hutton (1998), Poorkaj (1998), Poorkaj, (1998), Clark (1998)
43305	MAPT	C	T	CC	Frontotemporal dementia, with parkinsonism	Reed (1997), Reed , (1997), Hutton (1998), van Swieten , (1999), Connell (2001), Connell (2001), Tatebayashi (2002), Saito (2002), Rademakers (2003), Rademakers (2003)
43313	MAPT	C	T	CC	Frontotemporal dementia, with parkinsonism	Dark, (1997), Hutton (1998), Poorkaj , (2001), Janssen (2002), Pickering-Brown (2002), Janssen , (2002), Lantos , (2002), Pickering-Brown (2004), Pickering-Brown (2004)
43288	MAPT	T	G	TT	Frontotemporal dementia, with parkinsonism	Wszolek (1992), Clark (1998), Clark , (1998), Delisle (1999), Yasuda (1999), Yasuda (1999), Delisle , (1999), Yasuda , (1999), Arima (2000), Wszolek (2000), Arima , (2000), Tsuboi (2002), Tsuboi (2002), Tsuboi (2002), Rademakers (2004)
43283	MAPT	A	C	AA	Frontotemporal dementia, with parkinsonism	Pickering-Brown (2000)
43284	MAPT	A	G	AA	Frontotemporal dementia, with parkinsonism	Hutton (2001)
43286	MAPT	G	T	GG	Frontotemporal dementia, with parkinsonism	Heutink (1997), Hutton (1998)
43287	MAPT	G	A	GG	Frontotemporal dementia, with parkinsonism	der Zee (2006)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
43288	MAPT	T	G	TT	Frontotemporal dementia, with parkinsonism	Wszolek (1992), Clark (1998), Clark (1998), Delisle (1999), Yasuda (1999), Yasuda (1999), Delisle (1999), Yasuda (1999), Arima (2000), Wszolek (2000), Arima (2000), Tsuboi (2002), Tsuboi (2002), Tsuboi (2002), Rademakers (2004)
43289	MAPT	A	C	AA	Frontotemporal dementia, with parkinsonism	Iseki (2001)
43291	MAPT	C	T	CC	Frontotemporal dementia, with parkinsonism	Heutink (1997), Hutton (1998), Poorkaj (1998), Hutton (1998), Poorkaj (1998), Poorkaj, (1998), Clark (1998)
43295	MAPT	A	T	AA	Frontotemporal dementia, with parkinsonism	Zarranz (2005), Zarranz (2005)
43299	MAPT	G	A	GG	Frontotemporal dementia, with parkinsonism	Poorkaj (1998), Hutton (1998), Poorkaj (1998), Poorkaj, (1998)
43302	MAPT	A	T	AA	Frontotemporal dementia, with parkinsonism	Neumann (2001)
43305	MAPT	C	T	CC	Frontotemporal dementia, with parkinsonism	Reed (1997), Reed (1997), Hutton (1998), van Swieten (1999), Connell (2001), Connell (2001), Tatebayashi (2002), Saito (2002), Rademakers (2003), Rademakers (2003)
43309	MAPT	A	G	AA	Frontotemporal dementia, with parkinsonism	Hutton (1998), Pickering-Brown (2002)
43312	MAPT	C	T	CC	Frontotemporal dementia, with parkinsonism	Wilhelmsen (1994), Hutton (1998)
43313	MAPT	C	T	CC	Frontotemporal dementia, with parkinsonism	Dark, (1997), Hutton (1998), Poorkaj (2001), Janssen (2002), Pickering-Brown (2002), Janssen (2002), Lantos (2002), Pickering-Brown (2004), Pickering-Brown (2004)
43314	MAPT	G	A	GG	Frontotemporal dementia, with parkinsonism	Spillantini (1998)
43316	MAPT	T	C	TT	Frontotemporal dementia, with parkinsonism	D'Souza (1999)
43319	MAPT	T	C	TT	Frontotemporal dementia, with parkinsonism	Miyamoto (2001)
43322	MAPT	C	T	CC	Frontotemporal dementia, association with	Sobrido (2003)
43282	MAPT	G	A	GG	Frontotemporal dementia ?	Stanford (2004)
42393	GRN	C	T	CC	Frontotemporal Dementia	Gass (2006), Rademakers (2007)
42384	GRN	G	C	GG	Frontotemporal Dementia	der Zee , (2006), Cruts (2006)
78906	CHMP2B	G	T	GG	Frontotemporal Dementia	Skibinski (2005)
39337	PSEN1	C	T	CC	Frontotemporal dementia	Zekanowski (2006)
42384	GRN	G	C	GG	Frontotemporal Dementia	der Zee (2006), Cruts (2006)
42385	GRN	T	C	TT	Frontotemporal Dementia	Baker (2006), Cruts (2006)
42386	GRN	G	A	GG	Frontotemporal Dementia	Cruts (2006)
42387	GRN	C	A	CC	Frontotemporal Dementia	Mukherjee (2006)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
42388	GRN	C	T	CC	Frontotemporal Dementia	Rademakers (2002), Baker (2006), Cruts (2006), Baker (2006)
42389	GRN	G	A	GG	Frontotemporal Dementia	Gass (2006)
42390	GRN	G	A	GG	Frontotemporal Dementia	Baker (2006)
42391	GRN	C	T	CC	Frontotemporal Dementia	Baker (2006)
42392	GRN	C	T	CC	Frontotemporal Dementia	Baker (2006)
42393	GRN	C	T	CC	Frontotemporal Dementia	Gass (2006), Rademakers (2007)
43280	MAPT	G	A	GG	Frontotemporal dementia	Hayashi (2002)
43285	MAPT	C	G	CC	Frontotemporal dementia	Kobayashi (2003)
43290	MAPT	C	T	CC	Frontotemporal dementia	Sperfeld (1999), Bugiani (1999), Yasuda (2000), Lossos (2003), Werber (2003), Lossos (2003), Werber (2003), Yasuda (2005), Yasuda (2005), Yasuda (2005)
43293	MAPT	G	A	GG	Frontotemporal dementia	Iijima (1999)
43294	MAPT	T	G	TT	Frontotemporal dementia	van Herpen (2003)
43296	MAPT	C	T	CC	Frontotemporal dementia	Rosso (2002)
43297	MAPT	G	T	GG	Frontotemporal dementia	Neumann (2005)
43298	MAPT	A	G	AA	Frontotemporal dementia	Pickering-Brown (2004)
43300	MAPT	A	T	AA	Frontotemporal dementia	Lippa (2000), Lippa (2000)
43303	MAPT	G	A	GG	Frontotemporal dementia	Murrell (1999), Pickering-Brown (2000)
43304	MAPT	G	C	GG	Frontotemporal dementia	Murrell (1999)
43307	MAPT	C	T	CC	Frontotemporal dementia	Giaccone (2005)
43308	MAPT	G	A	GG	Frontotemporal dementia	Bird (2005)
43310	MAPT	C	G	CC	Frontotemporal dementia	Stanford (2003)
43311	MAPT	C	T	CC	Frontotemporal dementia	Yasuda (2000)
43315	MAPT	G	A	GG	Frontotemporal dementia	Stanford (2003)
43317	MAPT	T	C	TT	Frontotemporal dementia	Brown (1996), Spillantini (2000)
43320	MAPT	G	T	GG	Frontotemporal dementia	Malkani (2006)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
43323	MAPT	G	A	GG	Frontotemporal dementia	Rizzu (1999)
78906	CHMP2B	C	A	CC	Frontotemporal Dementia	Skibinski (2005)
78908	CHMP2B	G	C	GG	Frontotemporal Dementia	Skibinski (2005), Skibinski (2005), Cannon (2006)
64517	FLNA	T	G	TT	Frontometaphyseal dysplasia	Robertson (2003)
64547	FLNA	C	T	CC	Frontometaphyseal dysplasia	Robertson (2006)
64548	FLNA	G	C	GG	Frontometaphyseal dysplasia	Robertson (2006)
64556	FLNA	G	A	GG	Frontometaphyseal dysplasia	Robertson (2006)
64558	FLNA	G	A	GG	Frontometaphyseal dysplasia	Robertson (2003)
64559	FLNA	G	A	GG	Frontometaphyseal dysplasia	Robertson (2006)
64561	FLNA	A	G	AA	Frontometaphyseal dysplasia	Robertson (2006)
64563	FLNA	C	A	CC	Frontometaphyseal dysplasia	Zenker (2006)
64565	FLNA	A	C	AA	Frontometaphyseal dysplasia	Robertson (2006)
64567	FLNA	A	G	AA	Frontometaphyseal dysplasia	Robertson (2006)
20341	SLC4A1	C	T	CC	Froese Blood Group Antigen	McManus (2000)
37855	TTPA	C	T	CC	Friedreich-Like Ataxia with Isolated Vitamin E Deficiency	Hentati (1996)
37949	FXN	G	A	GG	Friedreich Ataxia	Zuhlke (1998)
37969	FXN	G	T	GG	Friedreich Ataxia	Bidichandani (1997), Koutnikova (1998), Delatycki (1999)
37960	FXN	T	G	TT	Friedreich Ataxia	Cossee (1999)
37949	FXN	G	A	GG	Friedreich Ataxia	Zuhlke (1998)
37960	FXN	T	G	TT	Friedreich Ataxia	Cossee (1999)
37961	FXN	T	C	TT	Friedreich Ataxia	Cossee (1999)
37962	FXN	G	T	GG	Friedreich Ataxia	Cossee (1997)
37963	FXN	A	C	AA	Friedreich Ataxia	Cossee (1999)
37964	FXN	A	T	AA	Friedreich Ataxia	Schwan (2004)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
37966	FXN	T	C	TT	Friedreich Ataxia	Bartolo (1998)
37967	FXN	T	G	TT	Friedreich Ataxia	Campuzano (1996)
37968	FXN	G	T	GG	Friedreich Ataxia	Cossee (1999)
37969	FXN	G	T	GG	Friedreich Ataxia	Bidichandani (1997), Koutnikova (1998), Delatycki (1999)
37970	FXN	C	G	CC	Friedreich Ataxia	Zuhlke (2004)
37971	FXN	A	T	AA	Friedreich Ataxia	Campuzano (1996), Koutnikova (1998)
37972	FXN	T	C	TT	Friedreich Ataxia	Labuda (1999)
37973	FXN	G	A	GG	Friedreich Ataxia	Castro (2000)
37974	FXN	T	C	TT	Friedreich Ataxia	Cossee (1999)
37975	FXN	G	C	GG	Friedreich Ataxia	De Michele (2000)
37976	FXN	C	T	CC	Friedreich Ataxia	Forrest (1998)
37977	FXN	C	T	CC	Friedreich Ataxia	Forrest (1998)
37978	FXN	T	A	TT	Friedreich Ataxia	Cossee (1999)
37979	FXN	A	G	AA	Friedreich Ataxia	Cossee (1999)
37980	FXN	T	G	TT	Friedreich Ataxia	Zuhlke (2004)
37982	FXN	G	C	GG	Friedreich Ataxia	McCormack (2000)
37983	FXN	A	G	AA	Friedreich Ataxia	Campuzano (1996)
37984	FXN	G	A	GG	Friedreich Ataxia	Doudney (1997)
37985	FXN	T	G	TT	Friedreich Ataxia	Forrest (1998)
48073	WT1	G	A	GG	Frasier Syndrome	Kohsaka (1999)
48093	WT1	G	A	GG	Frasier syndrome	Frasier , (1964), Blanchet , (1977), Haning , (1985), Kinberg , (1987), Haber , (1991), Barbaux (1997), Klamt (1998), Barbosa (1999), Melo (2002)
65315	FMR1	T	A	T	Fragile X Syndrome	Boulle (1993), Verheij (1995), Feng (1997), Darnell (2005)
70755	PRPH2	G	C	GG	Foveomacular Dystrophy, Adult-onset, with Choroidal Neovascularization	Gass (1974), Vine (1980), Jackson (1993)
66148	ACTN4	C	T	CC	Focal Segmental Glomerulosclerosis	Kaplan (2000)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
66150	ACTN4	T	C	TT	Focal Segmental Glomerulosclerosis	Weins (2005)
52878	LCAT	G	A	GG	Fish-eye Disease	Prydz (1992)
8158	ANTXR2	C	T	CC	Fibromatosis, juvenile hyaline	Dowling (2003)
8163	ANTXR2	T	A,C	TT	Fibromatosis, juvenile hyaline	Hanks (2003)
68366	MYH9	G	A	GG	Fechtner Syndrome	Consortium (2000), Epstein (1972), Heath (2001)
41358	MASS1	C	A	CC	Febrile and afebrile seizures	Nakayama (2002)
45819	BRIP1	G	A	GG	Fanconi Anemia, Complementation Group J	Levran (2005), Levitus (2005)
800981	BRIP1	G	C	GG	Fanconi Anemia, Complementation Group J	Levran (2005), Levitus (2005)
18542	FANCE	C	T	CC	Fanconi Anemia, Complementation Group E	Winter (2000)
18545	FANCE	G	A	GG	Fanconi Anemia, Complementation Group E	Winter (2000)
18567	FANCC	A	G	AA	Fanconi Anemia, Complementation Group C	T-to-C transition at base (1913), Strathdee (1992), Gavish (1992), Gavish (1993), Gavish (1993), Youssoufian (1996), Youssoufian (1996)
18568	FANCC	T	A	TT	Fanconi Anemia, Complementation Group C	Whitney (1993), Whitney (1994), Verlander (1995), Verlinsky (2001)
72208	FANCC	I	D	II	Fanconi Anemia, Complementation Group C	Strathdee (1992), Yamashita (1996), Pang (2001)
18736	FANCF	G	A,C	GG	Fanconi Anemia, Complementation Group A	Joenje , (1997), Winter (2000)
18738	FANCF	G	A,T	GG	Fanconi Anemia, Complementation Group A	Chandra (2005)
100851	LYZ	T	A	TT	Familial Visceral Amyloidosis	Granel (2002), Granel (2002), Granel (2005), Grateau (2006), Granel (2006)
2176	LMNA	G	A	GG	Familial Partial Lipodystrophy, Type II	Cao (2000), Shackleton (2000), Hegele (2000)
66792	NPHP1	C	A,T	CC	Familial Juvenile Nephronophthisis	Betz (2000)
2837	PRKAG2	C	T	CC	Familial Hypertrophic Cardiomyopathy with Wolff-Parkinson-White Syndrome|Wolff-Parkinson-White syndrome	Gollob (2001), Arad (2002)
800982	PRKAG2	C	T	CC	Wolff-Parkinson-White syndrome	Gollob (2001), Arad (2002)
2824	PRKAG2	G	T	GG	Familial Hypertrophic Cardiomyopathy with Wolff-Parkinson-White Syndrome	Arad (2002), Arad (2002)
2838	PRKAG2	T	C	TT	Familial Hypertrophic Cardiomyopathy with Wolff-Parkinson-White Syndrome	Blair (2001)
2840	PRKAG2	T	A	TT	Familial Hypertrophic Cardiomyopathy with Wolff-Parkinson-White Syndrome	Arad (2002)
100690	PRKAG2	D	I	DD	Familial Hypertrophic Cardiomyopathy with Wolff-Parkinson-White Syndrome	Blair (2001)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
3419	LDLR	A	C	AA	Familial Hypercholesterolemia	Jensen (1997), Jensen (1997)
3427	LDLR	C	T	CC	Familial Hypercholesterolemia	Knight (1989), Soutar (1989), Knight (1989), Soutar (1991), Rubinsztein (1992), Defesche (1992)
3410	LDLR	G	A	GG	Familial Hypercholesterolemia	Leitersdorf , (1989), Kotze (1991), Schuster (1993), Defesche (1993)
73605	LDLR	I	D	II	Familial Hypercholesterolemia	Koivisto (1992), Vuorio (1997)
16187	BAAT	T	C	TT	Familial Hypercholanemia	Carlton (2003)
38955	CACNA1A	C	T	CC	Familial Hemiplegic Migraine|Familial Hemiplegic Migraine with Progressive Cerebellar Ataxia|Sporadic Hemiplegic Migraine|Spinocerebellar Ataxia 6	Battistini (1999), Terwindt (2002), Alonso (2003), Alonso (2003)
38956	CACNA1A	G	A	GG	Familial Hemiplegic Migraine|Familial Hemiplegic Migraine with Progressive Cerebellar Ataxia|Sporadic Hemiplegic Migraine with Progressive Cerebellar Ataxia	Ophoff (1996), Ducros (1999), Friend (1999), Terwindt (2002), Kors (2003)
39977	ATP1A2	G	A	GG	Familial Hemiplegic Migraine 2	Terwindt (1997), Vanmolkot (2003), Segall (2005), Segall (2005)
39976	ATP1A2	T	C	TT	Familial Hemiplegic Migraine 2	Vanmolkot (2003), Segall (2005), Segall (2005)
38948	CACNA1A	C	T	CC	Familial Hemiplegic Migraine	Ophoff (1996)
40859	SCN1A	G	T	GG	Familial Hemiplegic Migraine	Dichgans (2005), Dichgans (2005)
48803	VHL	C	G	CC	Familial Erythrocytosis	Neumann (1995), Zbar (1996), Weirich (2002), Pastore (2003)
800855	VHL	C	G	CC	Von Hippel-Lindau Syndrome	Neumann (1995), Zbar (1996), Weirich (2002), Pastore (2003)
800856	VHL	C	G	CC	Pheochromocytoma	Neumann (1995), Zbar (1996), Weirich (2002), Pastore (2003)
36975	IKBKAP	G	A	GG	Familial Dysautonomia	Blumenfeld (1999), Leyne (2003)
62079	CIAS1	T	C	TT	Familial Cold Autoinflammatory Syndrome 1	Hoffman (2003)
66037	GSN	G	A	GG	Familial Amyloidosis, Finnish Type	Meretoja (1973), Sack (1981), Purcell (1983), Maury (1990), Maury, (1990), Ghiso (1990), Levy (1990), Hiltunen (1991), Maury (1991), Maury (1991), Maury (1991), Gorevic (1991), Paunio (1992), Haltia (1992), Chapelle (1992), Maury (1992), Maury, (1993), Sunada (1993), Paunio (1995), Steiner (1995), Aula (2002)
39705	PSEN2	A	T	AA	Familial Alzheimer Disease 4	Levy-Lahad (1995), Rogaev (1995), Tomita (1997), Tomita (1997), Marambaud (1998), Wijsman (2005), Wijsman (2005)
39735	APP	G	A	GG	Familial Alzheimer Disease 1	Kumar-Singh (2000), Edwards-Lee (2005)
39741	APP	C	A,G,T	CC	Familial Alzheimer Disease 1	Goate (1991), Naruse (1991), Yoshioka (1991), van Duijn (1991), Schellenberg (1991), Goate (1991), Karlinsky (1992), Karlinsky (1992), George-Hyslop (1994), George-Hyslop (1994), Maruyama (1996)
56538	PER2	T	C	TT	Familial Advanced Sleep Phase Syndrome	Jones (1999), Toh (2001)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
20984	F12	C	A,G,T	CC	Factor XII Deficiency	Schloesser (1995)
6129	F10	C	T	CC	Factor X Deficiency	James (1991)
6099	F10	G	A	GG	Factor X Deficiency	Marchetti (1995)
6127	F10	T	C	TT	Factor X Deficiency	Marchetti (1995)
4903	F7	C	T	CC	Factor VII Deficiency	Bernardi (1994), Herrmann (2000)
22361	MCFD2	C	T	CC	Factor V and Factor VIII Deficiency, Combined	Oeri , (1954), Zhang (2003)
22362	MCFD2	C	T	CC	Factor V and Factor VIII Deficiency, Combined	Zhang (2003)
29871	GLA	T	C	T	Fabry Disease, Cardiac Variant	Eng (1993), Davies (1993), Eng (1993)
29957	GLA	C	A,T	C	Fabry Disease, Cardiac Variant	Sakuraba (1990), Sawada (1996), Kase (2000)
29710	GLA	G	A	G	Fabry Disease	Verovnik (2004)
29825	GLA	C	G	C	Fabry Disease	Branton (2002)
29885	GLA	C	G,T	C	Fabry Disease	Eng (1993)
29886	GLA	G	A	G	Fabry Disease	Eng (1993), Davies (1993)
39825	TH	A	T	AA	Extrapyramidal Movement Disorder	Janssen (2000), Janssen (2000)
78080	EXT2	G	A	GG	Exostoses, Multiple, Type II	Philippe (1997)
801953	HBB	T	A	TT	Sickle Cell Anemia	Ingram (1956), Ingram (1959), Serjeant (1968), Dozy (1978), Mears (1981), Kan (1982), Orkin (1982), Antonarakis (1984), Pagnier (1984), Nagel (1985), Kulozik (1986), Jenkins (1987), Embury (1987), Ragusa (1988), Sammarco (1988), Barany (1991), Lapoumeroulie (1992), Zeng (1994), Currat (2002), Romero (2002), Kwiatkowski (2005), Serjeant (2007), Modiano (2008), Center for Pediatric Genomic Medicine of Children's Mercy Hospital and Clinics (2014), Emory Genetics Laboratory of Emory University (2014), Genetic Services Laboratory of the University of Chicago (2015), GeneDx (2016), Division of Human Genetics of Children's Hospital of Philadelphia (2016)
801954	HBB	T	G	TT	Hemoglobin G (Makassar)	Blackwell (1970)
1049	ESR1	C	T	CC	Estrogen Resistance	Smith (1994), Sudhir (1997)
73953	CHRNG	C	T	CC	Escobar Syndrome	Hoffmann (2006)
78168	SLC16A1	T	C	TT	Erythrocyte Lactate Transporter Defect	Fishbein (1986), Merezhinskaya (2000)
78169	SLC16A1	C	T	CC	Erythrocyte Lactate Transporter Defect	Merezhinskaya (2000)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
40242	SCN9A	A	T	AA	Erythermalgia, primary	Yang (2004), Cummins (2004), Rush (2006)
68367	MYH9	C	T	CC	Epstein Syndrome	Heath (2001), Seri (2002), Epstein (1972)
68368	MYH9	G	A	GG	Epstein Syndrome	Arrondel (2002), Utsch (2006)
800898	CACNA1A	C	T	CC	Episodic Ataxia, Type 2	Yue (1997), Yue (1997), Yue (1997), Wan (2005)
40300	KCNA1	G	A	GG	Episodic ataxia / myokymia	Scheffer (1998), Eunson (2000)
40302	KCNA1	C	T	CC	Episodic ataxia	Eunson (2000), Eunson (2000)
42513	CSTB	C	T	CC	Epilepsy, Progressive Myoclonus	Joensuu (2007)
42516	CSTB	T	C	TT	Epilepsy, Progressive Myoclonus	Lalioti (1997)
42549	EPM2A	G	A,C	GG	Epilepsy, Progressive Myoclonus	Minassian (1998), Minassian (1998), Gomez-Garre (2000)
42550	EPM2A	C	A,T	CC	Epilepsy, Progressive Myoclonus	Minassian (1998)
38441	KCNQ2	C	G,T	CC	Epilepsy, Benign Neonatal	Wang , (1996), Singh (1998)
801978	KCNQ2	C	G	CC	Epilepsy, Benign Neonatal	GeneDx (2016)
8580	KRT9	G	A	GG	Epidermolytic Palmoplantar Keratoderma associated with Knuckle Pads	Lu (2003), Lu (2003)
8561	KRT10	C	A,G,T	CC	Epidermolytic Hyperkeratosis|Nevus, Epidermal, Epidermolytic Hyerkeratotic Type	Rothnagel (1992), Cheng (1992), Cheng (1992), Cheng (1992), Chipev (1994), Paller (1994)
7146	KRT5	A	C,T	AA	Epidermolysis bullosa, Weber-Cockayne	Chan (1993), Ehrlich (1995)
7172	KRT5	A	G,T	AA	Epidermolysis bullosa, Weber-Cockayne	Chan (1994)
73015	KRT5	I	D	II	Epidermolysis Bullosa, Weber-Cockayne	Sprecher (2003)
7143	KRT5	G	A	GG	Epidermolysis bullosa, Weber-Cockayne	Muller (1998)
7144	KRT5	T	A	TT	Epidermolysis bullosa, Weber-Cockayne	Yasukawa (2006)
7145	KRT5	A	T	AA	Epidermolysis bullosa, Weber-Cockayne	Muller (1998)
7146	KRT5	A	C,T	AA	Epidermolysis bullosa, Weber-Cockayne	Chan (1993), Ehrlich (1995)
7147	KRT5	C	G,T	CC	Epidermolysis bullosa, Weber-Cockayne	Ciubotaru (2003)
7155	KRT5	T	C	TT	Epidermolysis bullosa, Weber-Cockayne	Liovic (2004)
7164	KRT5	G	C	GG	Epidermolysis bullosa, Weber-Cockayne	Humphries (1996)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
7165	KRT5	T	A,C,G	TT	Epidermolysis bullosa, Weber-Cockayne	Xu (2004)
7167	KRT5	A	C,G	AA	Epidermolysis bullosa, Weber-Cockayne	Ciubotaru (2003)
7169	KRT5	A	T	AA	Epidermolysis bullosa, Weber-Cockayne	Ciubotaru (2003)
7171	KRT5	A	T	AA	Epidermolysis bullosa, Weber-Cockayne	Chan (1994)
7172	KRT5	A	G,T	AA	Epidermolysis bullosa, Weber-Cockayne	Chan (1994)
7173	KRT5	T	A,C	TT	Epidermolysis bullosa, Weber-Cockayne	Li (2004)
7174	KRT5	T	A	TT	Epidermolysis bullosa, Weber-Cockayne	Matsuki (1995)
7175	KRT5	G	A,T	GG	Epidermolysis bullosa, Weber-Cockayne	Liovic (2000)
7176	KRT5	C	G	CC	Epidermolysis bullosa, Weber-Cockayne	Muller (1998)
7178	KRT5	G	A,T	GG	Epidermolysis bullosa, Weber-Cockayne	Chan (1994)
7179	KRT5	G	A,C,T	GG	Epidermolysis bullosa, Weber-Cockayne	Rugg (1993)
7182	KRT5	T	C	TT	Epidermolysis bullosa, Weber-Cockayne	Schuilenga-Hut (2003)
7185	KRT5	G	T	GG	Epidermolysis bullosa, Weber-Cockayne	Schuilenga-Hut (2003)
7232	KRT14	C	G	CC	Epidermolysis bullosa, Weber-Cockayne	Sorensen (1999)
7234	KRT14	C	T	CC	Epidermolysis bullosa, Weber-Cockayne	Chen (1995), Hu (1997), Shemanko (1998)
7250	KRT14	C	A,G,T	CC	Epidermolysis bullosa, Weber-Cockayne	Csikos (2004)
7255	KRT14	T	G	TT	Epidermolysis bullosa, Weber-Cockayne	Hovnanian (1993)
7256	KRT14	G	A,C	GG	Epidermolysis bullosa, Weber-Cockayne	Wood (2003)
7257	KRT14	A	T	AA	Epidermolysis bullosa, Weber-Cockayne	Chan (1994)
7260	KRT14	C	T	CC	Epidermolysis bullosa, Weber-Cockayne	Rugg (1993)
7263	KRT14	T	C	TT	Epidermolysis bullosa, Weber-Cockayne	Muller (1998)
7264	KRT14	G	T	GG	Epidermolysis bullosa, Weber-Cockayne	Chen (1995)
7266	KRT14	A	T	AA	Epidermolysis bullosa, Weber-Cockayne	Chen (1995)
7269	KRT14	G	A,C	GG	Epidermolysis bullosa, Weber-Cockayne	Chen (1995)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
7270	KRT14	C	G,T	CC	Epidermolysis bullosa, Weber-Cockayne	Ciubotaru (2003)
7272	KRT14	G	A,T	GG	Epidermolysis bullosa, Weber-Cockayne	Schuilenga-Hut (2003)
7276	KRT14	T	C	TT	Epidermolysis bullosa, Weber-Cockayne	Ciubotaru (2003)
7527	COL7A1	G	A	GG	Epidermolysis Bullosa, Pretibial	Gardella (2002)
7630	COL7A1	C	A	CC	Epidermolysis Bullosa, Pretibial	Christiano (1995)
7527	COL7A1	G	A	GG	Epidermolysis Bullosa, Pretibial	Gardella (2002)
7630	COL7A1	C	A	CC	Epidermolysis Bullosa, Pretibial	Christiano (1995)
7666	COL7A1	C	G	CC	Epidermolysis Bullosa, Pretibial	Gardella (2002)
6759	DSP	C	T	CC	Epidermolysis Bullosa, Lethal Acantholytic	Jonkman (2005)
6759	DSP	C	T	CC	Epidermolysis Bullosa, Lethal Acantholytic	Jonkman (2005)
7259	KRT14	G	T	GG	Epidermolysis bullosa, Koebner	Premaratne (2002)
7268	KRT14	A	G	AA	Epidermolysis bullosa, Koebner	Bonifas (1991), Bonifas (1991), Vassar (1991)
7282	KRT14	T	G	TT	Epidermolysis bullosa, Koebner	Jonkman (1996), Jonkman (1996)
7142	KRT5	A	T	AA	Epidermolysis bullosa, Koebner	Yasukawa (2006)
7150	KRT5	C	T	CC	Epidermolysis bullosa, Koebner	Yasukawa (2002)
7152	KRT5	C	G	CC	Epidermolysis bullosa, Koebner	Stephens (1995)
7160	KRT5	C	A,T	CC	Epidermolysis bullosa, Koebner	Yasukawa (2006)
7161	KRT5	C	A,T	CC	Epidermolysis bullosa, Koebner	Liovic (2001), Liovic (2001)
7168	KRT5	A	G	AA	Epidermolysis bullosa, Koebner	Galligan (1998)
7170	KRT5	A	G	AA	Epidermolysis bullosa, Koebner	Sorensen (1999)
7183	KRT5	C	T	CC	Epidermolysis bullosa, Koebner	Yasukawa (2002)
7186	KRT5	A	G	AA	Epidermolysis bullosa, Koebner	Ryynanen (1991), Dong (1993)
7196	KRT5	C	T	CC	Epidermolysis bullosa, Koebner	Yasukawa (2006)
7197	KRT5	C	G	CC	Epidermolysis bullosa, Koebner	Schuilenga-Hut (2003)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
7235	KRT14	T	C	TT	Epidermolysis bullosa, Koebner	Cummins (2001)
7237	KRT14	G	A	GG	Epidermolysis bullosa, Koebner	Yamanishi (1994)
7252	KRT14	C	G	CC	Epidermolysis bullosa, Koebner	Rugg (2000)
7254	KRT14	A	G	AA	Epidermolysis bullosa, Koebner	Sorensen (1999)
7259	KRT14	G	T	GG	Epidermolysis bullosa, Koebner	Premaratne (2002)
7261	KRT14	A	C,G	AA	Epidermolysis bullosa, Koebner	Schneider (2005)
7262	KRT14	A	C,G	AA	Epidermolysis bullosa, Koebner	Humphries (1993)
7265	KRT14	C	T	CC	Epidermolysis bullosa, Koebner	Corden (1998)
7268	KRT14	A	G	AA	Epidermolysis bullosa, Koebner	Bonifas (1991), Bonifas (1991), Vassar (1991)
7271	KRT14	G	A,C	GG	Epidermolysis bullosa, Koebner	Ciubotaru (2003)
7273	KRT14	C	A,T	CC	Epidermolysis bullosa, Koebner	Gu (2002)
7282	KRT14	T	G	TT	Epidermolysis bullosa, Koebner	Jonkman (1996), Jonkman (1996)
7283	KRT14	C	A,G,T	CC	Epidermolysis bullosa, Koebner	Schuilenga-Hut (2003)
74611	ITGA6	I	D	II	Epidermolysis bullosa, junctional, with pyloric atresia	Ruzzi (1997)
7444	LAMC2	C	T	CC	Epidermolysis Bullosa, Junctional, Herlitz Type	Aberdam (1994)
7724	LAMA3	C	T	CC	Epidermolysis Bullosa, Junctional, Herlitz Type	McGrath (1995), McGrath (1996)
7370	LAMB3	G	A	GG	Epidermolysis Bullosa, Junctional, Herlitz Type	McGrath (1995), Kivirikko (1996), Mellerio (1998)
7392	LAMB3	G	A	GG	Epidermolysis Bullosa, Junctional, Herlitz Type	Pulkkinen (1994), Ashton (1997), Pulkkinen (1997), Bruckner-Tuderman (1999), Muhle (2005), Muhle (2005)
7370	LAMB3	G	A	GG	Epidermolysis Bullosa, Junctional, Herlitz Type	McGrath (1995), Kivirikko (1996), Mellerio (1998)
7392	LAMB3	G	A	GG	Epidermolysis Bullosa, Junctional, Herlitz Type	Pulkkinen (1994), Ashton (1997), Pulkkinen (1997), Bruckner-Tuderman (1999), Muhle (2005), Muhle (2005)
7444	LAMC2	C	T	CC	Epidermolysis Bullosa, Junctional, Herlitz Type	Aberdam (1994)
7450	LAMC2	C	G	CC	Epidermolysis Bullosa, Junctional, Herlitz Type	Baudoin (1994)
7451	LAMC2	C	A	CC	Epidermolysis Bullosa, Junctional, Herlitz Type	Takizawa (2000)
7460	LAMC2	G	A	GG	Epidermolysis Bullosa, Junctional, Herlitz Type	Pulkkinen (1994), Pulkkinen (1994)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
7724	LAMA3	C	T	CC	Epidermolysis Bullosa, Junctional, Herlitz Type	McGrath (1995), McGrath (1996)
7721	LAMA3	A	G	AA	Epidermolysis bullosa, junctional	Posteraro (2004)
7443	LAMC2	C	T	CC	Epidermolysis bullosa, junctional	Nakano (2002)
7446	LAMC2	C	T	CC	Epidermolysis bullosa, junctional	Posteraro (2004)
7455	LAMC2	C	T	CC	Epidermolysis bullosa, junctional	Posteraro (2004)
7728	LAMA3	C	T	CC	Epidermolysis bullosa, junctional	Nakano (2002)
7843	ITGA6	C	T	CC	Epidermolysis bullosa, junctional	Gache (1998), Aloka (2000)
7726	LAMA3	G	T	GG	Epidermolysis bullosa, junctional	
7730	LAMA3	G	A	GG	Epidermolysis bullosa, junctional	Scaturro (2003)
7735	LAMA3	G	A	GG	Epidermolysis bullosa, junctional	Nakano (2002)
7844	ITGA6	G	T	GG	Epidermolysis bullosa, junctional	This mutation, (1856), Pulkkinen (1997)
7376	LAMB3	C	T	CC	Epidermolysis bullosa, junctional	McGrath (1995), Posteraro (1998), Mellerio (1998), Mavilio (2006)
7381	LAMB3	T	G	TT	Epidermolysis bullosa, junctional	Christiano (1996)
7383	LAMB3	G	A,C	GG	Epidermolysis bullosa, junctional	Varki (2006)
7371	LAMB3	G	A	GG	Epidermolysis bullosa, junctional	Mellerio (1998)
7373	LAMB3	C	G,T	CC	Epidermolysis bullosa, junctional	Posteraro (2004)
7376	LAMB3	C	T	CC	Epidermolysis bullosa, junctional	McGrath (1995), Posteraro (1998), Mellerio (1998), Mavilio (2006)
7379	LAMB3	A	T	AA	Epidermolysis bullosa, junctional	Nakano (2002)
7380	LAMB3	A	T	AA	Epidermolysis bullosa, junctional	Pulkkinen (1998)
7381	LAMB3	T	G	TT	Epidermolysis bullosa, junctional	Christiano (1996)
7382	LAMB3	A	G	AA	Epidermolysis bullosa, junctional	Nakano (2002)
7383	LAMB3	G	A,C	GG	Epidermolysis bullosa, junctional	Varki (2006)
7385	LAMB3	G	C	GG	Epidermolysis bullosa, junctional	Varki (2006)
7395	LAMB3	G	A	GG	Epidermolysis bullosa, junctional	Pulkkinen (1995)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
7397	LAMB3	G	A	GG	Epidermolysis bullosa, junctional	McGrath (1999)
7398	LAMB3	G	A	GG	Epidermolysis bullosa, junctional	Nakano (2002)
7402	LAMB3	T	C	TT	Epidermolysis bullosa, junctional	Varki (2006)
7404	LAMB3	G	A	GG	Epidermolysis bullosa, junctional	Nakano (2000)
7406	LAMB3	C	T	CC	Epidermolysis bullosa, junctional	Nakano (2002)
7408	LAMB3	T	C	TT	Epidermolysis bullosa, junctional	Takizawa (2000)
7409	LAMB3	C	T	CC	Epidermolysis bullosa, junctional	Cserhalmi-Friedman (1998)
7411	LAMB3	G	A	GG	Epidermolysis bullosa, junctional	Buchroithner (2004)
7412	LAMB3	C	T	CC	Epidermolysis bullosa, junctional	Nakano (2002)
7414	LAMB3	C	A	CC	Epidermolysis bullosa, junctional	Varki (2006)
7415	LAMB3	G	T	GG	Epidermolysis bullosa, junctional	Varki (2006)
7417	LAMB3	C	G	CC	Epidermolysis bullosa, junctional	Kivirikko (1996)
7419	LAMB3	C	T	CC	Epidermolysis bullosa, junctional	Pulkinnen (1995)
7443	LAMC2	C	T	CC	Epidermolysis bullosa, junctional	Nakano (2002)
7446	LAMC2	C	T	CC	Epidermolysis bullosa, junctional	Posteraro (2004)
7455	LAMC2	C	T	CC	Epidermolysis bullosa, junctional	Posteraro (2004)
7721	LAMA3	A	G	AA	Epidermolysis bullosa, junctional	Posteraro (2004)
7726	LAMA3	G	T	GG	Epidermolysis bullosa, junctional	
7728	LAMA3	C	T	CC	Epidermolysis bullosa, junctional	Nakano (2002)
7730	LAMA3	G	A	GG	Epidermolysis bullosa, junctional	Scaturro (2003)
7731	LAMA3	G	T	GG	Epidermolysis bullosa, junctional	Nakano (2002)
7732	LAMA3	G	A	GG	Epidermolysis bullosa, junctional	Posteraro (2004)
7735	LAMA3	G	A	GG	Epidermolysis bullosa, junctional	Nakano (2002)
7843	ITGA6	C	T	CC	Epidermolysis bullosa, junctional	Gache (1998), Aloka (2000)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
7844	ITGA6	G	T	GG	Epidermolysis bullosa, junctional	This mutation, (1856), Pulkkinen (1997)
7448	LAMC2	C	T	CC	Epidermolysis bullosa, Herlitz	Varki (2006)
7449	LAMC2	C	T	CC	Epidermolysis bullosa, Herlitz	Takizawa (1998)
7453	LAMC2	C	T	CC	Epidermolysis bullosa, Herlitz	Klausegger (2001)
7723	LAMA3	C	T	CC	Epidermolysis bullosa, Herlitz	Nakano (2002)
7459	LAMC2	G	T	GG	Epidermolysis bullosa, Herlitz	Vailly (1995)
7733	LAMA3	T	G	TT	Epidermolysis bullosa, Herlitz	Nakano (2002)
7375	LAMB3	G	A,C	GG	Epidermolysis bullosa, Herlitz	Takizawa (1998)
7391	LAMB3	C	T	CC	Epidermolysis bullosa, Herlitz	Takizawa (1998)
7399	LAMB3	G	A	GG	Epidermolysis bullosa, Herlitz	Engel, (1993), Engel, (1995), Benlian , (1996), Takizawa (1998), Takizawa (1998)
7369	LAMB3	C	T	CC	Epidermolysis bullosa, Herlitz	Cserhalmi-Friedman (2000)
7372	LAMB3	C	T	CC	Epidermolysis bullosa, Herlitz	Kon (1998)
7374	LAMB3	G	A	GG	Epidermolysis bullosa, Herlitz	Kivirikko (1996)
7375	LAMB3	G	A,C	GG	Epidermolysis bullosa, Herlitz	Takizawa (1998)
7377	LAMB3	G	A	GG	Epidermolysis bullosa, Herlitz	Kivirikko (1996)
7378	LAMB3	A	T	AA	Epidermolysis bullosa, Herlitz	Pulkkinen (1997)
7384	LAMB3	G	A	GG	Epidermolysis bullosa, Herlitz	Nakano (2000)
7386	LAMB3	C	A,T	CC	Epidermolysis bullosa, Herlitz	Varki (2006)
7387	LAMB3	G	A,T	GG	Epidermolysis bullosa, Herlitz	Varki (2006)
7388	LAMB3	G	A	GG	Epidermolysis bullosa, Herlitz	Varki (2006)
7389	LAMB3	G	A	GG	Epidermolysis bullosa, Herlitz	Kivirikko (1996)
7390	LAMB3	G	A	GG	Epidermolysis bullosa, Herlitz	Nakano (2002)
7391	LAMB3	C	T	CC	Epidermolysis bullosa, Herlitz	Takizawa (1998)
7394	LAMB3	G	A,T	GG	Epidermolysis bullosa, Herlitz	Christiano (1997)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
7396	LAMB3	G	T	GG	Epidermolysis bullosa, Herlitz	Schneider (2005)
7399	LAMB3	G	A	GG	Epidermolysis bullosa, Herlitz	Engel, (1993), Engel, (1995), Benlian (1996), Takizawa (1998), Takizawa (1998)
7400	LAMB3	G	A	GG	Epidermolysis bullosa, Herlitz	Christiano (1997)
7401	LAMB3	G	A	GG	Epidermolysis bullosa, Herlitz	Pulkkinen (1997)
7403	LAMB3	G	A	GG	Epidermolysis bullosa, Herlitz	Hata (2005)
7405	LAMB3	T	C	TT	Epidermolysis bullosa, Herlitz	Cserhalmi-Friedman (2000)
7407	LAMB3	T	G	TT	Epidermolysis bullosa, Herlitz	Takizawa (1998)
7410	LAMB3	C	T	CC	Epidermolysis bullosa, Herlitz	Pulkinnen (1997)
7413	LAMB3	A	T	AA	Epidermolysis bullosa, Herlitz	Nakano (2000)
7416	LAMB3	T	C,G	TT	Epidermolysis bullosa, Herlitz	Pulkinnen (1997)
7418	LAMB3	C	A	CC	Epidermolysis bullosa, Herlitz	Nakano (2000)
7445	LAMC2	C	T	CC	Epidermolysis bullosa, Herlitz	Takizawa (1998)
7448	LAMC2	C	T	CC	Epidermolysis bullosa, Herlitz	Varki (2006)
7449	LAMC2	C	T	CC	Epidermolysis bullosa, Herlitz	Takizawa (1998)
7452	LAMC2	A	T	AA	Epidermolysis bullosa, Herlitz	Kon (1998)
7453	LAMC2	C	T	CC	Epidermolysis bullosa, Herlitz	Klausegger (2001)
7454	LAMC2	C	T	CC	Epidermolysis bullosa, Herlitz	Spirito (2001)
7456	LAMC2	G	A	GG	Epidermolysis bullosa, Herlitz	Pulkkinen (1997)
7458	LAMC2	T	G	TT	Epidermolysis bullosa, Herlitz	Spirito (2001)
7459	LAMC2	G	T	GG	Epidermolysis bullosa, Herlitz	Vailly (1995)
7722	LAMA3	G	T	GG	Epidermolysis bullosa, Herlitz	Muhle (2005)
7723	LAMA3	C	T	CC	Epidermolysis bullosa, Herlitz	Nakano (2002)
7725	LAMA3	C	T	CC	Epidermolysis bullosa, Herlitz	Nakano (2002)
7727	LAMA3	A	T	AA	Epidermolysis bullosa, Herlitz	Muhle (2005)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
7733	LAMA3	T	G	TT	Epidermolysis bullosa, Herlitz	Nakano (2002)
7734	LAMA3	G	A	GG	Epidermolysis bullosa, Herlitz	Nakano (2002)
7447	LAMC2	C	T	CC	Epidermolysis Bullosa, Generalized Atrophic Benign	Nakano (2002)
7457	LAMC2	G	A	GG	Epidermolysis Bullosa, Generalized Atrophic Benign	Castiglia (2001)
7447	LAMC2	C	T	CC	Epidermolysis Bullosa, Generalized Atrophic Benign	Nakano (2002)
7457	LAMC2	G	A	GG	Epidermolysis Bullosa, Generalized Atrophic Benign	Castiglia (2001)
7729	LAMA3	C	T	CC	Epidermolysis Bullosa, Generalized Atrophic Benign	Nakano (2002)
7648	COL7A1	A	T	AA	Epidermolysis Bullosa, Dystrophica	Christiano (1993)
7159	KRT5	T	A	TT	Epidermolysis bullosa, Dowling-Meara ?	Pfendner (2003)
7158	KRT5	A	G	AA	Epidermolysis bullosa, Dowling-Meara	Shemanko (2000)
7195	KRT5	C	A,T	CC	Epidermolysis bullosa, Dowling-Meara	Muller (1999)
7153	KRT5	G	A	GG	Epidermolysis bullosa, Dowling-Meara	Nomura (1996)
7154	KRT5	T	C	TT	Epidermolysis bullosa, Dowling-Meara	Stephens (1997)
7156	KRT5	A	G	AA	Epidermolysis bullosa, Dowling-Meara	Stephens (1997)
7157	KRT5	G	T	GG	Epidermolysis bullosa, Dowling-Meara	Hamada (2005)
7158	KRT5	A	G	AA	Epidermolysis bullosa, Dowling-Meara	Shemanko (2000)
7187	KRT5	A	G	AA	Epidermolysis bullosa, Dowling-Meara	Irvine (1997)
7190	KRT5	T	A	TT	Epidermolysis bullosa, Dowling-Meara	Livingston (2001)
7191	KRT5	T	C	TT	Epidermolysis bullosa, Dowling-Meara	Bonifas (1991), Ryynanen (1991), Lane (1992)
7192	KRT5	C	T	CC	Epidermolysis bullosa, Dowling-Meara	Schuilenga-Hut (2003)
7194	KRT5	C	A,T	CC	Epidermolysis bullosa, Dowling-Meara	Stephens (1997)
7195	KRT5	C	A,T	CC	Epidermolysis bullosa, Dowling-Meara	Muller (1999)
7198	KRT5	C	T	CC	Epidermolysis bullosa, Dowling-Meara	Rugg (1999)
7207	KRT14	A	G	AA	Epidermolysis Bullosa, Dowling-Meara	Chen (1995), Hu (1997), Shemanko (1998)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
7236	KRT14	T	C,G	TT	Epidermolysis bullosa, Dowling-Meara	Chen (1995)
7238	KRT14	T	C	TT	Epidermolysis bullosa, Dowling-Meara	Sorensen (1999)
7239	KRT14	A	T	GA	Epidermolysis bullosa, Dowling-Meara	Csikos (2004)
7240	KRT14	G	T	GG	Epidermolysis bullosa, Dowling-Meara	Chen (1995)
7241	KRT14	G	C	GG	Epidermolysis bullosa, Dowling-Meara	Csikos (2004)
7242	KRT14	G	A,C	GG	Epidermolysis bullosa, Dowling-Meara	Coulombe (1991), Coulombe (1991), Sasaki (1999), Ma (2001)
7243	KRT14	C	A,G,T	CC	Epidermolysis bullosa, Dowling-Meara	Coulombe (1991), Shemanko (2000)
7244	KRT14	C	G	CC	Epidermolysis bullosa, Dowling-Meara	Morley (2003)
7245	KRT14	A	C	AA	Epidermolysis bullosa, Dowling-Meara	Chan (1996)
7247	KRT14	A	G	AA	Epidermolysis bullosa, Dowling-Meara	Schuilenga-Hut (2003)
7248	KRT14	C	A,G,T	CC	Epidermolysis bullosa, Dowling-Meara	Hamada (2005)
7275	KRT14	A	G	AA	Epidermolysis bullosa, Dowling-Meara	Hut (2000)
7277	KRT14	C	G	CC	Epidermolysis bullosa, Dowling-Meara	Wood (2003)
7327	ITGB4	G	A	GG	Epidermolysis Bullosa without Pyloric Atresia|Epidermolysis Bullosa, Generalized Atrophic Benign	Inoue (2000)
7327	ITGB4	G	A	GG	Epidermolysis Bullosa without Pyloric Atresia|Epidermolysis Bullosa, Generalized Atrophic Benign	Inoue (2000)
7331	ITGB4	G	A	GG	Epidermolysis Bullosa with Pyloric Atresia	Mellerio (1998)
7311	ITGB4	T	C	TT	Epidermolysis Bullosa with Pyloric Atresia	Pulkkinen (1998)
7306	ITGB4	T	C	TT	Epidermolysis Bullosa with Pyloric Atresia	Mellerio (1998)
7307	ITGB4	C	T	CC	Epidermolysis Bullosa with Pyloric Atresia	Varki (2006)
7308	ITGB4	G	A	GG	Epidermolysis Bullosa with Pyloric Atresia	Pulkkinen (1998)
7309	ITGB4	C	T	CC	Epidermolysis Bullosa with Pyloric Atresia	Pulkkinen (1998)
7310	ITGB4	G	T	GG	Epidermolysis Bullosa with Pyloric Atresia	Nakano (2001)
7311	ITGB4	T	C	TT	Epidermolysis Bullosa with Pyloric Atresia	Pulkkinen (1998)
7312	ITGB4	T	G	TT	Epidermolysis Bullosa with Pyloric Atresia	Pulkkinen (1998)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
7313	ITGB4	G	A	GG	Epidermolysis Bullosa with Pyloric Atresia	Varki (2006)
7314	ITGB4	C	T	CC	Epidermolysis Bullosa with Pyloric Atresia	Pulkkinen (1998)
7315	ITGB4	G	A	GG	Epidermolysis Bullosa with Pyloric Atresia	Nakano (2001)
7316	ITGB4	C	T	CC	Epidermolysis Bullosa with Pyloric Atresia	Nakano (2001)
7317	ITGB4	T	A	TT	Epidermolysis Bullosa with Pyloric Atresia	Nakano (2001)
7318	ITGB4	T	C	TT	Epidermolysis Bullosa with Pyloric Atresia	Nakano (2001)
7319	ITGB4	G	T	GG	Epidermolysis Bullosa with Pyloric Atresia	Pfendner (2003)
7320	ITGB4	A	C	AA	Epidermolysis Bullosa with Pyloric Atresia	Masunaga (2004)
7321	ITGB4	C	T	CC	Epidermolysis Bullosa with Pyloric Atresia	Pulkkinen (1998)
7322	ITGB4	T	C	TT	Epidermolysis Bullosa with Pyloric Atresia	Pulkkinen (1998)
7323	ITGB4	T	C	TT	Epidermolysis Bullosa with Pyloric Atresia	Pulkkinen (1998)
7324	ITGB4	C	A	CC	Epidermolysis Bullosa with Pyloric Atresia	Varki (2006)
7325	ITGB4	C	A	CC	Epidermolysis Bullosa with Pyloric Atresia	Pulkkinen (1998)
7326	ITGB4	C	T	CC	Epidermolysis Bullosa with Pyloric Atresia	Varki (2006)
7328	ITGB4	G	A	GG	Epidermolysis Bullosa with Pyloric Atresia	Nakano (2001), Koster (2001)
7329	ITGB4	C	T	CC	Epidermolysis Bullosa with Pyloric Atresia	Pulkkinen (1998), Koster (2001)
7330	ITGB4	G	T	GG	Epidermolysis Bullosa with Pyloric Atresia	Varki (2006)
7331	ITGB4	G	A	GG	Epidermolysis Bullosa with Pyloric Atresia	Mellerio (1998)
7332	ITGB4	C	T	CC	Epidermolysis Bullosa with Pyloric Atresia	Nakano (2001)
7333	ITGB4	A	G	AA	Epidermolysis Bullosa with Pyloric Atresia	Ashton (2001)
7334	ITGB4	C	T	CC	Possibly associated with Epidermolysis Bullosa with Pyloric Atresia	Nakano (2001)
7335	ITGB4	G	A	GG	Epidermolysis Bullosa with Pyloric Atresia	Varki (2006)
7336	ITGB4	G	A	GG	Epidermolysis Bullosa with Pyloric Atresia	Mellerio (1998)
7337	ITGB4	T	A	TT	Epidermolysis Bullosa with Pyloric Atresia	Chavanas (1999), Chavanas (1999)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
7807	PLEC1	G	A	GG	Epidermolysis bullosa with pyloric atresia	Pfendner (2005)
7809	PLEC1	G	A	GG	Epidermolysis bullosa with pyloric atresia	Nakamura (2005)
7823	PLEC1	G	A	GG	Epidermolysis bullosa with pyloric atresia	Nakamura (2005)
7824	PLEC1	G	A,C	GG	Epidermolysis bullosa with pyloric atresia	Pfendner (2005)
7826	PLEC1	C	T	CC	Epidermolysis bullosa with pyloric atresia	Nakamura (2005)
7817	PLEC1	G	A,C,T	GG	Epidermolysis Bullosa Simplex, Ogna Type	Gedde-Dahl (1971), Koss-Harnes (2002)
7817	PLEC1	G	A,C,T	GG	Epidermolysis Bullosa Simplex, Ogna Type	Gedde-Dahl (1971), Koss-Harnes (2002)
7141	KRT5	G	A	GG	Epidermolysis bullosa Simplex with Mottled Pigmentation	Lersch (1989), Uttam (1996), Irvine (1997), Until (1999), Moog (1999), Irvine (2001), Irvine (2001)
7181	KRT5	G	A,T	GG	Epidermolysis Bullosa Simplex	Yasukawa (2006)
7148	KRT5	C	T	CC	Epidermolysis Bullosa Simplex	Muller (2006)
7149	KRT5	C	G	CC	Epidermolysis Bullosa Simplex	Muller (2006)
7151	KRT5	T	A,C	TT	Epidermolysis Bullosa Simplex	Rugg (2007)
7162	KRT5	C	T	CC	Epidermolysis Bullosa Simplex	Muller (2006)
7163	KRT5	T	G	TT	Epidermolysis Bullosa Simplex	Yasukawa (2006)
7166	KRT5	T	A,C,G	TT	Epidermolysis Bullosa Simplex	
7177	KRT5	T	C	TT	Epidermolysis Bullosa Simplex	Rugg (2007)
7180	KRT5	C	T	CC	Epidermolysis Bullosa Simplex	Muller (2006)
7181	KRT5	G	A,T	GG	Epidermolysis Bullosa Simplex	Yasukawa (2006)
7184	KRT5	G	A	GG	Epidermolysis Bullosa Simplex	Rugg (2007)
7188	KRT5	T	G	TT	Epidermolysis Bullosa Simplex	Rugg (2007)
7189	KRT5	T	G	TT	Epidermolysis Bullosa Simplex	Muller (2006)
7193	KRT5	C	G,T	CC	Epidermolysis Bullosa Simplex	
7233	KRT14	T	A,C	TT	Epidermolysis Bullosa Simplex	Rugg (2007)
7246	KRT14	T	C	TT	Epidermolysis Bullosa Simplex	Rugg (2007)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
7249	KRT14	C	G	CC	Epidermolysis Bullosa Simplex	Rugg (2007)
7251	KRT14	A	G	AA	Epidermolysis Bullosa Simplex	Rugg (2007)
7253	KRT14	G	A	GG	Epidermolysis Bullosa Simplex	Indelman (2005)
7258	KRT14	C	G	CC	Epidermolysis Bullosa Simplex	Muller (2006)
7267	KRT14	A	G,T	AA	Epidermolysis Bullosa Simplex	Rugg (2007)
7278	KRT14	C	G	CC	Epidermolysis Bullosa Simplex	Muller (2006)
7279	KRT14	G	C	GG	Epidermolysis Bullosa Simplex	Rugg (2007)
7280	KRT14	A	T	AA	Epidermolysis Bullosa Simplex	Hut (2000)
7281	KRT14	C	G,T	CC	Epidermolysis Bullosa Simplex	Hut (2000)
7811	PLEC1	G	A	GG	Epidermolysis Bullosa Simplex	Bauer (2001)
7813	PLEC1	G	A	GG	Epidermolysis Bullosa Simplex	Kunz (2000)
7820	PLEC1	G	A	GG	Epidermolysis Bullosa Simplex	Kunz (2000)
7520	COL7A1	G	A	GG	Epidermolysis Bullosa Pruriginosa	Drera (2006)
7536	COL7A1	C	T	CC	Epidermolysis Bullosa Pruriginosa	Mellerio (1999)
7584	COL7A1	C	A	CC	Epidermolysis Bullosa Pruriginosa	Drera (2006)
7614	COL7A1	C	A	CC	Epidermolysis Bullosa Pruriginosa	Chuang (2004)
7615	COL7A1	C	T	CC	Epidermolysis Bullosa Pruriginosa	Mellerio (1999)
7639	COL7A1	C	G	CC	Epidermolysis Bullosa Pruriginosa	Mellerio (1999)
7687	COL7A1	C	T	CC	Epidermolysis Bullosa Pruriginosa	Mellerio (1999)
7702	COL7A1	T	C	TT	Epidermolysis Bullosa Pruriginosa	Jiang (2002)
7705	COL7A1	C	T	CC	Epidermolysis Bullosa Pruriginosa	Gardella (1996), Gardella (1996), Gardella (1996), Drera (2006)
7510	COL7A1	C	G	CC	Epidermolysis Bullosa Dystrophica, Localisata Variant	Terracina (1998)
7670	COL7A1	T	C	TT	Epidermolysis Bullosa Dystrophica, Localisata Variant	Gardella (1996), Kahofer (2003), Drera (2006), Centre for Mendelian Genomics of the University Medical Centre Ljubljana (2017), GeneDx (2017)
7692	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica, Localisata Variant	Terracina (1998)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
7549	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica ?	Mallipeddi (2003)
7477	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Christiano (1996)
7478	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Gardella (2000)
7479	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Pfendner (2003)
7480	COL7A1	A	G	AA	Epidermolysis Bullosa Dystrophica	Jiang (2005)
7481	COL7A1	G	C	GG	Epidermolysis Bullosa Dystrophica	Drera (2006)
7482	COL7A1	C	G	CC	Epidermolysis Bullosa Dystrophica	Varki (2007)
7483	COL7A1	C	A	CC	Epidermolysis Bullosa Dystrophica	Pfendner (2003)
7484	COL7A1	G	A,T	GG	Epidermolysis Bullosa Dystrophica	Sawamura (2005)
7485	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Hovnanian (1997)
7486	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7487	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Christiano (1996)
7489	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Christiano (1996)
7490	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Hovnanian (1997)
7492	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7493	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Dunnill (1994)
7494	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Varki (2007)
7495	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Sawamura (2005)
7496	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Cserhalmi-Friedman (1998)
7497	COL7A1	G	A,T	GG	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7498	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Christiano (1996)
7500	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Kern (2006)
7501	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Kern (2006)
7502	COL7A1	C	G	CC	Epidermolysis Bullosa Dystrophica	Posteraro (2005)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
7503	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Pfendner (2003)
7504	COL7A1	C	A	CC	Epidermolysis Bullosa Dystrophica	Varki (2007)
7505	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	
7507	COL7A1	T	G	TT	Epidermolysis Bullosa Dystrophica	Ryoo (2001)
7511	COL7A1	C	A,G,T	CC	Epidermolysis Bullosa Dystrophica	Kern (2006)
7512	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Kern (2006)
7514	COL7A1	C	G	CC	Epidermolysis Bullosa Dystrophica	Kern (2006)
7515	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7516	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Christiano (1996)
7518	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7519	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Kern (2006)
7521	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7522	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Cserhalmi-Friedman (1997)
7523	COL7A1	C	G	CC	Epidermolysis Bullosa Dystrophica	Gardella (2002)
7524	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Sawamura (2005)
7525	COL7A1	G	C	GG	Epidermolysis Bullosa Dystrophica	Kim (2003)
7526	COL7A1	G	A	GG	Possibly associated with Epidermolysis Bullosa Dystrophica	Pfendner (2003), Kern (2005), Vahidnezhad (2016)
7528	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7529	COL7A1	C	G	CC	Epidermolysis Bullosa Dystrophica	Kern (2006)
7530	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7531	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Posteraro (2005)
7532	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7533	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7534	COL7A1	C	G,T	CC	Epidermolysis Bullosa Dystrophica	Christiano (1996)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
7535	COL7A1	C	A	CC	Epidermolysis Bullosa Dystrophica	Kern (2006)
7537	COL7A1	C	G	CC	Epidermolysis Bullosa Dystrophica	Masunaga (2000)
7540	COL7A1	G	A,T	GG	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7541	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Varki (2007)
7542	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Sawamura (2005)
7543	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7544	COL7A1	C	A	CC	Epidermolysis Bullosa Dystrophica	Hovnanian (1997)
7547	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Hammami-Hauasli (1998)
7548	COL7A1	C	G	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7550	COL7A1	G	C	GG	Epidermolysis Bullosa Dystrophica	Hovnanian (1997)
7551	COL7A1	G	A,C	GG	Epidermolysis Bullosa Dystrophica	Kon (1998)
7552	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Winberg (1997)
7553	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Matsuba (2002)
7554	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Gardella (2002)
7555	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Hammami-Hauasli (1998)
7556	COL7A1	C	G	CC	Epidermolysis Bullosa Dystrophica	Hovnanian (1997)
7557	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Pfendner (2003)
7558	COL7A1	C	G,T	CC	Epidermolysis Bullosa Dystrophica	Lee (2000)
7559	COL7A1	C	G	CC	Epidermolysis Bullosa Dystrophica	Murata (2000)
7560	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Nordal (2001)
7561	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Fine (1989), Kon (1997), Hammami-Hauasli (1998), Mecklenbeck (1999), Mecklenbeck (1999), Martinez-Mir (2002), Martinez-Mir (2002)
7562	COL7A1	C	A	CC	Epidermolysis Bullosa Dystrophica	Rouan (1998)
7563	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Kern (2006)
7564	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Jonkman (1999), Sawamura (2006)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
7565	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Iwata (2006)
7566	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7567	COL7A1	C	A	CC	Epidermolysis Bullosa Dystrophica	Rouan (1998)
7568	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Christiano (1994)
7569	COL7A1	C	A,T	CC	Epidermolysis Bullosa Dystrophica	Christiano (1995), Mellerio (1998), Mellerio (1998), Mellerio (1998)
7570	COL7A1	C	A,T	CC	Epidermolysis Bullosa Dystrophica	Mecklenbeck (1999)
7571	COL7A1	C	A	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7572	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Hovnanian (1997)
7573	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Christiano (1996)
7574	COL7A1	T	C	TT	Epidermolysis Bullosa Dystrophica	Kern (2006)
7575	COL7A1	G	C	GG	Epidermolysis Bullosa Dystrophica	Hovnanian (1997)
7576	COL7A1	G	A,C	GG	Epidermolysis Bullosa Dystrophica	Hovnanian (1997)
7577	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Sawamura (2005)
7578	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Rouan (1998)
7579	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Posteraro (2005)
7580	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Kahofer (2003)
7581	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Gardella (2002)
7582	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Zhang (2003)
7583	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Dunnill (1996)
7585	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Kon (1997)
7586	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Kon (1997)
7587	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Christiano (1999)
7588	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Suzuki (2006)
7589	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
7590	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Kern (2006)
7591	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7592	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Kim (2003)
7593	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7594	COL7A1	C	A	CC	Epidermolysis Bullosa Dystrophica	Hovnanian (1997)
7595	COL7A1	A	T	AA	Epidermolysis Bullosa Dystrophica	Ryoo (2001)
7596	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Ryoo (2001)
7597	COL7A1	G	C	GG	Epidermolysis Bullosa Dystrophica	Ryoo (2001)
7598	COL7A1	C	A	CC	Epidermolysis Bullosa Dystrophica	Ryoo (2001)
7599	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7600	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7601	COL7A1	C	G,T	CC	Epidermolysis Bullosa Dystrophica	Lee (1997), Mellerio (1999)
7603	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Sato-Matsumura (2003)
7604	COL7A1	C	A	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7606	COL7A1	C	A	CC	Epidermolysis Bullosa Dystrophica	Posteraro (2005)
7607	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Shimizu (1999)
7608	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	
7609	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Pfendner (2003)
7611	COL7A1	C	G	CC	Epidermolysis Bullosa Dystrophica	Cserhalmi-Friedman (1999)
7612	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Hashimoto (1999)
7613	COL7A1	C	A	CC	Epidermolysis Bullosa Dystrophica	Sawamura (2005)
7616	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7617	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Varki (2007)
7618	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Kern (2006)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
7619	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7620	COL7A1	T	C	TT	Epidermolysis Bullosa Dystrophica	Varki (2007)
7621	COL7A1	G	A,C	GG	Epidermolysis Bullosa Dystrophica	Varki (2007)
7622	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Christiano (1996)
7623	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Varki (2007)
7624	COL7A1	G	A,T	GG	Epidermolysis Bullosa Dystrophica	Gardella (2002)
7625	COL7A1	C	G	CC	Epidermolysis Bullosa Dystrophica	Christiano (1996)
7626	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Shimizu (1996)
7627	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Hovnanian (1997)
7628	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica, Recessive	Gardella (2002), Cuadrado-Corrales (2010), Wertheim-Tysarowska (2012), van den Akker (2013)
801990	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Brittingham (2005), Steplewski (2012), GeneDx (2015)
7629	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Sawamura (2006)
7631	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Posteraro (2005)
7632	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Christiano (1996)
7633	COL7A1	C	A	CC	Epidermolysis Bullosa Dystrophica	Kon (1997)
7634	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7635	COL7A1	C	G	CC	Epidermolysis Bullosa Dystrophica	Christiano (1996)
7636	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Gardella (1999)
7637	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Kern (2006)
7640	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Rouan (1998)
7641	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Varki (2007)
7642	COL7A1	C	G	CC	Epidermolysis Bullosa Dystrophica	Kern (2006)
7643	COL7A1	C	G	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7644	COL7A1	C	A,T	CC	Epidermolysis Bullosa Dystrophica	Christiano (1996)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
7645	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Kon (1998)
7646	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7649	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Christiano (1996)
7650	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Sato-Matsumura (2002)
7651	COL7A1	C	A,G,T	CC	Epidermolysis Bullosa Dystrophica	Shimizu (1996)
7652	COL7A1	C	A	CC	Epidermolysis Bullosa Dystrophica	Sawamura (2005)
7653	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Varki (2007)
7654	COL7A1	C	G	CC	Epidermolysis Bullosa Dystrophica	Pfendner (2003)
7655	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Pfendner (2003)
7656	COL7A1	C	G	CC	Epidermolysis Bullosa Dystrophica	Hovnanian (1997)
7657	COL7A1	T	C	TT	Epidermolysis Bullosa Dystrophica	Kon (1998)
7658	COL7A1	A	T	AA	Epidermolysis Bullosa Dystrophica	Natsuga (2005)
7660	COL7A1	C	A	CC	Epidermolysis Bullosa Dystrophica	Sawamura (2005)
7661	COL7A1	T	C	TT	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7662	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7663	COL7A1	C	G	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7664	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Kern (2006)
7665	COL7A1	G	C	GG	Epidermolysis Bullosa Dystrophica	Hovnanian (1997)
7667	COL7A1	A	C,G	AA	Epidermolysis Bullosa Dystrophica	Kern (2006)
7668	COL7A1	A	C	AA	Epidermolysis Bullosa Dystrophica	Kern (2006)
7669	COL7A1	G	C	GG	Epidermolysis Bullosa Dystrophica	Kon (1998)
7671	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Kern (2006)
7672	COL7A1	C	A	CC	Epidermolysis Bullosa Dystrophica	Wessagowit (2005)
7673	COL7A1	T	C	TT	Epidermolysis Bullosa Dystrophica	Kern (2006)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
7675	COL7A1	G	A,T	GG	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7676	COL7A1	G	C	GG	Epidermolysis Bullosa Dystrophica	Pfendner (2003)
7677	COL7A1	G	T	GG	Epidermolysis Bullosa Dystrophica	Kon (1998)
802232	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Mellerio (1999)
7678	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7679	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7680	COL7A1	C	G,T	CC	Epidermolysis Bullosa Dystrophica	Hovnanian (1997)
7681	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Masse (2005)
7682	COL7A1	G	A	GG	Epidermolysis Bullosa Dystrophica	Gardella (2002)
7683	COL7A1	C	A	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7684	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7685	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7686	COL7A1	T	C	TT	Epidermolysis Bullosa Dystrophica	Horev (2003)
7689	COL7A1	T	G	TT	Epidermolysis Bullosa Dystrophica	Kon (1998)
7691	COL7A1	G	T	GG	Epidermolysis Bullosa Dystrophica	Csikos (2005)
7693	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7694	COL7A1	C	A	CC	Epidermolysis Bullosa Dystrophica	Kon (1998)
7695	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Christiano (1996)
7696	COL7A1	C	G	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7697	COL7A1	C	A	CC	Epidermolysis Bullosa Dystrophica	Gardella (2002)
7698	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7699	COL7A1	C	G	CC	Epidermolysis Bullosa Dystrophica	Tamai (1997)
7700	COL7A1	T	A	TT	Epidermolysis Bullosa Dystrophica	Christiano (1996)
7701	COL7A1	T	C	TT	Epidermolysis Bullosa Dystrophica	

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
7703	COL7A1	T	C	TT	Epidermolysis Bullosa Dystrophica	Whittock (1999)
7704	COL7A1	C	T	CC	Epidermolysis Bullosa Dystrophica	Titeux (2006)
7488	COL7A1	G	A	GG	Epidermolysis Bullosa	Hovnanian (1994)
7499	COL7A1	C	T	CC	Epidermolysis Bullosa	Christiano (1994)
7506	COL7A1	G	A	GG	Epidermolysis Bullosa	Pulkkinen (1999)
7508	COL7A1	G	A	GG	Epidermolysis Bullosa	Hovnanian (1994)
7509	COL7A1	G	A	GG	Epidermolysis Bullosa	Hovnanian (1994)
7539	COL7A1	T	G	TT	Epidermolysis Bullosa	Pulkkinen (1999)
7545	COL7A1	G	A,C	GG	Epidermolysis Bullosa	Pulkkinen (1999)
7610	COL7A1	G	A,T	GG	Epidermolysis Bullosa	Pulkkinen (1999)
7638	COL7A1	C	T	CC	Epidermolysis Bullosa	Pulkkinen (1999)
7647	COL7A1	G	A	GG	Epidermolysis Bullosa	Pulkkinen (1999)
7659	COL7A1	C	G	CC	Epidermolysis Bullosa	Pulkkinen (1999)
7690	COL7A1	C	A	CC	Epidermolysis Bullosa	Pulkkinen (1999)
52369	NR2E3	C	T	CC	Enhanced S-cone Syndrome	Haider (2000)
49725	MSH6	G	A	GG	Endometrial cancer	Wiest (2006)
71697	SERPINI1	A	C	AA	Encephalopathy, Familial, with Neuroserpin Inclusion Bodies	Davis (1999), Davis (1999), Davis (1999), Takao (2000), Davis (2002)
30409	ETHE1	G	A,C	GG	Encephalopathy, Ethylmalonic	Tiranti (2004)
19387	SPTA1	G	A,T	GG	Elliptocytosis, Rhesus-Unlinked Type	Lecomte (1987), Garbarz (1989), Coetzer (1991), Gallagher (1991), Coetzer (1991), Coetzer (1991), Baklouti (1991), Coetzer (1991), Gallagher (1991), Coetzer (1991), Lorenzo (1993)
19387	SPTA1	G	A,T	GG	Elliptocytosis, Rhesus-Unlinked Type	Lecomte (1987), Garbarz (1989), Coetzer (1991), Gallagher (1991), Coetzer (1991), Coetzer (1991), Baklouti (1991), Coetzer (1991), Gallagher (1991), Coetzer (1991), Lorenzo (1993)
19436	SPTA1	C	T	CC	Elliptocytosis, Rhesus-Unlinked Type	Alloisio (1988), Alloisio (1993), Alloisio (1993)
21508	SPTB	A	C	AA	Elliptocytosis|Hemolytic Anemia, Neonatal Nonimmune, Fatal and Near-fatal	Gallagher (1997), Gallagher (1997)
20400	EPB41	T	G	TT	Elliptocytosis	The propositus, born in (1948), Venezia (1992), Venezia (1992), Venezia (1992)
34108	COL1A2	A	G	AA	Ehlers-Danlos Syndrome, Type VII-B	Byers (1997)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
34110	COL1A2	G	C	GG	Ehlers-Danlos Syndrome, Type VII-B	Chiodo (1992), Carr (1994)
34112	COL1A2	G	A	GG	Ehlers-Danlos Syndrome, Type VII-B	Minor (1986), Viljoen (1987), Vasan (1991), Vasan (1991), Nicholls (1991), Watson (1992), Lehmann (1994)
34114	COL1A2	T	C	TT	Ehlers-Danlos Syndrome, Type VII-B	Wirtz (1987), Weil (1988), Weil (1988), Ho (1994)
35918	PLOD1	C	T	CC	Ehlers-Danlos Syndrome, Type VIA and Nevo Syndrome	Barnes, (1985), Hyland (1992), Hyland , (1992), Steinmann (1993), Steinmann (1993), Steinmann (1995), Al-Gazali (1997), Giunta (2005), Giunta (2005)
35921	PLOD1	C	G	CC	Ehlers-Danlos Syndrome, Type VIA	The male patient was born in (1989), Walker (1997), Walker, (1999), Walker (1999)
35928	PLOD1	G	A	GG	Ehlers-Danlos Syndrome, Type VIA	The male patient was born in (1989), Walker (1997), Walker, (1999), Walker (1999)
35348	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35361	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35381	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35385	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Anderson (1997), Pepin (2000)
35431	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Palmeri (2003)
35433	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Richards (1991), Nuytinck (1992), Nuytinck (1992)
35348	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35349	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Smith (1997)
35350	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35351	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35352	COL3A1	G	C	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35353	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Giunta (2000)
35354	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35355	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Giunta (2000)
35356	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35357	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35358	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Smith (1997)
35359	COL3A1	G	C	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
35360	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35361	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35362	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35363	COL3A1	G	C	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35364	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35365	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35366	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35367	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Giunta (2000)
35368	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35369	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Kroes (2003)
35370	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Tromp (1993)
35371	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35372	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Giunta (2000)
35373	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35374	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35375	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35376	COL3A1	G	C	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35377	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Hassan (2002)
35378	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35379	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35380	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35381	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35382	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35383	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
35384	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Mackay (1996)
35385	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Anderson (1997)
35386	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35387	COL3A1	G	C	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35388	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35389	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35390	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pope (1988), McGrory (1996)
35393	COL3A1	G	C	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35394	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Bateman (1998)
35395	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35396	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Gilchrist (1999)
35397	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35398	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Madhatheri (1994)
35399	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Mackay (1996)
35402	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Richards (1993)
35403	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35404	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35405	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35406	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35407	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Watanabe (2002)
35408	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35409	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35410	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Johnson (1995)
35411	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
35412	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35413	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35414	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	De Paepe (1993)
35415	COL3A1	G	C	GG	Ehlers-Danlos Syndrome, Type IV	Smith (1997)
35416	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Johnson (1995)
35417	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35418	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Tromp (1989)
35419	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Tromp (1995)
35420	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35421	COL3A1	G	C	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35423	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35424	COL3A1	G	C	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35425	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35426	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Fox (1988), Richards (1992)
35427	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35428	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35429	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Nishiyama (2001)
35430	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Tromp (1989)
35431	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Palmeri (2003)
35432	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35433	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Richards (1991), Nuytinck (1992)
35434	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35435	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Giunta (2000)
35436	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
35437	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	McGrory (1996)
35438	COL3A1	G	C	GG	Ehlers-Danlos Syndrome, Type IV	Goldstein (1994)
35439	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35440	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35441	COL3A1	G	C	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35442	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35443	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Johnson (1995)
35444	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Johnson (1995)
35447	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Johnson (1992)
35448	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Nuytinck (1994)
35449	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Leroy (1993)
35450	COL3A1	G	C	GG	Ehlers-Danlos Syndrome, Type IV	Smith (1997)
35451	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pepin (2000)
35452	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Kontusaari (1992)
35453	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Smith (1997)
35454	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Narcisi (1993)
35455	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pope (1996)
35456	COL3A1	C	T	CC	Ehlers-Danlos Syndrome, Type IV	Schwarze (2001)
35457	COL3A1	A	C	AA	Ehlers-Danlos Syndrome, Type IV	Pope (1996)
35458	COL3A1	T	A	TT	Ehlers-Danlos Syndrome, Type IV	Giunta (2000)
35459	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Schwarze (1997)
35460	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Kuivaniemi (1990)
35461	COL3A1	G	C	GG	Ehlers-Danlos Syndrome, Type IV	Schwarze (1997)
35462	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Schwarze (1997)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
35463	COL3A1	T	G	TT	Ehlers-Danlos Syndrome, Type IV	Chiodo (1995)
35465	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pope (1996)
35466	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Lee (1991)
35467	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Thakker-Varia (1995)
35468	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Schwarze (1997)
35469	COL3A1	G	C	GG	Ehlers-Danlos Syndrome, Type IV	Schwarze (1997)
35470	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Schwarze (1997)
35471	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Kuivaniemi (1995)
35472	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Schwarze (1997)
35473	COL3A1	T	C	TT	Ehlers-Danlos Syndrome, Type IV	Schwarze (1997)
35474	COL3A1	G	C	GG	Ehlers-Danlos Syndrome, Type IV	Giunta (2000)
35475	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Wu (1991)
35476	COL3A1	T	C	TT	Ehlers-Danlos Syndrome, Type IV	Richards (1994)
35477	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Schwarze (1997)
35478	COL3A1	G	T	GG	Ehlers-Danlos Syndrome, Type IV	Schwarze (1997)
35479	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Cole (1990), Sillence (1991)
35480	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Kuivaniemi (1990)
35482	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pope (1996)
35483	COL3A1	T	C	TT	Ehlers-Danlos Syndrome, Type IV	Schwarze (1997)
35484	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Pope (1996), Smith (1997), Schwarze (1997), Pepin (2000)
35485	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Schwarze (1997)
35486	COL3A1	A	C	AA	Ehlers-Danlos Syndrome, Type IV	Schwarze (1997)
35487	COL3A1	A	T	AA	Ehlers-Danlos Syndrome, Type IV	Schwarze (1997)
35488	COL3A1	G	C	GG	Ehlers-Danlos Syndrome, Type IV	Schwarze (1997)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
35489	COL3A1	T	C	TT	Ehlers-Danlos Syndrome, Type IV	Lloyd (1993)
35491	COL3A1	A	G	AA	Ehlers-Danlos Syndrome, Type IV	Giunta (2000)
35492	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type IV	Schwarze (1997)
35493	COL3A1	T	A	TT	Ehlers-Danlos Syndrome, Type IV	Schwarze (1997), Pepin (2000)
35494	COL3A1	T	G	TT	Ehlers-Danlos Syndrome, Type IV	Schwarze (1997), Pepin (2000)
35401	COL3A1	G	A	GG	Ehlers-Danlos Syndrome, Type III	Narcisi (1994)
33823	COL5A1	G	T	GG	Ehlers-Danlos Syndrome, Type II, Atypical	Nicholls (1996)
33820	COL5A1	T	G	TT	Ehlers-Danlos Syndrome, Type II	Burrows (1998), Burrows (1998), Burrows (1998), Burrows (1998)
35516	COL5A2	C	G	CC	Ehlers-Danlos Syndrome, Type II	Richards (1998)
33820	COL5A1	T	G	TT	Ehlers-Danlos Syndrome, Type II	Burrows (1998)
35516	COL5A2	C	G	CC	Ehlers-Danlos Syndrome, Type II	Richards (1998)
33822	COL5A1	A	G	AA	Ehlers-Danlos Syndrome, Type I	Takahara (2002)
33815	COL5A1	G	C	GG	Ehlers-Danlos Syndrome, Type I	De Paepe (1997)
35518	COL5A2	C	A	CC	Ehlers-Danlos Syndrome, Type I	Michalickova (1998)
100830	COL5A1	I	D	II	Ehlers-Danlos Syndrome, Type I	Wenstrup (1996)
33810	COL5A1	C	T	CC	Ehlers-Danlos Syndrome, Type I	Wenstrup (2000)
33815	COL5A1	G	C	GG	Ehlers-Danlos Syndrome, Type I	De Paepe (1997)
33822	COL5A1	A	G	AA	Ehlers-Danlos Syndrome, Type I	Takahara (2002)
35518	COL5A2	C	A	CC	Ehlers-Danlos Syndrome, Type I	Michalickova (1998)
68639	SLC39A13	I	D	II	Ehlers-Danlos Syndrome, Spondylocheiro Dysplastic Form	Giunta (2008)
34720	B4GALT7	C	T	CC	Ehlers-Danlos Syndrome, Progeroid Form	Faiyaz-Ul-Haque (2004)
34731	B4GALT7	C	A	CC	Ehlers-Danlos Syndrome, Progeroid Form	Kresse (1987), Okajima (1999), Almeida (1999)
34732	B4GALT7	T	C	TT	Ehlers-Danlos Syndrome, Progeroid Form	Kresse (1987), Okajima (1999), Almeida (1999)
34720	B4GALT7	C	T	CC	Ehlers-Danlos Syndrome, Progeroid Form	Faiyaz-Ul-Haque (2004)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
34731	B4GALT7	C	A	CC	Ehlers-Danlos Syndrome, Progeroid Form	Kresse (1987), Okajima (1999), Almeida (1999)
34732	B4GALT7	T	C	TT	Ehlers-Danlos Syndrome, Progeroid Form	Kresse (1987), Okajima (1999), Almeida (1999)
35318	TNXB	C	T	CC	Possibly associated with Ehlers-Danlos Syndrome, Hypermobility Type	Zweers (2005), GeneDx (2017)
34073	COL1A2	G	A	GG	Ehlers-Danlos Syndrome, Cardiac Valvular Form	Byers (2002), Byers (2002), McKusick, (2002), Byers (2002), Schwarze (2004), Schwarze (2004)
34088	COL1A2	G	A	GG	Ehlers-Danlos Syndrome, Cardiac Valvular Form	Hata (1988), Hata (1988), Schwarze (2004)
34072	COL1A2	A	G	AA	Ehlers-Danlos Syndrome, Cardiac Valvular Form	Hata (1988)
34073	COL1A2	G	A	GG	Ehlers-Danlos Syndrome, Cardiac Valvular Form	Byers (2002)
34088	COL1A2	G	A	GG	Ehlers-Danlos Syndrome, Cardiac Valvular Form	Hata (1988)
34089	COL1A2	G	C	GG	Ehlers-Danlos Syndrome, Cardiac Valvular Form	Byers (2002)
35883	COL1A1	C	A,G,T	CC	Ehlers-Danlos Syndrome VII-A	Byers (1997)
34070	COL1A2	G	T	GG	Ehlers-Danlos syndrome VII	Schwarze (2004)
34109	COL1A2	G	A	GG	Ehlers-Danlos syndrome VII	Byers (1997)
34111	COL1A2	G	A	GG	Ehlers-Danlos syndrome VII	Weil (1989)
34113	COL1A2	G	T	GG	Ehlers-Danlos syndrome VII	Byers (1997)
35885	COL1A1	C	T	CC	Ehlers-Danlos Syndrome VII	Chu , (1984), Cole (1986), Weil (1989), Weil (1989), Weil (1989), D'Alessio (1991)
35445	COL3A1	G	T	GG	Ehlers-Danlos syndrome IV	Pepin (2000)
33816	COL5A1	A	G	AA	Ehlers-Danlos syndrome II	Bouma (2001)
33817	COL5A1	G	T	GG	Ehlers-Danlos syndrome I	Wenstrup (2000)
33818	COL5A1	G	A	GG	Ehlers-Danlos syndrome I	Wenstrup (2000)
64535	FLNA	G	A	GG	Ehlers-Danlos syndrome & heterotopia, periventricular	Gomez-Garre (2006)
64536	FLNA	C	A	CC	Ehlers-Danlos syndrome & heterotopia, periventricular	Gomez-Garre (2005)
64560	FLNA	G	A	GG	Ehlers-Danlos syndrome & heterotopia, periventricular	Sheen (2005)
33814	COL5A1	G	A	GG	Ehlers-Danlos syndrome	Steinmann (2000), Giunta, (2000), Steinmann (2000), Steinmann (2000)
35317	TNXB	G	A	GG	Ehlers-Danlos Syndrome	Zweers (2005)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
71836	COL5A2	T	C	TT	Ehlers-Danlos Syndrome	Malfait (2005)
33805	COL5A1	C	T	CC	Ehlers-Danlos syndrome	Malfait (2005)
33808	COL5A1	C	T	CC	Ehlers-Danlos syndrome	Malfait (2005)
33809	COL5A1	G	T	GG	Ehlers-Danlos syndrome	Malfait (2005)
33811	COL5A1	C	T	CC	Ehlers-Danlos syndrome	Malfait (2005)
33812	COL5A1	G	T	GG	Ehlers-Danlos syndrome	Malfait (2005)
33813	COL5A1	G	T	GG	Ehlers-Danlos syndrome	Malfait (2005)
33814	COL5A1	G	A	GG	Ehlers-Danlos syndrome	Steinmann (2000)
33819	COL5A1	T	G	TT	Ehlers-Danlos syndrome	Malfait (2005)
33821	COL5A1	G	T	GG	Ehlers-Danlos syndrome	Malfait (2005)
33824	COL5A1	G	A	GG	Ehlers-Danlos syndrome	Malfait (2005)
33825	COL5A1	G	A	GG	Ehlers-Danlos syndrome	Malfait (2005)
34115	COL1A2	G	C	GG	Ehlers-Danlos syndrome	Marini (2007)
35422	COL3A1	G	T	GG	Ehlers-Danlos syndrome	Demirogullari (2006)
35557	COL1A1	C	G,T	CC	Ehlers-Danlos syndrome	Cabral (2005)
35560	COL1A1	C	T	CC	Ehlers-Danlos syndrome	Marini (2007)
35565	COL1A1	C	A	CC	Ehlers-Danlos syndrome	Cabral (2005)
35567	COL1A1	C	T	CC	Ehlers-Danlos syndrome	Cabral (2005)
35581	COL1A1	C	T	CC	Ehlers-Danlos syndrome	Cabral (2005)
35598	COL1A1	G	A	GG	Ehlers-Danlos syndrome	Nuytinck (2000)
35881	COL1A1	T	C	TT	Ehlers-Danlos syndrome	Symoens (2004)
35887	COL1A1	T	A	TT	Ehlers-Danlos syndrome	Cabral (2005)
23895	NFKBIA	C	A,T	CC	Ectodermal Dysplasia, Anhidrotic, with T-call Immunodeficiency	Courtois (2003)
41456	PARK7	G	A	GG	Early-onset Parkinson Disease	Abou-Sleiman (2003)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
41455	PARK7	T	C	TT	Early-onset Parkinson Disease	Bonifati (2003)
11995	POMC	G	T	GG	Early-onset Obesity, Adrenal Insufficiency and Red Hair	Krude (1998), Krude (1998), Krude (2003), Krude (2003)
11998	POMC	T	A	TT	Early-onset Obesity, Adrenal Insufficiency and Red Hair	Krude (2003)
42877	TOR1A	I	D	II	Dystonia, Torsion	Ozelius (1997), Ikeuchi (1999), Kamm (1999), Hewett (2000), Hjermind (2002), Ikeuchi (2002), Grundmann (2003), Grundmann (2003), Dauer (2004), Naismith (2004), Torres (2004), Wong (2005), Wong (2005), Wong (2005), Dauer (2006)
800987	TOR1A	I	D	II	Dystonia, Torsion	Ozelius (1997), Ikeuchi (1999), Kamm (1999), Hewett (2000), Hjermind (2002), Ikeuchi (2002), Grundmann (2003), Grundmann (2003), Dauer (2004), Naismith (2004), Torres (2004), Wong (2005), Wong (2005), Wong (2005), Dauer (2006)
800900	TH	C	T	CC	Dystonia, L-DOPA-responsive	Heuvel (1998), Heuvel (1998)
43039	GCH1	T	C,G	TT	Dystonia, Dopa-responsive|Dystonia with Motor Delay	Leuzzi (2002), Leuzzi (2002)
43049	GCH1	T	C	TT	Dystonia, dopa-responsive	Weber (1997)
42904	ATP1A3	C	T	CC	Dystonia 12	Aguiar (2004)
42906	ATP1A3	A	C	AA	Dystonia 12	Dobyns (1993), Aguiar (2004)
42908	ATP1A3	C	A,G,T	CC	Dystonia 12	Brashear (1997), Aguiar (2004)
19004	TERC	G	C	GG	Dyskeratosis Congenita	Vulliamy (2001)
19005	TERC	T	C	TT	Dyskeratosis Congenita	Vulliamy (2006)
19006	TERC	T	A,C	TT	Dyskeratosis Congenita	Ly (2005)
19013	TERC	C	T	CC	Dyskeratosis Congenita	Vulliamy (2004)
19110	TERT	C	G	CC	Dyskeratosis Congenita	Armanios (2005)
64816	DKC1	C	T	C	Dyskeratosis Congenita with Hoyeraal-Hreidarsson Syndrome	Vulliamy (2006)
64827	DKC1	C	T	C	Dyskeratosis Congenita with Hoyeraal-Hreidarsson Syndrome	Vulliamy (2006)
64828	DKC1	C	A	C	Dyskeratosis congenita with Hoyeraal-Hreidarsson syndrome	Vulliamy (2006)
64832	DKC1	A	G	A	Dyskeratosis Congenita with Hoyeraal-Hreidarsson Syndrome	Vulliamy (2006)
64840	DKC1	G	A	G	Dyskeratosis Congenita with Hoyeraal-Hreidarsson Syndrome	Vulliamy (2006)
64815	DKC1	C	T	C	Dyskeratosis Congenita	Knight (1999)
64817	DKC1	C	G	C	Dyskeratosis Congenita	Wong (2004)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
64818	DKC1	T	G	T	Dyskeratosis Congenita	Heiss (1998)
64820	DKC1	A	G	A	Dyskeratosis Congenita	Knight (1999)
64821	DKC1	C	G	C	Dyskeratosis Congenita	Heiss (1998)
64822	DKC1	G	A	G	Dyskeratosis Congenita	Knight (1999)
64823	DKC1	A	G	A	Dyskeratosis Congenita	Heiss (2001)
64825	DKC1	G	C	G	Dyskeratosis Congenita	Knight (1999)
64826	DKC1	A	G	A	Dyskeratosis Congenita	Hassock (1999)
64830	DKC1	C	T	C	Dyskeratosis Congenita	Knight (2001)
64831	DKC1	A	C	A	Dyskeratosis Congenita	Knight (2001)
64833	DKC1	C	G	C	Dyskeratosis Congenita	Knight (1999)
64834	DKC1	T	C	T	Dyskeratosis Congenita	Knight (1999)
64835	DKC1	G	A	G	Dyskeratosis Congenita	Knight (1999)
64836	DKC1	G	C	G	Dyskeratosis Congenita	Kraemer (2003)
800962	DKC1	C	T	C	Dyskeratosis Congenita	Knight (1999), Yaghmai (2000), Yaghmai (2000), Heiss (2001)
64838	DKC1	G	A	G	Dyskeratosis Congenita	Vulliamy (2006)
64839	DKC1	C	T	C	Dyskeratosis Congenita	Knight (2001)
64841	DKC1	G	A	G	Dyskeratosis Congenita	Heiss (1998)
64842	DKC1	G	A	G	Dyskeratosis Congenita	Knight (1999)
64843	DKC1	C	T	C	Dyskeratosis Congenita	Vulliamy (2006)
64844	DKC1	C	T	C	Dyskeratosis Congenita	Ding (2004)
64845	DKC1	C	G	C	Dyskeratosis Congenita	Knight (2001), Salowsky (2002)
64846	DKC1	A	G	A	Dyskeratosis Congenita	Vulliamy (2006)
64847	DKC1	C	G	C	Dyskeratosis Congenita	Knight (1999)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
22466	FGA	G	A	GG	Dysfibrinogenemia|Fibrinogen Bergamo 1|Fibrinogen Hershey 2|Fibrinogen Homburg 2|Fibrinogen Homburg 3|Fibrinogen Kawaguchi 1|Fibrinogen Leogan|Fibrinogen Metz 1|Fibrinogen New Albany|Fibrinogen Osaka 1|Fibrinogen Schwarzach 1|Fibrinogen Stony Brook 1|Fib	Henschen , (1981), Southan (1982), Henschen (1983), Reber (1985), Miyashita (1985), Miyata (1987), Galanakis (1989), Galanakis (1989), Lee (1991), Galanakis (1993), Bolliger-Stucki (2001), Bolliger-Stucki (2001), Biot, (2001), Flood (2006), Flood (2006)
22020	FGG	T	A	TT	Dysfibrinogenemia|Thrombophilia, Dysfibrinogenemic	Reber (1986), Reber (1986), Lounes (2000), Lounes (2000)
21999	FGG	G	A	GG	Dysfibrinogenemia	Matsuda (1983), Bell (1985), Yoshida (1988), Terukina (1988), Terukina (1988), Schmelzer (1989), Mosesson (1995)
22465	FGA	C	T	CC	Dysfibrinogenaemia|Fibrinogen Amiens 1 |Fibrinogen Amiens 2|Fibrinogen Bergamo 3|Fibrinogen Bern 2|Fibrinogen Bicetre 1|Fibrinogen Birmingham 1|Fibrinogen CHapel Hill 2|Fibrinogen Clermont-Ferrand 1|Fibrinogen Giessen 1|Fibrinogen Leitchfield|Fibrinogen	Shafer (1981), Carrell (1983), Qureshi (1983), Galanakis (1983), Southan (1985), Ebert (1986), Henschen (1987), Reber (1987), Siebenlist (1988), Galanakis (1993)
36991	IKBKAP	C	G	CC	Dysautonomia, Familial	Slaugenhaupt (2001), Anderson (2001), Dong (2002)
36992	IKBKAP	A	G	AA	Dysautonomia, Familial	Slaugenhaupt (2001), Anderson (2001), Ibrahim (2007), Counsyl (2015), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2015), Invitae (2017), GeneDx (2017)
21351	ALB	G	A	GG	Dysalbuminemic Hyperthyroxinaemia, Familial	Petersen (1994), Pohlenz (2001)
26947	ABCC2	A	T	AA	Dubin-Johnson Syndrome	Mor-Cohen (2001)
26944	ABCC2	C	T	CC	Dubin-Johnson Syndrome	Wada (1998), Toh (1999), Lage (2003)
26946	ABCC2	G	A	GG	Dubin-Johnson Syndrome	Mor-Cohen (2001)
43380	DBH	G	T	GG	Dopamine Beta Hydroxylase Deficiency	Deinum (2004)
43382	DBH	T	C	TT	Dopamine Beta Hydroxylase Deficiency	Kim (2002), Kim (2002), Kim (2002), Zabetian (2003), Deinum (2004), Kim (2011), Laboratory for Molecular Medicine of Partners HealthCare Personalized Medicine (2014), GeneDx (2015), GeneReviews (2015), Emory Genetics Laboratory of Emory University (2016), Illumina Clinical Services Laboratory (2016), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2016)
22059	ART4	G	A,C	GG	Dombrock-null Phenotype	Rios (2002)
66098	ATP6V1B1	C	T	CC	Distal Renal Tubular Acidosis with Progressive Deafness	Karet (1999)
66020	ATP6V0A4	A	T	AA	Distal Renal Tubular Acidosis with Late-onset Sensorineural Hearing Loss	Stover (2002)
20356	SLC4A1	C	T	CC	Distal Renal Tubular Acidosis	Wrong (1972), Wrong (1993), Bruce (1997), Karet (1998)
20370	SLC4A1	C	T	CC	Distal Renal Tubular Acidosis	Tanphaichitr (1998), Bruce (2000), Yenchitsomanus (2002), Yenchitsomanus (2003)
36136	TNNI2	G	A	GG	Distal Arthrogryposis Syndrome 2b	Sung (2003)
36153	TNNT3	G	A	GG	Distal Arthrogryposis Syndrome 2b	Varnava (1999), Sung (2003)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
36118	TPM2	G	C	GG	Distal Arthrogryposis Syndrome 1	Sung (2003)
73028	IL12B	D	I	DD	Disseminated Infection with BCG and Salmonella enteritidis	Picard (2002), Barbier (2008)
60965	POR	G	C	GG	Disordered Steroidogenesis Due to POR Deficiency	Fluck (2004), Kelley (2002), Huang (2005), Arlt (2004)
60968	POR	G	A	GG	Disordered Steroidogenesis Due to POR Deficiency	Fluck (2004), Arlt (2004)
100733	MTND6	G	A	?	Leber Optic Atrophy	Jun (1994), Shoffner (1995), Kirby (2000), Gropman (2004), Watanabe (2006)
70736	MTND6	T	C	?	Leber Optic Atrophy	Bernier (1970), Canadians dated from (1970), Anderson , (1981), Brown , (1992), Howell, (1992), Johns , (1992), Brown (1997), Carelli (1998), Carelli (1999), Carelli (1999), Macmillan (2000), Macmillan (2000), Howell (2003), Nishioka (2003), Howell (2003), Howell (2003), Laberge (2005), GeneReviews (2014), Stanford Center for Inherited Cardiovascular Disease of Stanford University (2015), ARUP Laboratories (2017)
39034	MYF6	C	A	CC	Dilatative Myopathy	Kerst (1998)
33222	QDPR	C	T	CC	Dihydropteridine Reductase Deficiency	Dianzani (1993)
33240	QDPR	T	C	TT	Dihydropteridine Reductase Deficiency	Ikeda (1997)
67955	SLC26A2	T	C	TT	Diastrophic Dysplasia	Hastbacka (1999)
79310	NEUROG3	C	A,T	CC	Diarrhea, Malabsorptive, Congenital	Wang (2006)
79311	NEUROG3	G	T	GG	Diarrhea, Malabsorptive, Congenital	Wang (2006)
14173	HNF4A	T	C	TT	Diabetes, MODY1	Thomas (2001)
11494	INSR	C	T	CC	Diabetes Mellitus, Insulin-Resistant, with Acanthosis Nigricans	Moller (1994)
12581	AQP2	A	C	AA	Diabetes Insipidus, Nephrogenic	Lin (2002)
11665	AVPR2	C	T	C	Diabetes Insipidus, Nephrogenic	Crawford (1969), Crawford, (1969), Miettinen (1970), Holtzman (1993)
11735	AVPR2	G	A	G	Diabetes Insipidus, Nephrogenic	Sadeghi (1997), Sadeghi (1997)
12593	AQP2	G	T	GG	Diabetes Insipidus, Nephrogenic	Lin (2002)
12725	AVP	C	T	CC	Diabetes Insipidus, Central	Copeland (1988), Ito (1993), McLeod (1993), McLeod (1993), Krishnamani (1993), Siggaard (1999)
9908	FOXP2	C	T	CC	Developmental Verbal Dyspraxia	MacDermot (2005)
9909	FOXP2	G	A	GG	Developmental Verbal Dyspraxia	Hurst (1990), Lai (2001)
31988	DHCR24	T	G	TT	Desmosterolosis	Waterham (2001)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
70301	TPH2	A	G	AA	Depression	Haghighi (2008)
48007	WT1	C	A,T	CC	Denys-Drash Syndrome|Mesangial Sclerosis, Isolated Diffuse	Pelletier (1991), Baird (1992), Little (1993), Jeanpierre (1998)
48057	WT1	G	A	GG	Denys-Drash syndrome	Clarkson (1993), Little (1993), Little (1993), Kaplinsky (1996), Kaplinsky (1996)
48075	WT1	G	A	GG	Denys-Drash syndrome	Denys (1967), McCoy (1983), Pelletier (1991), Pelletier (1991), Bruening (1992), Baird (1992), Coppes (1992), Little (1993)
34086	COL1A2	G	A	GG	Dentinogenesis Imperfecta, Severe, with Very Mild Osteogenesis Imperfecta	Nicholls (1996), Nicholls (1996)
66731	CLCN5	G	A	G	Dent disease	Ludwig (2006)
38788	GDAP1	C	G	CC	Demyelinating Charcot-Marie-Tooth Disease 4A|Axonal Neuropathy with Vocal Cord Paresis|Axonal Charcot-Marie-Tooth Disease, Type 2K	Baxter (2002), Nelis (2002), Cuesta (2002), Birouk (2003)
42421	SNCB	C	G,T	CC	Dementia with Lewy Bodies	Ohtake (2004)
42422	SNCB	G	T	GG	Dementia with Lewy Bodies	Ohtake (2004)
800957	ALAD	G	A	GG	Delta-aminolevulinate Dehydratase Porphyria	Doss (1986), Fujita (1987), Sassa (1991), Ishida (1992)
55393	ALAD	C	T	CC	Delta-aminolevulinate Dehydratase Porphyria	Doss (1986), Fujita (1987), Sassa (1991), Ishida (1992)
41888	AANAT	G	A	GG	Delayed Sleep Phase Syndrome	Hohjoh (2003)
43632	EGR2	G	A	GG	Dejerine-Sottas Neuropathy|Charcot-Marie-Tooth Disease, Type 1D	Warner (1999), Boerkoel (2001), Chung (2005), Chung (2005)
22614	F13A1	C	A	CC	Deficiency of Factor XIII, A Subunit	Mikkola (1996), Garcia (2001)
22631	F13A1	C	T	CC	Deficiency of Factor XIII, A Subunit	Castle (1981), Coggan (1995), Coggan (1995), Coggan (1995)
22636	F13A1	G	A	GG	Deficiency of Factor XIII, A Subunit	Mikkola (1994)
30973	HSD3B2	C	A	CC	Deficiency of 3-Beta-hydroxysteroid Dehydrogenase, Type II	Alos (2000)
30983	HSD3B2	G	A	GG	Deficiency of 3-Beta-hydroxysteroid Dehydrogenase, Type II	Rheaume (1992), Zachmann (1979), Rheaume (1991)
13451	AR	A	G	A	Defective Spermatogenesis	Ghadessy (1999)
56034	DRD5	T	G	TT	Decreased Binding of Risperidone	Cravchik (1999)
25791	MYO6	A	G	AA	Deafness, Sensorineural, with Hypertrophic Cardiomyopathy	Mohiddin (2004), Mohiddin (2004)
26080	ACTG1	G	A	GG	Deafness, Progressive	Van Wijk (2003)
26129	ACTG1	G	A	GG	Deafness, Progressive	DeWan (2003), Zhu (2003)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
26131	ACTG1	A	G	AA	Deafness, Progressive	Teig (1968), Rendtorff (2006)
26103	ACTG1	G	A	GG	Deafness, Progressive	Zhu (2003)
26128	ACTG1	T	A	TT	Deafness, Progressive	Zhu (2003)
26130	ACTG1	G	C	GG	Deafness, Progressive	Smith (2000), Zhu (2003)
25773	SLC26A4	A	G	AA	Deafness, non-syndromic, autosomal recessive	Lopez-Bigas (2002)
25684	SLC26A4	C	T	CC	Deafness, non-syndromic, autosomal recessive	Prasad (2004)
25689	SLC26A4	C	T	CC	Deafness, non-syndromic, autosomal recessive	Lopez-Bigas (2001)
25729	SLC26A4	G	A	GG	Deafness, non-syndromic, autosomal recessive	Li (1998)
25672	SLC26A4	T	C	TT	Deafness, non-syndromic, autosomal recessive	Prasad (2004)
25674	SLC26A4	C	G	CC	Deafness, non-syndromic, autosomal recessive	Prasad (2004)
25675	SLC26A4	C	A	CC	Deafness, non-syndromic, autosomal recessive	Park (2003)
25678	SLC26A4	C	A	CC	Deafness, non-syndromic, autosomal recessive	Park (2003)
25680	SLC26A4	A	G	AA	Deafness, non-syndromic, autosomal recessive	Prasad (2004)
25681	SLC26A4	C	T	CC	Deafness, non-syndromic, autosomal recessive	Park (2003)
25688	SLC26A4	C	T	CC	Deafness, non-syndromic, autosomal recessive	Tsukamoto (2003)
25695	SLC26A4	A	G	AA	Deafness, non-syndromic, autosomal recessive	Tsukamoto (2003)
25702	SLC26A4	T	A	TT	Deafness, non-syndromic, autosomal recessive	Park (2003)
25703	SLC26A4	T	C	TT	Deafness, non-syndromic, autosomal recessive	Park (2003)
25708	SLC26A4	A	G	AA	Deafness, non-syndromic, autosomal recessive	Usami (1999)
25713	SLC26A4	A	T	AA	Deafness, non-syndromic, autosomal recessive	Park (2003)
25714	SLC26A4	T	C	TT	Deafness, non-syndromic, autosomal recessive	Hutchin (2005)
25716	SLC26A4	G	C	GG	Deafness, non-syndromic, autosomal recessive	Park (2003)
25719	SLC26A4	A	G	AA	Deafness, non-syndromic, autosomal recessive	Prasad (2004)
25725	SLC26A4	C	A	CC	Deafness, non-syndromic, autosomal recessive	Park (2003)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
25740	SLC26A4	G	C	GG	Deafness, non-syndromic, autosomal recessive	Iwasaki (2006)
25747	SLC26A4	C	T	CC	Deafness, non-syndromic, autosomal recessive	Tsukamoto (2003)
25751	SLC26A4	T	A	TT	Deafness, non-syndromic, autosomal recessive	Park (2003)
25752	SLC26A4	T	C	TT	Deafness, non-syndromic, autosomal recessive	Prasad (2004)
25755	SLC26A4	C	T	CC	Deafness, non-syndromic, autosomal recessive	Usami (1999)
25757	SLC26A4	A	G	AA	Deafness, non-syndromic, autosomal recessive	Prasad (2004)
25763	SLC26A4	G	C	GG	Deafness, non-syndromic, autosomal recessive	Prasad (2004)
25783	SLC26A4	A	G	AA	Deafness, non-syndromic, autosomal recessive	Park (2003)
800890	GJB3	C	A	CC	Deafness, non-syndromic, autosomal recessive	Uyguner (2003)
60564	MYO7A	G	C	GG	Deafness, non-syndromic, autosomal recessive	Liu (1997)
60580	MYO7A	G	A	GG	Deafness, non-syndromic, autosomal recessive	Weil (1997)
60647	MYO7A	A	G	AA	Deafness, non-syndromic, autosomal recessive	Liu (1997)
60559	MYO7A	C	T	CC	Deafness, non-syndromic, autosomal dominant	Di Leva (2006)
60576	MYO7A	A	T	AA	Deafness, non-syndromic, autosomal dominant	Luijendijk (2004)
60589	MYO7A	G	C	GG	Deafness, non-syndromic, autosomal dominant	Street (2004)
60596	MYO7A	C	T	CC	Deafness, non-syndromic, autosomal dominant	Bolz (2004)
26497	DSPP	G	T	GG	Deafness, Nonsyndromic Sensorineural with Dentinogenesis Imperfecta, Type I|Dentinogenesis Imperfecta, Shields Type II|Dentinogenesis Imperfecta, Shields Type III	Xiao (2001), Xiao (2001), Kim (2005), Kim (2005), Kim (2005)
26494	DSPP	C	A	CC	Deafness, Nonsyndromic Sensorineural with Dentinogenesis Imperfecta, Type I	Xiao (2001)
25821	TECTA	A	G	AA	Deafness, Nonsyndromic Sensorineural 8	Verhoeven (1998), Verhoeven (1998)
7953	GJB6	G	A	GG	Deafness, Nonsyndromic Sensorineural 3	Grifa (1999)
25826	TECTA	T	A	TT	Deafness, Nonsyndromic Sensorineural 12	Balciuniene (1998), Balciuniene (1999)
25810	TECTA	G	C	GG	Deafness, Nonsyndromic Sensorineural 12	Alloisio (1999)
26020	MYH14	C	A	CC	Deafness, Nonsyndromic Sensorineural	Donaudy (2004), Kim (2005)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
26022	MYH14	C	T	CC	Deafness, Nonsyndromic Sensorineural	Donaudy (2004)
26038	MYH14	C	A	CC	Deafness, Nonsyndromic Sensorineural	Donaudy (2004)
26039	MYH14	C	T	CC	Deafness, Nonsyndromic Sensorineural	Yang (2005)
26233	COCH	C	T	CC	Deafness, Nonsyndromic Sensorineural	Kok (1999), De Kok (1999)
26285	GJB3	C	T	CC	Deafness, Nonsyndromic Sensorineural	Xia (1998)
25804	MYO6	G	A	GG	Deafness, Nonsyndromic Sensorineural	Melchionda (2001), Sato (2004)
26247	COCH	G	T	GG	Deafness, Nonsyndromic Sensorineural	Collin (2006)
26248	COCH	G	A	GG	Deafness, Nonsyndromic Sensorineural	Robertson (1998)
26251	COCH	G	A	GG	Deafness, Nonsyndromic Sensorineural	Usami (2003)
26252	COCH	G	T	GG	Deafness, Nonsyndromic Sensorineural	Street (2005), Street (2005)
26286	GJB3	G	A	GG	Deafness, Nonsyndromic Sensorineural	Xia (1998), Xia (1998)
26302	KCNQ4	G	C	GG	Deafness, Nonsyndromic Sensorineural	Coucke (1999), Akita (2001), Van Camp (2002), Van Camp (2002), Nie (2008)
26305	KCNQ4	G	T	GG	Deafness, Nonsyndromic Sensorineural	Kubisch (1999), Coucke (1999), Kubisch (1999)
26306	KCNQ4	G	A	GG	Deafness, Nonsyndromic Sensorineural	Coucke (1999)
26246	COCH	T	G	TT	Deafness, Nonsyndromic Sensorineural	Manolis (1996), Robertson (1998)
26250	COCH	T	C	TT	Deafness, Nonsyndromic Sensorineural	Robertson (1998)
25899	GJB2	C	G	CC	Deafness, Nonsyndromic Sensorineural	Denoyelle , (1998), Tekin (2001)
25927	GJB2	G	A,C	GG	Deafness, Nonsyndromic Sensorineural	Kelsell (1997), Richard (1998), Janecke (2001), Kudo (2003), Kudo (2003)
26008	GJB2	C	A	CC	Deafness, Nonsyndromic Sensorineural	Morle (2000)
26230	DIAPH1	C	A	CC	Deafness, Nonsyndromic Sensorineural	Lynch (1997), Lynch (1997)
100736	MTND6	T	C	?	Leigh Syndrome due to Mitochondrial Complex I Deficiency	Funalot (2002), Solano (2003), Ugalde (2003), Thorburn (2003), Bugiani (2004)
26288	GJB3	C	A	CC	Deafness, Nonsyndromic	Uyguner (2003)
26058	CRYM	T	A	TT	Deafness, Nonsyndromic	Abe (2003)
26373	OTOF	G	C	GG	Deafness, Nonsyndromic	Migliosi (2002)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
26391	OTOF	G	A	GG	Deafness, Nonsyndromic	Migliosi (2002), Migliosi (2002), Varga (2006)
71955	OTOF	A	T	AA	Deafness, Nonsyndromic	Yasunaga (1999)
26160	ESPN	G	A	GG	Deafness, Neurosensory without Vestibular Involvement	Donaudy (2006)
25835	TECTA	G	A	GG	Deafness, Neurosensory 21	Mustapha (1999), Mustapha (1999)
26350	TMIE	A	C	AA	Deafness, Neurosensory	Santos (2006)
26347	TMIE	C	T	CC	Deafness, Neurosensory	Naz (2002)
26348	TMIE	C	T	CC	Deafness, Neurosensory	Naz (2002)
26349	TMIE	C	T	CC	Deafness, Neurosensory	Naz (2002)
25881	GJB2	C	T	CC	Deafness, Neurosensory	Kelsell (1997), Maheshwari (2003), Alvarez (2005)
25930	GJB2	C	T	CC	Deafness, Neurosensory	Brown , (1996), Kelsell (1997)
25948	GJB2	A	G	AA	Deafness, Neurosensory	Loffler (2001)
26003	GJB2	C	G	CC	Deafness, Neurosensory	Denoyelle (1997), Hujirat (2004), Hujirat (2004)
71952	GJB2	I	D	II	Deafness, Neurosensory	Abe (2000), Kudo (2000), Liu (2002), Yan (2003), Dai (2007)
801006	GJB2	I	D	II	Deafness, Neurosensory	Abe (2000), Kudo (2000), Liu (2002), Yan (2003), Dai (2007)
801981	GJB2	D	I	DD	Deafness, autosomal recessive 1	Rabionet (2000), Emory Genetics Laboratory of Emory University (2012), Counsyl (2015)
71953	GJB2	I	D	II	Deafness, Neurosensory	Ziprkowski (1964), Marres (1989), Carrasquillo (1997), Zelante (1997), Denoyelle (1997), Estivill (1998), Antoniadi (1999), Green (1999), Green (2000), Salvador (2000), Pampanos (2000), Abe (2000), Kudo (2000), Gasparini (2000), Rabionet (2000), Anichkina (2001), Genetic Services Laboratory of the University of Chicago (2013), Laboratory for Molecular Medicine of Partners HealthCare Personalized Medicine (2014), Genetics Diagnostic Laboratory of the Children's Hospital of Eastern Ontario (2015), The Children's Hospital of Philadelphia (2015), Centre for Mendelian Genomics of the University Medical Centre Ljubljana (2015), GeneDx (2016), Counsyl (2016), Emory Genetics Laboratory of Emory University (2016), Knight Diagnostic Laboratories of Oregon Health and Sciences University (2016), Athena Diagnostics Inc (2016), GeneDx (2017), ARUP Laboratories (2017), The Children's Hospital of Philadelphia (2017), Shahid Beheshti University of Medical Sciences (2018), Athena Diagnostics Inc (2018), ClinGen Hearing Loss Expert Panel (2018)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
850031	GJB2	D	I	DD	Deafness, Neurosensory	Estivill (1998), Rabionet (2000), Hamelmann (2001), D'Andrea (2002), Snoeckx (2005), Declau (2008), Dai (2009), Clinical Molecular Genetics Laborator of Johns Hopkins All Children's Hospital (2009), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2012), Counsyl (2015), Laboratory for Molecular Medicine of Partners HealthCare Personalized Medicine (2016), Athena Diagnostics Inc (2017), Integrated Genetics of Laboratory Corporation of America (2018), CeGaT Praxis fuer Humangenetik Tuebingen (2018), GeneDx (2018), Invitae (2019), Genetic Testing Center for Deafness of the Institute of Otolaryngology, Chinese PLA General Hospital (2019)
25872	GJB2	C	A,T	CC	Deafness, autosomal recessive 1	Rabionet (2000), GeneReviews (2011), Laboratory for Molecular Medicine of Partners HealthCare Personalized Medicine (2012), Genetic Services Laboratory of the University of Chicago (2013), Emory Genetics Laboratory of Emory University (2013), Counsyl (2016)
801007	GJB2	I	D	II	Deafness, Neurosensory	Ziprkowski (1964), Marres (1989), Carrasquillo (1997), Zelante (1997), Denoyelle (1997), Estivill (1998), Antoniadi (1999), Green (1999), Green (2000), Salvador (2000), Pampanos (2000), Abe (2000), Kudo (2000), Gasparini (2000), Anichkina (2001), The Children's Hospital of Philadelphia (2015), Emory Genetics Laboratory (2016), Knight Diagnostic Laboratories of Oregon Health and Sciences University (2016), Counsyl (2016), GeneDx (2016)
71954	GJB2	I	D	II	Deafness, Neurosensory	Morell (1998), Zelante (1997)
25845	GJB2	T	A	TT	Deafness, Neurosensory	Gualandi (2002)
25902	GJB2	C	A,T	CC	Deafness, Neurosensory	Denoyelle (1997)
25941	GJB2	C	G	CC	Deafness, Neurosensory	Kenna (2001), Beltramello (2005)
25976	GJB2	G	A	GG	Deafness, Neurosensory	Brobby (1998), Emory Genetics (2015), Partners HealthCare Personalized Medicine (2016), GeneDx (2016)
26016	GJB2	C	T	CC	Deafness, Neurosensory	Denoyelle (1999)
26369	FGF3	G	A	GG	Deafness, Congenital, with Inner Ear Agenesis, Microtia, and Microdontia	Tekin (2007)
25790	MYO6	A	T	AA	Deafness, Congenital Neurosensory	Ahmed (2003)
25805	MYO6	C	T	CC	Deafness, Congenital Neurosensory	Ahmed (2003)
2921	JAG1	C	T	CC	Deafness, Congenital Heart Defects, and Posterior Embryotoxon	Le Caignec (2002)
25637	TMPRSS3	G	A	GG	Deafness, Childhood-onset Neurosensory	Masmoudi (2001), Masmoudi , (2001), Wattenhofer (2005)
801979	TMPRSS3	G	A	GG	Deafness, Congenital Neurosensory	Masmoudi (2001), Masmoudi , (2001), Wattenhofer (2005)
25659	TMPRSS3	C	T	CC	Deafness, Childhood-onset Neurosensory	Scott (2001), Scott (2001)
25655	TMPRSS3	C	A,T	CC	Deafness, Childhood-onset Neurosensory	Wattenhofer (2005)
25864	GJB2	T	A,C	TT	Deafness, autosomal recessive 1	Estivill (1998)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
25878	GJB2	C	G	CC	Deafness, autosomal recessive 1	Rabionet (2000)
25885	GJB2	G	A,T	GG	Deafness, autosomal recessive 1	Prasad (2000)
25892	GJB2	G	C,T	GG	Deafness, autosomal recessive 1	Feldmann (2004)
25893	GJB2	G	C	GG	Deafness, autosomal recessive 1	Sironi (2005)
25898	GJB2	C	G,T	CC	Deafness, autosomal recessive 1	Green (1999)
25911	GJB2	G	A	GG	Deafness, autosomal recessive 1	Wilcox (1999)
25919	GJB2	G	T	GG	Deafness, autosomal recessive 1	Roux (2005)
25921	GJB2	G	C	GG	Deafness, autosomal recessive 1	Estivill (1998)
25928	GJB2	A	G	AA	Deafness, autosomal recessive 1	Carrasquillo (1997)
25932	GJB2	A	C,G	AA	Deafness, autosomal recessive 1	Hamelmann (2001)
25934	GJB2	T	C	TT	Deafness, autosomal recessive 1	Uyguner (2003)
25935	GJB2	T	A	TT	Deafness, autosomal recessive 1	Najmabadi (2005)
25936	GJB2	G	T	GG	Deafness, autosomal recessive 1	Kalay (2005)
25937	GJB2	G	A	GG	Deafness, autosomal recessive 1	Hutchin (2005)
25950	GJB2	C	T	CC	Deafness, autosomal recessive 1	Kelley (1998)
25951	GJB2	C	T	CC	Deafness, autosomal recessive 1	Green (1999)
25952	GJB2	G	A	GG	Deafness, autosomal recessive 1	Green (1999)
25954	GJB2	T	A	TT	Deafness, autosomal recessive 1	Primignani (2005)
25955	GJB2	T	C	TT	Deafness, autosomal recessive 1	Jun (2000)
25957	GJB2	A	C	AA	Deafness, autosomal recessive 1	Kelley (1998)
25958	GJB2	C	A	CC	Deafness, autosomal recessive 1	Rabionet (2000)
25960	GJB2	T	A	TT	Deafness, autosomal recessive 1	Green (1999)
25962	GJB2	G	A	GG	Deafness, autosomal recessive 1	Scott (1998)
25970	GJB2	C	T	CC	Deafness, autosomal recessive 1	Primignani (2005)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
25971	GJB2	G	T	GG	Deafness, autosomal recessive 1	Fuse (1999), Abe (2007)
25973	GJB2	C	T	CC	Deafness, autosomal recessive 1	Marlin (2001)
25978	GJB2	C	T	CC	Deafness, autosomal recessive 1	Frei (2004)
25979	GJB2	C	A	CC	Deafness, autosomal recessive 1	Vijaya (2003)
25980	GJB2	C	A,T	CC	Deafness, autosomal recessive 1	Rabionet (2000)
25994	GJB2	C	G,T	CC	Deafness, autosomal recessive 1	Posukh (2005)
25995	GJB2	G	C	GG	Deafness, autosomal recessive 1	Rabionet (2000)
25996	GJB2	G	A	GG	Deafness, autosomal recessive 1	Kalay (2005)
25997	GJB2	A	G	AA	Deafness, autosomal recessive 1	Primignani (2005)
25998	GJB2	G	T	GG	Deafness, autosomal recessive 1	Denoyelle (1999)
25999	GJB2	A	G	AA	Deafness, autosomal recessive 1	Hamelmann (2001)
26001	GJB2	G	A	GG	Deafness, autosomal recessive 1	Wilcox (2000)
26002	GJB2	C	G,T	CC	Deafness, autosomal recessive 1	Hamelmann (2001)
26005	GJB2	C	A,T	CC	Deafness, autosomal recessive 1	Hamelmann (2001)
26006	GJB2	G	A	GG	Deafness, autosomal recessive 1	Green (1999)
26010	GJB2	A	G	AA	Deafness, autosomal recessive 1	Leshinsky-Silver (2005)
26013	GJB2	A	T	AA	Deafness, autosomal recessive 1	Feldmann (2004)
26014	GJB2	A	G	AA	Deafness, autosomal recessive 1	Hamelmann (2001)
26015	GJB2	T	G	TT	Deafness, autosomal recessive 1	Antoniadi (2000)
62350	WFS1	A	C	AA	Deafness, Autosomal Dominant 6	Komatsu (2002)
62279	WFS1	C	T	CC	Deafness, Autosomal Dominant 6	Bespalova (2001), Cryns (2002)
62281	WFS1	G	A	GG	Deafness, Autosomal Dominant 6	Bespalova (2001), Cryns (2002)
62286	WFS1	G	A	GG	Deafness, Autosomal Dominant 6	Young (2001)
62348	WFS1	G	T	GG	Deafness, Autosomal Dominant 6	Yuhe (2005)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
62389	WFS1	T	C	TT	Deafness, Autosomal Dominant 6	Bespalova (2001)
25910	GJB2	T	G	TT	Deafness, autosomal dominant 3	Gualandi (2004)
25989	GJB2	T	C,G	TT	Deafness, autosomal dominant 3	Fialho (2003)
26301	KCNQ4	T	A	TT	Deafness, autosomal dominant 2	Van Hauwe (2000)
25831	TECTA	G	A	GG	Deafness, autosomal dominant 12	Verhoeven (1998)
25828	TECTA	T	G	TT	Deafness, autosomal dominant 12	Pfister (2004)
25829	TECTA	C	T	CC	Deafness, autosomal dominant 12	Verhoeven (1998)
26283	GJB3	A	G	AA	Deafness	Liu (2000)
25825	TECTA	C	A	CC	Deafness	Hutchin (2005)
25827	TECTA	G	A	GG	Deafness	Hutchin (2005)
25834	TECTA	G	A	GG	Deafness	Iwasaki (2002)
25832	TECTA	T	G	TT	Deafness	Moreno-Pelayo (2001)
79294	GRHL2	D	I	DD	Deafness	Peters (2002)
801009	GRHL2	D	I	DD	Deafness	Peters (2002)
9334	ATP2A2	A	G	AA	Darier Disease	Ruiz-Perez (1999)
9286	ATP2A2	G	A	GG	Darier Disease	Sakuntabhai (1999), Ringpfeil (2001), Chao (2002)
28461	LAMP2	C	T	C	Danon Disease	Balmer (2005)
28464	LAMP2	G	A	G	Danon Disease	Charron (2004)
28465	LAMP2	C	T	C	Danon Disease	Arad (2005), Bertini (2005)
28466	LAMP2	A	G	A	Danon Disease	Musumeci (2005)
28469	LAMP2	C	T	C	Danon Disease	Nishino (2000), Danon (1981), Riggs (1983)
71906	LAMP2	G	T	G	Danon Disease	Nadeau (2007)
5003	COX15	A	C,G	AA	Cytochrome C Oxidase Deficiency	Bugiani (2005)
30705	SCO2	C	T	CC	Cytochrome C Oxidase Deficiency	Papadopoulou (1999), Jaksch (2001), Salviati (2002)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
67246	SLC7A9	C	T	CC	Cystinuria, non-type I	Consortium (1999), Font-Llitjos (2005), Font-Llitjos (2005), Di Perna (2008)
67251	SLC7A9	C	T	CC	Cystinuria, non-type I	Consortium (1999), Colombo (2000), Font-Llitjos (2005)
67252	SLC7A9	C	A,T	CC	Cystinuria, non-type I	Consortium (1999), Font-Llitjos (2005)
67269	SLC7A9	G	A	GG	Cystinuria, non-type I	Consortium (2001)
67147	SLC3A1	C	T	CC	Cystinuria	Bisceglia (1996), Di Perna (2008)
67154	SLC3A1	C	T	CC	Cystinuria	Pras (1995)
67185	SLC3A1	C	G	CC	Cystinuria	Bisceglia (2001), Di Perna (2008)
67179	SLC3A1	T	C	TT	Cystinuria	Calonge (1994), Bisceglia (1996), Harnevik (2001), Harnevik (2003), Font-Llitjos (2005), Font-Llitjos (2005), Di Perna (2008)
5539	SCNN1B	A	G	AA	Cystic Fibrosis, Non-classic	Sheridan (2005)
5524	SCNN1B	C	T	CC	Cystic Fibrosis, Non-classic	Sheridan (2005)
5525	SCNN1B	G	A	GG	Cystic Fibrosis, Non-classic	Sheridan (2005)
5526	SCNN1B	G	A	GG	Cystic Fibrosis, Non-classic	Sheridan (2005)
101456	CFTR	G	T	GG	Cystic Fibrosis	Rozen (1992)
56741	CFTR	I	D	II	Cystic Fibrosis	Kerem (1989), European Working Group on CF Genetics (1990), ozen (1990), Braekeleer (1991), Daigneault (1991), Rozen (1992), Kerem (1990), Wauters (1991), Gille (1991), Lerer (1992), Casals (1992), Ballabio (1990), Grebe (1992), Casals (1997), Russo (1995), Grebe (1994), Rose (2005), Wainwright (2015), Children's Hospital of Philadelphia (2016), Mendelics (2018), FRIGE's Institute of Human Genetics (2019), Invitae (2019), Johns Hopkins University (2019), Klinikum rechts der Isar (2020)
850038	**CFTR**	**I**	**D**	**ID**	**Cystic Fibrosis**	**Kerem (1989), European Working Group on CF Genetics (1990), ozen (1990), Braekeleer (1991), Daigneault (1991), Rozen (1992), Kerem (1990), Wauters (1991), Gille (1991), Lerer (1992), Casals (1992), Ballabio (1990), Grebe (1992), Casals (1997), Russo (1995), Grebe (1994), Rose (2005), Wainwright (2015), Children's Hospital of Philadelphia (2016), Mendelics (2018), FRIGE's Institute of Human Genetics (2019), Invitae (2019), Johns Hopkins University (2019), Klinikum rechts der Isar (2020)**
56710	CFTR	I	D	II	Possibly Associated With Cystic Fibrosis	Li (2010)
101454	CFTR	I	D	II	Cystic Fibrosis	Schwartz , 1994
56722	CFTR	C	T	CC	Cystic Fibrosis	Yoshimura (1999)
56735	CFTR	G	A	GG	Cystic Fibrosis	Marechal (2001)
56736	CFTR	C	G	CC	Cystic Fibrosis	Mercier (1993)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
56737	CFTR	A	C	AA	Cystic Fibrosis	Ferec (1995)
56738	CFTR	C	T	CC	Cystic Fibrosis	Elahi (2006)
56739	CFTR	C	G	CC	Cystic Fibrosis	Chevalier (1994)
56742	CFTR	G	A	GG	Cystic Fibrosis	Telleria (1999)
56750	CFTR	G	A,C	GG	Cystic Fibrosis	Jezequel (1995)
56752	CFTR	C	T	CC	Cystic Fibrosis	
56753	CFTR	T	A	TT	Cystic Fibrosis	Claustres (1993)
56754	CFTR	T	C	TT	Cystic Fibrosis	Bienvenu (2005)
56756	CFTR	G	A	GG	Cystic Fibrosis	Axton (1994)
56757	CFTR	G	T	GG	Cystic Fibrosis	Hughes (1996)
56758	CFTR	A	G	AA	Cystic Fibrosis	Cheadle (1993)
56759	CFTR	C	T	CC	Cystic Fibrosis	Savov (1994)
56760	CFTR	A	T	AA	Cystic Fibrosis	Savov (1994)
56761	CFTR	C	A	CC	Cystic Fibrosis	Glavac (1993)
56762	CFTR	C	T	CC	Cystic Fibrosis	Casals (1997)
56763	CFTR	C	G	CC	Cystic Fibrosis	Visich (2002)
56764	CFTR	G	T	GG	Cystic Fibrosis	Wong (2003)
56765	CFTR	A	C	AA	Cystic Fibrosis	Hughes (1995)
56766	CFTR	C	T	CC	Cystic Fibrosis	Cao (1996)
56767	CFTR	A	T	AA	Cystic Fibrosis	Ferec (1993)
56768	CFTR	G	A	GG	Cystic Fibrosis	Chevalier-Porst (1998)
56769	CFTR	G	T	GG	Cystic Fibrosis	Macek (1997)
56770	CFTR	G	C	GG	Cystic Fibrosis	Seia (2004)
56771	CFTR	G	A	GG	Cystic Fibrosis	Ramirez (2006)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
56772	CFTR	G	T	GG	Cystic Fibrosis	Shackleton (1992)
56773	CFTR	G	A	GG	Cystic Fibrosis	Bienvenu (1994)
56774	CFTR	C	T	CC	Cystic Fibrosis	Chillon (1994)
56775	CFTR	G	T	GG	Cystic Fibrosis	Zielinski (1995)
56776	CFTR	G	A	GG	Cystic Fibrosis	Girodon (2003)
56777	CFTR	C	T	CC	Cystic Fibrosis	Cutting (1992)
56778	CFTR	C	T	CC	Cystic Fibrosis	Ferec (1995)
56779	CFTR	A	G	AA	Cystic Fibrosis	Fanen (1992)
56780	CFTR	C	A	CC	Cystic Fibrosis	Tzetis (1997)
56781	CFTR	T	C	TT	Cystic Fibrosis	Casals (2000)
56782	CFTR	C	A	CC	Cystic Fibrosis	Zielenski (1997)
56784	CFTR	T	C	TT	Cystic Fibrosis	Malone (1997)
56785	CFTR	T	G	TT	Cystic Fibrosis	Brancolini (1995)
56786	CFTR	G	A	GG	Cystic Fibrosis	Audrezet (1993)
56787	CFTR	G	A	GG	Cystic Fibrosis	Marechal (1999)
56788	CFTR	A	G	AA	Cystic Fibrosis	Claustres (2000)
56790	CFTR	G	A	GG	Cystic Fibrosis	Claustres (2000)
56791	CFTR	G	T	GG	Cystic Fibrosis	Tsui (1992)
56792	CFTR	A	G	AA	Cystic Fibrosis	Girodon (2003)
56793	CFTR	A	G	AA	Cystic Fibrosis	Cashman (2003)
56794	CFTR	C	T	CC	Cystic Fibrosis	Claustres (1993)
56795	CFTR	A	T	AA	Cystic Fibrosis	Doerk (1992)
56796	CFTR	A	G	AA	Cystic Fibrosis	Kilinc (2002)
56797	CFTR	C	A	CC	Cystic Fibrosis	Gall (1996)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
56798	CFTR	G	A	GG	Cystic Fibrosis	Pacheco (1999)
56799	CFTR	G	A	GG	Cystic Fibrosis	Schrijver (2005)
56803	CFTR	C	T	CC	Cystic Fibrosis	Doerk (1994)
56804	CFTR	T	C	TT	Cystic Fibrosis	Macek (1993)
56805	CFTR	G	A	GG	Cystic Fibrosis	Ferec (1993)
56806	CFTR	G	A	GG	Cystic Fibrosis	Harris (1991), Zielenski (1991), Faucz (2007)
56807	CFTR	G	T	GG	Cystic Fibrosis	Casals (1997)
56808	CFTR	T	C	TT	Cystic Fibrosis	Bienvenu (1994)
56809	CFTR	T	A	TT	Cystic Fibrosis	Savov (1994)
56810	CFTR	T	C	TT	Cystic Fibrosis	Hughes (1996)
56811	CFTR	T	G	TT	Cystic Fibrosis	Macek (1992)
56812	CFTR	A	G	AA	Cystic Fibrosis	Seia (1999)
56813	CFTR	T	C	TT	Cystic Fibrosis	Ferec (1998)
56814	CFTR	G	A	GG	Cystic Fibrosis	Guillermit (1993)
56815	CFTR	A	T	AA	Cystic Fibrosis	Girodon (2004)
56816	CFTR	G	A	GG	Cystic Fibrosis	Shackleton (1992), Nunes (1993)
56817	CFTR	G	T	GG	Cystic Fibrosis	Will (1994), Will (1994)
56818	CFTR	C	A	CC	Cystic Fibrosis	Ferec (1998)
56819	CFTR	C	T	CC	Cystic Fibrosis	Malone (1998)
56821	CFTR	A	G	AA	Cystic Fibrosis	Romey (1995)
56822	CFTR	C	T	CC	Cystic Fibrosis	Schwartz (1992)
56824	CFTR	T	G	TT	Cystic Fibrosis	Casals (1996)
56825	CFTR	T	C	TT	Cystic Fibrosis	Baudis (2001)
56826	CFTR	G	T	GG	Cystic Fibrosis	Edkins (1992)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
56827	CFTR	T	A	TT	Cystic Fibrosis	Claustres (2000)
56828	CFTR	C	T	CC	Cystic Fibrosis	Seydewitz (1995)
56829	CFTR	T	A	TT	Cystic Fibrosis	Schaedel (1998)
56830	CFTR	A	G	AA	Cystic Fibrosis	Schaedel (1994)
56831	CFTR	T	A	TT	Cystic Fibrosis	Feuillet-Fieux (2004)
56832	CFTR	C	A	CC	Cystic Fibrosis	Padoan (2002)
56834	CFTR	G	C	GG	Cystic Fibrosis	Orita (1989), Dean (1990)
56835	CFTR	G	T	GG	Cystic Fibrosis	Casals (2000)
56836	CFTR	C	G	CC	Cystic Fibrosis	Ferec (1996)
56838	CFTR	A	T	AA	Cystic Fibrosis	Bonizzato (2004)
56839	CFTR	G	T	GG	Cystic Fibrosis	Banjar (1999)
56840	CFTR	G	A	GG	Cystic Fibrosis	Schrijver (2005)
56841	CFTR	G	C	GG	Cystic Fibrosis	Walker (2000)
56842	CFTR	C	T	CC	Cystic Fibrosis	Doerk (1994)
56844	CFTR	G	C	GG	Cystic Fibrosis	Reiss (1993)
56845	CFTR	G	T	GG	Cystic Fibrosis	Ferec (1995)
56846	CFTR	A	G	AA	Cystic Fibrosis	Schwarz (2002)
56847	CFTR	G	A	GG	Cystic Fibrosis	Chillon (1994)
56849	CFTR	T	A	TT	Cystic Fibrosis	Chevalier (1992), Sermet-Gaudelus (2007)
56851	CFTR	T	C	TT	Cystic Fibrosis	Kilinc (2002)
56852	CFTR	G	A	GG	Cystic Fibrosis	Wagner (1994)
56853	CFTR	T	G	TT	Cystic Fibrosis	Ferec (1996)
56854	CFTR	T	A	TT	Cystic Fibrosis	Wallace (1996)
56855	CFTR	T	C	TT	Cystic Fibrosis	Patrinos (2005)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
56856	CFTR	T	G	TT	Cystic Fibrosis	Chevalier-Porst (1997)
56857	CFTR	A	G	AA	Cystic Fibrosis	Ferec (1995)
56858	CFTR	A	T	AA	Cystic Fibrosis	Banjar (1999)
56859	CFTR	C	T	CC	Cystic Fibrosis	Tzetis (1998)
56860	CFTR	C	T	CC	Cystic Fibrosis	Ferec (1994)
56861	CFTR	C	A	CC	Cystic Fibrosis	Gouya (1997)
56865	CFTR	G	A	GG	Cystic Fibrosis	Mercier (1995)
56866	CFTR	T	A	TT	Cystic Fibrosis	Kammesheidt (2006)
56868	CFTR	C	T	CC	Cystic Fibrosis	Shackleton (1992)
56869	CFTR	T	G	TT	Cystic Fibrosis	Morokawa (2000)
56870	CFTR	A	G	AA	Cystic Fibrosis	Wong (2004)
56871	CFTR	G	C	GG	Cystic Fibrosis	Zielenski (1997)
56875	CFTR	T	A	TT	Cystic Fibrosis	Andrew (2000)
56876	CFTR	T	C	TT	Cystic Fibrosis	Casals (2002)
56877	CFTR	A	C	AA	Cystic Fibrosis	Andrew (1999)
56878	CFTR	A	G	AA	Cystic Fibrosis	Hirtz (2004)
56879	CFTR	T	A	TT	Cystic Fibrosis	Georges (2004)
56880	CFTR	T	G	TT	Cystic Fibrosis	Strandvik (2001)
56881	CFTR	A	G	AA	Cystic Fibrosis	Tzetis (1998)
56882	CFTR	T	C	TT	Cystic Fibrosis	Quint (2005)
56883	CFTR	A	G	AA	Cystic Fibrosis	Macek (1997)
56884	CFTR	C	G	CC	Cystic Fibrosis	Claustres (1992)
56885	CFTR	C	T	CC	Cystic Fibrosis	Ferec (1994)
56886	CFTR	A	G	AA	Cystic Fibrosis	Romey (1994)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
56887	CFTR	T	C	TT	Cystic Fibrosis	Bienvenu (1996)
56888	CFTR	G	A	GG	Cystic Fibrosis	Soldatova (2004)
56889	CFTR	G	A	GG	Cystic Fibrosis	Zielenski (1991)
56890	CFTR	C	A	CC	Cystic Fibrosis	Wong (2004)
56891	CFTR	C	A	CC	Cystic Fibrosis	Claustres (1998)
56892	CFTR	C	A	CC	Cystic Fibrosis	Gaia (2002)
56893	CFTR	C	A	CC	Cystic Fibrosis	Li (2006)
56894	CFTR	T	G	TT	Cystic Fibrosis	Groman (2002)
56895	CFTR	A	G	AA	Cystic Fibrosis	Audrezet (1994)
56896	CFTR	G	A	GG	Cystic Fibrosis	Hubert (2004)
56897	CFTR	G	A	GG	Cystic Fibrosis	Mercier (1995)
56898	CFTR	G	T	GG	Cystic Fibrosis	Ferec (1994)
56900	CFTR	G	T	GG	Cystic Fibrosis	Ferec (1998)
56901	CFTR	G	C	GG	Cystic Fibrosis	Walker (1999)
56902	CFTR	C	T	CC	Cystic Fibrosis	Doerk (1994)
56903	CFTR	A	G	AA	Cystic Fibrosis	D'Apice (2004)
56904	CFTR	T	G	TT	Cystic Fibrosis	Tsui (1992)
56906	CFTR	G	A	GG	Cystic Fibrosis	Bernardio (2000)
56907	CFTR	G	A	GG	Cystic Fibrosis	Casals (2003)
56909	CFTR	C	G	CC	Cystic Fibrosis	Ferec (2002)
56910	CFTR	C	T	CC	Cystic Fibrosis	Chillon (1993)
56911	CFTR	T	G	TT	Cystic Fibrosis	Claustres (1993), Rozen (1995), Clain (2005), Clain (2005), Clain (2005)
56912	CFTR	G	T	GG	Cystic Fibrosis	Ferec (1997)
56913	CFTR	C	T	CC	Cystic Fibrosis	Zielenski (1993)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
56914	CFTR	G	T	GG	Cystic Fibrosis	Ferec (1998)
56915	CFTR	T	C	TT	Cystic Fibrosis	Girodon (2003)
56916	CFTR	G	T	GG	Cystic Fibrosis	Ferec (2002)
56917	CFTR	G	A	GG	Cystic Fibrosis	Yoshimura (1999)
56918	CFTR	A	G	AA	Cystic Fibrosis	Zielenski (1997), Lee (2003)
56919	CFTR	T	A	TT	Cystic Fibrosis	Zielenski (1996)
56920	CFTR	C	T	CC	Cystic Fibrosis	Shackleton (1994)
56921	CFTR	A	G	AA	Cystic Fibrosis	Ferec (1998)
56922	CFTR	T	C	TT	Cystic Fibrosis	Fanen (1992)
56923	CFTR	T	A	TT	Cystic Fibrosis	Mercier (1996)
56924	CFTR	T	G	TT	Cystic Fibrosis	Dequeker (2000)
56925	CFTR	T	A	TT	Cystic Fibrosis	Casals (1997)
56926	CFTR	C	G	CC	Cystic Fibrosis	Hubert (2004)
56927	CFTR	G	A	GG	Cystic Fibrosis	Bienvenu (1995)
56928	CFTR	G	A	GG	Cystic Fibrosis	Marechal (2001)
56929	CFTR	A	C	AA	Cystic Fibrosis	Yoshimura (1999)
56931	CFTR	C	G	CC	Cystic Fibrosis	Tzetis (1997)
56933	CFTR	G	T	GG	Cystic Fibrosis	Kammesheidt (2006)
56934	CFTR	A	G	AA	Cystic Fibrosis	Mercier (1995)
56936	CFTR	C	G	CC	Cystic Fibrosis	Bernardio (2000)
56937	CFTR	C	A	CC	Cystic Fibrosis	Ferec (1995)
56938	CFTR	T	C	TT	Cystic Fibrosis	Ramirez (2006)
56939	CFTR	T	C	TT	Cystic Fibrosis	Casals (2002)
56941	CFTR	A	T	AA	Cystic Fibrosis	Shrimpton (1997)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
56942	CFTR	C	T	CC	Cystic Fibrosis	Ferec (1995)
56943	CFTR	G	A	GG	Cystic Fibrosis	Bienvenu (1996)
56944	CFTR	C	A	CC	Cystic Fibrosis	Ferec (2002)
56947	CFTR	G	A	GG	Cystic Fibrosis	Ferec (1999)
56949	CFTR	C	G	CC	Cystic Fibrosis	Ferec (1996)
56950	CFTR	G	A	GG	Cystic Fibrosis	Onay (1998)
56952	CFTR	C	A	CC	Cystic Fibrosis	Ferrari (1994)
56953	CFTR	C	G	CC	Cystic Fibrosis	Cartault (1998)
56955	CFTR	T	C	TT	Cystic Fibrosis	Ferec (1992)
56956	CFTR	G	C	GG	Cystic Fibrosis	Nasr (1996)
56957	CFTR	G	A	GG	Cystic Fibrosis	Tsui (1992)
56958	CFTR	G	T	GG	Cystic Fibrosis	Chevalier-Porst (1997)
56959	CFTR	T	C	TT	Cystic Fibrosis	Ferec (1993)
56960	CFTR	T	A	TT	Cystic Fibrosis	Iron (2001)
56961	CFTR	A	T	AA	Cystic Fibrosis	Macek (1994)
56963	CFTR	T	C	TT	Cystic Fibrosis	Seia (2004)
56964	CFTR	T	C	TT	Cystic Fibrosis	Ferec (1994)
56966	CFTR	G	T	GG	Cystic Fibrosis	Macek (1997)
56967	CFTR	C	T	CC	Cystic Fibrosis	Gasparini (1991), Antinolo (1997), Faucz (2007)
56968	CFTR	G	A	GG	Cystic Fibrosis	Ferec (1995)
56970	CFTR	T	A	TT	Cystic Fibrosis	Cuppens (1993)
56971	CFTR	C	T	CC	Cystic Fibrosis	Saba (1993), Leoni (1995)
56973	CFTR	T	A	TT	Cystic Fibrosis	Ferec (2002)
56974	CFTR	T	C	TT	Cystic Fibrosis	Boteva (1994)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
56975	CFTR	G	A	GG	Cystic Fibrosis	Audrezet (1993)
56976	CFTR	G	C	GG	Cystic Fibrosis	Dean (1990)
56977	CFTR	G	T	GG	Cystic Fibrosis	Audrezet (1993), Audrezet (1993)
56978	CFTR	C	T	CC	Cystic Fibrosis	Ferec (1993)
56981	CFTR	C	T	CC	Cystic Fibrosis	Audrezet (1993)
56982	CFTR	G	A	GG	Cystic Fibrosis	Gasparini (1993)
56983	CFTR	C	G	CC	Cystic Fibrosis	Girodon (2003)
56984	CFTR	C	T	CC	Cystic Fibrosis	Schrijver (2005)
56985	CFTR	C	T	CC	Cystic Fibrosis	Seia (2000)
56986	CFTR	G	C	GG	Cystic Fibrosis	Barbolina (2004)
56987	CFTR	G	A	GG	Cystic Fibrosis	Ellis (1998)
56988	CFTR	A	G	AA	Cystic Fibrosis	Ferec (1999)
56989	CFTR	C	G	CC	Cystic Fibrosis	Ferec (1998)
56990	CFTR	T	A	TT	Cystic Fibrosis	Telleria (1998)
56991	CFTR	T	C	TT	Cystic Fibrosis	Bienvenu (1993)
56992	CFTR	T	G	TT	Cystic Fibrosis	Visich (2002)
56993	CFTR	T	C	TT	Cystic Fibrosis	Macek (1997)
56994	CFTR	T	C	TT	Cystic Fibrosis	Casals (2000)
56995	CFTR	A	C	AA	Cystic Fibrosis	Jezequel (1996)
56996	CFTR	A	G	AA	Cystic Fibrosis	Tzetis (2002)
56998	CFTR	G	T	GG	Cystic Fibrosis	Glaeser (2000)
56999	CFTR	T	C	TT	Cystic Fibrosis	Casals (2000)
57002	CFTR	T	G	TT	Cystic Fibrosis	Zielenski (1997)
57003	CFTR	T	G	TT	Cystic Fibrosis	Ferec (1998)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
57004	CFTR	C	A	CC	Cystic Fibrosis	Casals (2002)
57005	CFTR	C	T	CC	Cystic Fibrosis	Yoshimura (2000)
57006	CFTR	G	A	GG	Cystic Fibrosis	Schrijver (2005)
57007	CFTR	G	A	GG	Cystic Fibrosis	Cuppens (1993)
57008	CFTR	G	C	GG	Cystic Fibrosis	Ferec (1999)
57009	CFTR	A	T	AA	Cystic Fibrosis	Zielenski (1999)
57010	CFTR	C	T	CC	Cystic Fibrosis	Doerk (1994)
57011	CFTR	A	G	AA	Cystic Fibrosis	Sava (1994)
57012	CFTR	G	A	GG	Cystic Fibrosis	Bienvenu (2000)
57013	CFTR	C	G	CC	Cystic Fibrosis	Leymarie (1998)
57014	CFTR	C	T	CC	Cystic Fibrosis	Schrijver (2005)
57016	CFTR	T	G	TT	Cystic Fibrosis	Zielenski (1999)
57017	CFTR	G	T	GG	Cystic Fibrosis	McGinniss (2005)
57018	CFTR	A	C	AA	Cystic Fibrosis	Claustres (1997)
57019	CFTR	C	A	CC	Cystic Fibrosis	Kerem (1990), Rozen (1992)
57020	CFTR	G	T	GG	Cystic Fibrosis	Doerk (1994)
57021	CFTR	T	C	TT	Cystic Fibrosis	McCormick (2002)
57022	CFTR	G	T	GG	Cystic Fibrosis	Cuppens (1990), Tsui (1992)
57023	CFTR	G	A	GG	Cystic Fibrosis	Groman (2002)
57024	CFTR	G	T	GG	Cystic Fibrosis	Bieth (2003)
57025	CFTR	C	A	CC	Cystic Fibrosis	Kammesheidt (2006)
57026	CFTR	C	A	CC	Cystic Fibrosis	Mittre (1996)
57027	CFTR	C	G	CC	Cystic Fibrosis	Deufel (1994)
57031	CFTR	G	A	GG	Cystic Fibrosis	Girodon (1999)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
57032	CFTR	G	A	GG	Cystic Fibrosis	Kawasoe (2001)
57033	CFTR	G	T	GG	Cystic Fibrosis	Smit (1995)
57034	CFTR	G	A	GG	Cystic Fibrosis	Hawworth (1995)
57036	CFTR	A	G	AA	Cystic Fibrosis	Ferec (1998)
57038	CFTR	A	T	AA	Cystic Fibrosis	Andrew (1999)
57039	CFTR	G	T	GG	Cystic Fibrosis	Hughes (1995)
57040	CFTR	T	G	TT	Cystic Fibrosis	Korytina (2003)
57041	CFTR	C	A	CC	Cystic Fibrosis	Plouvier (1997)
57042	CFTR	T	C	TT	Cystic Fibrosis	Chevalier-Porst (1998)
57043	CFTR	C	T	CC	Cystic Fibrosis	Ferec (1992)
57044	CFTR	A	C	AA	Cystic Fibrosis	Kilinc (2002)
57045	CFTR	A	G	AA	Cystic Fibrosis	Savov (1994)
57046	CFTR	C	T	CC	Cystic Fibrosis	Kerem (1990)
57047	CFTR	G	A	GG	Cystic Fibrosis	Balassopoulou (1994)
57050	CFTR	A	G	AA	Cystic Fibrosis	Schrijver (2005)
57051	CFTR	T	A	TT	Cystic Fibrosis	Hinks (2002)
57052	CFTR	T	C	TT	Cystic Fibrosis	Chevalier-Porst (1998)
57053	CFTR	G	C	GG	Cystic Fibrosis	Tsui (1992)
57054	CFTR	G	T	GG	Cystic Fibrosis	Casals (1997)
57055	CFTR	T	C	TT	Cystic Fibrosis	Desgeorges (1995)
57056	CFTR	T	G	TT	Cystic Fibrosis	Deufel (1994)
57062	CFTR	T	C	TT	Cystic Fibrosis	Mittre (2002)
57063	CFTR	A	G	AA	Cystic Fibrosis	Arduino (2002)
57064	CFTR	A	G	AA	Cystic Fibrosis	Arduino (1997)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
57065	CFTR	G	A	GG	Cystic Fibrosis	Bienvenu (2005)
57066	CFTR	G	T	GG	Cystic Fibrosis	Jones (1992)
57067	CFTR	A	T	AA	Cystic Fibrosis	Patrinos (2005)
57068	CFTR	C	A	CC	Cystic Fibrosis	Jones (1992)
57069	CFTR	C	T	CC	Cystic Fibrosis	Shackleton (1994)
57070	CFTR	G	C	GG	Cystic Fibrosis	Byrne (1997)
57072	CFTR	G	C	GG	Cystic Fibrosis	Ferec (1998)
57073	CFTR	T	C	TT	Cystic Fibrosis	Kambouris (2000)
57074	CFTR	C	A	CC	Cystic Fibrosis	Audrezet (1993)
57076	CFTR	T	C	TT	Cystic Fibrosis	Chomel (1995)
57077	CFTR	G	T	GG	Cystic Fibrosis	Kerem (1990), Cuppens (1990), Lerer (1992), Gasparini (1993), Grebe (1994), Savov (1995), Castaldo (1997), Loirat (1997), Casals (1997)
57078	CFTR	G	A	GG	Cystic Fibrosis	Ferec (1994)
57081	CFTR	G	A	GG	Cystic Fibrosis	Cutting (1990)
57082	CFTR	G	T	GG	Cystic Fibrosis	Kerem (1990)
57083	CFTR	T	G	TT	Cystic Fibrosis	Kerem (1990), Sangiuolo (1991), Romey (1999), Romey (1999), Romey (2000)
57084	CFTR	A	C	AA	Cystic Fibrosis	Sangiuolo (1991)
57085	CFTR	G	A	GG	Cystic Fibrosis	Ferec (1995)
57086	CFTR	G	T	GG	Cystic Fibrosis	Deiman (1992)
57087	CFTR	G	A	GG	Cystic Fibrosis	Strong (1991)
57088	CFTR	G	A	GG	Cystic Fibrosis	Cutting (1990), Curtis (1991), Burger (1991), Hamosh (1992), Hamosh (1992), Delaney (1996), Bobadilla (2002)
57090	CFTR	C	A	CC	Cystic Fibrosis	Faucz (1996)
57091	CFTR	C	T	CC	Cystic Fibrosis	Gasparini (1993)
57092	CFTR	C	G	CC	Cystic Fibrosis	Schrijver (2005)
57093	CFTR	C	T	CC	Cystic Fibrosis	Cutting (1990), Cutting (1990), Cutting (1990), Bal (1991), Hamosh (1991), Hamosh (1991), Cheadle (1992), Chen (2005), Chen (2005), Faucz (2007)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
57094	CFTR	G	A	GG	Cystic Fibrosis	Dork (1991), Stern (1997)
57095	CFTR	C	A	CC	Cystic Fibrosis	Audrezet (1993)
57096	CFTR	A	G	AA	Cystic Fibrosis	Zielenski (1999)
57098	CFTR	T	C	TT	Cystic Fibrosis	Tsui (1992)
57099	CFTR	G	A	GG	Cystic Fibrosis	Cutting (1990)
57100	CFTR	C	A	CC	Cystic Fibrosis	Girodon (1999)
57101	CFTR	A	G	AA	Cystic Fibrosis	Casals (2002)
57102	CFTR	G	A	GG	Cystic Fibrosis	Vidaud (1989), Ferec (1992)
57103	CFTR	G	C	GG	Cystic Fibrosis	Kerem (1990)
57104	CFTR	A	C	AA	Cystic Fibrosis	Malone (1998)
57105	CFTR	A	T	AA	Cystic Fibrosis	Liechti (1999)
57106	CFTR	C	A	CC	Cystic Fibrosis	Mendes (2003)
57107	CFTR	G	C	GG	Cystic Fibrosis	Hughes (1996)
57108	CFTR	T	A	TT	Cystic Fibrosis	Kerem (1990)
57109	CFTR	T	G	TT	Cystic Fibrosis	Macek (1997)
57110	CFTR	A	G	AA	Cystic Fibrosis	Delhaize (1996)
57111	CFTR	C	A	CC	Cystic Fibrosis	Elahi (2006)
57113	CFTR	G	A	GG	Cystic Fibrosis	Wallace (2003)
57114	CFTR	T	A	TT	Cystic Fibrosis	Macek (1997), Counsyl (2016), Laboratory Corporation of America (2017)
57116	CFTR	T	C	TT	Cystic Fibrosis	Kabra (2000)
57117	CFTR	T	G	TT	Cystic Fibrosis	Malone (1998)
57118	CFTR	A	G	AA	Cystic Fibrosis	Petreska (1996)
57119	CFTR	T	A	TT	Cystic Fibrosis	Ferec (1993)
57120	CFTR	T	C	TT	Cystic Fibrosis	Casals (1997)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
57121	CFTR	G	A	GG	Cystic Fibrosis	Verlingue (1995)
57123	CFTR	C	A	CC	Cystic Fibrosis	Kerem (1990)
57125	CFTR	G	T	GG	Cystic Fibrosis	Girodon (1996)
57126	CFTR	A	T	AA	Cystic Fibrosis	Pagani (2003)
57127	CFTR	G	T	GG	Cystic Fibrosis	Harris (1994)
57128	CFTR	A	C	AA	Cystic Fibrosis	Pacheco (1998)
57129	CFTR	A	G	AA	Cystic Fibrosis	Brancolini (1995)
57131	CFTR	C	G	CC	Cystic Fibrosis	Chillon (1994)
57132	CFTR	C	T	CC	Cystic Fibrosis	Georges (2004)
57133	CFTR	G	T	GG	Cystic Fibrosis	Cremonesi (1992)
57135	CFTR	A	T	AA	Cystic Fibrosis	Schrijver (2005)
57136	CFTR	G	T	GG	Cystic Fibrosis	Spitzer (2002)
57137	CFTR	G	A	GG	Cystic Fibrosis	Stanziale (2005)
57138	CFTR	G	C	GG	Cystic Fibrosis	Kinnunen (2005)
57140	CFTR	A	G	AA	Cystic Fibrosis	Hubert (2004)
57141	CFTR	T	C	TT	Cystic Fibrosis	Bienvenu (2005)
57142	CFTR	A	T	AA	Cystic Fibrosis	Vankeerberghen (1998)
57143	CFTR	G	T	GG	Cystic Fibrosis	Strandvik (2001)
57144	CFTR	C	G	CC	Cystic Fibrosis	Schrijver (2005)
57146	CFTR	A	G	AA	Cystic Fibrosis	Kilinc (2002)
57147	CFTR	A	G	AA	Cystic Fibrosis	Padoan (2002)
57148	CFTR	T	C	TT	Cystic Fibrosis	Vankeerberghen (1998)
57149	CFTR	G	A	GG	Cystic Fibrosis	Vankeerberghen (1998)
57150	CFTR	A	G	AA	Cystic Fibrosis	Audrezet (1993)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
57151	CFTR	G	T	GG	Cystic Fibrosis	Girodon (1999)
57152	CFTR	T	C	TT	Cystic Fibrosis	Macek (1997)
57153	CFTR	T	C	TT	Cystic Fibrosis	Doerk (1994)
57154	CFTR	A	C	AA	Cystic Fibrosis	Vankeerberghen (1998)
57155	CFTR	T	G	TT	Cystic Fibrosis	Vankeerberghen (1998)
57156	CFTR	G	A	GG	Cystic Fibrosis	Vankeerberghen (1998)
57157	CFTR	G	A	GG	Cystic Fibrosis	Fanen (1992)
57158	CFTR	G	C	GG	Cystic Fibrosis	Cuppens (1993)
57159	CFTR	T	C	TT	Cystic Fibrosis	Vankeerberghen (1998)
57160	CFTR	C	T	CC	Cystic Fibrosis	Michel-Calemard (1996)
57161	CFTR	T	C	TT	Cystic Fibrosis	Bombieri (1998)
57162	CFTR	C	T	CC	Cystic Fibrosis	Hirtz (2004)
57163	CFTR	G	T	GG	Cystic Fibrosis	Aulehla-Scholz (2002)
57164	CFTR	A	T	AA	Cystic Fibrosis	Gasparini (1993)
57165	CFTR	G	A	GG	Cystic Fibrosis	Bombieri (1998)
57167	CFTR	G	T	GG	Cystic Fibrosis	Claustres (1997)
57168	CFTR	G	T	GG	Cystic Fibrosis	Clavel (1997)
57169	CFTR	A	T	AA	Cystic Fibrosis	Messaoud (1996)
57170	CFTR	G	T	GG	Cystic Fibrosis	Doerk (1994)
57171	CFTR	G	A	GG	Cystic Fibrosis	Walker (1999)
57172	CFTR	A	G	AA	Cystic Fibrosis	Chevalier-Porst (2000)
57173	CFTR	C	T	CC	Cystic Fibrosis	Audrezet (1994)
57175	CFTR	C	T	CC	Cystic Fibrosis	Girodon (2003)
57176	CFTR	G	T	GG	Cystic Fibrosis	Casals (1997)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
57177	CFTR	T	C	TT	Cystic Fibrosis	Audrezet (1993)
57179	CFTR	A	G	AA	Cystic Fibrosis	Ferec (1997)
57180	CFTR	C	T	CC	Cystic Fibrosis	Bonizzato (1995)
57181	CFTR	G	A	GG	Cystic Fibrosis	Wagner (2002)
57182	CFTR	A	T	AA	Cystic Fibrosis	Gasparini (1993)
57183	CFTR	C	G	CC	Cystic Fibrosis	Ferec (2002)
57184	CFTR	A	T	AA	Cystic Fibrosis	Audrezet (1993)
57185	CFTR	T	A	TT	Cystic Fibrosis	Doerk (1994)
57186	CFTR	C	T	CC	Cystic Fibrosis	Malone (1999)
57188	CFTR	G	A	GG	Cystic Fibrosis	Georges (2004)
57189	CFTR	G	T	GG	Cystic Fibrosis	Cuppens (1993)
57190	CFTR	T	G	TT	Cystic Fibrosis	Tzetis (1997)
57191	CFTR	G	A	GG	Cystic Fibrosis	Benga (2001)
57193	CFTR	C	T	CC	Likely associated with Cystic Fibrosis	Orozco (2000), Center for Pediatric Genomic Medicine of Children's Mercy Hospital and Clinics (2017), Counsyl (2017), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2017), Laboratory Corporation of America (2017), ARUP Laboratories (2017), Illumina Clinical Services Laboratory (2018), Mendelics (2018)
57194	CFTR	G	C	GG	Cystic Fibrosis	Ferec (2002)
57196	CFTR	C	T	CC	Cystic Fibrosis	Zielenski (1999)
57197	CFTR	C	T	CC	Cystic Fibrosis	Hughes (1996)
57199	CFTR	C	G	CC	Cystic Fibrosis	Georges (2004)
57200	CFTR	C	T	CC	Cystic Fibrosis	Zhou (1997)
57201	CFTR	C	A	CC	Cystic Fibrosis	Girodon (2002)
57202	CFTR	C	T	CC	Cystic Fibrosis	Wallace (1994)
57204	CFTR	C	T	CC	Cystic Fibrosis	Claustres (1993)
57205	CFTR	C	G	CC	Cystic Fibrosis	Mercier (1995)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
57208	CFTR	T	C	TT	Cystic Fibrosis	Tzetis (2002)
57209	CFTR	C	T	CC	Cystic Fibrosis	Ahmed (2003)
57210	CFTR	G	A	GG	Cystic Fibrosis	Mercier (1993)
57211	CFTR	G	T	GG	Cystic Fibrosis	Tzetis (1997)
57212	CFTR	G	T	GG	Cystic Fibrosis	Tzetis (1998)
57213	CFTR	G	A	GG	Cystic Fibrosis	Vankeerberghen (1998)
57214	CFTR	G	T	GG	Cystic Fibrosis	Ferec (1992)
57215	CFTR	A	G	AA	Cystic Fibrosis	Ferec (2002)
57216	CFTR	A	T	AA	Cystic Fibrosis	Mercier (1993)
57217	CFTR	G	T	GG	Cystic Fibrosis	Hughes (1996)
57219	CFTR	G	T	GG	Cystic Fibrosis	Georges (2004)
57221	CFTR	G	A	GG	Cystic Fibrosis	Cheadle (1993)
57222	CFTR	G	A	GG	Cystic Fibrosis	Vidaud (1990)
57223	CFTR	C	G	CC	Cystic Fibrosis	Castaldo (1999)
57224	CFTR	G	T	GG	Cystic Fibrosis	Casals (1997)
57225	CFTR	C	T	CC	Cystic Fibrosis	White (1991)
57227	CFTR	G	A	GG	Cystic Fibrosis	Tsui (1992)
57228	CFTR	T	A	TT	Cystic Fibrosis	Haworth (1996)
57229	CFTR	G	A	GG	Cystic Fibrosis	Ferec (1996)
57230	CFTR	A	G	AA	Cystic Fibrosis	Casals (2000)
57231	CFTR	C	T	CC	Cystic Fibrosis	Ghanem (1994)
57232	CFTR	A	G	AA	Cystic Fibrosis	Kilinc (1998)
57234	CFTR	C	T	CC	Cystic Fibrosis	Lazaro (2000)
57236	CFTR	C	A	CC	Cystic Fibrosis	Hammerle (2000)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
57237	CFTR	C	A	CC	Cystic Fibrosis	Saba (1993)
57239	CFTR	A	G	AA	Cystic Fibrosis	Vidaud (1990)
57240	CFTR	T	A	TT	Cystic Fibrosis	Schrijver (2005)
57241	CFTR	T	A	TT	Cystic Fibrosis	Kammesheidt (2006)
57242	CFTR	A	G	AA	Cystic Fibrosis	Hughes (1996)
57243	CFTR	T	G	TT	Cystic Fibrosis	Schwarz (1996)
57244	CFTR	A	G	AA	Cystic Fibrosis	Savov (1994)
57245	CFTR	G	A	GG	Cystic Fibrosis	Steffan (1998)
57247	CFTR	T	C	TT	Cystic Fibrosis	Hermans (1994)
57248	CFTR	T	C	TT	Cystic Fibrosis	Ferec (1999)
57252	CFTR	A	G	AA	Cystic Fibrosis	Ferec (1996)
57253	CFTR	C	G	CC	Cystic Fibrosis	Ferec (1993)
57254	CFTR	C	T	CC	Cystic Fibrosis	Claustres (1993)
57255	CFTR	A	T	AA	Cystic Fibrosis	Haworth (1996)
57256	CFTR	C	T	CC	Cystic Fibrosis	Ghanem (1994)
57257	CFTR	A	G	AA	Cystic Fibrosis	Ferec (1995)
57259	CFTR	T	C	TT	Cystic Fibrosis	Casals (2000)
57260	CFTR	G	C	GG	Cystic Fibrosis	Onay (1998)
57262	CFTR	G	T	GG	Cystic Fibrosis	Malone (2000)
57263	CFTR	G	C	GG	Cystic Fibrosis	Cuppens (1993)
57264	CFTR	G	A	GG	Cystic Fibrosis	Wagner (1999)
57265	CFTR	T	C	TT	Cystic Fibrosis	Ferec (1998)
57266	CFTR	T	C	TT	Cystic Fibrosis	Hughes (1996)
57267	CFTR	C	T	CC	Cystic Fibrosis	Ferec (1997)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
57269	CFTR	A	T	AA	Cystic Fibrosis	Plouvier (1997)
57270	CFTR	T	A	TT	Cystic Fibrosis	Bienvenu (1996)
57271	CFTR	G	C	GG	Cystic Fibrosis	Claustres (1998)
57272	CFTR	G	T	GG	Cystic Fibrosis	Bienvenu (1994)
57274	CFTR	G	T	GG	Cystic Fibrosis	Georges (2004)
57275	CFTR	T	G	TT	Cystic Fibrosis	Claustres (1998)
57276	CFTR	G	T	GG	Cystic Fibrosis	Ferec (2000)
57277	CFTR	T	G	TT	Cystic Fibrosis	Doerk (1994)
57278	CFTR	C	A	CC	Cystic Fibrosis	Ferec (1995)
57279	CFTR	T	A	TT	Cystic Fibrosis	Casals (1998)
57280	CFTR	G	A	GG	Cystic Fibrosis	Schrijver (2005)
57282	CFTR	C	T	CC	Cystic Fibrosis	Onay (1998)
57283	CFTR	A	G	AA	Cystic Fibrosis	Bozon (2000)
57284	CFTR	T	C	TT	Cystic Fibrosis	Alper (2004)
57285	CFTR	C	G	CC	Cystic Fibrosis	Tzetis (2002)
57288	CFTR	T	G	TT	Cystic Fibrosis	Lazaro (2000)
57289	CFTR	G	T	GG	Cystic Fibrosis	Onay (1998)
57290	CFTR	T	A	TT	Cystic Fibrosis	Seia (2004)
57292	CFTR	C	T	CC	Cystic Fibrosis	Bogdanova (2004)
57293	CFTR	C	A	CC	Cystic Fibrosis	McGinniss (2005)
57294	CFTR	C	T	CC	Cystic Fibrosis	Ferec (1998)
57295	CFTR	T	G	TT	Cystic Fibrosis	Messaoud (2005)
57296	CFTR	G	C	GG	Cystic Fibrosis	Ferec (2002)
57297	CFTR	A	G	AA	Cystic Fibrosis	Pasquet (2004)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
57298	CFTR	T	G	TT	Cystic Fibrosis	Mercier (1993)
57299	CFTR	C	T	CC	Cystic Fibrosis	Bienvenu (1998)
57300	CFTR	C	G	CC	Cystic Fibrosis	Mercier (1994)
57302	CFTR	A	G	AA	Cystic Fibrosis	Fiorentino (2005)
57303	CFTR	T	G	TT	Cystic Fibrosis	Doerk (1994)
57304	CFTR	A	C	AA	Cystic Fibrosis	Casals (1995)
57305	CFTR	G	A	GG	Cystic Fibrosis	Bienvenu (1996)
57306	CFTR	G	C	GG	Cystic Fibrosis	Mercier (1993)
57307	CFTR	G	A	GG	Cystic Fibrosis	Fanen (1992)
57308	CFTR	C	T	CC	Cystic Fibrosis	Tzetis (1998)
57309	CFTR	T	C	TT	Cystic Fibrosis	Ghanem (1994)
57310	CFTR	T	G	TT	Cystic Fibrosis	Casals (1998)
57311	CFTR	G	A	GG	Cystic Fibrosis	Ferec (1992), Ferec (1992)
57312	CFTR	G	T	GG	Cystic Fibrosis	Mercier (1993)
57313	CFTR	C	A	CC	Cystic Fibrosis	Schrijver (2005)
57314	CFTR	C	T	CC	Cystic Fibrosis	Ferec (1992)
57315	CFTR	C	A	CC	Cystic Fibrosis	Girodon (1999)
57316	CFTR	G	A	GG	Cystic Fibrosis	Ferec (1992)
57317	CFTR	G	A	GG	Cystic Fibrosis	Savov (1994)
57318	CFTR	C	T	CC	Cystic Fibrosis	Jezequel (1995)
57319	CFTR	G	A	GG	Cystic Fibrosis	Mercier (1993)
57320	CFTR	G	C	GG	Cystic Fibrosis	Shrimpton (1997)
57321	CFTR	A	C	AA	Cystic Fibrosis	Ghanem (1994)
57322	CFTR	G	T	GG	Cystic Fibrosis	Claustres (2000)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
57324	CFTR	C	T	CC	Cystic Fibrosis	Bombieri (1998)
57326	CFTR	T	A	TT	Cystic Fibrosis	Casals (1997)
57327	CFTR	T	C	TT	Cystic Fibrosis	Tsui (1992)
57329	CFTR	A	G	AA	Cystic Fibrosis	Gasparini (1993)
57330	CFTR	C	T	CC	Cystic Fibrosis	Bienvenu (1996)
57331	CFTR	A	G	AA	Cystic Fibrosis	Girodon (2003)
57333	CFTR	A	G	AA	Cystic Fibrosis	Strandvik (2001)
57334	CFTR	T	C	TT	Cystic Fibrosis	Zielinski (1995)
57335	CFTR	G	A	GG	Cystic Fibrosis	Shoshani (1994)
57336	CFTR	T	C	TT	Cystic Fibrosis	Egan (1997)
57337	CFTR	A	G	AA	Cystic Fibrosis	Trujillo-Tiebas (2004)
57338	CFTR	C	A	CC	Cystic Fibrosis	Gasparini (1993)
57339	CFTR	C	G	CC	Cystic Fibrosis	Shrijver (2005)
57340	CFTR	T	C	TT	Cystic Fibrosis	Yee (1999)
57341	CFTR	T	G	TT	Cystic Fibrosis	Claustres (1998)
57342	CFTR	T	C	TT	Cystic Fibrosis	Schrijver (2005)
57343	CFTR	G	A	GG	Cystic Fibrosis	Chillon (1994)
57344	CFTR	G	A	GG	Cystic Fibrosis	Michel-Calemard (1996)
57345	CFTR	G	T	GG	Cystic Fibrosis	Orozco (2000)
57346	CFTR	C	A	CC	Cystic Fibrosis	McGinniss (2005)
57347	CFTR	A	C	AA	Cystic Fibrosis	Chillon (1994)
57348	CFTR	T	A	TT	Cystic Fibrosis	Zielenski (1993)
57349	CFTR	T	G	TT	Cystic Fibrosis	Mercier (1993)
57350	CFTR	A	T	AA	Cystic Fibrosis	Claustres (2004)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
57351	CFTR	G	T	GG	Cystic Fibrosis	Zielinski (1995)
57352	CFTR	T	G	TT	Cystic Fibrosis	Scotet (2003)
57353	CFTR	C	G	CC	Cystic Fibrosis	Zielenski (1999)
57354	CFTR	C	T	CC	Cystic Fibrosis	Ferec (1998)
57355	CFTR	G	C	GG	Cystic Fibrosis	Hughes (1996)
57356	CFTR	G	A	GG	Cystic Fibrosis	Bienvenu (1994)
57357	CFTR	A	T	AA	Cystic Fibrosis	Ghanem (1996)
57360	CFTR	A	G	AA	Cystic Fibrosis	Giorgi (1999)
57361	CFTR	G	A	GG	Cystic Fibrosis	Ferec (2000)
57362	CFTR	T	G	TT	Cystic Fibrosis	Hirtz (2004)
57363	CFTR	A	G	AA	Cystic Fibrosis	Zielenski (1993)
57364	CFTR	A	G	AA	Cystic Fibrosis	Teng (1994)
57365	CFTR	T	A	TT	Cystic Fibrosis	Ferec (1998)
57367	CFTR	C	T	CC	Cystic Fibrosis	Schwarz (1996)
57368	CFTR	G	A	GG	Cystic Fibrosis	Seia (2000)
57369	CFTR	G	A	GG	Cystic Fibrosis	Kilinc (2002)
57370	CFTR	C	A	CC	Cystic Fibrosis	Casals (2000)
57371	CFTR	G	C	GG	Cystic Fibrosis	Chillon (1995)
57374	CFTR	C	T	CC	Cystic Fibrosis	Ronchetto (1992)
57375	CFTR	T	C	TT	Cystic Fibrosis	Macek (1993)
57376	CFTR	C	T	CC	Cystic Fibrosis	Hirtz (2004)
57377	CFTR	C	G	CC	Cystic Fibrosis	Marechal (2001)
57378	CFTR	C	T	CC	Cystic Fibrosis	Gasparini (1991), Gasparini (1992), Faucz (2007)
57380	CFTR	T	G	TT	Cystic Fibrosis	Messaoud (2005)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
57381	CFTR	A	G	AA	Cystic Fibrosis	Macek (1993)
57382	CFTR	A	G	AA	Cystic Fibrosis	Baralle (1994)
57383	CFTR	A	T	AA	Cystic Fibrosis	Banjar (1999)
57384	CFTR	C	G	CC	Cystic Fibrosis	Wallace (1994)
57385	CFTR	C	T	CC	Cystic Fibrosis	Haworth (1996)
57386	CFTR	T	A	TT	Cystic Fibrosis	Glavac (1994)
57387	CFTR	A	C	AA	Cystic Fibrosis	Savov (2002)
57388	CFTR	C	G	CC	Cystic Fibrosis	Ivaschenko (1993)
57389	CFTR	A	G	AA	Cystic Fibrosis	Fanen (1992)
57390	CFTR	A	G	AA	Cystic Fibrosis	Cutting (1992)
57391	CFTR	G	A	GG	Cystic Fibrosis	Ghanem (1994)
57392	CFTR	C	G	CC	Cystic Fibrosis	Ferec (1996)
57393	CFTR	T	A	TT	Cystic Fibrosis	Seia (2004)
57394	CFTR	G	A	GG	Cystic Fibrosis	Nukiwa (1993)
57395	CFTR	G	A	GG	Cystic Fibrosis	Macek (1993)
57396	CFTR	T	G	TT	Cystic Fibrosis	Korytina (2003)
57397	CFTR	T	C	TT	Cystic Fibrosis	Jezequel (2000)
57399	CFTR	T	C	TT	Cystic Fibrosis	Claustres (1997)
57400	CFTR	A	G	AA	Cystic Fibrosis	Gasparini (1993)
57402	CFTR	G	A	GG	Cystic Fibrosis	Casals (2000)
57403	CFTR	A	G	AA	Cystic Fibrosis	Ferec (1993)
57404	CFTR	C	T	CC	Cystic Fibrosis	Audrezet (1993)
57405	CFTR	T	G	TT	Cystic Fibrosis	Zielenski (1999)
57406	CFTR	G	A	GG	Cystic Fibrosis	Devoto (1991)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
57407	CFTR	G	T	GG	Cystic Fibrosis	Savov (1995), Clain (2005)
57409	CFTR	G	A	GG	Cystic Fibrosis	Casals (1996)
57410	CFTR	G	A	GG	Cystic Fibrosis	Dijkstra (1994)
57411	CFTR	G	A	GG	Cystic Fibrosis	Greil (1994)
57412	CFTR	G	A	GG	Cystic Fibrosis	Kaelin (1992), Gasparini (1993)
57413	CFTR	A	C	AA	Cystic Fibrosis	Schrijver (2005)
57414	CFTR	T	A	TT	Cystic Fibrosis	Hughes (1996)
57415	CFTR	T	C	TT	Cystic Fibrosis	Lissens (1992), Gasparini (1993)
57416	CFTR	C	A	CC	Cystic Fibrosis	Cutting (1990)
57417	CFTR	C	T	CC	Cystic Fibrosis	Cartault (1998)
57420	CFTR	T	G	TT	Cystic Fibrosis	Seia (2002)
57422	CFTR	T	A	TT	Cystic Fibrosis	McDowell (1995)
57423	CFTR	G	A	GG	Cystic Fibrosis	Zielenski (1999)
57424	CFTR	C	T	CC	Cystic Fibrosis	Casals (1997)
57425	CFTR	T	C	TT	Cystic Fibrosis	Ivaschenko (1993)
57426	CFTR	T	G	TT	Cystic Fibrosis	Faucz (1996)
57427	CFTR	G	A	GG	Cystic Fibrosis	Vidaud (1990), Vidaud (1990), Hamosh (1991), Shoshani (1992), Shoshani (1994), Faucz (2007)
57428	CFTR	G	T	GG	Cystic Fibrosis	Ferec (1996)
57429	CFTR	G	A	GG	Cystic Fibrosis	Chevalier (1994)
57430	CFTR	G	T	GG	Cystic Fibrosis	Cheadle (1992)
57431	CFTR	T	C	TT	Cystic Fibrosis	Dorval (1993)
57434	CFTR	C	T	CC	Cystic Fibrosis	Feldmann (2001)
57435	CFTR	A	G	AA	Cystic Fibrosis	Doerk (1994)
57436	CFTR	G	C	GG	Cystic Fibrosis	Jones (1992)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
57437	CFTR	G	A	GG	Cystic Fibrosis	Ferec (1997)
57438	CFTR	C	T	CC	Cystic Fibrosis	Liechti (1999)
57439	CFTR	T	C	TT	Cystic Fibrosis	Poncin (1996)
57440	CFTR	A	C	AA	Cystic Fibrosis	Claustres (1992), Gasparini (1993)
57441	CFTR	A	T	AA	Cystic Fibrosis	Lissens (1995)
57442	CFTR	C	G	CC	Cystic Fibrosis	Osborne (1991), Osborne (1992), Osborne (1992), Faucz (2007)
57443	CFTR	T	A	TT	Cystic Fibrosis	Claustres (1997)
57445	CFTR	T	A	TT	Cystic Fibrosis	Zielenski (1999)
57446	CFTR	G	T	GG	Cystic Fibrosis	Ferec (1997)
57447	CFTR	G	A	GG	Cystic Fibrosis	Tsui (1992)
57448	CFTR	C	A	CC	Cystic Fibrosis	Malone (1996)
57449	CFTR	C	T	CC	Cystic Fibrosis	Audrezet (1993)
57450	CFTR	G	A	GG	Cystic Fibrosis	Cutting (1990)
57451	CFTR	T	C	TT	Cystic Fibrosis	Ferec (1998)
57452	CFTR	G	C	GG	Cystic Fibrosis	Ferec (1994)
57453	CFTR	T	C	TT	Cystic Fibrosis	Bienvenu (2005)
57454	CFTR	T	C	TT	Cystic Fibrosis	Zielenski (1997)
57457	CFTR	G	A	GG	Cystic Fibrosis	Tsui (1992)
57458	CFTR	G	A	GG	Cystic Fibrosis	Yoshimura (1999)
57461	CFTR	A	T	AA	Cystic Fibrosis	Ferec (1999)
57463	CFTR	T	C	TT	Cystic Fibrosis	Ferec (1999)
57465	CFTR	G	T	GG	Cystic Fibrosis	Cutting (1992)
57467	CFTR	C	T	CC	Cystic Fibrosis	Schrijver (2005)
57469	CFTR	C	A	CC	Cystic Fibrosis	Casals (2002)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
57470	CFTR	C	T	CC	Cystic Fibrosis	Claustres (1998)
57472	CFTR	T	A	TT	Cystic Fibrosis	Petreska (1994)
57473	CFTR	A	C	AA	Cystic Fibrosis	Valaskova (2002)
57474	CFTR	G	T	GG	Cystic Fibrosis	Strandvik (2001)
57476	CFTR	A	T	AA	Cystic Fibrosis	Claustres (1993)
57479	CFTR	C	T	CC	Cystic Fibrosis	Hughes (1996)
57480	CFTR	G	T	GG	Cystic Fibrosis	Girodon (2004)
57481	CFTR	C	T	CC	Cystic Fibrosis	Claustres (1997)
57483	CFTR	T	C	TT	Cystic Fibrosis	Ferec (1999)
57484	CFTR	C	T	CC	Cystic Fibrosis	Schrijver (2005)
57485	CFTR	G	A	GG	Cystic Fibrosis	Schrijver (2005)
57488	CFTR	C	T	CC	Cystic Fibrosis	Claustres (1999)
57489	CFTR	A	G	AA	Cystic Fibrosis	Bienvenu (2003)
57492	CFTR	G	T	GG	Cystic Fibrosis	Romey (1999)
57493	CFTR	C	T	CC	Cystic Fibrosis	Strandvik (2001)
57498	CFTR	A	T	AA	Cystic Fibrosis	Culard (1994)
57499	CFTR	G	T	GG	Cystic Fibrosis	Bienvenu (2005)
57500	CFTR	A	G	AA	Cystic Fibrosis	Strandvik (2001)
57501	CFTR	G	A	GG	Cystic Fibrosis	Savor (1994)
57502	CFTR	G	A	GG	Cystic Fibrosis	Kerem (1990), Guillermit (1990), American College of Medical Genetics and Genomics (2004), Center for Pediatric Genomic Medicine of Children's Mercy Hospital and Clinics (2015), ARUP Laboratories (2016), GeneDx (2016), Counsyl (2016), Invitae (2017), GeneDx (2017)
57503	CFTR	T	A	TT	Cystic Fibrosis	Vouk (1999)
57507	CFTR	G	A	GG	Cystic Fibrosis	Jordanova (1996)
57509	CFTR	T	C	TT	Cystic Fibrosis	Claustres (1996)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
57510	CFTR	G	A	GG	Cystic Fibrosis	Chillon (1994)
57515	CFTR	G	A	GG	Cystic Fibrosis	Schrijver (2005)
57517	CFTR	G	C	GG	Cystic Fibrosis	Petreska (1996)
57518	CFTR	T	C	TT	Cystic Fibrosis	Kambouris (2000)
57519	CFTR	A	C	AA	Cystic Fibrosis	Mercier (1995)
57520	CFTR	A	G	AA	Cystic Fibrosis	Casals (1997)
57522	CFTR	G	A	GG	Cystic Fibrosis	Strong (1992)
57523	CFTR	G	A	GG	Cystic Fibrosis	Georges (2004)
57524	CFTR	G	C	GG	Cystic Fibrosis	Cuppens (1993)
57526	CFTR	G	T	GG	Cystic Fibrosis	Crawford (1995)
57527	CFTR	G	T	GG	Cystic Fibrosis	Zielenski (1995)
57529	CFTR	A	G	AA	Cystic Fibrosis	Scotet (2003)
57530	CFTR	G	A	GG	Cystic Fibrosis	Audrezet (1993), Audrezet (1993)
57531	CFTR	G	T	GG	Cystic Fibrosis	Scotet (2003)
57535	CFTR	G	C	GG	Cystic Fibrosis	Dubourg (1999)
57536	CFTR	G	T	GG	Cystic Fibrosis	Ferec (1995)
57540	CFTR	T	A	TT	Cystic Fibrosis	Tzetis (1997)
57542	CFTR	A	G	AA	Cystic Fibrosis	Marigo (1995)
57544	CFTR	G	A	GG	Cystic Fibrosis	Patrinos (2005)
57545	CFTR	G	C	GG	Cystic Fibrosis	Schwartz (1993)
57546	CFTR	G	T	GG	Cystic Fibrosis	Bienvenu (1994)
57547	CFTR	G	T	GG	Cystic Fibrosis	Groman (2002)
57549	CFTR	G	A	GG	Cystic Fibrosis	Highsmith (1990)
57551	CFTR	G	A	GG	Cystic Fibrosis	Malone (1996)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
57553	CFTR	T	C	TT	Cystic Fibrosis	Poncin (1996)
57555	CFTR	A	G	AA	Cystic Fibrosis	Macek (1997)
57556	CFTR	A	T	AA	Cystic Fibrosis	Ferec (1995)
57557	CFTR	C	G	CC	Cystic Fibrosis	Bienvenu (2005)
57558	CFTR	G	A	GG	Cystic Fibrosis	Guillermit (1990)
57559	CFTR	G	A	GG	Cystic Fibrosis	Zielenski (1994)
57560	CFTR	G	A	GG	Cystic Fibrosis	Macek (1997), Dork (1998)
57561	CFTR	A	G	AA	Cystic Fibrosis	Fanen (1992)
57562	CFTR	A	G	AA	Cystic Fibrosis	Girodon (2003)
57563	CFTR	A	G	AA	Cystic Fibrosis	Kanavakis (1995)
57564	CFTR	A	G	AA	Cystic Fibrosis	Reiss (1993)
57567	CFTR	A	G	AA	Cystic Fibrosis	Schrijver (2005)
57568	CFTR	G	A	GG	Cystic Fibrosis	Mercier (1993)
57569	CFTR	G	A	GG	Cystic Fibrosis	Mercier (1994)
57570	CFTR	A	G	AA	Cystic Fibrosis	Schrijver (2005)
57571	CFTR	A	T	AA	Cystic Fibrosis	Wong (2001)
57575	CFTR	T	C	TT	Cystic Fibrosis	Creegan (1994)
57576	CFTR	T	G	TT	Cystic Fibrosis	Ferec (2002)
57577	CFTR	A	G	AA	Cystic Fibrosis	Doerk (1993)
57581	CFTR	G	A	GG	Variant of Unknown Significance (questionable association with Cystic Fibrosis)	Zielenski (1994)
57582	CFTR	G	A	GG	Cystic Fibrosis	Steffann (1998)
57584	CFTR	G	A	GG	Cystic Fibrosis	Audrezet (1993)
57585	CFTR	T	G	TT	Cystic Fibrosis	Doerk (1993)
57586	CFTR	A	G	AA	Cystic Fibrosis	Ronchetto (1992)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
57587	CFTR	A	G	AA	Cystic Fibrosis	Ferec (2002)
57589	CFTR	G	A	GG	Cystic Fibrosis	Cutting (1992)
57590	CFTR	G	A	GG	Cystic Fibrosis	Greil (1993)
57591	CFTR	G	A	GG	Cystic Fibrosis	Kilinc (2002)
57593	CFTR	A	G	AA	Cystic Fibrosis	Visich (2002)
57594	CFTR	C	A	CC	Cystic Fibrosis	Strandvik (2001)
57595	CFTR	C	T	CC	Cystic Fibrosis	Bienvenu (1994)
57596	CFTR	G	A	GG	Cystic Fibrosis	Orozco (2000)
57600	CFTR	G	A	GG	Cystic Fibrosis	Wagner (2002)
57601	CFTR	G	C	GG	Cystic Fibrosis	Tzetis (1997)
57602	CFTR	G	T	GG	Cystic Fibrosis	Walker (2000)
57603	CFTR	T	A	TT	Cystic Fibrosis	Alper (2004)
57604	CFTR	T	C	TT	Cystic Fibrosis	Ferec (1994)
57605	CFTR	T	C	TT	Cystic Fibrosis	Malone (1998)
57608	CFTR	G	A	GG	Cystic Fibrosis	Visich (2002)
57612	CFTR	G	A	GG	Cystic Fibrosis	Ferec (1992)
57613	CFTR	G	C	GG	Cystic Fibrosis	Jones (1992)
57614	CFTR	T	C	TT	Cystic Fibrosis	Boman (1997)
57616	CFTR	G	A	GG	Cystic Fibrosis	Desgeorges (1997)
57617	CFTR	G	A	GG	Cystic Fibrosis	Mittre (1999)
57618	CFTR	T	A	TT	Cystic Fibrosis	Bieth (2003)
57619	CFTR	G	A	GG	Cystic Fibrosis	Feuillet-Fieux (2004)
57620	CFTR	A	C	AA	Cystic Fibrosis	Tang (2002)
57623	CFTR	G	C	GG	Cystic Fibrosis	Chevalier-Porst (1999)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
57625	CFTR	G	A	GG	Cystic Fibrosis	Fanen (1992)
57626	CFTR	G	T	GG	Cystic Fibrosis	Doerk (1993)
57628	CFTR	A	C	AA	Cystic Fibrosis	Costes (1994)
57629	CFTR	A	G	AA	Cystic Fibrosis	El-Harith (1997)
57631	CFTR	G	A	GG	Cystic Fibrosis	Orozco (2000)
57632	CFTR	G	C	GG	Cystic Fibrosis	Bonizatto (1992)
57633	CFTR	G	T	GG	Cystic Fibrosis	Bienvenu (1995)
57635	CFTR	T	C	TT	Cystic Fibrosis	Kilinc (2002)
57636	CFTR	A	C	AA	Cystic Fibrosis	Macek (1997)
57637	CFTR	A	G	AA	Cystic Fibrosis	Ghanem (1994)
57638	CFTR	G	A	GG	Cystic Fibrosis	Doerk (1993)
57639	CFTR	A	C	AA	Cystic Fibrosis	Cuppens (1993)
57640	CFTR	A	G	AA	Cystic Fibrosis	Ramirez (2006)
57644	CFTR	G	A	GG	Cystic Fibrosis	Macek (1997)
57645	CFTR	G	T	GG	Cystic Fibrosis	Zielenski (1991)
57646	CFTR	T	C	TT	Cystic Fibrosis	Malone (1998)
57647	CFTR	T	G	TT	Cystic Fibrosis	Claustres (1993)
57648	CFTR	G	T	GG	Cystic Fibrosis	Casals (1997)
57649	CFTR	A	C	AA	Cystic Fibrosis	Macek (1994)
57650	CFTR	A	G	AA	Cystic Fibrosis	Petreska (1994)
57652	CFTR	A	T	AA	Cystic Fibrosis	Casals (1997)
57653	CFTR	G	A	GG	Cystic Fibrosis	Banjar (1999)
57654	CFTR	G	A	GG	Cystic Fibrosis	Bisceglia (1994)
57655	CFTR	G	T	GG	Cystic Fibrosis	Zielenski (1991)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
57657	CFTR	G	A	GG	Cystic Fibrosis	Casals (1997)
57658	CFTR	G	C	GG	Cystic Fibrosis	Zielenski (1993)
57659	CFTR	T	G	TT	Cystic Fibrosis	Macek (1997)
57662	CFTR	G	A	GG	Cystic Fibrosis	Kambouris (2000)
57663	CFTR	G	C	GG	Cystic Fibrosis	Marechal (2001)
57664	CFTR	A	C	AA	Cystic Fibrosis	Doerk (1993)
57666	CFTR	G	A	GG	Cystic Fibrosis	Telleria (1999)
57667	CFTR	G	A	GG	Cystic Fibrosis	Doerk (1996)
57669	CFTR	A	G	AA	Cystic Fibrosis	Marechal (2001)
57672	CFTR	G	A	GG	Cystic Fibrosis	Seki (1999)
57673	CFTR	G	A	GG	Cystic Fibrosis	Doerk (1993)
57674	CFTR	G	A	GG	Cystic Fibrosis	Aulehla-Scholz (2001)
6319	CTH	C	T	CC	Possibly associated with Cystathioninuria	Wang (2003), Espinós (2010), Illumina Clinical Services Laboratory (2016), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2018)
67297	INSL3	G	A	GG	Cryptorchidism	Marin (2001), Ferlin (2003)
34911	FGFR3	C	A	CC	Crouzon Syndrome with Acanthosis Nigricans|Crouzonodermoskeletal Syndrome	Reddy , (1985), Breitbart , (1989), Breitbart (1989), Orlow, (1992), Jabs (1994), Meyers (1995), Meyers (1995), Meyers (1995), Meyers (1995), Meyers (1995), Meyers (1995), Superti-Furga , (1996), Superti-Furga , (1996), Cohen (1999), Cohen (1999), Schweitzer (2001), Schweitzer (2001)
65058	FGFR2	A	C,G	AA	Crouzon syndrome	Park (1995)
65080	FGFR2	C	A,G,T	CC	Crouzon syndrome	Reardon (1994), Rutland (1995), Steinberger (1995)
65097	FGFR2	T	C	TT	Crouzon syndrome	Ravel (2005), McGillivray (2005)
15027	UGT1A1	C	T	CC	Crigler-Najjar Syndrome, Type 1|Gilbert Syndrome	Ritter , (1992), Moghrabi , (1993), Moghrabi (1993), Mackenzie , (1997), Maruo (2003)
15000	UGT1A1	T	G	TT	Crigler-Najjar syndrome 2	Seppen (1996), Emi (2003)
15029	UGT1A1	A	G	AA	Crigler-Najjar syndrome 1	Francoual (2002), Francoual (2002)
37164	PRNP	G	A	GG	Creutzfeldt-Jakob Syndrome	Goldgaber (1989), Goldfarb (1990), Goldfarb (1990), Mitrova (1990)
37170	PRNP	G	A	GG	Creutzfeldt-Jakob Syndrome	Meggendorfer, (1930), Goldfarb (1991), Nieto (1991), Haltia , (1991), Goldfarb (1992), Brown (1992), Brown (1992), Brown (1992), Laplanche (1992), Kretzschmar (1995), Riek (1998)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
34934	FGFR3	C	G	CC	Craniosynostosis|Muenke Syndrome	Bellus (1996), Muenke (1997), Moloney (1997)
35026	ANKH	C	T	CC	Craniometaphyseal Dysplasia	Nurnberg (2001)
100832	ANKH	I	D	II	Craniometaphyseal Dysplasia	Nurnberg (2001), Reichenberger (2001)
100833	ANKH	I	D	II	Craniometaphyseal Dysplasia	Nurnberg (2001), Reichenberger (2001)
100834	ANKH	I	D	II	Craniometaphyseal Dysplasia	Reichenberger (2001)
64651	EFNB1	G	A	G	Craniofrontonasal Syndrome	Twigg (2004), Wieland (2005), Twigg (2006), Wallis (2009)
33412	CPT2	C	T	CC	CPT2 Deficiency, inducable with anesthesia	Vladutiu (1998, 2000)
33394	CPT2	A	G	AA	CPT2 Deficiency	Orngreen (2005)
33364	CPT2	C	A	CC	CPT2 Deficiency	Verderio (1995), Vladutiu (2002)
33385	CPT2	C	T	CC	CPT2 Deficiency	Taroni (1993), DiDonato (1978), Bonnefont (1996), Handig (1996), Martin (1999)
33419	CPT2	C	T	CC	CPT2 Deficiency	Taroni (1992), Taroni (1993)
33414	CPT2	G	A	GG	CPT2 Deficiency	Taggart (1999)
100583	CPT2	I	D	II	CPT2 Deficiency	Taggart (1999), Elpeleg (2001), Vladutiu (2002)
100587	CPT2	D	I	DD	CPT2 Deficiency	Albers (2001), Vladutiu (2002), Elpeleg (2001)
801011	CPT2	D	I	DD	CPT2 Deficiency	Albers (2001), Vladutiu (2002), Elpeleg (2001)
30865	CPT1A	T	C	TT	CPT1A Deficiency	Yamamoto (2000), Ogawa (2002), Prip-Buus (2001)
30875	CPT1A	C	T	CC	CPT1A Deficiency	Prip-Buus (2001)
31152	CPOX	G	A	GG	Cpox Deficiency	Lamoril (2001), Wiman (2002)
31154	CPOX	T	C	TT	Cpox Deficiency	Lamoril (1998)
46687	PTEN	C	T	CC	Cowden Disease|Bannayan-Riley-Ruvalcaba Syndrome|Macrocephaly/Autism Syndrome	Nelen (1997), Zori (1998), Zori (1998), Herman (2007), Herman (2007)
46717	PTEN	C	T	CC	Cowden Disease|Bannayan-Riley-Ruvalcaba Syndrome	Gorlin (1992), Gorlin (1992), Liaw (1997), Marsh (1997)
62859	HRAS	C	A,G,T	CC	Costello Syndrome	Aoki (2005), Kerr (2006)
62863	HRAS	C	A,G,T	CC	Costello Syndrome	Aoki (2005)
62864	HRAS	T	C	TT	Costello Syndrome	Kerr (2006), Hoornaert (2006)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
14236	SERPINA6	C	T	C	Corticosteroid-binding Globulin Deficiency	Emptoz-Bonneton (2000)
14248	SERPINA6	C	T	C	Corticosteroid-binding Globulin Deficiency	Torpy (2001)
1066	MEF2A	G	A	GG	Possibly associated with Coronary Artery Disease and Myocardial Infarction	Bhagavatula (2004)
34273	FBN2	A	C	AA	Contractural Arachnodactyly, Congenital	Belleh (2000), Belleh (2000), Belleh (2000)
34295	FBN2	C	G	CC	Contractural Arachnodactyly, Congenital	Babcock (1998)
34307	FBN2	A	C	AA	Contractural Arachnodactyly, Congenital	Wang , (1995), Maslen (1997), Maslen (1997), Maslen (1997)
38654	PLP1	C	T	C	Connatal Pelizaeus-Merzbacher Disease	Lazzarini (1996), Yamamoto (1998)
52743	PLG	A	G	AA	Ligneous Conjunctivitis due to Plasminogen Deficiency, Type I	Schuster (1999), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2015), Knight Diagnostic Laboratories of Oregon Health and Sciences University (2016)
52747	PLG	G	A	GG	Ligneous Conjunctivitis due to Plasminogen Deficiency, Type I	Schuster (1997), Schuster (1999)
52755	PLG	G	A	GG	Ligneous Conjunctivitis due to Plasminogen Deficiency, Type I	Schuster (1997)
53947	GNAT1	G	A	GG	Congenital Stationary Nightblindness	Dryja (1996)
42064	CHRNB1	C	A	CC	Congenital Slow Channel Myasthenic Syndrome	Gomez (1996)
42065	CHRNB1	G	A	GG	Congenital Slow Channel Myasthenic Syndrome	Engel (1996)
38471	CHRNE	G	A,T	GG	Congenital Slow Channel Myasthenic Syndrome	Croxen (2002)
38479	CHRNE	G	A	GG	Congenital Slow Channel Myasthenic Syndrome	Uchitel (1993), Ohno (1996)
38494	CHRNE	G	A	GG	Congenital Slow Channel Myasthenic Syndrome	Gammack (1995), Engel (1996)
41975	CHRNA1	C	T	CC	Congenital Slow Channel Myasthenic Syndrome	Bady (1994), Sine (1995), Sine (1995), Croxen (1997)
1812	SCN5A	C	T	CC	Congenital Sick Sinus Syndrome	Kyndt (2001), Benson (2003)
1814	SCN5A	G	A	GG	Congenital Sick Sinus Syndrome	Benson (2003)
65682	NPHS1	G	A	GG	Congenital Nephrosis 1, Finnish Type	Kestila (1998), Koziell (2002)
100923	NPHS1	I	D	II	Congenital Nephrosis 1, Finnish Type	Kestila (1998)
42003	RAPSN	T	C	TT	Congenital Myasthenic Syndrome associated with Acetylcholine Receptor Deficiency	Goldhammer (1990), Ohno (2003), Ohno (2003)
42008	RAPSN	G	T	GG	Congenital Myasthenic Syndrome associated with Acetylcholine Receptor Deficiency	Ohno (2002), Maselli (2003), Muller (2003), Burke (2003), Muller , (2004), Muller (2004)
42046	CHRND	G	A	GG	Congenital myasthenic syndrome	Muller (2006)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
41996	RAPSN	G	T	GG	Congenital Myasthenic Syndrome	Muller (2003)
38357	FKRP	C	A	CC	Congenital Muscular Dystrophy, Type 1C, with Neurologic Abnormalities	Topaloglu (2003), Louhichi (2004), Louhichi (2004)
41154	LAMA2	C	A	CC	Congenital Merosin Deficient Muscular Dystrophy	Guicheney , (1998), Di Blasi (2005)
11953	STAR	C	A,T	CC	Congenital lipoid adrenal hyperplasia	Bose (1996), Bose (2000)
11961	STAR	G	A	GG	Congenital lipoid adrenal hyperplasia	Bose (1996), Bose (1996), Bose (2000)
100893	NTRK1	I	D	II	Congenital Insensitivity to Pain Syndrome	Indo (1996), Indo (1996), Miura (2000), Miura (2000)
6579	ZIC3	A	G	A	Congenital Heart Disease Heterotaxy	Ware (2004)
6338	GATA4	T	C	TT	Congenital heart defects	Reamon-Buettner (2005)
6348	GATA4	T	C	TT	Congenital heart defects	Reamon-Buettner (2005)
38501	CHRNE	C	G	CC	Congenital Fast-channel Myasthenic Syndrome	Wang (2000), Wang (2000)
42043	CHRND	C	A	CC	Congenital Fast Channel Myasthenic Syndrome	Shen (2002)
8635	UROS	A	C,G	AA	Congenital Erythropoietic Porphyria	Deybach (1990), Warner (1990), Warner (1992), Boulechfar (1992), Tanigawa (1995), Frank (1998)
8647	UROS	G	A	GG	Congenital Erythropoietic Porphyria	Warner (1992), Warner (1992), Boulechfar (1992)
32194	ALG1	C	G	CC	Congenital disorders of glycosylation type 1K	Grubenmann (2004)
32195	ALG1	C	T	CC	Congenital disorders of glycosylation type 1K	Schwarz (2004), Kranz (2004)
32632	ALG12	C	T	CC	Congenital disorders of glycosylation type 1G	Grubenmann (2002)
32636	ALG12	G	A	GG	Congenital disorders of glycosylation type 1G	Eklund (2005)
32206	ALG3	A	G	AA	Congenital disorders of glycosylation type 1D	Schollen (2002)
32207	ALG3	C	T	CC	Congenital disorders of glycosylation type 1D	Stibler (1995), Korner (1999)
32208	ALG3	G	A	GG	Congenital disorders of glycosylation type 1D	Schollen (2005)
75056	MGAT2	A	G	AA	Congenital Disorder of Glycosylation, Type IIa	Tan (1996)
75057	MGAT2	A	G	AA	Congenital Disorder of Glycosylation, Type IIa	Cormier-Daire (2000)
75055	MGAT2	C	T	CC	Congenital Disorder of Glycosylation, Type IIa	Tan (1996)
75058	MGAT2	T	A	TT	Congenital Disorder of Glycosylation, Type IIa	Cormier-Daire (2000)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
31175	MPI	A	G	AA	Congenital Disorder of Glycosylation, Type Ib	Lonlay (1999)
31167	MPI	C	T	CC	Congenital Disorder of Glycosylation, Type Ib	Jaeken (1998)
31172	MPI	G	A	GG	Congenital Disorder of Glycosylation, Type Ib	Schollen (2000)
31173	MPI	G	A	GG	Congenital Disorder of Glycosylation, Type Ib	Niehues (1998), Schollen (2000)
31174	MPI	G	A	GG	Congenital Disorder of Glycosylation, Type Ib	Schollen (2000)
31178	MPI	G	A	GG	Congenital Disorder of Glycosylation, Type Ib	Babovic-Vuksanovic (1999)
31166	MPI	T	C	TT	Congenital Disorder of Glycosylation, Type Ib	Schollen (2000)
31170	MPI	T	C	TT	Congenital Disorder of Glycosylation, Type Ib	Jaeken (1998)
31171	MPI	T	C	TT	Congenital Disorder of Glycosylation, Type Ib	Westphal (2001)
31177	MPI	T	C	TT	Congenital Disorder of Glycosylation, Type Ib	Lonlay (1999)
31168	MPI	A	G	AA	Congenital Disorder of Glycosylation, Type Ib	Schollen (2002)
30559	PMM2	C	A	CC	Congenital Disorder of Glycosylation, Type Ia	Matthijs (1998), Kjaergaard (1998)
30545	PMM2	G	T	GG	Congenital Disorder of Glycosylation, Type Ia	Matthijs (1998)
30571	PMM2	G	A	GG	Congenital Disorder of Glycosylation, Type Ia	Matthijs (1997), Kjaergaard (1998), Matthijs (1999), Schollen (2000), Vuillaumier-Barrot (2000), Bohles (2001)
30611	PMM2	G	C	GG	Congenital Disorder of Glycosylation, Type Ia	Matthijs (1999), Vuillaumier-Barrot (2000), Grunewald (2001)
30541	PMM2	T	C	TT	Congenital Disorder of Glycosylation, Type Ia	Matthijs (1998)
32153	SLC35C1	C	T	CC	Congenital Disorder of Glycosylatio, Type IIc	Luhn (2001)
32154	SLC35C1	C	G	CC	Congenital Disorder of Glycosylatio, Type IIc	Lubke (2001), Etzioni (2002)
78781	GJA3	C	A	CC	Congenital Cataract	Guleria (2007)
22494	FGA	C	A,G	CC	Congenital Afibrinogenemia|Congenital Hypofibrinogenemia	Neerman-Arbez (2000), Lefebvre (2004)
10679	CYP11B1	T	C	TT	Congenital Adrenal Hyperplasia due to Steroid-11 Beta-hydroxylase Deficiency	Chabre (2000)
52299	GUCY2D	C	T	CC	Cone-rod Dystrophy 6	Kelsell (1998)
51927	ABCA4	C	A	CC	Cone-rod Dystrophy 3 and Retinitis Pigmentosa 19	Cremers (1998), Cremers (1998)
52300	GUCY2D	C	T	CC	Cone-rod dystrophy	Perrault (1998)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
52302	GUCY2D	G	C	GG	Cone-rod Dystrophy	
64203	BCS1L	C	T	CC	Complex III deficiency	de Lonlay (2001)
42197	NDUFS3	C	T	CC	Complex 1 Deficiency	Benit (2004)
42198	NDUFS3	C	T	CC	Complex 1 Deficiency	Benit (2004)
65530	APRT	T	A	TT	Complete Adenine Phosphoribosyltransferase Deficiency, Icelandic Type	Chen (1990)
23640	C5	G	A	GG	Complement C5 Deficiency	Wang (1995)
23641	C5	G	A	GG	Complement C5 Deficiency	Wang (1995)
23842	C3	C	T	CC	Complement C3 Deficiency	Singer (1994)
23844	C3	G	A	GG	Complement C3 Deficiency	Reis (2004)
23667	C2	G	A	GG	Complement C2 Deficiency, Type II	Wetsel (1996)
32981	PSAP	T	A	TT	Combined Saposin Deficiency	Harzer (1989), Schnabel (1992), Paton (1992), Bradova (1993), Potier (1990)
32984	PSAP	G	A	GG	Combined Saposin Deficiency	Wenger (1989), Rafi (1990)
30806	GFM1	C	T	CC	Combined Oxidative Phosphorylation Deficiency 1	Valente (2007)
30809	GFM1	T	G	TT	Combined Oxidative Phosphorylation Deficiency 1	Valente (2007)
54912	VKORC1	G	A	GG	Combined Deficiency of Vitamin K-dependent Clotting Factors, Type 2	Oldenburg , (2000), Rost (2004), Rost (2004)
50173	OGG1	G	A	GG	Colorectal cancer, association with ?	Kim (2004)
15966	MUTYH	C	A	CC	Colorectal cancer	Eliason (2005)
15978	MUTYH	G	A	GG	Colorectal cancer	Eliason (2005)
47401	TP53	C	A,G,T	CC	Colorectal cancer	Achatz (2007)
47432	TP53	G	A	GG	Colorectal cancer	Tsuchiya (1997)
52475	OPN1MW	A	C	A	Colorblindness, Deutan	Ueyama (2002)
47397	TP53	G	A	GG	Colon Tumors, Concurrent Multiple Primary	Miyaki (2003)
15801	ABCB1	C	A	CC	Colchicine Resistance	Choi , (1988), Safa (1990)
68596	VPS13B	I	D	II	Cohen Syndrome	Kolehmainen (2003)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
71995	RUNX2	G	A	GG	Cleidocranial Dysplasia	Quack (1999)
26416	TBX22	A	T	A	Cleft Palate and Ankyloglossia	Marcano (2004)
26441	TBX22	G	C	G	Cleft Palate and Ankyloglossia	Braybrook (2001)
14725	PVRL1	C	T	CC	Cleft Lip/Palate-Ectodermal Dysplasia Syndrome	Suzuki (2000), Suzuki (2000), Sozen (2001)
26460	MSX1	A	T	AA	Cleft Lip/Palate, Nonsyndromic	Jezewski (2003)
26463	MSX1	G	A	GG	Cleft Lip/Palate, Nonsyndromic	Jezewski (2003)
26470	MSX1	C	A	CC	Cleft lip and palate	Suzuki (2004), Vieira (2005), Tongkobpetch (2006), Tongkobpetch (2006)
58447	DNAI1	G	A	GG	Ciliary dyskinesia, primary	Zariwala (2006)
78559	TXNDC3	T	A	TT	Ciliary Dyskinesia, Primary	Duriez (2007)
16138	SAR1B	C	T	CC	Chylomicron Retention Disease	Jones (2003)
44249	GABRB3	C	T	CC	Chronic Insomnia	Laposky , (2001), Buhr (2002), Buhr (2002)
22865	CYBB	C	A	C	Chronic Granulomatous Disease, Cytochrome b-positive, Chronic	Stasia (2002), Stasia (2002), Stasia (2002), Bionda (2004), Bionda (2004)
22833	CYBA	C	T	CC	Chronic Granulomatous Disease, Cytochrome b-Negative	Yamada (2000), Yamada (2000)
23032	CYBB	T	C	T	Chronic Granulomatous Disease, Chronic	Ezekowitz (1988), Ezekowitz (1988), Ezekowitz (1988), Rae (1998), Newburger (2000)
49182	PRF1	A	G	AA	Chronic Active Epstein-Barr Virus Infection	Katano (2004)
49203	PRF1	C	G	CC	Chronic Active Epstein-Barr Virus Infection	Katano (2004)
77944	CHM	G	A	G	Choroideremia	van Bokhoven (1994)
77954	CHM	G	T	G	Choroideremia	Schwartz (1993), van Bokhoven (1994)
38253	VPS13A	C	T	CC	Choreoacanthocytosis	Rampoldi (2001), Dobson-Stone (2002)
64997	ARSE	C	T	C	Chondrodysplasia Punctata 1	Brunetti-Pierri (2003)
34687	EBP	C	T	C	Chondrodysplasia Punctata	Derry (1999), Braverman (1999)
34671	EBP	G	A	G	Chondrodysplasia Punctata	Braverman (1999), Aughton (2003)
35027	ANKH	G	T	GG	Chondrocalcinosis 2	Williams (2003)
5208	LIPA	A	G	AA	Cholesterol Ester Storage Disease	Illingworth (1993)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
35169	SH3BP2	G	A	GG	Cherubism	Ueki (2001)
63190	LYST	G	A,C	GG	Chediak-Higashi Syndrome, Childhood Type	Barbosa (1997)
63192	LYST	G	A	GG	Chediak-Higashi Syndrome, Childhood Type	Barbosa (1997)
36367	CHD7	C	T	CC	CHARGE Syndrome	Lalani (2006)
36428	CHD7	G	A	GG	CHARGE Syndrome	Lalani (2006)
36396	CHD7	T	G	TT	CHARGE Syndrome	Vissers (2004)
79111	FGD4	C	T	CC	Charcot-Marie-Tooth, Type 4H	Stendel (2007)
79113	FGD4	G	T	GG	Charcot-Marie-Tooth, Type 4H	Stendel (2007)
79112	FGD4	T	G	TT	Charcot-Marie-Tooth, Type 4H	Stendel (2007), Delague (2007)
78926	KIAA0274	C	T	CC	Charcot–Marie–Tooth Disorder, Type 4J	Chow (2007)
78925	KIAA0274	T	C	TT	Charcot–Marie–Tooth Disorder, Type 4J	Chow (2007)
43689	PRX	G	A	GG	Charcot-Marie-Tooth Disease, Type 4F	Kijima (2004), Kijima (2004), Otagiri (2006)
101088	SH3TC2	G	A	GG	Charcot-Marie-Tooth Disease, Type 4C	Senderek (2003), Azzedine (2006), Gosselin (2008), Houlden (2009), Azzedine (2010)
101223	SH3TC2	G	A	GG	Charcot-Marie-Tooth Disease, Type 4C	Gooding (2005), Claramunt (2007)
101225	SH3TC2	C	T	CC	Charcot-Marie-Tooth Disease, Type 4C	Senderek (2003), GeneReviews (2008), Athena Diagnostics Inc (2013)
101226	SH3TC2	I	D	II	Charcot-Marie-Tooth Disease, Type 4C	Senderek (2003)
43520	MPZ	G	A,T	GG	Charcot-Marie-Tooth Disease, Type 2J	Schiavon (1998), De Jonghe (1999), De Jonghe (1999), Chapon (1999), De Jonghe (1999), Misu (2000), Senderek (2000), Misu (2000), Baloh (2004), Baloh (2004), Triggs (2006)
802228	MPZ	G	A,T	GG	Charcot-Marie-Tooth Disease, Type 1B	Schiavon (1998), De Jonghe (1999), De Jonghe (1999), Chapon (1999), De Jonghe (1999), Misu (2000), Senderek (2000), Misu (2000), Baloh (2004), Baloh (2004), Triggs (2006)
37919	PMP22	G	A	GG	Variant of Unknown Significance (possibly associated with Charcot-Marie-Tooth Disease, Type 1A)	Roa (1993), Bathke (1996), Nelis (1997), Shy (2006), GeneReviews (2012), Genetic Services Laboratory of the University of Chicago (2015), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2015), Center for Pediatric Genomic Medicine of Children's Mercy Hospital and Clinics (2016), Praxis fuer Humangenetik Tuebingen (2016), Illumina Clinical Services Laboratory (2016), Invitae (2017), GeneDx (2017), ARUP Laboratories (2017)
2234	LMNA	C	T	CC	Charcot-Marie-Tooth Disease, Axonal, Type 2B1	De Sandre-Giovannoli (2002)
79119	PRPS1	A	C	A	Charcot-Marie-Tooth Disease, 5	Kim (2007)
79120	PRPS1	T	C	T	Charcot-Marie-Tooth Disease, 5	Young (2001)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
43502	MPZ	C	G	CC	Charcot-Marie-Tooth disease 1b	Hayasaka (1993), Rouger (1996), Rouger (1996)
63439	ABHD5	A	C	AA	Chanarin-Dorfman Syndrome	Lefevre (2001)
63443	ABHD5	G	A	GG	Chanarin-Dorfman Syndrome	Lefevre (2001)
65410	BRAF	T	C	TT	CFC Syndrome	Niihori (2006)
65413	BRAF	C	A,G,T	CC	CFC Syndrome	Niihori (2006)
65417	BRAF	C	G,T	CC	CFC Syndrome	Niihori (2006), Razzaque (2007)
53820	MAF	T	C	TT	Cerulean Cataract, Congenital	Vanita (2006)
71917	CLN6	I	D	II	Neuronal Ceroid Lipofuscinoses 6	Wheeler (2002), Teixeira (2003)
33467	CYP27A1	G	A	GG	Cerebrotendinous Xanthomatosis	Chen (1997)
100829	CYP27A1	G	A	GG	Cerebrotendinous Xanthomatosis	Leitersdorf (1993)
100828	CYP27A1	I	D	II	Cerebrotendinous Xanthomatosis	Leitersdorf (1993)
39784	CST3	A	T	AA	Cerebroarterial Amyloidosis, Icelandic-type	Abrahamson (1987), Palsdottir (1988), Grubb (1994), Grubb (1994)
39057	CCM2	C	T	CC	Cerebral cavernous malformations	Denier (2004)
41536	NOTCH3	G	A	GG	Cerebral Arteriopathy with Subcortical Infarcts and Leukoencephalopathy	Joutel (1996), Joutel (2000), Joutel (2000)
41537	NOTCH3	G	A	GG	Cerebral Arteriopathy with Subcortical Infarcts and Leukoencephalopathy	Joutel (1996), Joutel (1997)
41571	NOTCH3	G	A,T	GG	Cerebral Arteriopathy with Subcortical Infarcts and Leukoencephalopathy	Joutel (1997), Mykkanen (2004)
37944	ATCAY	G	T	GG	Cerebellar Ataxia, Cayman Type	Bomar (2003)
72826	MTTE	T	C	?	Myopathy, Mitochondrial, with Diabetes Mellitus	Hudgson (1972), Hao (1995), Hanna (1995), McFarland (2004)
39023	MYF6	G	T	GG	Centronuclear Myopathy|Becker Muscular Dystrophy	Kerst (2000)
79159	BIN1	C	A	CC	Centronuclear Myopathy	Nicot (2007)
79160	BIN1	C	T	CC	Centronuclear Myopathy	Nicot (2007)
47965	PHOX2B	T	C	TT	Central hypoventilation syndrome	Trochet (2005)
47968	PHOX2B	T	G	TT	Central hypoventilation syndrome	Trochet (2005)
800954	CRYGC	G	A	GG	Cataract, Primary Congenital	Santhiya (2002), Gonzalez-Huerta (2007)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
53819	MAF	C	G	CC	Cataract, Ocular Anterior Dysgenesis and Coloboma	Jamieson (2002)
53447	HSF4	C	T	CC	Cataract, Marner Type	Marner (1949), Eiberg (1988), Bu (2002), Francis (2004), Ke (2006), Enoki (2010)
52292	CRYGC	T	G	TT	Cataract, Coppock-like	Brakenhoff (1994), Heon (1999), Heon (1999), Liang (2003)
52274	CRYGC	G	A	GG	Cataract, Congenital Lamellar	Santhiya (2002), Gonzalez-Huerta (2007)
79282	PITX3	C	T	CC	Cataract, Congenital	Semina (1998)
53848	CRYBB2	G	T	GG	Cataract, Central Nuclear	Santhiya (2004)
23740	CASP8	C	T	CC	Caspase-8 Deficiency	Chun (2002), Chun (2002)
33514	RMRP	T	C	TT	Cartilage-Hair Hypoplasia	Ridanpaa (2001), Sulisalo (1997), Ridanpaa (2003), Bonafe (2002)
30889	SLC25A20	G	A	GG	Carnitine-acylcarnitine Translocase Deficiency	Costa (1999)
30894	SLC25A20	T	C	TT	Carnitine-acylcarnitine Translocase Deficiency	Aqeel (2003), Iacobazzi (2004)
14867	SLC22A5	A	G	AA	Carnitine Deficiency, Systemic Primary	Pereira (1988), Scholte (1990), Vaz (1999)
65244	MYH8	C	T	CC	Carney Complex Variant	Veugelers (2004), Stratakis (2004), (Chaudron (1992), Toydemir (2006), Hoof (1974), Chen (1992)
47303	PRKAR1A	A	G	AA	Carney complex	Kirschner (2000)
1501	TAZ	G	A	G	Cardiomyopathy, X-linked infantile	Brady (2006)
1507	TAZ	G	T	G	Cardiomyopathy, X-linked infantile	Bissler (2002)
1516	TAZ	G	A	G	Cardiomyopathy, X-linked infantile	D'Adamo (1997)
62900	MAP2K1	T	C	TT	Cardiofaciocutaneous Syndrome	Rodriguez-Viciana (2006)
62888	MAP2K2	A	C,T	AA	Cardiofaciocutaneous Syndrome	Rodriguez-Viciana (2006)
62901	MAP2K1	A	G	AA	Cardiofaciocutaneous Syndrome	Rodriguez-Viciana (2006)
47492	TP53	C	A,T	CC	Carcinoma of Ampulla of Vater	Achatz (2007)
65497	CA2	G	A	GG	Carbonic Anhydrase Deficiency	Hu (1992)
9856	RASA1	C	T	CC	Capillary Malformation-Arteriovenous Malformation	Eerola (2003)
27461	ASPA	A	G	AA	Canavan disease ?	Tacke (2005)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
27430	ASPA	C	A	CC	Canavan Disease	Kaul (1994), Kaul (1996), Propheta (1998), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2014), Laboratory for Molecular Medicine of Partners HealthCare Personalized Medicine (2014), Invitae (2016), Illumina Clinical Services Laboratory (2016)
27433	ASPA	C	A	CC	Canavan Disease	Kaul (1994), Shaag (1995)
27432	ASPA	A	C	AA	Canavan Disease	Kaul (1993), Elpeleg (1994), Feigenbaum (2004), Laboratory for Molecular Medicine of Partners HealthCare Personalized Medicine (2015), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2016), Invitae (2016), GeneDx (2016)
27434	ASPA	T	G	TT	Canavan Disease	Zeng (2006)
27435	ASPA	T	C	TT	Canavan Disease	Kaul (1996)
27436	ASPA	A	C	AA	Canavan Disease	Sistermans (2000)
27437	ASPA	A	G	AA	Canavan Disease	Zeng (2002)
27438	ASPA	G	A	GG	Canavan Disease	Kaul (1996)
27439	ASPA	G	A	GG	Canavan Disease	Sistermans (2000)
27440	ASPA	A	C	AA	Canavan Disease	Zeng (2002)
27441	ASPA	G	A	GG	Canavan Disease	Janson (2006)
27442	ASPA	T	G	TT	Canavan Disease	Elpeleg (1999)
27443	ASPA	C	A	CC	Canavan Disease	Kaul (1996)
27444	ASPA	G	T	GG	Canavan Disease	Olsen (2002)
27445	ASPA	A	T	AA	Canavan Disease	Zeng (2006)
27446	ASPA	G	A	GG	Canavan Disease	Kaul (1996)
27447	ASPA	T	C	TT	Canavan Disease	Kobayashi (1998)
27448	ASPA	A	T	AA	Canavan Disease	Zeng (2006)
27449	ASPA	T	C	TT	Canavan Disease	Kaul (1995)
27450	ASPA	G	A	GG	Canavan Disease	Kaul (1996)
27451	ASPA	C	G	CC	Canavan Disease	Zeng (2002)
27452	ASPA	C	T	CC	Canavan Disease	Zeng (2006)
27453	ASPA	G	A	GG	Canavan Disease	Sistermans (2000)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
27454	ASPA	C	T	CC	Canavan Disease	Kaul (1996)
27455	ASPA	C	T	CC	Canavan Disease	Zeng (2006)
27456	ASPA	C	A	CC	Canavan Disease	Sistermans (2000)
27457	ASPA	C	A	CC	Canavan Disease	Elpeleg (1999)
27458	ASPA	C	T	CC	Canavan Disease	Zeng (2002)
27459	ASPA	G	T	GG	Canavan Disease	Elpeleg (1999)
27466	ASPA	A	T	AA	Canavan Disease	Zeng (2006)
27467	ASPA	A	T	AA	Canavan Disease	Zeng (2002)
27468	ASPA	T	C	TT	Canavan Disease	Zeng (2006)
27469	ASPA	G	A	GG	Canavan Disease	Shaag (1995)
27470	ASPA	C	T	CC	Canavan Disease	Elpeleg (1999)
27471	ASPA	C	T	CC	Canavan Disease	Elpeleg (1999)
27472	ASPA	G	A	GG	Canavan Disease	Elpeleg (1999)
27473	ASPA	A	G	AA	Canavan Disease	Surendran (2003)
27474	ASPA	T	C	TT	Canavan Disease	Shaag (1995)
27475	ASPA	A	G	AA	Canavan Disease	Zeng (2002)
27476	ASPA	A	T	AA	Canavan Disease	Zeng (2002)
27477	ASPA	A	G	AA	Canavan Disease	Kaul (1994)
27478	ASPA	G	C	GG	Canavan Disease	Zeng (2006)
27479	ASPA	G	T	GG	Canavan Disease	Rady (2000)
56675	TGFB1	A	G	AA	Camurati-Engelmann Disease	Kinoshita (2000), Janssens (2000), Saito (2001)
56698	TGFB1	G	A	GG	Camurati-Engelmann Disease	II receptor (1901), Kinoshita (2000), Janssens (2000), McGowan (2003), Kinoshita (2004)
35753	COL1A1	G	A	GG	Caffey Disease	Gensure (2005)
64111	CUL4B	G	A	G	Cabezas Syndrome	Tarpey (2007), Wei (1993), Zou (2007), Zou (2007)

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Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
64112	CUL4B	G	A	G	Cabezas Syndrome	Tarpey (2007), Cabezas (2000)
78497	CD96	C	T	CC	C Syndrome	Kaname (2007)
13265	AR	G	A	G	Breast Cancer, Male, with Reifenstein Syndrome	Wooster (1992), Weidemann (1998)
78879	SMARCB1	G	A	GG	Brain Tumor, Posterior Fossa, of Infancy, Familial	Taylor (2000)
34749	HOXD13	C	G	CC	Brachydactyly, Type E	Johnson (2003)
800812	HOXD13	C	G	CC	Brachydactyly, Type D	Johnson (2003)
34750	HOXD13	A	C	AA	Brachydactyly, Type E	Johnson (2003)
800813	HOXD13	A	C	AA	Brachydactyly, Type D	Johnson (2003)
35146	BMPR1B	G	A	GG	Brachydactyly, Type C	Lehmann (2006)
800814	BMPR1B	G	A	GG	Brachydactyly, Type 2A	Lehmann (2006)
800815	BMPR1B	G	A	GG	Symphalangism, Type 1	Lehmann (2006)
34884	ROR2	C	T	CC	Brachydactyly, Type B1	Oldridge (2000)
34886	ROR2	G	A,T	GG	Brachydactyly, Type B1	Oldridge (2000)
34887	ROR2	G	A	GG	Brachydactyly, Type B	Schwabe (2000)
35144	BMPR1B	T	A	TT	Brachydactyly, Type A2	Lehmann (2003)
35145	BMPR1B	C	T	CC	Brachydactyly, Type A2	Lehmann (2003)
34844	IHH	C	T	CC	Brachydactyly, Type A1	Farabee (1903), Drinkwater (1908), Drinkwater (1914), McCready (2002), Giordano (2003), McCready (2005)
34845	IHH	T	C	TT	Brachydactyly, Type A1	Gao (2001), Kirkpatrick (2003)
34847	IHH	C	T	CC	Brachydactyly, Type A1	Gao (2001)
34848	IHH	G	T	GG	Brachydactyly, Type A1	Gao (2001)
34849	IHH	C	T	CC	Brachydactyly, Type A1	Gao (2001)
34850	IHH	G	A	GG	Brachydactyly, Type A1	Liu (2006)
34645	NOG	C	T	CC	Brachydactyl, Type B	Mangino (2002)
800808	NOG	C	T	CC	Symphalangism, Proximal	Mangino (2002)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
34647	NOG	C	G	CC	Brachydactyl, Type B	Gong (1999), Dixon (2001), Lehmann (2007)
800809	NOG	C	G	CC	Symphalangism, Proximal	Gong (1999), Dixon (2001), Lehmann (2007)
800810	NOG	C	G	CC	Tarsal-Carpal Coalition Syndrome	Gong (1999), Dixon (2001), Lehmann (2007)
34649	NOG	G	A	GG	Brachydactyl, Type B	Kosaki (2004)
800811	NOG	G	A	GG	Symphalangism, Proximal, and Premature Ovarian Failure	Kosaki (2004)
52217	RLBP1	G	A,C	GG	Bothnia Retinal Dystrophy	Burstedt (1999), Burstedt (2001), Granse (2001)
800807	RLBP1	G	A,C	GG	Retinitis Punctata Albescens	Morimura (1999)., Burstedt (1999), Burstedt (2001), Granse (2001)
64027	PHF6	T	C	T	Borjeson-Forssman-Lehmann Syndrome	Lower (2002)
64028	PHF6	A	T	A	Borjeson-Forssman-Lehmann Syndrome	Lower (2002)
64029	PHF6	G	A	G	Borjeson-Forssman-Lehmann Syndrome	Lower (2002)
64030	PHF6	G	T	G	Borjeson-Forssman-Lehmann Syndrome	Lower (2002)
64031	PHF6	A	G	A	Borjeson-Forssman-Lehmann Syndrome	Lower (2002)
64032	PHF6	A	G	A	Borjeson-Forssman-Lehmann Syndrome	Lower (2002)
64033	PHF6	A	G	A	Borjeson-Forssman-Lehmann Syndrome	Lower (2002), Vallee (2004)
64034	PHF6	A	G	A	Borjeson-Forssman-Lehmann Syndrome	Crawford (2006)
64035	PHF6	C	T	C	Borjeson-Forssman-Lehmann Syndrome	Borjeson (1962), Lower (2002), Lower (2004)
64036	PHF6	A	G	A	Borjeson-Forssman-Lehmann Syndrome	Vallee (2004)
64158	FLNB	T	G	TT	Boomerang dysplasia	Bicknell (2005)
64169	FLNB	T	C	TT	Boomerang dysplasia	Bicknell (2005)
52480	OPN1LW	T	C	T	Blue-cone Monochromacy	Winderickx (1992)
52502	OPN1LW	C	T	C	Blue-cone Monochromacy	Nathans (1993)
52503	OPN1LW	C	T	C	Blue-cone Monochromacy	Nathans (1993)
61732	RECQL3	A	T	AA	Bloom Syndrome	Ellis (1995)
61733	RECQL3	A	T	AA	Bloom Syndrome	Ellis (1997)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
61734	RECQL3	C	T	CC	Bloom Syndrome	Ellis (1997)
61735	RECQL3	A	G	AA	Bloom Syndrome	Ellis (1995)
61736	RECQL3	C	T	CC	Bloom Syndrome	Ellis (1997)
61737	RECQL3	C	T	CC	Bloom Syndrome	No author listed (1999)
61738	RECQL3	T	C	TT	Bloom Syndrome	Ellis (1995)
61739	RECQL3	T	C	TT	Bloom Syndrome	Barakat (2000)
61740	RECQL3	G	A	GG	Bloom Syndrome	Ellis (1997)
61741	RECQL3	C	T	CC	Bloom Syndrome	Ellis (1997)
61742	RECQL3	G	A	GG	Bloom Syndrome	Ellis (1997)
61743	RECQL3	G	T	GG	Bloom Syndrome	Foucault (1997)
61744	RECQL3	G	C	GG	Bloom Syndrome	Ellis (1995)
8656	FOXL2	A	C	AA	Blepharophimosis, Ptosis, and Epicanthus Inversus, Type I	The first reported case in this family was born in (1841), Dollfus (2003)
14787	CARD15	G	A	GG	Blau Syndrome	Miceli-Richard (2001)
75043	BTD	I	D	II	Biotinidase Deficiency	Pomponio (1995)
30130	BTD	G	A	GG	Biotinidase Deficiency, late-onset	Cole (2994), Norrgard (1997), Wolf (1997), Norrgard (1998), Norrgard (1999), Wolf (2000), GeneReviews (2011), Center for Pediatric Genomic Medicine of Children's Mercy Hospital and Clinics (2014), Counsyl (2014), GeneDx (2016)
30134	BTD	T	G	TT	Biotinidase Deficiency	Norrgard (1999)
30143	BTD	G	A	GG	Variant of Unknown Significance (possibly associated with Biotinidase Deficiency)	Norrgard (1999), Genetic Services Laboratory of the University of Chicago (2014), GeneDx (2016), Emory Genetics Laboratory of Emory University (2016)
30144	BTD	G	T	GG	Biotinidase Deficiency	Milankovics (2007)
30145	BTD	C	A	CC	Biotinidase Deficiency	Wolf (2005)
30146	BTD	G	T	GG	Biotinidase Deficiency	Wolf (2005)
30147	BTD	T	G	TT	Biotinidase Deficiency	Pomponio (2000)
30148	BTD	G	A	GG	Biotinidase Deficiency	Muhl (2001)
30149	BTD	G	T	GG	Biotinidase Deficiency	Norrgard (1999)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
30150	BTD	G	A	GG	Biotinidase Deficiency	Wolf (2002)
30151	BTD	T	C	TT	Biotinidase Deficiency	Laszlo (2003)
30152	BTD	C	T	CC	Biotinidase Deficiency	Pomponio (2000)
30153	BTD	G	A	GG	Biotinidase Deficiency	Laszlo (2003)
30154	BTD	C	A	CC	Biotinidase Deficiency	Laszlo (2003)
30155	BTD	C	T	CC	Biotinidase Deficiency	Pomponio (1997)
30156	BTD	A	G	AA	Biotinidase Deficiency	Wolf (2002)
30157	BTD	C	T	CC	Biotinidase Deficiency	Wolf (2005)
30158	BTD	G	A	GG	Biotinidase Deficiency	Norrgard (1999)
30159	BTD	G	A	GG	Biotinidase Deficiency, Partial	Wolf (2002)
30160	BTD	G	T	GG	Biotinidase Deficiency	Norrgard (1999)
30161	BTD	T	G	TT	Biotinidase Deficiency, Partial	Muhl (2001)
30162	BTD	G	A	GG	Biotinidase Deficiency	Laszlo (2003)
30163	BTD	G	C	GG	Biotinidase Deficiency	Pomponio (1997)
30164	BTD	G	T	GG	Biotinidase Deficiency	Wolf (2005)
30165	BTD	A	G	AA	Biotinidase Deficiency	Norrgard (1999)
30166	BTD	T	G	TT	Biotinidase Deficiency	Swango (1998)
30167	BTD	T	C	TT	Biotinidase Deficiency	Norrgard (1999)
30168	BTD	A	C	AA	Biotinidase Deficiency	Milankovics (2007)
30169	BTD	C	T	CC	Biotinidase Deficiency	Pomponio (2000)
30170	BTD	G	A	GG	Biotinidase Deficiency	Pomponio (1997)
30171	BTD	C	T	CC	Biotinidase Deficiency	Milankovics (2007)
30172	BTD	C	T	CC	Biotinidase Deficiency	Norrgard (1999)
30173	BTD	A	G	AA	Biotinidase Deficiency, Partial	Muhl (2001)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
30174	BTD	G	T	GG	Biotinidase Deficiency	Norrgard (1999)
30175	BTD	G	A	GG	Biotinidase Deficiency	Pomponio (2000)
30176	BTD	A	G	AA	Biotinidase Deficiency	Milankovics (2007)
30177	BTD	A	G	AA	Biotinidase Deficiency	Pomponio (1997)
30178	BTD	C	G	CC	Biotinidase Deficiency	Pomponio (2000)
30179	BTD	G	A	GG	Biotinidase Deficiency	Wolf (2002)
30180	BTD	A	G	AA	Biotinidase Deficiency	Pomponio (1997)
30181	BTD	C	T	CC	Biotinidase Deficiency	Norrgard (1999)
30182	BTD	A	G	AA	Biotinidase Deficiency	Wolf (2005)
30183	BTD	C	T	CC	Biotinidase Deficiency	Pomponio (1997)
30184	BTD	G	C	GG	Biotinidase Deficiency	Wolf (2005)
30185	BTD	G	C	GG	Biotinidase Deficiency	Laszlo (2003)
30186	BTD	G	T	GG	Biotinidase Deficiency	Swango (1998)
30187	BTD	G	A	GG	Biotinidase Deficiency	Wolf (2005)
30188	BTD	A	G	AA	Biotinidase Deficiency	Wolf (1997)
30189	BTD	C	T	CC	Biotinidase Deficiency	Wolf (2005)
30190	BTD	A	T	AA	Biotinidase Deficiency	Wolf (2002)
30191	BTD	C	G	CC	Biotinidase Deficiency	Laszlo (2003)
30192	BTD	T	C	TT	Biotinidase Deficiency	Pomponio (1997)
30193	BTD	T	G	TT	Biotinidase Deficiency, Partial	Muhl (2001)
30194	BTD	G	C	GG	Biotinidase Deficiency	Norrgard (1999)
30195	BTD	T	G	TT	Biotinidase Deficiency	Wolf (2002)
30196	BTD	G	A	GG	Biotinidase Deficiency	Pomponio (2000)
30197	BTD	G	A	GG	Biotinidase Deficiency	Norrgard (1999)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
30198	BTD	G	A	GG	Biotinidase Deficiency	Pomponio (1997)
30199	BTD	G	A	GG	Biotinidase Deficiency	Wolf (2002)
30200	BTD	A	G	AA	Biotinidase Deficiency	Swango (1998)
30201	BTD	C	T	CC	Biotinidase Deficiency, Partial	Wolf (2002)
30202	BTD	G	A	GG	Biotinidase Deficiency	Norrgard (1999)
30208	BTD	G	C	GG	Biotinidase Deficiency	Muhl (2001)
30209	BTD	G	A	GG	Biotinidase Deficiency	Wolf (2005)
30210	BTD	G	C	GG	Biotinidase Deficiency	Wolf (2005)
30211	BTD	A	G	AA	Biotinidase Deficiency	Wolf (2002)
30212	BTD	T	A	TT	Biotinidase Deficiency	Neto (2004)
30213	BTD	G	T	GG	Biotinidase Deficiency	Pomponio (1997)
30214	BTD	G	A	GG	Biotinidase Deficiency	Swango (1998)
30215	BTD	A	G	AA	Biotinidase Deficiency	Wolf (2005)
30216	BTD	A	C	AA	Biotinidase Deficiency	Wolf (1997)
30217	BTD	G	A	GG	Biotinidase Deficiency	Pomponio (1997)
30218	BTD	G	A	GG	Biotinidase Deficiency	Wolf (2002)
30219	BTD	T	C	TT	Biotinidase Deficiency	Pomponio (2000)
30220	BTD	G	A	GG	Biotinidase Deficiency	Pomponio (1997)
30221	BTD	A	C	AA	Biotinidase Deficiency	Pomponio (1998)
30222	BTD	C	T	CC	Biotinidase Deficiency	Norrgard (1999)
30223	BTD	T	A	TT	Biotinidase Deficiency	Laszlo (2003)
30224	BTD	C	G	CC	Biotinidase Deficiency	Wolf (2005)
30225	BTD	C	T	CC	Biotinidase Deficiency	Pomponio (2000)
30226	BTD	G	T	GG	Biotinidase Deficiency	Wolf (2002)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
30227	BTD	C	T	CC	Biotinidase Deficiency, late-onset	Pomponio (1997)
30228	BTD	A	G	AA	Biotinidase Deficiency	Norrgard (1999)
30229	BTD	G	C	GG	Biotinidase Deficiency	Muhl (2001)
30230	BTD	G	A	GG	Biotinidase Deficiency	Muhl (2001)
30231	BTD	G	A	GG	Biotinidase Deficiency, late-onset	Pomponio (1997)
30232	BTD	G	T	GG	Biotinidase Deficiency	Milankovics (2007)
30233	BTD	G	A	GG	Biotinidase Deficiency	Pomponio (1997)
2991	JAG1	C	T	CC	Biliary Atresia, Extrahepatic	Kohsaka (2002)
78872	AMACR	A	G	AA	Bile Acid Synthesis Defect, Congenital, 4	Sequeira (1998), Ferdinandusse (2000)
41491	COL6A1	G	T	GG	Bethlem Myopathy	Jobsis (1996)
41492	COL6A1	A	G	AA	Bethlem myopathy	Scacheri (2002)
41493	COL6A1	G	A	GG	Bethlem myopathy	Lucioli (2005)
41494	COL6A1	C	T	CC	Bethlem myopathy	Lampe (2005)
41495	COL6A1	G	A	GG	Bethlem myopathy	Lucioli (2005)
41498	COL6A1	G	A	GG	Bethlem myopathy	Lampe (2005)
41500	COL6A1	G	T	GG	Bethlem myopathy	Jobsis (1996)
41501	COL6A1	G	A	GG	Bethlem myopathy	Scacheri (2002)
41502	COL6A1	G	T	GG	Bethlem myopathy	Lucioli (2005)
41503	COL6A1	A	C	AA	Bethlem myopathy	Lampe (2005)
41508	COL6A1	G	A	GG	Bethlem Myopathy	Lamande (1999), Pan (2003), Lucioli (2005)
27519	HEXA	G	A	GG	Beta-hexosaminidase pseudodeficiency	Triggs-Raine (1992), Mules (1992), Tomczak (1993), Cao (1993), Cao (1994), Triggs-Raine (1995), Park (2010)
20018	HBB	T	A	TT	Beta Thalassemia	Chang (1979), Kan (1979), Steger (1993), Krawczak (2000), Ding (2004)
20033	HBB	C	A,G,T	CC	Beta Thalassemia	Ingram (1961), Shibata (1962), Blackwell (1970), Fairbanks (1980), Benz (1981), Kazazian (1984), Orkin (1982), Antonarakis (1982), Thein (1987), Rey (1991), Rees (1996), Chotivanich (2002), Ohashi (2004), Flatz (2004), Edison (2005), Premawardhena (2001),

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
20064	HBB	G	A,C,T	GG	Beta Thalassemia	Chehab (1986), Trecartin (1981), (Epstein (1963), Witkowski (1990), Edgar (1966), Rosatelli (1992), Rosatelli (1987)
20220	HBB	G	A,C,T	GG	Beta Thalassemia	Rund (1989, 1991)
20229	HBB	T	C,G	TT	Beta Thalassemia	Orkin (1983), Ding (2004)
20236	HBB	T	C	TT	Beta Thalassemia	Kazazian (1988), Rund (1992)
20251	HBB	C	G	CC	Beta Thalassemia	Renda (1992)
20273	HBB	A	G,T	AA	Beta Thalassemia	Orkin (1982)
72216	HBB	D	I	DD	Beta Thalassemia	Kazazian (1984), Villegas (1998), Baig (2006), Ahmed (2007), Usman (2009), Al-Allawi (2010), Integrated Genetics of Laboratory Corporation of America (2011), Teh (2014), GeneReviews (2015), Illumina Clinical Services Laboratory (2016), Knight Diagnostic Laboratories of Oregon Health and Sciences University (2016), GeneDx (2016), Counsyl (2016), Illumina Clinical Services Laboratory (2017), Invitae (2018)
72217	HBB	I	D	II	Beta Thalassemia	Kazazian (1984), Kimura (1983), Lau (1997), Chiu (2002)
72218	HBB	I	D	II	Beta Thalassemia	Kazazian (1983), Bouhass (1990), Rosatelli (1992), Romey (1993)
75059	HBB	C	T	CC	Beta Thalassemia	Spritz (1981), Williamson (1981), Kaplan (1990)
75061	HBB	I	D	II	Beta Thalassemia	Rund (1989), Rund (1991)
53273	BEST1	T	C	TT	Best Macular Dystrophy	Petrukhin (1998)
53274	BEST1	G	C	GG	Best Macular Dystrophy	Nordstrom (1977), Petrukhin (1998)
53277	BEST1	T	A	TT	Best Macular Dystrophy	Petrukhin (1998)
800955	BEST1	C	T	CC	Best Macular Dystrophy	Kramer (2000)
800806	BEST1	C	T	CC	Vitelliform Macular Dystrophy	Kramer (2000)
53279	BEST1	C	T	CC	Best Macular Dystrophy	Allikmets (1999)
53280	BEST1	G	C	GG	Best Macular Dystrophy	Caldwell (1999)
53294	BEST1	C	G	CC	Best Macular Dystrophy	Lotery (2000)
800956	BEST1	A	C	AA	Best Macular Dystrophy	Petrukhin (1998), Kramer (2000)
53296	BEST1	A	G	AA	Best Macular Dystrophy	Apushkin (2006)
53297	BEST1	G	A	GG	Best Macular Dystrophy	Marquardt (1998)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
53298	BEST1	T	C	TT	Best macular dystrophy?	Petrukhin (1998)
53299	BEST1	C	T	CC	Best Macular Dystrophy	Bakall (1999)
53300	BEST1	G	A	GG	Best Macular Dystrophy	Marquardt (1998)
53301	BEST1	A	T	AA	Best Macular Dystrophy	Kramer (2003)
53302	BEST1	G	A	GG	Best Macular Dystrophy	Caldwell (1999)
53303	BEST1	C	T	CC	Best Macular Dystrophy	Marchant (2001)
53304	BEST1	T	G	TT	Best Macular Dystrophy	Lotery (2000)
53305	BEST1	C	G	CC	Best Macular Dystrophy	White (2000)
53306	BEST1	G	T	GG	Best Macular Dystrophy	Marquardt (1998)
53307	BEST1	G	A	GG	Best Macular Dystrophy	Marquardt (1998)
53308	BEST1	C	T	CC	Best Macular Dystrophy	Kramer (2000)
53309	BEST1	G	C	GG	Best Macular Dystrophy	Kramer (2003)
53310	BEST1	C	G	CC	Best Macular Dystrophy	Kramer (2000)
53311	BEST1	T	C	TT	Best Macular Dystrophy	Kramer (2003)
53313	BEST1	C	G	CC	Best macular dystrophy	Schatz (2006)
53314	BEST1	A	G	AA	Best macular dystrophy	Lotery (2000)
53315	BEST1	T	C	TT	Best macular dystrophy	Kramer (2003)
53317	BEST1	A	T	AA	Best macular dystrophy	Kramer (2000)
53318	BEST1	T	A	TT	Best macular dystrophy	Marchant (2001)
53319	BEST1	C	A	CC	Best macular dystrophy	Lotery (2000)
53320	BEST1	C	G	CC	Best macular dystrophy	Bakall (1999)
53322	BEST1	T	C	TT	Best macular dystrophy	Eksandh (2001)
53323	BEST1	C	T	CC	Best macular dystrophy	Lotery (2000)
53324	BEST1	C	A	CC	Best macular dystrophy	Kramer (2000)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
53325	BEST1	C	T	CC	Best macular dystrophy	Bakall (1999)
53326	BEST1	G	A	GG	Best macular dystrophy	Marchant (2001)
53327	BEST1	G	C	GG	Best macular dystrophy	Bakall (1999)
53328	BEST1	C	A	CC	Best macular dystrophy	White (2000)
53329	BEST1	T	G	TT	Best macular dystrophy	Kramer (2000)
53330	BEST1	C	A	CC	Best macular dystrophy	Lotery (2000)
53331	BEST1	T	C	TT	Best macular dystrophy	Kramer (2003)
53332	BEST1	G	C	GG	Best macular dystrophy	Kramer (2003)
53333	BEST1	C	A	CC	Best macular dystrophy?	Petrukhin (1998)
53335	BEST1	C	G	CC	Best macular dystrophy	Li (2004)
53336	BEST1	C	G	CC	Best macular dystrophy	Lotery (2000)
53337	BEST1	G	A	GG	Best macular dystrophy	Bakall (1999)
53338	BEST1	T	G	TT	Best macular dystrophy	Lotery (2000)
53339	BEST1	G	A	GG	Best macular dystrophy	Kramer (2000)
53342	BEST1	C	T	CC	Best macular dystrophy	Lotery (2000)
53343	BEST1	T	C	TT	Best macular dystrophy	Lotery (2000)
53345	BEST1	G	A	GG	Best macular dystrophy	White (2000)
53348	BEST1	G	A	GG	Best macular dystrophy	Lotery (2000)
53349	BEST1	C	A	CC	Best macular dystrophy	Bakall (1999)
53350	BEST1	C	T	CC	Best macular dystrophy	Marquardt (1998)
53351	BEST1	T	G	TT	Best macular dystrophy	Palomba (2000)
53353	BEST1	C	A	CC	Best macular dystrophy	Kramer (2000)
53354	BEST1	T	C	TT	Best macular dystrophy	Lotery (2000)
53355	BEST1	A	G	AA	Best macular dystrophy	Marquardt (1998)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
53356	BEST1	T	G	TT	Best macular dystrophy	White (2000)
53357	BEST1	G	A	GG	Best macular dystrophy	Marquardt (1998)
53358	BEST1	G	C	GG	Best macular dystrophy	Marchant (2001)
53360	BEST1	C	G	CC	Best macular dystrophy	White (2000)
53362	BEST1	C	A	CC	Best macular dystrophy	Kramer (2003)
53363	BEST1	G	A	GG	Best macular dystrophy	Lotery (2000)
53365	BEST1	C	G	CC	Best macular dystrophy	Lotery (2000)
53367	BEST1	C	A	CC	Best macular dystrophy	Bakall (1999)
53368	BEST1	C	G	CC	Best macular dystrophy	Kramer (2003)
53369	BEST1	T	C	TT	Best macular dystrophy	Yanagi (2002)
53370	BEST1	A	G	AA	Best macular dystrophy	Marchant (2001)
53371	BEST1	A	C	AA	Best macular dystrophy	Lotery (2000)
53373	BEST1	C	G	CC	Best macular dystrophy	Marquardt (1998)
53374	BEST1	T	C	TT	Best macular dystrophy	Kramer (2003)
53375	BEST1	G	A	GG	Best macular dystrophy	Petrukhin (1998)
53376	BEST1	G	A	GG	Best macular dystrophy	White (2000)
53377	BEST1	T	G	TT	Best macular dystrophy	Caldwell (1999)
53378	BEST1	G	A	GG	Best macular dystrophy	Kramer (2000)
53379	BEST1	A	G	AA	Best macular dystrophy	Lotery (2000)
53380	BEST1	A	T	AA	Best macular dystrophy	Lotery (2000)
53381	BEST1	G	C	GG	Best macular dystrophy	Marchant (2002)
53382	BEST1	T	A	TT	Best macular dystrophy	Marchant (2002)
53383	BEST1	T	C	TT	Best macular dystrophy	Marquardt (1998)
53384	BEST1	A	G	AA	Best macular dystrophy	Lotery (2000)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
53385	BEST1	G	C	GG	Best macular dystrophy	Lotery (2000)
53386	BEST1	C	T	CC	Best macular dystrophy	Caldwell (1999)
53387	BEST1	A	G	AA	Best macular dystrophy	Lotery (2000)
53388	BEST1	A	G	AA	Best macular dystrophy	Marchant (2002)
53389	BEST1	T	C	TT	Best macular dystrophy	White (2000)
53390	BEST1	T	G	TT	Best macular dystrophy	White (2000)
53393	BEST1	G	C	GG	Best macular dystrophy	Kramer (2000)
22206	GP9	A	G	AA	Bernard-Soulier Syndrome, Type C	Clemetson (1994), Sachs (2003), Sachs (2003)
43653	BSCL2	C	G	CC	Berardinelli-Seip lipodystrophy	Magre (2001)
43654	BSCL2	G	A	GG	Berardinelli-Seip lipodystrophy	Ebihara (2004)
39066	TITF1	C	A,T	CC	Benign Hereditary Chorea	Breedveld (2002)
66836	COL4A4	C	T	CC	Benign Familial Hematuria	Lemmink (1996), Lemmink (1996)
65095	FGFR2	T	C	TT	Beare-Stevenson Cutis Gyrata Syndrome	Przylepa (1996), Wang (2002), Vargas (2003)
12259	CLCNKB	C	T	CC	Bartter Syndrome, Type 3	Simon (1997)
12460	SLC12A1	G	A	GG	Bartter Syndrome, Antenatal, Type 1	Kurtz (1997)
67026	KCNJ1	G	A,T	GG	Bartter Syndrome	Karolyi (1997)
1508	TAZ	G	A	G	Barth Syndrome and Noncompaction of the Left Ventricular Myocardium, Familial Isolated	D'Adamo (1997), Bleyl (1997)
1517	TAZ	A	G	A	Barth Syndrome	Ichida (2001)
1515	TAZ	C	T	C	Barth Syndrome	Johnston (1997)
1518	TAZ	G	A	G	Barth Syndrome	Kelley (1991), Johnston (1997)
1521	TAZ	G	A	G	Barth Syndrome	Bione (1996)
1506	TAZ	T	G	T	Barth Syndrome	Cantlay (1999)
1512	TAZ	T	G	T	Barth Syndrome	Cantlay (1999)
1523	TAZ	T	G	T	Barth Syndrome	Johnston (1997)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
7546	COL7A1	C	T	CC	Bart Syndrome	Bart (1966), Christiano (1996)
800889	TAP1	G	A	GG	Bare Lymphocyte Syndrome, Type I	Dogu (2006)
63819	BBS4	C	A	CC	Bardet-Biedl Syndrome 4	Katsanis (2002)
68635	CEP290	C	A	CC	Bardet-Biedl Syndrome 14	Leitch (2008)
68626	MKS1	A	G	AA	Bardet-Biedl Syndrome 13	Leitch (2008)
68627	MKS1	C	T	CC	Bardet-Biedl Syndrome 13	Leitch (2008)
68628	MKS1	T	C	TT	Bardet-Biedl Syndrome 13	Leitch (2008)
75037	BBS10	D	I	DD	Bardet-Biedl Syndrome 10	Stoetzel (2006), Claus (2008)
62751	BBS1	T	G	TT	Bardet-Biedl Syndrome 1	Mykytyn (2002), Mykytyn (2003), Beales (2003), Fan (2004)
62757	BBS1	T	C	TT	Bardet-Biedl Syndrome	Mykytyn (2003)
68658	USP9Y	I	D	I	Azoospermia, Nonobstructive	Sun (1999)
38786	GDAP1	C	T	CC	Axonal Neuropathy with Vocal Cord Paresis	Cuesta (2002), Boerkoel (2003), Claramunt (2005)
38780	GDAP1	C	T	CC	Axonal Charcot-Marie-Tooth Disease, Type 2K	Claramunt (2005), Claramunt (2005)
37994	NEFL	G	A,T	GG	Axonal Charcot-Marie-Tooth Disease, Type 2E	Georgiou (2002), Fabrizi (2004)
41699	GARS	G	C	GG	Axonal Charcot-Marie-Tooth Disease, Type 2D|Distal Hereditary Motor Neuropathy, Type V	Ionasescu (1996), Pericak-Vance (1997), Antonellis (2003)
40927	MFN2	G	A	GG	Axonal Charcot-Marie-Tooth Disease, Type 2A2	Saito (1997), Zuchner (2004)
40934	MFN2	C	T	CC	Axonal Charcot-Marie-Tooth Disease, Type 2A12|Hereditary Motor and Sensory Neuropathy, Type VI	Zuchner (2004), Zuchner (2006)
13743	AIRE	A	G	AA	Autoimmune Polyendocrinopathy-Candidiasis-Ectodermal Dystrophy	Nagamine (1997)
13747	AIRE	C	T	CC	Autoimmune Polyendocrinopathy-Candidiasis-Ectodermal Dystrophy	Rosatelli (1998)
13751	AIRE	C	T	CC	Autoimmune Polyendocrinopathy-Candidiasis-Ectodermal Dystrophy	Nagamine (1997), Consortium (1997), Nagamine (1997), Wang (1998), Halonen (2002), Stolarski (2006), Zaidi (2009), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2014), Counsyl (2016), GeneDx (2017)
48437	CASP10	C	T	CC	Autoimmune Lymphoproliferative Syndrome, Type IIA	Wang (1999)
70246	NLGN4X	D	I	D	Autism|Mental Retardation	Laumonnier (2004), Laumonnier (2004)
70245	NLGN4X	D	I	D	Autism|Asperger Syndrome	Jamain (2003), Jamain (2003), Chih (2004)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
56069	NLGN3	C	T	C	Autism Spectrum Disorder|Asperger Syndrome	Jamain (2003), Jamain (2003), Jamain (2003), Comoletti (2004), Chih (2004)
73014	SLC9A9	G	A	GG	Autism	Morrow (2008)
71956	OTOF	A	G	AA	Auditory Neuropathy, Nonsyndromic|Auditory Neuropathy, Temperature-sensitive	Starr (1998), Mirghomizadeh (2002), Mirghomizadeh (2002), Varga (2006), Varga (2006)
24170	RAG1	C	T	CC	Atypical SCID/Omenn syndrome|Alpha/Beta T-Cell Lymphopenia with Gamma/Delta T-Cell Expansion, Severe CMV Infection, and Autoimmunity	Villa (2001), De Villartay (2005), De Villartay (2005)
800902	PANK2	C	T	CC	Atypical Pantothenate Kinase-associated Neurodegeneration	Zhou (2001), Hayflick (2003)
800901	PANK2	G	A	GG	Atypical Pantothenate Kinase-associated Neurodegeneration	Zhou (2001)
19868	ATRX	G	A	G	ATRX Syndrome	Wada (2000), Badens (2006), Badens (2006)
6023	CRELD1	G	A	GG	Atrioventricular Septal Defect	Maslen (2006)
5969	NKX2-5	C	A	CC	Atrioventricular Block, Idiopathic Second-Degree	Benson (1999)
9509	HR	G	A	GG	Atrichia with Papular Lesions	Hashimoto (1998), Ahmad (1999)
1850	SCN5A	A	G,T	AA	Atrial standstill ?	Makita (2005)
5953	NKX2-5	G	A	GG	Atrial Septal Defect with Atrioventricular Conduction Defects	Schott (1998), Hirayama-Yamada (2005)
5959	NKX2-5	G	A	GG	Atrial Septal Defect with Atrioventricular Conduction Defects	Hirayama-Yamada (2005)
6356	GATA4	G	A	GG	Atrial Septal Defect	Garg (2003)
78195	TBX20	G	C	GG	Atrial Septal Defect	Kirk (2007)
78196	TBX20	G	A	GG	Atrial Septal Defect	Kirk (2007)
2160	LMNA	G	A	GG	Atrial Fribrillation	Sebillon (2003)
25421	CCL5	T	C	TT	Atopic Dermatitis	Nickel (2000)
67940	SLC26A2	C	T	CC	Atelosteogenesis, Type II|Epiphyseal Dysplasia 4, Multiple|Diastrophic Dysplasia	Hastbacka (1996), Rossi (1996), Rossi (1996), Superti-Furga (1999), Huber (2001), Czarny-Ratajczak (2001), Macias-Gomez (2004), Macias-Gomez (2004), Ambry Genetics (2014), Counsyl (2015), Laboratory Corporation of America (2016), Illumina Clinical Services Laboratory (2016), GeneDx (2017), Invitae (2018), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2018), Fulgent Genetics (2018)
67937	SLC26A2	C	T	CC	Atelosteogenesis, Type IB	Superti-Furga (1996), Macias-Gomez (2004), Macias-Gomez (2004)
802233	SLC26A2	C	T	CC	Diastrophic Dysplasia	Superti-Furga (1996), Macias-Gomez (2004), Macias-Gomez (2004)
41087	SETX	G	A	GG	Ataxia-ocular apraxia 2	Moreira (2004)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
41091	SETX	A	C	AA	Ataxia-ocular Apraxia 2	Duquette (2005)
37874	TTPA	G	A	GG	Ataxia, isolated vitamin E deficiency	Cavalier (1998)
41286	MRE11A	C	G	CC	Ataxia telangiectasia-like disease	Fernet (2005)
70826	ATM	I	D	II	Ataxia Telangiectasia|T-cell Prolymphocytic Leukemia, Somatic	Wright (1996), Vorechovsky , (1996), Vorechovsky (1997), Spring (2002)
47175	ATM	C	T	CC	Ataxia Telangiectasia without Immunodeficiency	Toyoshima (1998)
46990	ATM	T	G	TT	Ataxia Telangiectasia Variant	McConville (1996), Vorechovsky (1997), Stankovic (1998)
46997	ATM	T	G	TT	Ataxia Telangiectasia Variant	McConville (1996)
47221	ATM	A	G	AA	Ataxia telangiectasia	McConville (1996), Sutton (2004)
47022	ATM	C	T	CC	Ataxia telangiectasia	Gilad (1996), Gilad (1996)
47215	ATM	G	A	GG	Ataxia telangiectasia	Sandoval (1999), Sandoval (1999)
70828	ATM	G	A	GG	Ataxia Telangiectasia	Sandoval (1999), Sandoval (1999)
47185	ATM	T	G	TT	Ataxia telangiectasia	Stankovic (1998)
47213	ATM	T	A	TT	Ataxia telangiectasia	Sasaki (1998)
70825	ATM	D	I	DD	Ataxia Telangiectasia	Laake (1998), Laake (2000)
70827	ATM	I	D	II	Ataxia Telangiectasia	Sasaki (1998)
37872	TTPA	A	C	AA	Ataxia and Retinitis Pigmentosa with Isolated Vitamin E Deficiency	Yokota , (1987), Gotoda (1995), Yokota (1996)
32141	ASL	G	A	GG	ASL Deficiency	Linnebank (2002)
43619	ALS2	G	A,T	GG	Ascending Spastic Paralysis, Infantile-onset	Devon (2003)
65370	VPS33B	G	A,T	GG	Arthrogryposis, Renal Dysfunction, and Cholestasis 1	Gissen (2004), Eastham (2001), Horslen (1994), Gissen (2006)
36078	MYH3	G	A,T	GG	Arthrogryposis, Distal, Type 2A	Toydemir (2006)
36085	MYH3	C	T	CC	Arthrogryposis, Distal, Type 2A	Toydemir (2006)
36086	MYH3	G	A	GG	Arthrogryposis, Distal, Type 2A	Toydemir (2006)
35464	COL3A1	G	A	GG	Arterial Aneurysms, Familial	Kontusaari (1990), Kuivaniemi (1990)
35490	COL3A1	G	A	GG	Arterial Aneurysms, Familial	Pinto (2000), Byers (2000)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
32036	ARG1	C	T	CC	Argininemia	Cardoso (1999)
32035	ARG1	T	C	TT	Argininemia	Snyderman (1979), Uchino (1995)
22167	AQP1	C	T	CC	Aquaporin-1 Deficiency|Colton-Null	Preston (1994)
65509	APRT	A	G	AA	APRT Deficiency, Japanese Type	Hidaka (1988), Kamatani (1989), Kamatani (1989), Kamatani (1990), Sahota (1991), Kamatani (1992), Kamatani (1996)
100872	APRT	D	I	DD	APRT Deficiency	Kamatani (1992)
5121	APOA2	A	G	AA	Apolipoprotein A2 Deficiency	Yazaki (2003)
65045	FGFR2	G	C	GG	Apert Syndrome	Wilkie (1995), Slaney (1996), Passo-Buenos (1998), Mantilla-Capacho (2005), Ibrahimi (2001)
65047	FGFR2	G	C	GG	Apert Syndrome	Wilkie (1995), Slaney (1996), Ibrahimi (2001), Andreou (2006)
67725	TGFBR1	G	A	GG	Aortic Aneurysm, Thoracic	Matyas (2006)
67696	MYH11	C	A	CC	Aortic Aneurysm, Familial Thoracic 5	Van Kien (2004), Zhu (2006)
67775	TGFBR2	G	A	GG	Aortic Aneurysm, Familial Thoracic	Pannu (2005), Disabella (2006)
35400	COL3A1	G	A	GG	Aortic Aneurysm	Kontusaari (1990), Kuivaniemi (1991)
800877	FGFR1	A	G	AA	Antley-Bixler Syndrome	Kress (2000), Hurley (2004), Huang (2005)
39530	SERPINF2	G	A	GG	Antiplasmin Alpha 2 Deficiency	Thorsen (1999)
39531	SERPINF2	G	A	GG	Antiplasmin Alpha 2 Deficiency	Yoshinaga (2000)
67034	KCNJ1	G	A,C	GG	Antenatal Bartter Syndrome, Type 2	Simon (1996), Schwalbe (1998)
53066	PAX6	G	A	GG	Aniridia	Martha (1995), van Heyningen (1998)
61754	UBE3A	A	G	AA	Angelman Syndrome	Rapakko (2004)
61766	UBE3A	T	C	TT	Angelman Syndrome	Kishino (1997)
13188	AR	G	A	G	Androgen Insensitivity, Partial	McPhaul (1992), Beitel (1994)
13206	AR	T	G	T	Androgen Insensitivity, Partial	Holterhus (1997), Holterhus (1997), Gottlieb (1999)
13395	AR	G	A	G	Androgen Insensitivity, Complete	Marcelli (1990)
800878	AR	A	G	A	Androgen Insensitivity Syndrome	Hiort (1996), Giwercman (2001)
13313	AR	C	T	C	Androgen Insensitivity Syndrome	Holterhus (2000), Holterhus (2000)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
13246	AR	G	A	G	Androgen Insensitivity Syndrome	Nguyen (2001)
16140	SAR1B	C	T	CC	Anderson Disease	Jones (2003)
6643	KCNJ2	C	T	CC	Andersen Cardiodysrhythmic Periodic Peralysis	Andelfinger (2002)
70542	SLC12A6	G	A,T	GG	Andermann Syndrome	Howard (2002), Salin-Cantegrel (2007)
70543	SLC12A6	I	D	II	Andermann Syndrome	Howard (2002)
70741	MTCYB	G	A	?	Exercise Intolerance	Andreu (1999), Andreu (1999)
78907	CHMP2B	A	C	AA	Amyotrphic Lateral Sclerosis	Parkinson (2006)
37230	DCTN1	C	T	CC	Amyotrophic Lateral Sclerosis|Frontotemporal Dementia	Munch (2005), Munch (2005)
37349	SOD1	A	C	AA	Amyotrophic Lateral Sclerosis	Andersen (1995), Andersen (1995), Robberecht (1996), Al-Chalabi (1998), Aguirre (1999), Khoris (2000), Hand (2001), Hand (2001)
37356	SOD1	G	T	GG	Amyotrophic Lateral Sclerosis	Rosen (1993), Regal (2006)
37375	SOD1	T	C	TT	Amyotrophic Lateral Sclerosis	Rosen (1993), Jones , (1993), Jones (1993), Jones (1995), Hayward (1996), Kikugawa (1997), Brock (1998)
37228	DCTN1	A	G	AA	Amyotrophic Lateral Sclerosis	Munch (2004)
37300	SOD1	C	T	CC	Amyotrophic Lateral Sclerosis	Brown (1951), Deng (1993), Rosen (1994)
37123	TTR	C	A	CC	Amyloidotic Polyneuropathy, German-American Type	Wallace (1986), Wallace (1988), Satier (1990), Blanco-Jerez (1998), Blanco-Jerez (1998)
37143	TTR	C	A	CC	Amyloidotic Polyneuropathy, Cardiac or Denmark Type|Amyloidosis III	The mother, who died in the influenza epidemic of (1918), They had available stored, frozen serum samples obtained in (1959), Frederiksen (1962), Frederiksen (1962), Derrick (1969), Allensworth (1969), Husby (1986), Nordvag (1992), Ranlov (1992), Benson (1992), Nordvag (1993), Nordvag (1995)
37031	TTR	G	A	GG	Amyloid Polyneuropathy, Andrade or Portuguese Type|Amyloidosis I, Hereditary Neuropathic|Amyloidosis I	Stern (1929), Treble (1938), Thomas, (1959), Poussaint (1962), Swedish patients were reported in (1968), Araki (1968), Andersson, (1970), Andrade (1971), Kito (1973), Hofer, (1974), Cohen (1977), Cohen, (1977), Costa (1978), Costa (1978), Benson (1980), Benson (1980), Kito (1980), Kito (1980), Lourenco, (1980), Benson (1981), Gorevic , (1982), Tawara (1983), Saraiva (1983), Benson (1984), Libbey (1984), Whitehead (1984), Sequeiros (1984), Sasaki (1984), Nakazato (1984), Benson (1984), Koeppen (1985), Saraiva (1985), Sasaki (1985), Nakazato (1985), Dwulet (1985), Saraiva (1986), Benson (1986), Ochiai (1986), Yoshioka (1986), Saraiva (1986), Maeda (1986), Nakazato (1986), Yoshioka (1986), Yamada (1987), Saraiva (1987), Ikeda (1987), Furuya (1987), Staunton (1987), Ueno (1988), Holmgren (1988), Tanaka (1988), Holmgren (1988), Saraiva (1988), Yoshioka (1989), Holt (1989), Imaizumi (1989), Sequeiros (1989), Skare (1990), Skare (1990), Sandgren (1991), Sandgren (1991), Staunton (1991), Holmgren (1992), Drugge (1993), Drugge (1993), Ducla-Soares (1994)
42690	ST3GAL5	G	A	GG	Amish Infantile Epilepsy Syndrome	Simpson (2004)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
39372	PSEN1	A	C	AA	Alzheimer disease	Group (1995), Lemere (1996), Lemere (1996), Lopera (1997), Johnson (2001)
39402	PSEN1	C	A	CC	Alzheimer disease	Yescas (2006), Murrell (2006)
39320	PSEN1	G	C	GG	Alzheimer disease	Athan (2001)
101916	ALMS1	I	D	II	Alström Syndrome	Hearn (2002), Marshall (2007)
66531	COL4A5	G	A	G	Alport syndrome	Barker (1997), Ohkubo (2003)
66892	COL4A3	G	A	GG	Alport syndrome	Nagel (2005)
66529	COL4A5	T	G	T	Alport syndrome	Barker (1996), Barker (1996)
40071	CRYAB	T	C	TT	Alpha-B Crystallinopathy with Cataract	Fardeau (1978), Vicart (1998), Liang (2003), Zobel (2003), Zobel (2003)
100861	POLG	G	A	GG	Alpers-like Hepatocerebral Syndrome	Kurt (2010)
68643	SLC16A2	T	C	TT	Allan-Herndon-Dudley Syndrome	Dumitrescu (2004)
68645	SLC16A2	T	C	TT	Allan-Herndon-Dudley Syndrome	Allan (1944), Schwartz (2005)
68644	SLC16A2	I	D	II	Allan-Herndon-Dudley Syndrome	Dumitrescu (2004)
75038	HGD	D	I	DD	Alkaptonuria	Zatkova (2000)
30342	HGD	A	T	AA	Alkaptonuria	Phornphutkul (2002)
30343	HGD	A	G	AA	Alkaptonuria	Phornphutkul (2002)
30344	HGD	A	G	AA	Alkaptonuria	Felbor (1999)
30345	HGD	T	G	TT	Alkaptonuria	Beltran-Valero (1998)
30346	HGD	G	A,T	GG	Alkaptonuria	Zatkova (2000)
30347	HGD	G	A	GG	Alkaptonuria	Rodriguez (2000)
30348	HGD	A	C	AA	Alkaptonuria	Beltran-Valero (1999)
30349	HGD	A	G	AA	Alkaptonuria	Phornphutkul (2002)
30350	HGD	T	C	TT	Alkaptonuria	Beltran-Valero (1999)
30351	HGD	G	T	GG	Alkaptonuria	Phornphutkul (2002)
30352	HGD	A	C	AA	Alkaptonuria	Beltran-Valero (1998)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
30353	HGD	A	C,G	AA	Alkaptonuria	Jorge (2002)
30354	HGD	G	T	GG	Alkaptonuria	Beltran-Valero (1999)
30355	HGD	G	A	GG	Alkaptonuria	Ladjouze-Rezig (2006)
30356	HGD	A	T	AA	Alkaptonuria	Phornphutkul (2002)
30357	HGD	T	A	TT	Alkaptonuria	Phornphutkul (2002)
30358	HGD	T	C	TT	Alkaptonuria	Beltran-Valero (1998)
30359	HGD	G	C	GG	Alkaptonuria	Phornphutkul (2002)
30360	HGD	C	T	CC	Alkaptonuria	Gehrig (1997), Muller (1999), Zatkova (2000)
30361	HGD	C	T	CC	Alkaptonuria	Higashino (1998)
30362	HGD	C	A	CC	Alkaptonuria	Rodriguez (2000)
30363	HGD	C	A	CC	Alkaptonuria	Beltran-Valero (1998)
30364	HGD	C	T	CC	Alkaptonuria	Mannoni (2004)
30365	HGD	A	G	AA	Alkaptonuria	Beltran-Valero (1998)
30366	HGD	C	G,T	CC	Alkaptonuria	Beltran-Valero (1998)
30367	HGD	C	A	CC	Alkaptonuria	Phornphutkul (2002)
30368	HGD	A	G	AA	Alkaptonuria	Beltran-Valero (1998)
30369	HGD	G	T	GG	Alkaptonuria	Beltran-Valero (1999)
30370	HGD	G	A	GG	Alkaptonuria	Fernandez-Canon (1996), Ramos (1998)
30371	HGD	T	C	TT	Alkaptonuria	Porfirio (2000)
30372	HGD	T	G	TT	Alkaptonuria	Phornphutkul (2002)
30373	HGD	C	A,T	CC	Alkaptonuria	Elcioglu (2003)
30374	HGD	G	A,T	GG	Alkaptonuria	Beltran-Valero (1999)
30375	HGD	T	C	TT	Alkaptonuria	Rodriguez (2000)
30376	HGD	A	C,G	AA	Alkaptonuria	Fernandez-Canon (1996)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
30377	HGD	G	A	GG	Alkaptonuria	Phornphutkul (2002)
30378	HGD	G	A	GG	Alkaptonuria	Rodriguez (2000)
30379	HGD	A	G	AA	Alkaptonuria	Rodriguez (2000)
30380	HGD	C	A,G,T	CC	Alkaptonuria	Bernabe (1999)
30381	HGD	C	G	CC	Alkaptonuria	Porfirio (2000)
30382	HGD	C	T	CC	Alkaptonuria	Phornphutkul (2002)
30383	HGD	T	C	TT	Alkaptonuria	Bernabe (1998), Bernabe (1999)
30384	HGD	T	C	TT	Alkaptonuria	Bernabe (1999)
30385	HGD	C	G	CC	Alkaptonuria	Mannoni (2004)
30387	HGD	C	T	CC	Alkaptonuria	Muller (1999)
30388	HGD	C	A,T	CC	Alkaptonuria	Rodriguez (2000)
30389	HGD	T	C	TT	Alkaptonuria	Phornphutkal (2002)
30390	HGD	C	A,T	CC	Alkaptonuria	Muller (1999)
30391	HGD	C	A,T	CC	Alkaptonuria	Beltran-Valero (1998)
30392	HGD	C	T	CC	Alkaptonuria	Porfirio (2000)
30393	HGD	A	G	AA	Alkaptonuria	Phornphutkal (2002)
30394	HGD	T	G	TT	Alkaptonuria	Phornphutkal (2002)
30395	HGD	C	T	CC	Alkaptonuria	Beltran-Valero (1998)
30396	HGD	C	T	CC	Alkaptonuria	Beltran-Valero (1998)
27701	GFAP	G	A,T	GG	Alexander Disease	Rodriguez (2001)
27709	GFAP	G	A,C	GG	Alexander Disease	Brenner (2001), Shiroma (2001), Rodriguez (2001), Li (2005)
27710	GFAP	C	A,G,T	CC	Alexander Disease	Brenner (2001), Li (2005)
27728	GFAP	G	A,C,T	GG	Alexander Disease	Brenner (2001), Li (2005), Perng (2006)
21340	ALB	C	T	CC	Albumin Variant	Brennan (1990)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
21368	ALB	G	A	GG	Albumin Variant	Arai (1989), Galliano (1990)
74817	TYRP1	C	G	CC	Albinism, Oculocutaneous, Type III	Manga (1997), Manga (1997), Chiang (2008), Chiang (2008)
74816	TYRP1	I	D	II	Albinism, Oculocutaneous, Type III	Boissy (1996), Manga (1997), Manga (1997)
74820	TYRP1	I	D	II	Albinism, Oculocutaneous, Type III	Rooryck (2006)
74822	TYRP1	I	D	II	Albinism, Oculocutaneous, Type III	Chiang (2009)
70707	GPR143	D	I	D	Albinism, Ocular, Type I	Johnson , (1971), Krawczak, (1991), Bassi (1995)
65298	NOTCH2	C	T	CC	Alagille Syndrome 2	McDaniell (2006)
23527	BTK	C	T	C	Agammaglobulinemia and Isolated Growth Hormone Deficiency	DNA, and in vitro splicing assays to demonstrate an intronic point mutation, (1882), Duriez (1994), Duriez (1994), Duriez (1994), Fiorini (2004)
23221	BTK	C	T	C	Agammaglobulinemia	Ohta (1994), Weers (1994), Wood (2001)
23386	BTK	C	T	C	Agammaglobulinemia	Vetrie (1993), Ohta (1994)
23447	BTK	G	T	G	Agammaglobulinemia	Bradley (1994), Jones (1996)
23496	BTK	T	A	T	Agammaglobulinemia	Zhu (1994)
24218	IGLL1	G	A	GG	Agammaglobulinemia	Minegishi (1998)
40414	ARSA	G	A	GG	Adult Metachromatic Leukodystrophy	Polten (1991), Polten (1991), Polten (1991), Polten (1991), Polten , (1991), Barth , (1993), Gieselmann , (1994), Draghia (1997)
32086	ADSL	G	A	GG	ADSL Deficiency	Marie (1999), Kmoch (1997)
32095	ADSL	G	A	GG	ADSL Deficiency	Maaswinkel-Mooij (1997), Marie (1999), Kmoch (2000), Edery (2003)
32099	ADSL	T	C	TT	ADSL Deficiency	Marie (2002)
63628	PEX26	C	T	CC	Adrenoleukodystrophy, Autosomal Neonatal Form	Matsumoto (2003), Weller (2005), Steinberg (2004)
65148	PEX5	T	G	TT	Adrenoleukodystrophy, Autosomal Neonatal Form	Dodt (1995)
65175	PEX13	T	C	TT	Adrenoleukodystrophy, Autosomal Neonatal Form	Shimozawa (1999), Liu (1999), Shimozawa (1998)
800940	MEN1	C	T	CC	Adrenocortical Tumor	Gortz (1999), Turner (2002)
14397	CYP11A1	G	A	GG	Adrenal Insufficiency, Congenital	Katsumata (2002)
11790	NR0B1	T	A	T	Adrenal Hypoplasia, Congenital	Petersen (1982), Schwartz (1997), Lalli (2000)
11819	NR0B1	C	T	C	Adrenal Hypoplasia, Congenital	Muscatelli (1994)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
10580	CYP21A2	C	T	CC	Adrenal hyperplasia	Kharrat (2004), Merino (2007)
14326	ADIPOQ	C	T	CC	Adiponectin Deficiency	Vasseur (2002)
40661	AMPD1	G	A	GG	Adenosine Monophosphate Deaminase Deficiency	Morisaki (2000), Abe (2000), GeneDx (2017)
40671	AMPD1	C	A	CC	Adenosine Monophosphate Deaminase Deficiency	Gross (2002), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2015), GeneDx (2016)
40672	AMPD1	C	A	CC	Variant of Unknown Significance (possibly associated with Adenosine Monophosphate Deaminase Deficiency)	Toyama (2004), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2015), GeneDx (2016), Center for Pediatric Genomic Medicine of Children's Mercy Hospital and Clinics (2017)
40673	AMPD1	C	G,T	CC	Adenosine Monophosphate Deaminase Deficiency	Morisaki (2000), Abe (2000), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2014), Soonchunhyang University Bucheon Hospital of Soonchunhyang University Medical Center (2016)
40662	AMPD1	T	A	TT	Adenosine Monophosphate Deaminase Deficiency	Toyama (2004), Mayo Clinic Genetic Testing Laboratories (2016), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2018), Fulgent Genetics (2018)
22803	ADA	C	T	CC	Adenosine Deaminase Deficiency, Partial	Akeson (1987), Akeson (1988), Onodera (1998)
46369	APC	G	A	GG	Adenomatous polyposis coli	Spirio (1999)
65534	APRT	C	T	CC	Adenine Phosphoribosyltransferase Deficiency	Mimori (1991)
78830	CDKN1B	G	A	GG	Acute Lymphoblastic Leukemia	Takeuchi (2002)
47117	ATM	G	A	GG	Acute lymphoblastic leukaemia	Pause (2003)
55390	ALAD	C	T	CC	Acute Hepatic Porphyria, Severe Infantile-onset	Plewinska (1991)
55394	ALAD	C	T	CC	Acute Hepatic Porphyria, Severe Infantile-onset	Plewinska (1991)
36293	ACTA1	C	A,G,T	CC	Actin myopathy	Goebel (1997), Goebel , (1997), Nowak (1999), Nowak (1999)
34817	NPR2	C	T	CC	Acromesomelic Dysplasia, Maroteaux Type	Bartels (2004)
34803	NPR2	T	A	TT	Acromesomelic Dysplasia, Maroteaux Type	Bartels (2004)
51978	CNGB3	G	A	GG	Achromatopsia 3	Sundin (2000), Kohl (2000)
54192	GNAT2	G	A	GG	Achromatopsia	Kohl (2002)
54193	GNAT2	C	T	CC	Achromatopsia	Kohl (2002)
63490	FGD1	C	T	C	Aarskog-Scott syndrome with Attention Deficit Hyperactivity Disorder	Orrico (2005)
63493	FGD1	C	T	C	Aarskog-Scott syndrome	Schwartz (2000)
53442	OPA3	C	G	CC	3-methylglutaconic Aciduria, Type III	Anikster (2001)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
60216	HSD17B3	G	A	GG	17 Beta-hydroxysteroid Dehydrogenase 3 Deficiency	Bilbao (1998)
60217	HSD17B3	C	T	CC	17 Beta-hydroxysteroid Dehydrogenase 3 Deficiency	Kohn (1985), Eckstein (1989), Geissler (1994), Rosler (1996)
60230	HSD17B3	C	T	CC	17 Beta-hydroxysteroid Dehydrogenase 3 Deficiency	Akesode (1977), Geissler (1994), Can (1998)
60213	HSD17B3	C	T	CC	17 Beta-hydroxysteroid Dehydrogenase 3 Deficiency	Moghrabi (1998)
60215	HSD17B3	T	C,G	TT	17 beta-hydroxysteroid dehydrogenase 3 deficiency	Boehmer (1999)
60218	HSD17B3	T	C	TT	17 Beta-hydroxysteroid Dehydrogenase 3 Deficiency	Moghrabi (1998)
60220	HSD17B3	G	A	GG	17 beta-hydroxysteroid dehydrogenase 3 deficiency	Boehmer (1999)
60221	HSD17B3	G	A,T	GG	17 Beta-hydroxysteroid Dehydrogenase 3 Deficiency	Geissler (1994)
60225	HSD17B3	T	C	TT	17 Beta-hydroxysteroid Dehydrogenase 3 Deficiency	Geissler (1994)
60226	HSD17B3	C	T	CC	17 Beta-hydroxysteroid Dehydrogenase 3 Deficiency	Lindqvist (2001)
60228	HSD17B3	C	G	CC	17 Beta-hydroxysteroid Dehydrogenase 3 Deficiency	Geissler (1994)
60229	HSD17B3	T	A	TT	17 beta-hydroxysteroid dehydrogenase 3 deficiency	Boehmer (1999)
12796	CYP17A1	G	T	GG	17-alpha-hydroxylase	Mussig (2005)
12797	CYP17A1	T	G	TT	17-alpha-hydroxylase	Imai (1993)
12800	CYP17A1	C	T	CC	17-alpha-hydroxylase	Brooke (2006)
12820	CYP17A1	T	A	TT	17-alpha-hydroxylase	Schwab (2005)
12821	CYP17A1	G	A	GG	17-alpha-hydroxylase	Hong (2003)
12828	CYP17A1	G	A	GG	17-alpha-hydroxylase	Yang (2006)
12831	CYP17A1	A	G	AA	17-alpha-hydroxylase	Yang (2006)
12816	CYP17A1	C	T	CC	17,20-Lyase Deficiency, Isolated	Geller (1997)
12817	CYP17A1	C	T	CC	17,20-Lyase Deficiency, Isolated	Geller (1997)
12826	CYP17A1	A	C	AA	17,20-Lyase Deficiency, Isolated	Biason-Lauber (2000)
12806	CYP17A1	T	C	TT	17,20-lyase deficiency	Biason-Lauber (2000)
12833	CYP17A1	C	A,T	CC	17,20-lyase deficiency	Biason-Lauber (2000)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
12795	CYP17A1	C	T	CC	17 alpha-hydroxylase	Suzuki (1998)
12799	CYP17A1	A	C	AA	17 alpha-hydroxylase	Di Cerbo (2002)
12801	CYP17A1	G	A	GG	17 alpha-hydroxylase / 17,20-Lyase Deficiency, Combined Complete	Laflamme (1996), Martin (2003)
12802	CYP17A1	A	G	AA	17 alpha-hydroxylase / 17,20-Lyase Deficiency, Combined Complete	Lin (1991), Jones (1992)
12803	CYP17A1	A	C	AA	17 alpha-hydroxylase	Van den Akker (2002)
12804	CYP17A1	T	A	TT	17 alpha-hydroxylase	Van den Akker (2002)
12808	CYP17A1	A	T	AA	17 alpha-hydroxylase	Taniyama (2005)
12810	CYP17A1	G	A	GG	17 alpha-hydroxylase	Ahlgren (1992)
12812	CYP17A1	A	C,T	AA	17 alpha-hydroxylase	Martin (2003)
12814	CYP17A1	G	T	GG	17 alpha-hydroxylase	Ahlgren (1992)
12815	CYP17A1	G	A,T	GG	17 alpha-hydroxylase	Van den Akker (2002)
12818	CYP17A1	G	A	GG	17 alpha-hydroxylase / 17,20-Lyase Deficiency, Combined Complete	Martin (2003), Costa-Santos (2004)
12822	CYP17A1	A	G	AA	17 alpha-hydroxylase / 17,20-Lyase Deficiency, Combined Complete	Martin (2003), Costa-Santos (2004)
12827	CYP17A1	G	A	GG	17 alpha-hydroxylase / 17,20-Lyase Deficiency, Combined Complete	Martin (2003)
12836	CYP17A1	C	A	CC	17 alpha-hydroxylase	Suzuki (1998)
12837	CYP17A1	C	T	CC	17 alpha-hydroxylase	Yazaki (1982), Yamaguchi (1997)
59912	MEFV	G	C	GG	Familial Mediterranean Fever	Haverkamp (2005)
59913	MEFV	G	A	GG	Familial Mediterranean Fever	Haverkamp (2005)
59914	MEFV	G	A,T	GG	Familial Mediterranean Fever	Touitou (2001)
59915	MEFV	C	T	CC	Familial Mediterranean Fever	Cazeneuve (2004)
59916	MEFV	T	C,G	TT	Familial Mediterranean Fever	Medlej-Hashim (2005)
59917	MEFV	C	T	CC	Familial Mediterranean Fever	Rittore-Domingo (2005)
59918	MEFV	T	A	TT	Familial Mediterranean Fever	Touitou (2001)
59919	MEFV	C	G	CC	Familial Mediterranean Fever	Akar (2001)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
59920	MEFV	C	A	CC	Familial Mediterranean Fever	Lohse (2005)
59921	MEFV	T	A	TT	Variant of Unknown Significance (questionable association with Familial Mediterranean Fever)	Medlej-Hashim (2002)
59922	MEFV	T	C,G	TT	Familial Mediterranean Fever	Aldea (2004)
59923	MEFV	C	G	CC	Familial Mediterranean Fever	Bernot (1998)
59924	MEFV	G	A,T	GG	Familial Mediterranean Fever	Arostegui (2004)
59925	MEFV	G	A	GG	Familial Mediterranean Fever	Medlej-Hashim (2005)
59926	MEFV	C	A,T	CC	Familial Mediterranean Fever	Timmann (2003)
59927	MEFV	G	C	GG	Familial Mediterranean Fever	Arostegui (2005)
59928	MEFV	C	G,T	CC	Familial Mediterranean Fever	Touitou (2001)
59929	MEFV	C	A,G,T	CC	Familial Mediterranean Fever	Lohse (2002)
59930	MEFV	T	C	TT	Familial Mediterranean Fever	Haverkamp (2005)
59931	MEFV	G	A	GG	Familial Mediterranean Fever	Bernot (1998)
59932	MEFV	G	A,T	GG	Familial Mediterranean Fever	Aldea (2004)
59933	MEFV	G	C	GG	Familial Mediterranean Fever	Haverkamp (2003)
59934	MEFV	G	A,C	GG	Familial Mediterranean Fever	Danner (2005)
59935	MEFV	G	A,T	GG	Familial Mediterranean Fever	Corbani (2005)
59936	MEFV	C	T	CC	Likely harmless variant (previously associated with Familial Mediterranean Fever)	Lohse (2002)
59937	MEFV	G	A	GG	Familial Mediterranean Fever	Salem (2006)
59938	MEFV	C	T	CC	Familial Mediterranean Fever	Aldea (2004)
59939	MEFV	C	T	CC	Familial Mediterranean Fever	Lohse (2002)
59940	MEFV	G	A	GG	Familial Mediterranean Fever	Haverkamp (2006)
59941	MEFV	G	A	GG	Familial Mediterranean Fever	Tchernitchko (2003)
59942	MEFV	T	C,G	TT	Familial Mediterranean Fever	Haverkamp (2005)
59943	MEFV	G	A	GG	Familial Mediterranean Fever	Haverkamp (2006)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
59944	MEFV	C	T	CC	Familial Mediterranean Fever	Medlej-Hashim (2005)
59945	MEFV	G	A	GG	Familial Mediterranean Fever	Aldea (2004)
59946	MEFV	G	C	GG	Familial Mediterranean Fever	Bernot (1998)
59947	MEFV	C	G,T	CC	Familial Mediterranean Fever	Baffico (2002)
59948	MEFV	G	C	GG	Familial Mediterranean Fever	Baffico (2002)
59949	MEFV	G	A	GG	Familial Mediterranean Fever	Wildhardt (2003)
59950	MEFV	G	C	GG	Familial Mediterranean Fever	Rittore-Domingo (2005)
59951	MEFV	C	A,T	CC	Familial Mediterranean Fever	Goulielmos (2006)
59952	MEFV	G	C	GG	Familial Mediterranean Fever	Goulielmos (2006)
59953	MEFV	T	A	TT	Familial Mediterranean Fever	Goulielmos (2006)
59954	MEFV	G	A	GG	Familial Mediterranean Fever	Goulielmos (2006)
59955	MEFV	A	G	AA	Familial Mediterranean Fever	Goulielmos (2006)
59884	MEFV	C	T	CC	Familial Mediterranean Fever	Schaner (2001)
59956	MEFV	T	G	TT	Familial Mediterranean Fever	Touitou (2001)
59957	MEFV	C	T	CC	Familial Mediterranean Fever	Goulielmos (2006)
59958	MEFV	C	T	CC	Familial Mediterranean Fever	Dode (2000)
59959	MEFV	C	T	CC	Familial Mediterranean Fever	Touitou (2001)
59882	MEFV	C	G	CC	Familial Mediterranean Fever	Consortium (1997)
59883	MEFV	C	A,G,T	CC	Familial Mediterranean Fever	Consortium (1997)
59960	MEFV	T	C,G	TT	Familial Mediterranean Fever	Dode (2000)
59961	MEFV	G	A	GG	Familial Mediterranean Fever	Booth (1998)
59962	MEFV	T	C	TT	Familial Mediterranean Fever	Goulielmos (2006)
59963	MEFV	G	C	GG	Familial Mediterranean Fever	Notarnicola (2001)
59879	MEFV	C	T	CC	Familial Mediterranean Fever	Consortium (1997), Touitou (2001)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
59880	MEFV	T	A,C	TT	Familial Mediterranean Fever	Consortium (1997)
59964	MEFV	T	A	TT	Familial Mediterranean Fever	Lohse (2002)
59965	MEFV	T	C	TT	Likely associated with Familial Mediterranean Fever	Bernot (1998), Knight Diagnostic Laboratories of the Oregon Health and Sciences University (2015), Laboratory Corporation of America (2015), Centre for Mendelian Genomics of the University Medical Centre Ljubljana (2016), GeneReviews (2016), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2016), GeneDx (2017), Praxis fuer Humangenetik Tuebingen (2017), Invitae (2017), Counsyl (2017)
59966	MEFV	T	A	TT	Familial Mediterranean Fever	Goulielmos (2006)
59967	MEFV	G	A	GG	Familial Mediterranean Fever	Aksentijevich (2003)
59968	MEFV	C	T	CC	Familial Mediterranean Fever	Touitou (2001)
59969	MEFV	G	A	GG	Familial Mediterranean Fever	Goulielmos (2006)
59970	MEFV	G	C	GG	Familial Mediterranean Fever	Medlej-Hashim (2002)
59876	MEFV	A	G,T	AA	Familial Mediterranean Fever	International FMF Consortium (1997), LabCorp (2011), Center for Pediatric Genomic Medicine of Children's Mercy Hospital and Clinics (2015), Emory Genetics Laboratory of Emory University (2015), GeneReviews (2016), GeneDx (2016)
59971	MEFV	G	C	GG	Familial Mediterranean Fever	Goulielmos (2006)
59972	MEFV	C	A	CC	Familial Mediterranean Fever	Samuels (1998), Bernot (1998), Mikula (2008), Caglayan (2010), Shohat (2011), Nikibakhsh (2012), Yolbas (2012)
59973	MEFV	G	A	GG	Familial Mediterranean Fever	Goulielmos (2006)
59974	MEFV	C	T	CC	Familial Mediterranean Fever	Bernot (1998)
59975	MEFV	C	G	CC	Familial Mediterranean Fever	Takabe (2006)
59976	MEFV	G	T	GG	Familial Mediterranean Fever	Goulielmos (2006)
59979	MEFV	G	A	GG	Variant of Unknown Significance (questionable association with Familial Mediterranean Fever)	Dumont (2005)
59980	MEFV	G	C	GG	Variant of Unknown Significance (questionable association with Familial Mediterranean Fever)	Touitou (2004)
59983	MEFV	T	C,G	TT	Familial Mediterranean Fever	Lohse (2005)
802000	HNRNPH2	C	T	CC	Severe Neurodevelopmental Disorder: Bain type (DETECTION ALERT: Contact Dr. Wendy Chung of Columbia University by emailing altruist@sequencing.com)	Bain (2016), GeneDx (2017), Institute of Human Genetics of Klinikum rechts der Isar (2017)
802002	HNRNPH2	G	A	GG	Severe Neurodevelopmental Disorder: Bain type (DETECTION ALERT: Contact Dr. Wendy Chung of Columbia University by emailing altruist@sequencing.com)	Bain (2016), GeneDx (2017)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
802003	HNRNPH2	G	A	GG	Uncertain significance (DETECTION ALERT: Contact Dr. Wendy Chung of Columbia University in New York by emailing altruist@sequencing.com)	GeneDx (2017)
802004	HNRNPH2	I	D	II	Uncertain significance (DETECTION ALERT: Contact Dr. Wendy Chung of Columbia University in New York by emailing altruist@sequencing.com)	GeneDx (2017)
44966	BRCA1	A	C,G	AA	Increased Risk of Breast and/or Ovarian Cancer	Sekine (2001)
45022	BRCA1	G	A	GG	Increased Risk of Breast and/or Ovarian Cancer	Wang (2000)
45024	BRCA1	C	A,T	CC	Increased Risk of Breast and/or Ovarian Cancer	Sekine (2001)
45065	BRCA1	T	A	TT	Increased Risk of Breast and/or Ovarian Cancer	Pohlreich (2005)
45067	BRCA1	T	A	TT	Increased Risk of Breast and/or Ovarian Cancer	Gayther (1999)
45072	BRCA1	A	C	AA	Increased Risk of Breast and/or Ovarian Cancer	Xiaoman (1999)
45084	BRCA1	C	A,T	CC	Increased Risk of Breast and/or Ovarian Cancer	Tworek (1999)
45108	BRCA1	G	A	GG	Increased Risk of Breast and/or Ovarian Cancer	Kashima (2000)
45109	BRCA1	A	T	AA	Increased Risk of Breast and/or Ovarian Cancer	Pal (2005)
45111	BRCA1	G	A	GG	Increased Risk of Breast and/or Ovarian Cancer	Borg (1999)
45130	BRCA1	G	A,T	GG	Increased Risk of Breast and/or Ovarian Cancer	Majdak (2005)
45172	BRCA1	C	A	CC	Increased Risk of Breast and/or Ovarian Cancer	Risch (2001)
45207	BRCA1	C	A	CC	Increased Risk of Breast and/or Ovarian Cancer	Borg (1999)
45305	BRCA1	T	A,C,G	TT	Increased Risk of Breast and/or Ovarian Cancer	Sekine (2001)
45308	BRCA1	C	A,T	CC	Increased Risk of Breast and/or Ovarian Cancer	Perkowska (2003)
45309	BRCA1	C	A,G,T	CC	Increased Risk of Breast and/or Ovarian Cancer	Ozcelik (1999)
45317	BRCA1	A	C,G	AA	Increased Risk of Breast and/or Ovarian Cancer	Risch (2001)
45347	BRCA1	C	A,G,T	CC	Increased Risk of Breast and/or Ovarian Cancer	Kashima (2000)
45355	BRCA1	C	T	CC	Increased Risk of Breast and/or Ovarian Cancer	Ostrow (2001)
45506	BRCA2	T	C	TT	Increased Risk of Breast and/or Ovarian Cancer	Jakubowska (2003)
45594	BRCA2	C	A	CC	Increased Risk of Wilms Tumor	De Benedetti (1998), Reid (2005)

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Your Genetic Testing Data

Variant ID	Gene	No Risk	Risk	Your Genetic Makeup	Condition / Trait Assessed	Reference(s)
45597	BRCA2	C	G	CC	Increased Risk of Breast and/or Ovarian Cancer	Risch (2001)
45609	BRCA2	C	T	CC	Increased Risk of Breast and/or Ovarian Cancer	Pal (2005)
45635	BRCA2	A	G	AA	Increased Risk of Ocular melanoma	Scott (2002)
45662	BRCA2	G	T	GG	Increased Risk of Liver cancer	Katagiri (1996)
45673	BRCA2	G	A	GG	Increased Risk of Breast and/or Ovarian Cancer	Roth (1998)
45683	BRCA2	A	G	AA	Increased Risk of Fallopian tube cancer	Baudi (2003)
45686	BRCA2	G	A	GG	Increased Risk of Pancreatic cancer	Hahn (2003)
45724	BRCA2	A	G	AA	Increased Risk of Breast and/or Ovarian Cancer	Chen (2006)
800852	BRCA2	C	A	CC	Increased Risk of Glioblastoma	De Benedetti (1998), Reid (2005)
800853	BRCA2	C	A	CC	Increased Risk of Pre-B-Cell Acute Lymphoblastic Leukemia	De Benedetti (1998), Reid (2005)
800854	BRCA2	C	A	CC	Increased Risk of Medulloblastoma	De Benedetti (1998), Reid (2005)
801244	BRCA1	G	A	GG	Increased Risk of Prostate Cancer	Zuhlke (2004)
801268	BRCA2	I	D	II	Increased Risk of Wilms Tumor	Hirsch (2004), Reid (2005), Alter (2007)
16064	**ATP8B1**	**C**	**A,T**	**CT**	**Likely harmless variant (previously associated with Intrahepatic Cholestasis of Pregnancy)**	**Klomp (2004), Mullenbach (2005), EGL Genetic Diagnostics of Eurofins Clinical Diagnostics (2017), GeneDx (2017), Genomic Research Center of Shahid Beheshti University of Medical Sciences (2018)**
850038	**CFTR**	**I**	**D**	**ID**	**Cystic Fibrosis**	**Kerem (1989), European Working Group on CF Genetics (1990), ozen (1990), Braekeleer (1991), Daigneault (1991), Rozen (1992), Kerem (1990), Wauters (1991), Gille (1991), Lerer (1992), Casals (1992), Ballabio (1990), Grebe (1992), Casals (1997), Russo (1995), Grebe (1994), Rose (2005), Wainwright (2015), Children's Hospital of Philadelphia (2016), Mendelics (2018), FRIGE's Institute of Human Genetics (2019), Invitae (2019), Johns Hopkins University (2019), Klinikum rechts der Isar (2020)**

SEQUENCING

Analysis Alerts

This page contains important information about to the analysis of your genetic data. The alerts below are provided so that you are aware of any limitations that were identified during the analysis of your data. You may want to discuss the alerts with your healthcare professional.

Additional information about alerts: https://sequencing.com/knowledge-center/app-alerts

Ambiguity in genetic data

✓ **No ambiguity exists in the genetic data.**

Incomplete genetic data

While the genetic data file contained a lot of useful data, it did not contain enough data for a complete analysis.

As a generic example, this app may attempt to analyze 20 different genetic variants in-order to provide a risk assessment for condition X. While many genetic data files do contain data for all 20 variants, some genetic data files only contain data for 18 out of the 20. Some files may even only provide data on just two or three variants out of the 20.

The analysis conducted by this app will use whatever data is available and will also provide this alert if data isn't available for 100% of the variants that the app is configured to analyze.

The results may be different if more data was available. The genetic data was incomplete for the diseases, conditions and traits listed below. If not listed below then the data for that disease, condition or trait was complete.

Disease, Condition, Trait or Medication	**Available Data** *used for analysis*	**Your Analysis** *available variants*	**Required for Full Analysis** *total known variants*
Leigh Syndrome	96%	23	24
Deafness, Nonsyndromic Sensorineural	91%	21	23
Obesity, Early-onset	67%	2	3
Obesity	60%	6	10

No genetic data (analysis not possible)

There was no genetic data available, and no genetic analysis could occur, for the following diseases, conditions, traits or medications.

This table differs from the 'Incomplete genetic data' table above because genetic analysis was still performed for everything listed above. This was possible because for those diseases, conditions, traits or medications,, there was at least *some* genetic data available.

Disease, Condition, Trait or Med. ***Analysis not possible***	**Available Data**	**Your Analysis** *available variants*	**Required for Full Analysis** *total known variants*
Myopathy, Mitochondrial, Late-onset	0%	0	1
Leigh Syndrome Due To Mitochondrial Complex I Deficiency	0%	0	4
Possibly Associated With Leigh Syndrome	0%	0	1
Cerebellar Ataxia, Cataracts, And Diabetes Mellitus	0%	0	1
MERRF/MELAS Overlap Syndrome	0%	0	1
Pigmentary Retinopathy And Sensorineural Deafness	0%	0	1
Striatonigral Degeneration, Infantile, Mitochondrial	0%	0	1
Ataxia And Polyneuropathy, Adult-onset	0%	0	1
MERRF Syndrome	0%	0	2
Deafness, Aminoglycoside-induced And Mitochondrial Cytochrome C Oxidase Deficiency	0%	0	1
Deafness, Aminogycloside-induced	0%	0	1
Acquired Idiopathic Sideroblastic Anemia	0%	0	2
Myopathy, Mitochondrial, With Diabetes Mellitus	0%	0	1
Myopathy	0%	0	1

Disease, Condition, Trait or Med. ***Analysis not possible***	**Available Data**	**Your Analysis** *available variants*	**Required for Full Analysis** *total known variants*
Sensorineural Deafness And Migraine	0%	0	1
Dystonia, Adult-onset	0%	0	1
Leber Hereditary Optic Neuropathy	0%	0	3
Leber Hereditary Optic Neuropathy, Severe	0%	0	1
Obesity, Autosomal Dominant	0%	0	8
Obesity, Severe, Early-onset	0%	0	1
MELAS Syndrome	0%	0	2
Melas Syndrome	0%	0	1
Neonatal Death|Leigh Syndrome	0%	0	1
Leber Optic Atrophy	0%	0	3
Exercise Intolerance	0%	0	1