YOUR FAVORITE GENOTYPES

YOUR STARRED GENOTYPES

This list will be anonymously persisted across your sessions.

★ rs2464196(T;T) bad:4.42 10.09% **~2x increased lung cancer risk**

rs2464196 is a SNP that was originally associated with C-reactive protein levels, and given other findings that suggest C-reactive protein levels may be predictive of cancer risk, was thought to be linked to cancer risk.

A large study pooling data from 3 Finnish studies totaling over 18,000 individuals concluded that while this SNP is not likely to be causative (relative to cancer), it and one other CRP SNP (rs1169300) are associated with increased risk for lung cancer. The odds ratio for minor alleles of either SNP was about 1.5x and 2x for heterozygotes and homozygotes, respectively. One other CRP SNP (rs1892534) was associated with increased overall cancer risk. CRP SNPs were not associated with colorectal, prostate or breast cancer risk.

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Gene related topics:

cancer(5.00) · lung cancer(5.00) · UNKNOWN(2.00) · diabetes mellitus type 1 diabetes mellitus type 2(2.00) · breast cancer(1.00) · OTHER(1.00) · METABOLIC(0.64) · gamma glutamyltransferase(0.64) · diabetes mellitus type ii diabetes mellitus type 2(0.57) · IMMUNE(0.36) · inflammation(0.36) · diabetes type 2(0.28) · CARDIOVASCULAR(0.16) · c reactive protein(0.16) · diabetes mellitus type 2(0.09) · diabetes mellitus(0.07) · CANCER(0.03) · pancreatic neoplasms(0.03)

Relevant papers and external links:

PMID 20727736

PHARMGKB PA162411157

Evidence: PubMed ID:18439552In a candidate gene-based association analysis of 4333 European-descended, age 65 or older, subjects from the CHS (Cardiovascular Health Study), this was one of the SNPs most strongly associated with plasma C-reactive protein levels. The minor allele is part of a haplotype of frequency 30% which was associated with lower levels of CRP.

dbSNP: rs2464196 Genecard: HNF1A chromosome 12 SNPedia: rs2464196(T;T)

★ i3000001(D;I) bad:4.29 **Cystic Fibrosis carrier**

also known as rs113993960 you have one copy of the variation.

Gene related topics:

cystic fibrosis(5.00)

Relevant papers and external links:

SNPedia: i3000001(D;I)

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★ rs17602729(T;T) bad:3.84 0.15%(rare) **AMPD1 deficiency homozygous**

This is found in ~2% of all caucasians. The majority of people with the AMPD gene are asymptomatic, but in response to vigorous exercise, others have symptoms including early fatigue, muscle pain and muscle cramping.

External Link

External Link

External Link

External Link

This position is odd. Under the older reference genome build36, it was on the plus strand; in build37 it is on the minus strand. This makes it very prone to some ambiguous flip confusion in the literature, plus, 23andMe users have self-reported the flipped form. IN addition, instead of the usual SNP listed as having 2 alleles, all 4 alleles are reported in dbSNP, even if this is probably due to the strand changes. dbSNP currently says the normal allele is C, and T is the rare geno which causes a premature stop, early in the gene. We at SNPedia suspect that the A & G alleles are not common enough to have ever been genuinely reported, but for de-novo mutations each allele would be non-synonymous and result in a different amino acid. External Link

rs17602729, a SNP located in the AMPD1 gene and also known as 'C34T', has at times been called the "most prevalent genetic disease mutation", at least in Caucasians. Perhaps up to 10% of Caucasians and African-American carry one C34T allele (i.e. carry one rs17602729(A) allele) - and actually, most of them are unaware of any medically related issues since they don't typically have any particular symptoms that would warrant a trip to the doctor.

So what's the issue? The AMPD1 gene encodes the enzyme adenosine monophosphate deaminase, which is one of the key enzymes used to process the energy source ATP. The C34T variation causes a premature stop in the protein, leading to a nonfunctional AMPD1 enzyme. Some individuals - but by no means all or even a majority apparently - who are AMPD1 deficient get muscle cramps and pains when they exert themselves. The most common genotype bringing this about is rs17602729(A;A), i.e. the individuals who carry two copies of the C34T allele and therefore have no functioning AMPD1 allele. It is not known why only some of C34T homozygotes experience muscle myalgia.

Heterozygotes, i.e. rs17602729(A;G) individuals, do not seem to suffer any exercise or fitness related deficit. In fact, one of the best elite Caucasian distance runners is known to be a C34T heterozygote.

In fact, heterozygotes for rs17602729 may even have advantages over those lacking a C34T allele. The most striking study reported that C34T heterozygotes are 8.6 fold (CI: 3.05 - 23.87) more likely to survive for more than 5 years without needing a heart transplant after a congestive heart failure related hospitalization than the wild-type homozygotes (i.e. rs17602729(G;G) homozygotes). Similarly, C34T heterozygotes with coronary artery disease seem to fare better than wild-type homozygotes.

This is one of the SNPs reported by NutraHacker.

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Gene related topics:

pain(1.00) · heart failure(1.00) · coronary artery disease(1.00) · heart transplant(1.00) · METABOLIC(0.80) · glycogen storage disease type v(0.80) · CARDIOVASCULAR(0.67) · congestive heart failure(0.67) · NORMALVARIATION(0.36) · endurance performance(0.36)

Relevant papers and external links:

PMID 11331279
PMID 16505069
PMID 10086964
PMID 11028479
OMIM 102770

dbSNP: rs17602729 Genecard: AMPD1 chromosome 1 SNPedia: rs17602729(T;T)

★ rs3865444(T;T) good:3.60 4.46%(rare) **slight reduction in risk for Alzheimer's disease**

see citations at rs3865444

The less common allele for this SNP, rs3865444(T), is associated with a slight decrease in risk for Alzheimer's disease, on the order of 0.9x (i.e. a 10% decrease in risk). One visualization of such studies is available here on the AlzGene website.

Alzheimer's disease associated, based on large 2011 study

Established associations:

alzheimers disease **you have 0 copies of the risk allele G**

(risk allele=G, odds ratio/beta 1.06 [1.04-1.1] with pval 3E-6, pubmedid=24162737)

Relevant papers and external links:

PMID 24162737
Meta-analysis of 74,046 individuals identifies 11 new susceptibility loci for alzheimer's disease.

dbSNP: rs3865444 GWAS Catalog: rs3865444 Genecard: CD33 chromosome 19

SNPedia: rs3865444(T;T)

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★ rs2305480(T;T) **bad:3.44** **8.22%** **if 4 years old or younger, ~3x increased asthma risk if exposed to smoke**

rs2305480 is one of several SNPs from a region on chromosome 17q21 that has been linked to risk for asthma.

A total of 651 French patients with asthma, from among 1,621 subjects in 388 families, were analyzed for SNPs in the (17q21) region. An association was found between this SNP and early-onset asthma (but not with later onset asthma) which was significant after correction at $p < 0.001$. Upon further study, this association was seen primarily only in children exposed to tobacco smoke. The odds ratio was 2.77, based on the most likely assumption that this risk is associated only with the recessive homozygous state (and not the heterozygous state, which is still possible).

Established associations:

ulcerative colitis **you have 2 copies of the risk allele T**

(risk allele=T, odds ratio/beta 1.15 with pval 3E-8, pubmedid=20228799)

childhood onset asthma **you have 0 copies of the risk allele C**

(risk allele=C, odds ratio/beta 1.32 [1.23-1.39] with pval 6E-23, pubmedid=24241537)

asthma **you have 0 copies of the risk allele C**

(risk allele=C, odds ratio/beta 1.18 [1.11-1.23] with pval 1E-7, pubmedid=20860503)

Gene related topics:

asthma(5.00)

Relevant papers and external links:

PMID 20860503
A large-scale, consortium-based genomewide association study of asthma.

PMID 20228799
Genome-wide association identifies multiple ulcerative colitis susceptibility loci.

PMID 24241537
A genome-wide association study identifies cdhr3 as a susceptibility locus for early childhood asthma with severe exacerbations.

PMID 18923164

PHARMGKB PA162356827
Evidence: PubMed ID:18923164This variant in the 17q21 region was strongly associated (P<0.001) with early-onset asthma. The study shows that the risk increases further by early-life exposure to environmental tobacco smoke.

OMIM 611403

OMIM 612380

dbSNP: rs2305480 GWAS Catalog: rs2305480 Genecard: GSDMB chromosome 17

SNPedia: rs2305480(T;T)

★ gs191 **bad:3.43** **problem metabolizing NSAIDs**

impaired NSAID drug metabolism, which is a risk factor for gastrointestinal bleeding when taking any of these medications: aceclofenac, celecoxib, diclofenac, ibuprofen, indomethazine, lornoxicam, meloxicam, naproxen, piroxicam, tenoxicam and valdecoxib.

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You have one of these
CYP2C8*3 (rs11572080 and rs10509681)
CYP2C9*2 (rs1799853)
CYP2C9*3 (rs1057910)

Gene related topics:

metabolism(1.00) · gastrointestinal bleeding(1.00)

Relevant papers and external links:

PMID 19422321

SNPedia: gs191

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★ rs5031017(G;T) info:3.38 **One copy of CYP2A6*5 non-functioning variant; impaired nicotine metabolism**

rs5031017 in the CYP2A6 gene, also known as G479V, 1436G>T or 6582G>T, the T allele indicates the CYP2A6*5 inactive form of the gene.

reports that poor metabolizers (such as those with inactive forms of CYP2A6), among male smokers and compared to normal smokers, smoked fewer cigarettes, took longer to smoke regularly, and if they had stopped smoking, waited a shorter period of time before making the decision. Unfortunately, this group also had a lower likelihood of smoking cessation.

Smoking

Gene related topics:

CHEMDEPENDENCY(11.00) · nicotine(11.00) · metabolism(5.00) · METABOLIC(3.00) · enzyme activity(3.00) · cyp2a6(2.00) · smoking behavior(1.48) · CANCER(1.00) · lung cancer smoking behavior(1.00) · smoking cessation(1.00) · smoking(0.21) · OTHER(0.12) · INFECTION(0.10) · hiv infections(0.10) · gastric cancer(0.10) · neoplasms(0.03) · esophageal cancer(0.03)

Relevant papers and external links:

PMID 21205058

dbSNP: rs5031017 Genecard: CYP2A6 chromosome 19 SNPedia: rs5031017(G;T)

★ rs464049(T;T) bad:3.38 15.35% **increased risk of schizophrenia in limited study**

Associated with schizophrenia in a study of Croatians. Due to the limited number and diversity of the studied population, more study is needed to confirm any link. The T allele was associated with an increased risk of schizophrenia.

A network of dopaminergic gene variations implicated as risk factors for schizophrenia.

Family based association study of pediatric bipolar disorder and the dopamine transporter gene (SLC6A3).

Genome-wide and candidate gene association study of cigarette smoking behaviors.

Financial and psychological risk attitudes associated with two single nucleotide polymorphisms in the nicotine receptor (CHRNA4) gene.

Two loci control tuberculin skin test reactivity in an area hyperendemic for tuberculosis.

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Gene related topics:

PSYCH(9.95) · attention deficit hyperactivity disorder(9.95) · adhd attention deficit hyperactivity disorder(3.41) · schizophrenia methamphetamine abuse(2.00) · CHEMDEPENDENCY(1.25) · smoking behavior(1.25) · perceptual disorders(1.00) · bipolar disorder(1.00) · tuberculosis(1.00) · nicotine(1.00) · personality(0.98) · cocaine related disorders(0.67) · psychosis(0.60) · delirium(0.57) · impulsivity(0.57) · tourette syndrome(0.47) · adhd(0.40) · NEUROLOGICAL(0.40) · parkinson s disease(0.40) · REPRODUCTION(0.36) · prenatal exposure delayed effects(0.36) · schizophrenia(0.27) · substance related disorders(0.27) · heroin dependence(0.25) · bulimia(0.24) · alcohol(0.21) · dyslexia(0.19) · alcohol abuse(0.17) · alcoholism(0.16) · personality traits(0.12) · migraine disorders(0.08) · depression(0.07) · obsessive compulsive disorder(0.06) · autism(0.05) · smoking(0.05) · PHARMACOGENOMIC(0.02)

Relevant papers and external links:

PMID 19673036
PMID 18045777
PMID 18361424
PMID 19247474
PMID 19693267
PMID 19901083

dbSNP: rs464049 Genecard: SLC6A3 chromosome 5 SNPedia: rs464049(T;T)

★ rs6920220(A;G) bad:3.34 17.11% **1.2x risk Rheumatoid Arthritis**

rs6920220 has been reported in a large study to be associated with rheumatoid arthritis.

The risk allele (oriented to the dbSNP entry) is (A); the odds ratio associated with heterozygotes is 1.20 (CI 1.06-1.36), and for homozygotes, 1.72 (CI 1.33-2.22).

rheumatoid arthritis
rs6920220 [P= 2.6 x 10(-6), OR 1.22 (1.13-1.33)].
rs5029937
rs13207033 protective [P= 0.0001, OR 0.86 (0.8-0.93)] perfectly correlated with rs10499194
The combination of the carriage of both risk alleles of rs6920220 and rs5029937 together with the absence of the protective allele of rs13207033 was strongly associated with RA when compared to carriage of none [OR of 1.86 (95% CI) (1.51-2.29)]. This equates to an effect size of 1.50 (95% CI 1.21-1.85)

Lack of association or interactions between the IL-4, IL-4Ralpha and IL-13 genes, and rheumatoid arthritis.

Rheumatoid arthritis susceptibility loci at chromosomes 10p15, 12q13 and 22q13.

Gene variants influencing measures of inflammation or predisposing to autoimmune and inflammatory diseases are not associated with the risk of type 2 diabetes.

Rheumatoid arthritis: a view of the current genetic landscape.

Genetic variants near TNFAIP3 on 6q23 are associated with systemic lupus erythematosus.

Genome-wide scan reveals association of psoriasis with IL-23 and NF-kappaB pathways.

Genetic risk factors for rheumatoid arthritis differ in Caucasian and Korean populations.

Analysis of TNFAIP3, a feedback inhibitor of nuclear factor-kappaB and the neighbor intergenic 6q23 region in rheumatoid arthritis susceptibility.

6q23 polymorphisms in rheumatoid arthritis Spanish patients.

Meta-analysis and imputation identifies a 109 kb risk haplotype spanning TNFAIP3 associated with lupus nephritis and hematologic manifestations.

Complex genetic association of 6q23 with autoimmune rheumatic conditions.

Association of common polymorphisms in known susceptibility genes with rheumatoid arthritis in a Slovak population using osteoarthritis patients as controls.

Overlap of disease susceptibility loci for rheumatoid arthritis and juvenile idiopathic arthritis.

A large-scale replication study identifies TNIP1, PRDM1, JAZF1, UHRF1BP1 and IL10 as risk loci for systemic lupus erythematosus.

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Pathway analysis of GWAS provides new insights into genetic susceptibility to 3 inflammatory diseases.

Representation of genetic association via attributable familial relative risks in order to identify polymorphisms functionally relevant to rheumatoid arthritis.

Conditional analysis of the major histocompatibility complex in rheumatoid arthritis.

Lack of association between the rs6920220 (G/A) polymorphism of the 6q23 region and biopsy-proven giant cell arteritis.

Evaluation of the rheumatoid arthritis susceptibility loci HLA-DRB1, PTPN22, OLIG3/TNFAIP3, STAT4 and TRAF1/C5 in an inception cohort.

AntEpiSeeker: detecting epistatic interactions for case-control studies using a two-stage ant colony optimization algorithm.

Genetic variants in the prediction of rheumatoid arthritis.

Investigation of rheumatoid arthritis susceptibility genes identifies association of AFF3 and CD226 variants with response to anti-tumour necrosis factor treatment.

Association of TNFAIP3 polymorphism with susceptibility to systemic lupus erythematosus in a Japanese population.

Most common SNPs associated with rheumatoid arthritis in subjects of European ancestry confer risk of rheumatoid arthritis in African-Americans.

Meta-analysis of genome-wide association studies in celiac disease and rheumatoid arthritis identifies fourteen non-HLA shared loci.

A Tunisian case-control association study of a 6q polymorphism in rheumatoid arthritis.

Associations between TNFAIP3 gene polymorphisms and rheumatoid arthritis: a meta-analysis.

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Established associations:

rheumatoid arthritis **you have 1 copies of the risk allele A** (risk allele=A, odds ratio/beta 1.22 [1.16-1.29] with pval 9E-13, pubmedid=20453842)
ulcerative colitis **you have 1 copies of the risk allele A** (risk allele=A, odds ratio/beta 1.14 [1.09-1.20] with pval 8E-17, pubmedid=21297633)
inflammatory bowel disease **you have 1 copies of the risk allele A** (risk allele=A, odds ratio/beta 1.102 [1.064-1.141] with pval 1E-21, pubmedid=23128233)

Gene related topics:

arthritis(5.00) · rheumatoid arthritis(5.00) · celiac disease(1.00) · systemic lupus erythematosus(1.00) · giant cell arteritis(1.00) · lupus nephritis(1.00) · osteoarthritis(1.00) · lupus erythematosus(1.00) · type 2 diabetes(1.00) · psoriasis(1.00) · lupus(1.00) · inflammation(1.00)

Relevant papers and external links:

PMID 20453842
Genome-wide association study meta-analysis identifies seven new rheumatoid arthritis risk loci.

PMID 21297633
Meta-analysis identifies 29 additional ulcerative colitis risk loci, increasing the number of confirmed associations to 47.

PMID 23128233
Host-microbe interactions have shaped the genetic architecture of inflammatory bowel disease.

PMID 17554300

PMID 19417005

OMIM 612378

OMIM 180300

PHARMGKB PA161613785
Evidence: PubMed ID:17982455; PubMed ID:17982456This SNP has been associated with risk of rheumatoid arthritis by replicated GWAS.

PMID 18625055

PMID 18794857

PMID 18853133

PMID 18987647

PMID 19165918

PMID 19169254

PMID 19180477

PMID 19292917

PMID 19321514

PMID 19387456

PMID 19439038

PMID 19445664

PMID 19674979

PMID 19838195

PMID 19956648

PMID 20017963

PMID 20018027

PMID 20231195

PMID 20353580

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PMID 20426808
PMID 20439292
PMID 20444755
PMID 20617138
PMID 20973039
PMID 21383967
PMID 21833526
PMID 22402800

dbSNP: rs6920220 GWAS Catalog: rs6920220 Genecard: nan chromosome 6

SNPedia: rs6920220(A;G)

★ rs17782313(C;C) bad:3.24 5.76% **adults likely to be 0.44 BMI units higher**

A study of 60,000 adults indicates that rs17782313(C) alleles are associated with higher body mass index (BMI), with even greater effect in children. The average increase in BMI is 0.22 BMI units (p = 2.8 x 10-15).

Note that rs10871777 is considered an equivalent SNP in Caucasian and Asian populations. The rs17782313(C) allele is equivalent to the rs10871777(G) allele.

The rs17782313(C) allele is significantly associated with higher intakes of total energy and dietary fat. In addition, the SNP was related to greater long-term weight change and increased risk of diabetes in women.

rs17782313(C) more snacking, however rs1421085(C) did not influence eating behavior

obesity and type-2 diabetes
rs1121980
rs7498665
rs4752856
rs17782313
rs2815752
rs10938397

Established associations:

body height **you have 0 copies of the risk allele T**

(risk allele=T, odds ratio/beta 0.028 unit decrease with pval 4E-11, pubmedid=20881960; risk

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allele=T, odds ratio/beta 0.039 [0.025-0.053] unit decrease with pval 2E-8, pubmedid=25429064)

obesity **you have 2 copies of the risk allele C**

(risk allele=C, children odds ratio/beta 1.22 [1.05-1.40] with pval 5E-15, pubmedid=19151714)

body mass index **you have 2 copies of the risk allele C**

(risk allele=C, odds ratio/beta 0.2 [0.12-0.28] kg/m2 increase with pval 5E-18, pubmedid=19079261; risk allele=C, odds ratio/beta 0.05 [0.04-0.06] unit increase with pval 3E-15, pubmedid=18454148)

Gene related topics:

bmi(5.00) · METABOLIC(2.73) · obesity(2.73) · PSYCH(2.00) · binge eating(2.00) · body mass(1.00) · weight(1.00) · body mass index(1.00) · body weight obesity(0.71) · obesity morbid(0.33) · diabetes mellitus type 2 obesity(0.31) · diabetes mellitus type ii diabetes mellitus type 2(0.09)

Relevant papers and external links:

PMID 19079261
Six new loci associated with body mass index highlight a neuronal influence on body weight regulation.

PMID 20881960
Hundreds of variants clustered in genomic loci and biological pathways affect human height.

PMID 25429064
Meta-analysis of genome-wide association studies of adult height in east asians identifies 17 novel loci.

PMID 19151714
Genome-wide association study for early-onset and morbid adult obesity identifies three new risk loci in european populations.

PMID 18454148

PMID 18697794

PMID 19153581

PMID 19164386

OMIM 601665

OMIM 155541

PHARMGKB PA164740184
Evidence: PubMed ID:19079261; Web Resource:External Link results: Six new loci associated with body mass index highlight a neuronal influence on body weight regulation. (Initial Sample Size: 32,387 individuals; Replication Sample Size: 59,092 individuals); (Region: 18q21.32; Reported Gene(s): MC4R; Risk Allele: rs17782313-C); (p-value= 0.000000000000000005).This variant is associated with Body mass index.

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PHARMGKB PA162356421
Evidence: PubMed ID:18454148; Web Resource:External Link Results: Common variants near MC4R are associated with fat mass, weight and risk of obesity (Initial Sample Size: 16,876 individuals; Replication Sample Size: 60,352 individuals; Risk Allele: rs17782313-C). This variant is associated with Body mass index.

PHARMGKB PA164740070
Evidence: PubMed ID:19151714; Web Resource:External Link results: Genome-wide association study for early-onset and morbid adult obesity identifies three new risk loci in European populations. (Initial Sample Size: 695 obese adults, 685 obese children, 731 lean adults, 685 lean children; Replication Sample Size: 1,171 obese adults, 896 obese children, 1,114 lean adults, 1,297 lean children, 4,417 adults, 5,291 children); (Region: 18q21.32; Reported Gene(s): MC4R; Risk Allele: rs17782313-C); (p-value= 0.000000000000005).This variant is associated with Obesity.

PHARMGKB PA162565810
Evidence: PubMed ID:19174833The analyses of genome-wide association data from 1,380 Europeans with early-onset and morbid adult obesity and 1,416 age-matched normal-weight controls found a strong association of this variant (rs17782313) downstream of the MC4R for risk of pooled childhood and adult severe obesity.

dbSNP: rs17782313 GWAS Catalog: rs17782313 Genecard: nan chromosome 18

SNPedia: rs17782313(C;C)

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★ **rs2547231(T;T)** **info:3.21** **82.84%** **Increased blood metabolite levels**

5alpha-androstan-3beta,17beta-diol disulfate - 0.081 unit increase

X-12850 - 0.059 unit increase

X-12456 - 0.041 unit increase

X-11440/4-androsten-3beta,17beta-diol disulfate 2 - 0.141 unit increase

Established associations:

blood metabolite measurement **you have 2 copies of the risk allele T**

(risk allele=T, X-12456 odds ratio/beta 0.041 [0.029-0.053] unit increase with pval 3E-11, pubmedid=24816252; risk allele=T, X-12850 odds ratio/beta 0.059 [0.045-0.073] unit increase with pval 4E-16, pubmedid=24816252; risk allele=T, 5alpha-androstan-3beta,17beta-diol disulfate odds ratio/beta 0.081 [0.065-0.097] unit increase with pval 3E-22, pubmedid=24816252; risk allele=T, X-11440/4-androsten-3beta,17beta-diol disulfate 2 odds ratio/beta 0.141 [0.13-0.15] unit increase with pval 3E-191, pubmedid=24816252)

Relevant papers and external links:

PMID 24816252

dbSNP: rs2547231 GWAS Catalog: rs2547231 Genecard: SULT2A1 chromosome 19

SNPedia: rs2547231(T;T)

★ rs1801131(C;C) bad:3.15 6.22% **Number of risks. Complex.**

MTHFR rs1801131 is involved in converting 5-methylfolate (5MTHF) to tetrahydrofolate (THF). Unlike MTHFR C677T, this mutation does not lead to elevated homocysteine levels. rs1801131 is a SNP in the MTHFR gene, representing an A>C mutation at mRNA position 1298, resulting in a glu429-to-ala (E429A) substitution (hence this SNP is also known as A1298C or E429A).

A diplotype (according to the authors, but perhaps actually a genoset) of rs1801131 and rs1801133 has been linked to slightly increased risk for several types of brain cancer. The highest risk of meningioma was associated with heterozygosity for both MTHFR SNPs (odds ratio 2.11, CI: 1.42-3.12, p=0.002). The corresponding odds ratio for glioma was 1.23 (CI: 0.91-1.66, p=0.02. In general, risks were increased with genotypes associated with reduced MTHFR activity.

rs1801131 shows no consistent association with schizophrenia overall in 12 studies totaling 3,000+ patients

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Gene related topics:

METABOLIC(14.69) · hyperhomocysteinemia(14.69) · DEVELOPMENTAL(14.49) · neural tube defects(14.49) · down syndrome(11.36) · CARDIOVASCULAR(10.00) · coronary artery disease hyperhomocysteinemia(10.00) · folic acid deficiency(7.00) · homocysteine(6.92) · UNKNOWN(5.00) · chronic renal failure hyperhomocysteinemia kidney failure chronic(5.00) · coronary disease coronary heart disease hyperhomocysteinemia(5.00) · CANCER(4.00) · adenoma colorectal neoplasms neoplasm recurrence local(4.00) · hyperhomocysteinemia retinal vein occlusion(4.00) · spinal dysraphism(3.85) · colorectal cancer(3.74) · arteriosclerosis(3.57) · apoplexy stroke(3.12) · apoplexy brain ischemia hyperhomocysteinemia stroke(3.00) · cardiovascular diseases hyperhomocysteinemia(3.00) · congenital heart defects heart defects congenital(3.00) · diabetes mellitus type ii diabetes mellitus type 2 hyperhomocysteinemia(3.00) · hearing loss sudden(3.00) · hyperhomocysteinemia postoperative complications(3.00) · hyperhomocystinemia(3.00) · vascular diseases(3.00) · cardiovascular diseases(2.93) · apoplexy brain ischemia stroke(2.61) · REPRODUCTION(2.61) · pregnancy loss(2.61) · abortion spontaneous(2.58) · leukemia lymphocytic acute l1 precursor cell lymphoblastic leukemia lymphoma(2.58) · pregnancy complications(2.40) · precursor cell lymphoblastic leukemia lymphoma(2.16) · colonic neoplasms(2.13) · IMMUNE(2.00) · NEUROLOGICAL(2.00) · VISION(2.00) · abortion habitual hyperhomocysteinemia(2.00) · abortion habitual pregnancy complications hematologic thrombophilia(2.00) · abruptio placentae(2.00) · activated protein c resistance hyperhomocysteinemia thrombophilia venous thrombosis(2.00) · acute lymphocytic leukemia hyperhomocysteinemia precursor cell lymphoblastic leukemia lymphoma(2.00) · atherosclerosis thrombosis(2.00) · cardiovascular diseases chronic renal failure hyperhomocysteinemia kidney failure chronic(2.00) · cerebrovascular disease ischemic(2.00) · chronic ulcerative colitis colitis ulcerative folic acid deficiency hyperhomocysteinemia(2.00) · cleft lip cleft palate syndrome(2.00) · epilepsy hyperhomocysteinemia(2.00) · glaucoma angle closure

glaucoma open angle(2.00) · heart defects congenital(2.00) · hyperhomocysteinemia hyperlipidemias(2.00) · hyperhomocysteinemia hypertension(2.00) · hyperhomocysteinemia pre eclampsia(2.00) · retinal artery occlusion retinal vein occlusion(2.00) · hyperhomocysteinemia venous thrombosis(1.80) · venous thrombosis(1.78) · abortion habitual thrombophilia(1.71) · stomach neoplasms(1.71) · HEMATOLOGICAL(1.58) · thrombophilia(1.58) · thrombosis(1.44) · PHARMACOGENOMIC(1.39) · methotrexate toxicity(1.39) · thromboembolism venous(1.35) · anemia sickle cell sickle cell anemia(1.29) · cleft lip cleft palate(1.29) · adenocarcinoma stomach neoplasms(1.25) · acute lymphocytic leukemia precursor cell lymphoblastic leukemia lymphoma(1.25) · diabetes mellitus type ii diabetes mellitus type 2 diabetic nephropathies diabetic nephropathy(1.12) · coronary disease coronary heart disease(1.10) · pre eclampsia(1.05) · coronary artery disease(1.04) · antiphospholipid syndrome thrombophilia thrombosis(1.00) · brain ischemia(1.00) · brain neoplasms glioma meningeal neoplasms meningioma(1.00) · carcinoma squamous cell skin neoplasms squamous cell carcinoma(1.00) · cervical intraepithelial neoplasia cervical neoplasm papillomavirus infections uterine cervical neoplasms(1.00) · migraine with aura migraine without aura(1.00) · thrombophilia and vascular disease(1.00) · brain(1.00) · cancer(1.00) · glioma(1.00) · brain cancer(1.00) · breast cancer(0.93) · migraine disorders(0.92) · thrombosis deep vein(0.86) · RENAL(0.84) · diabetic nephropathy(0.84) · pregnancy loss recurrent(0.83) · pre eclampsia thrombophilia(0.82) · esophageal cancer stomach cancer(0.80) · abortion habitual(0.75) · preeclampsia(0.73) · arthritis rheumatoid rheumatoid arthritis(0.71) · PSYCH(0.67) · behcet syndrome venous thrombosis(0.67) · brain ischemia stroke thrombophilia(0.67) · cerebrovascular disorders(0.67) · congenital abnormalities(0.67) · dementia vascular(0.67) · diabetic nephropathies diabetic nephropathy(0.67) · aortic aneurysm abdominal(0.59) · insulin resistance polycystic ovarian syndrome polycystic ovary syndrome(0.57) · heart anomalies congenital(0.53) · leukemia(0.52) · atherosclerosis(0.51) · hypertension(0.51) · cardiovascular risk(0.50) · cerebrovascular disease(0.50) · colonic neoplasms rectal neoplasms(0.50) · thrombosis venous thrombosis(0.50) · chronic renal failure kidney failure chronic(0.49) · esophageal cancer(0.44) · fatty liver(0.44) · adenoma colorectal neoplasms(0.44) · migraine(0.43) · inflammatory bowel disease nos inflammatory bowel diseases(0.43) · OTHER(0.40) · chromosome aberrations dna damage(0.40) · chronic progressive chorea huntington disease(0.40) · infertility male(0.38) · venous thromboembolism(0.36) · alzheimer s disease(0.36) · intima media thickness(0.33) · migraine with aura(0.33) · brain ischemia stroke(0.31) · infertility female(0.31) · stroke ischemic(0.30) · schizophrenia(0.30) · multiple myeloma(0.29) · atherosclerosis coronary(0.29) · schizophrenia bipolar disorder(0.27) · colorectal neoplasms(0.26) · body weight obesity(0.26) · cardiovascular disease(0.25) · stroke(0.24) · bladder neoplasm urinary bladder neoplasms(0.24) · stomach cancer(0.23) · carcinoma squamous cell esophageal neoplasms oesophageal neoplasm squamous cell carcinoma(0.23) · diabetes type 2(0.23) · arthritis rheumatoid(0.23) · myocardial ischemia(0.21) · CHEMDEPENDENCY(0.21) · alcohol(0.21) · bladder cancer(0.20) · kidney transplant complications(0.19) · neoplasms(0.19) · myocardial infarction(0.17) · arthritis juvenile rheumatoid chronic childhood arthritis(0.17) · myocardial infarct(0.16) · cerebral infarction(0.15) · kidney failure chronic(0.14) · lymphoma(0.14) · kidney diseases(0.14) · choroidal neovascularization macular degeneration(0.13) · cleft lip with cleft palate cleft lip without cleft palate(0.12) · restenosis(0.12) · type 2 diabetes(0.12) · carcinoma hepatocellular lcc liver cell carcinoma liver neoplasms(0.11) · INFECTION(0.11) · helicobacter infections(0.11) · depression(0.11) · leukemia lymphocytic chronic b cell(0.11) · carotid artery diseases(0.10) · cerebral palsy(0.09) · AGING(0.08) · longevity(0.08) · cardiovascular(0.08) · carcinoma squamous cell head and neck neoplasms squamous cell carcinoma(0.08) · polycystic ovary syndrome(0.08) · graft vs host disease(0.06) · dna damage(0.06) · autism(0.05) · atrial fibrillation(0.05) · lymphoma non hodgkin(0.05) · NORMALVARIATION(0.04) · normal variation(0.04) · crohn s disease ulcerative colitis(0.04) · dementia(0.04) · osteoporosis(0.04) · coronary disease(0.02) · cervical cancer(0.02)

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Relevant papers and external links:

PMID 18483342

OMIM 607093

PHARMGKB PA165110385

Evidence: PubMed ID:18580170Risk or phenotype-associated allele(s): C/C. Phenotype:Patients with a homozygous MTHFR 1298A>C mutation (n = 25) developed higher plasma homocysteine concentrations (median [interquartile range], 14.9 [10.0-26.4] microm) than wild-type or heterozygous patients (9.3 [7.5-15.5] microm; n = 115). The change in homocysteine after nitrous oxide anesthesia was tripled in homozygous patients compared with wild-type (5.6 microm [+60%] vs. 1.8 microm [+22%]). Study size: 140. Study population/ethnicity: healthy patients undergoing elective surgery. Type of association: GN.

PHARMGKB PA161145164
Evidence: Web Resource:External Link studied, associated with multiple phenotypes.

PHARMGKB PA164918206
Evidence: PubMed ID:15643524In a case control study of Asian gastric cancer (n=633), individuals with 6 variant alleles of three MTHFR common variants (i.e. C677T, A1298C and G1793A) were at increased risk for gastric cardia adenocarcinoma (OR =4.64, 95% CI =1.34-16.01) compared with those having 0-2 variants.

PHARMGKB PA165106617
Evidence: PubMed ID:16572443At 6 months methotrexate and folic acid therapy, of early rheumatoid arthritis patients with the MTHFR 1298AA genotype showed good improvement relative to combined CA and AA genotypes (OR 2.3), while 1298C allele carriers developed more adverse drug events (OR 2.5) (e.g. pneumonitis, gastrointestinal ADEs, skin and mucosal ADEs, and elevated liver enzyme levels). Patients with MTHFR 1298AA / 677CC diplotype showed greater clinical improvement.

PHARMGKB PA165106622
Evidence: PubMed ID:19016697In 330 patients who completed 3 months methotrexate treatment for psoriasis, no significant genotypic associations were found between clinical outcome (e.g. efficacy, toxicity) and 50 SNPs in pathway genes for methotrexate metabolism (ATIC, FPGS, GGH, MTHFR), including 47 common (>5% minor allele frequency) haplotype-tagging SNPs (r(2) > 0.8) plus 3 additional SNPs.

PMID 21302350

dbSNP: rs1801131 Genecard: MTHFR chromosome 1 SNPedia: rs1801131(C;C)

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★ rs4149056(C;T) bad:3.10 16.00% **reduced breakdown of some drugs; 5x increased myopathy risk for statin users**

influences statin response
rs72559745 (SLCO1B1*1) AA AA
rs56061388 (SLCO1B1*3) TT TT
rs4149056 (SLCO1B1*5) TT TT
rs55901008 (SLCO1B1*6) TT TT
rs4149056, also known as 37041T>C or V174A, is a SNP in the SLCO1B1 gene, which encodes the 'organic anion transporting polypeptide 1B1' (OATP1B1) protein. This protein, found primarily in the liver, regulates the uptake of numerous drugs and natural compounds. The rs4149056(C) SNP defines the SLCO1B1*5 allele.

The rs4149056(C) allele gives rise to an amino acid change (from valine to alanine at residue 174) which has reduced uptake/transport activity. Therefore, drugs metabolized by OATP1B1 tend to build up to higher circulating concentrations than they would otherwise..

The drugs known (or in some cases, thought) to be transported less well by the variant OATP1B1 protein encoded by the rs4149056(C) allele include:

Several cholesterol lowering statins, generally leading to reduced inhibitory effects on liver cholesterol synthesis and possibly worse side effects, by such drugs as:
simvastatin
pravastatin
rosuvastatin
pitavastatin
fexofenadine
repaglinide
methotrexate
the SN-38 active metabolite of irinotecan
rifampicin
caspofungin
lopinavir

rs4363657 in nearly complete linkage disequilibrium with rs4149056 SNP (r2=0.97), which has been linked to statin metabolism. rs4149056(C) odds ratio for myopathy among 20,000 individuals taking either 40 or 80mg of simvastatin daily was 4.5 (CI: 2.6-7.7) per copy of the C allele, and 16.9 (CI: 4.7-61.1) in (C;C) as compared with (T;T) homozygotes.

The Gene Sherpa points out a 16x odds ratio for myopathy when taking statins and suggests fenofibrates might be a good alterative.

A study of ~500 individuals taking statins concluded that rs4149056(C), i.e. SLCO1B1*5, was associated with statin-induced side-effects, most likely somewhat correlated to the number of such alleles being carried.

A study of >4000 individuals with type 2 diabetes using routine prescribing data from the Electronic Medical Record in Tayside Scotland suggests high proportion rs4149056(C) homozygotes discontinue or reduce their doses of statins

23andMe blog discussion of this SNP

SLCO1B1 genetic variant associated with statin-induced myopathy: a proof-of-concept study using the clinical practice research datalink

Statin Response

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Established associations:

blood metabolite measurement **you have 1 copies of the risk allele T**

(risk allele=T, X-12456 odds ratio/beta 0.081 [0.069-0.093] unit decrease with pval 9E-44, pubmedid=24816252; risk allele=T, X-12063 odds ratio/beta 0.118 [0.1-0.13] unit decrease with

pval 1E-73, pubmedid=24816252; risk allele=T, 4-androsten-3beta,17beta-diol disulfate 2 odds ratio/beta 0.049 [0.037-0.061] unit decrease with pval 1E-18, pubmedid=24816252; risk allele=T, X-11491 odds ratio/beta 0.088 [0.072-0.104] unit decrease with pval 3E-32, pubmedid=24816252; risk allele=T, 1-arachidonoylglycerophosphoethanolamine odds ratio/beta 0.029 [0.023-0.035] unit decrease with pval 3E-18, pubmedid=24816252; risk allele=T, isoleucine/X-11529 odds ratio/beta 0.3 [0.28-0.32] unit increase with pval 6E-328, pubmedid=24816252; risk allele=T, X-11529 odds ratio/beta 0.295 [0.28-0.31] unit decrease with pval 6E-315, pubmedid=24816252)

sex hormone globulin binding measurement **you have 1 copies of the risk allele T**

(risk allele=T, Men + Women odds ratio/beta 0.029 [0.019-0.039] umol/L increase with pval 2E-8, pubmedid=22829776)

response to statin **you have 1 copies of the risk allele C**

(risk allele=C, odds ratio/beta 4.5 [2.60-7.70] with pval 2E-9, pubmedid=18650507)

bilirubin measurement **you have 1 copies of the risk allele C**

(risk allele=C, odds ratio/beta 0.05 [0.03-0.07] umol/l increase in logtbil with pval 7E-13, pubmedid=19414484)

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Gene related topics:

PHARMACOGENOMIC(3.00) · rosuvastatin pharmacokinetics(3.00) · OTHER(2.00) · pitavastatin pharmacokinetics(2.00) · repaglinide pharmacokinetics(2.00) · pravastatin kinetics(1.29) · cholesterol(1.00) · metabolism(1.00) · type 2 diabetes(1.00) · irinotecan pharmacokinetics(0.19) · HEMATOLOGICAL(0.17) · bilirubin(0.17) · METABOLIC(0.13) · hyperlipidemias(0.13)

Relevant papers and external links:

PMID 19414484
Genome-wide association meta-analysis for total serum bilirubin levels.

PMID 22829776
A genome-wide association meta-analysis of circulating sex hormone-binding globulin reveals multiple loci implicated in sex steroid hormone regulation.

PMID 24816252
An atlas of genetic influences on human blood metabolites.

PMID 16758257

PMID 18650507

PMID 19833260

PMID 21178985

PMID 23942138

OMIM 604843

PHARMGKB PA165109803

Evidence: PubMed ID:19890249Risk or phenotype-associated allele: SLCO1B1*15 defined by rs2306283 Asn130Asp (338A>G) and rs4149056 Val174Ala (521T>C). Phenotype: SLCO1B1*15 allele was associated with higher mycophenolic acid (MPA) AUC(0-12 hours) (p = 0.0246) and lower ratio of MPAG (MPA glucuronide)/MPA (p = 0.0267), but no association with MPAG AUC(0-9 h). Study size: 70. Study population/ethnicity: Caucasian cohort from the SOPHIE study cotreated with mycophenylate mofetil (MMF) and either sirolimus or tacrolimus. Significance metric(s): p < 0.03. Type of association: GN; PK

PHARMGKB PA161145159
Evidence: Web Resource:External Link with increased pravastatin plasma AUC.

PHARMGKB PA161748444
Evidence: PubMed ID:18466105Studies into the effect of the 521T>C (V174A) SNP in SLCO1B1 yielded discrepancies in the transport data of estradiol-17?-D-glucuronide and estrone-3-sulfate.

PHARMGKB PA162356046
Evidence: PubMed ID:18650507This variant in the SLCO1B1 gene is strongly associated with an increased risk of statin-induced myopathy. This study also showed an association between rs4149056 and the cholesterol-lowering effects of simvastatin.

PHARMGKB PA162356181
Evidence: PubMed ID:18794729The cholesterol synthesis rate was higher in CC individuals than in TT individuals. There was no association between this SNP and the short-term effects of statins on cholesterol synthesis rates.

PHARMGKB PA164740859
Evidence: PubMed ID:18650507; Web Resource:External Link results: SLCO1B1 Variants and Statin-Induced Myopathy--A Genomewide Study. (Initial Sample Size: 85 cases, 90 controls; Replication Sample Size: 19,856 individuals); (Region: 12p12.1; Reported Gene(s): SLCO1B1; Risk Allele: rs4149056-C); (p-value= 0.000000002).This variant is associated with Myopathy.

PHARMGKB PA164891482
Evidence: PubMed ID:11477075Variant with C allele showed reduced cell surface expression

PHARMGKB PA164891481
Evidence: PubMed ID:12811365; PubMed ID:17177112Variant with C allele had reduced transport, relative to variant with T allele (based upon lower plasma AUC) of pravastatin

PHARMGKB PA164891484
Evidence: PubMed ID:18187595Variant with C allele showed NO plasma AUC of nateglinide relative to T allele, in contrast to repaglinide

PHARMGKB PA164891483
Evidence: PubMed ID:18187595Variant with C allele showed increased plasma AUC of repaglinide relative to T

PHARMGKB PA164925732
Evidence: PubMed ID:19374892In a study of 16 subjects rifampicin increased atorvastatin plasma concentration in accordance with SLCO1B1 521T>C genotype. Rifampicin pharmacokinetics were not altered by SLCO1B1 genotype.

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PHARMGKB PA165110263
Evidence: PubMed ID:19833260Risk or phenotype-associated allele: SLCO1B1*5 allele, defined by rs4149056 (521T>C, Val174Ala). Phenotype: SLCO1B1*5 allele (rs4149056, V174A) was associated with the adverse events from statins (chi-square = 4.2, p = 0.03, FDR = 0.24), with greatest risk with simvastatin. There were significantly more females (66%) with adverse events than females without adverse events (50%) (p < 0.01). Among subjects with adverse effects (n = 99), females bore greater risk of adverse events (OR = 2.0, p = 0.004). In multivariate analysis adjusted for race, female sex (OR = 2.2, p = 0.001) and SLCO1B1*5 genotype (OR = 1.7, p = 0.03) were independently associated with adverse events to statins (p < 0.05 for both). Simvastatin plasma concentration was positively correlated with SLCO1B1*5 both at 20 mg (p = 0.006) and 80 mg (p = 0.03). Study size: 452. Study population/ethnicity: Hypercholesterolemia outpatients given 1 of 3 statins (atorvastatin, simvastatin, or pravastatin) between 2001 and 2002 Significance metric(s): OR (1.7 - 2.2), p < 0.05 Type of association: CO; GN; PK; ADR

PHARMGKB PA165110328
Evidence: PubMed ID:19901119Risk or phenotype-associated allele: C allele (174Ala). Phenotype: Genome-wide analysis of 398,699 germline SNPs showed association of the rs4149056 C allele (174Ala) with methotrexate (MTX) plasma clearance. Study size: 434 (discovery cohort), 206 (independent validation cohort). Study population/ethnicity: Multiethnic children (5.92 median age , 1.02-18.85 range) with ALL given 3,014 courses of methotrexate at 2-5 g/m(2) enrolled in Tennessee. Significance metric(s): p = 1.9 x 10(-7) (n = 434); p = 1.2 x 10(-7) (n = 206). Type of association: CO; GN; PK

OMIM 601816

dbSNP: rs4149056 GWAS Catalog: rs4149056 Genecard: SLCO1B1 chromosome 12

SNPedia: rs4149056(C;T)

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★ rs1801260(C;T) info:3.06 35.38% **Normal risk of ADHD symptoms.**

This is one normal and one rare form of the CLOCK gene. In addition to possibly affecting evening preference, it has been linked to normal Attention Deficit Hyperactivity Disorder symptom scores. rs1801260, a SNP in the CLOCK gene known as 3111 T/C, has been reported to influence sleep and activity patterns in patients affected by bipolar depression.

From this article's abstract:

"Compared to T/T homozygotes, carriers of the C allele had a similar degree of severity of depression, but showed higher activity levels in the evening, a delayed sleep onset (mean 79 min later), and a reduced amount of sleep during the night (mean 75 min less)."

Allele frequencies of T3111C SNP of hClock were significantly different between schizophrenics and controls (chi(2) = 19.738, P < 0.05). Schizophrenics had a significantly higher frequency of the C allele compared with controls (OR = 2.613, 95% CI = 1.693-4.034). (Han Chinese population.)

There was a strong, significant association (P < 0.001) between both self-rating and interview-based adult ADHD assessments and the rs1801260 polymorphism with at least one T-mutation being the risk allele.

rs1801260 variations associated with weight loss while on Mediterranean diet. Carriers of the G allele displayed greater difficulty in losing weight than non-carriers.

CLOCK 3111 T/C SNP was associated with activity levels in the second part of the day, neuropsychological performance and BOLD fMRI correlates (interaction of genotype and moral valence of the stimuli).

Association between polymorphisms in the Clock gene, obesity and the metabolic syndrome in man.

Clock genes may influence bipolar disorder susceptibility and dysfunctional circadian rhythm.

Genetic differences in human circadian clock genes among worldwide populations.

Genome-wide association scan for five major dimensions of personality.

Circadian polymorphisms associated with affective disorders.

CLOCK is suggested to associate with comorbid alcohol use and depressive disorders.

ARNTL (BMAL1) and NPAS2 gene variants contribute to fertility and seasonality.

Genotyping sleep disorders patients.

Association between CLOCK 3111T/C and preferred circadian phase in Korean patients with bipolar disorder.

SIRT1 and CLOCK 3111T>C combined genotype is associated with evening preference and weight loss resistance in a behavioral therapy treatment for obesity.

"Circadian locomotor output cycles kaput (CLOCK) molecule plays major roles in circadian rhythmicity and regulates daily physiological processes including digestive activity. Therefore, we hypothesized that the CLOCK 3111T/C single nucleotide polymorphism (SNP) might have adverse effects on the regulation of gastric motility."

"The clock molecule plays major roles in circadian rhythmicity and regulating lipid and glucose metabolism in peripheral organs. Disruption of the circadian rhythm can lead to cardiometabolic disorders."

"Here, we find that cholecystokinin (Cck) is a direct transcriptional target of CLOCK and levels of Cck are reduced in the ventral tegmental area (VTA) of ClockΔ19 mice. Selective knockdown of Cck expression via RNA interference in the VTA of wild-type mice produces a manic-like phenotype"

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" C genetic variants in CLOCK 3111 T/C are more obese and lose less weight in a dietary and behavioral treatment for obesity than TT carriers and they are also more evening-type subjects "

Attention-Deficit Hyperactivity Disorder

Gene related topics:

adhd(5.00) · OTHER(2.27) · sleep disorders(2.27) · circadian variability(2.00) · sleep(1.00) · bipolar disorder(1.00) · glucose(1.00) · metabolism(1.00) · affective disorder(1.00) · weight loss(1.00) · weight(1.00) · alcohol(1.00) · attention deficit hyperactivity disorder(1.00) · obesity(1.00) · depression(1.00) · PSYCH(0.06) · personality(0.06) · METABOLIC(0.04) · metabolic syndrome(0.04)

Relevant papers and external links:

PMID 17221848
PMID 20364331
PMID 17948273
PMID 20065968
PMID 17428266
PMID 18071340
PMID 18228528
PMID 18663240
PMID 18957941
PMID 19166596
PMID 20180986
PMID 20368993
PMID 20396431
PMID 20600471
PMID 22310473
PMID 22709985
PMID 20961464
PMID 23399917
PMID 24467926

dbSNP: rs1801260 Genecard: CLOCK chromosome 4 SNPedia: rs1801260(C;T)

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★ rs743777(G;G) bad:3.02 7.66% **; rheumatoid arthritis (you have the risk allele G)**

rs743777 has been reported in a large study to be associated with rheumatoid arthritis.

The risk allele (oriented to the dbSNP entry) is (G); the odds ratio associated with heterozygotes is 1.09 (CI 0.97-1.24), and for homozygotes, 1.72 (CI 1.40-2.11).

Lack of association or interactions between the IL-4, IL-4Ralpha and IL-13 genes, and rheumatoid arthritis.

Rheumatoid arthritis susceptibility loci at chromosomes 10p15, 12q13 and 22q13.

Representation of genetic association via attributable familial relative risks in order to identify polymorphisms functionally relevant to rheumatoid arthritis.

AntEpiSeeker: detecting epistatic interactions for case-control studies using a two-stage ant colony optimization algorithm.

A novel galectin-1 and interleukin 2 receptor beta haplotype is associated with autoimmune myasthenia gravis.

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Established associations:

type i diabetes mellitus **you have 2 copies of the risk allele G**
(risk allele=G, T1D odds ratio/beta 1.1 with pval 2E-6, pubmedid=21829393)

autoantibody measurement **you have 2 copies of the risk allele G**
(risk allele=G, T1D odds ratio/beta 1.1 with pval 2E-6, pubmedid=21829393)

rheumatoid arthritis **you have 2 copies of the risk allele G**
(risk allele=G, odds ratio/beta 1.09 [0.97-1.24] with pval 1E-6, pubmedid=17554300)

Gene related topics:

arthritis(1.00) · myasthenia gravis(1.00) · rheumatoid arthritis(1.00)

Relevant papers and external links:

PMID 21829393
Genome-wide association analysis of autoantibody positivity in type 1 diabetes cases.

PMID 17554300

PHARMGKB PA162356657

Evidence: PubMed ID:17554300; Web Resource:External Link Results: Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls (Initial Sample Size: 1,860 cases, 2,938 controls; Replication Sample Size: NR; Risk Allele: rs743777-G). This variant is associated with rheumatoid arthritis.

PMID 18625055
PMID 18794857
PMID 20017963
PMID 20426808
PMID 20728947

dbSNP: rs743777 GWAS Catalog: rs743777 Genecard: IL2RB chromosome 22

SNPedia: rs743777(G;G)

★ rs11052552(G;G) bad:3.02 **1.4x risk of type-1 diabetes**

rs11052552 has been reported in a large study to be associated with type-1 diabetes.

The risk allele (oriented to the dbSNP entry) is (G); the odds ratio associated with heterozygotes is 1.49 (CI 1.28-1.73), and for homozygotes, 1.43 (CI 1.21-1.69).

Established associations:

type i diabetes mellitus **you have 2 copies of the risk allele G**
(risk allele=G, odds ratio/beta 1.49 [1.28-1.73] with pval 7E-7, pubmedid=17554300)

Relevant papers and external links:

PMID 17554300
PHARMGKB PA162356637
Evidence: PubMed ID:17554300; Web Resource:External Link Results: Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls (Initial Sample Size: 1,963 cases, 2,938 controls; Replication Sample Size: (see Todd 2007); Risk Allele: rs11052552-G). This variant is associated with type 1 diabetes.

dbSNP: rs11052552 GWAS Catalog: rs11052552 Genecard: nan chromosome 12

SNPedia: rs11052552(G;G)

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★ rs8192678(A;G) bad:3.02 39.03% **higher blood pressure if <50**

rs8192678 encodes a SNP also known as Gly482Ser; the (A) allele encodes the Ser. Some reports have linked this SNP to risk for hypertension and systolic blood pressure (SBP).

However, a meta-analysis of 13,000+ individuals did not find any such association. However, gene-age interaction was apparent. For diastolic blood pressure (DBP), p(interaction)<0.0001; for systolic BP, p(interaction)=0.026. In younger individuals (<50yrs; n=2511) the rs8192678(A) allele was associated with higher DBP (p=4.2 x 10-12) and SBP (p=7.2 x 10-12), but no association was evident for individuals over 50yrs (n=5088).

Analysis of PGC-1alpha variants Gly482Ser and Thr612Met concerning their PPARgamma2-coactivation function.

PPARGC1A coding variation may initiate impaired NEFA clearance during glucose challenge.

Frequencies of single nucleotide polymorphisms in genes regulating inflammatory responses in a community-based population.

Medical sequencing at the extremes of human body mass.

Polymorphisms of the peroxisome proliferator-activated receptor-gamma coactivator-1alpha gene are associated with hypertrophic cardiomyopathy and not with hypertension hypertrophy.

PPARGC1A variation associated with DNA damage, diabetes, and cardiovascular diseases: the Boston Puerto Rican Health Study.

[Association study between PPARGC1A Thr394Thr/ Gly482Ser polymorphisms and type 2 diabetes].

Ser1369Ala variant in sulfonylurea receptor gene ABCC8 is associated with antidiabetic efficacy of gliclazide in Chinese type 2 diabetic patients.

Functional genetic polymorphisms and female reproductive disorders: Part I: Polycystic ovary syndrome and ovarian response.

Peroxisome proliferator-activated receptor gamma/Pro12Ala polymorphism and peroxisome proliferator-activated receptor gamma coactivator-1 alpha/Gly482Ser polymorphism in patients with sarcoidosis.

The gene coding for PGC-1alpha modifies age at onset in Huntington's Disease.

PGC-1alpha as modifier of onset age in Huntington disease.

Is there an optimum endurance polygenic profile?

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The Role of the PGC1alpha Gly482Ser Polymorphism in Weight Gain due to Intensive Diabetes Therapy.

Do PPARGC1A and PPARalpha polymorphisms influence sprint or endurance phenotypes?

Is there an interaction between PPARD T294C and PPARGC1A Gly482Ser polymorphisms and human endurance performance?

Localization of sequence variations in PGC-1alpha influence their modifying effect in Huntington disease.

Meta-analysis of association studies between five candidate genes and type 2 diabetes in Chinese Han population.

Gene related topics:

CARDIOVASCULAR(2.00) · ventricular dysfunction left(2.00) · cardiovascular disease(1.00) · glucose(1.00) · cardiomyopathy(1.00) · systolic blood pressure(1.00) · hypertrophic cardiomyopathy(1.00) · body mass(1.00) · hypertension(1.00) · cardiovascular(1.00) · diastolic blood pressure(1.00) · weight(1.00) · huntington disease(1.00) · dna damage(1.00) · cardiovascular diseases(1.00) · weight gain(1.00) · sarcoidosis(1.00) · NORMALVARIATION(0.36) · endurance performance(0.36) · METABOLIC(0.22) · diabetes gestational(0.22) · metabolic syndrome(0.09) · lipoproteins(0.08) · polycystic ovary syndrome(0.08) · type 2 diabetes(0.07) · blood pressure(0.05) · heart rate(0.04) · diabetes mellitus type 2(0.02)

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Relevant papers and external links:

PMID 18467552
PMID 17187763
PMID 17216277
PMID 17355643
PMID 17357083
PMID 17579564
PMID 18162502
PMID 18331997
PMID 18599530
PMID 18603647
PMID 19070258
PMID 19133136
PMID 19200361
PMID 19237423
PMID 19360113
PMID 19422653
PMID 19666693
PMID 21211002

PMID 22391941

dbSNP: rs8192678 Genecard: PPARGC1A chromosome 4 SNPedia: rs8192678(A;G)

★ rs9469220(A;A) bad:3.02 **1.5x risk of Crohn's disease**

rs9469220 has been reported in a large study to be associated with Crohn's disease.

The risk allele (oriented to the dbSNP entry) is (A); the odds ratio associated with heterozygotes is 1.14 (CI 0.98-1.32), and for homozygotes, 1.52 (CI 1.28-1.79).

Established associations:

serum ige measurement	**you have 0 copies of the risk allele G** (risk allele=G, odds ratio/beta 0.09 [0.05-0.12] unit decrease with pval 2E-7, pubmedid=23146381)
crohn's disease	**you have 2 copies of the risk allele A** (risk allele=A, odds ratio/beta 1.14 [0.98-1.32] with pval 2E-6, pubmedid=17554300)

Relevant papers and external links:

PMID 23146381
A meta-analysis of genome-wide association studies for serum total ige in diverse study populations.

PMID 17554300

PHARMGKB PA162356660
Evidence: PubMed ID:17554300; Web Resource:External Link Results: Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls (Initial Sample Size: 1,748 cases, 2,938 controls; Replication Sample Size: (see Parkes 2007); Risk Allele: rs9469220-A). This variant is associated with crohn's disease.

dbSNP: rs9469220 GWAS Catalog: rs9469220 SNPedia: rs9469220(A;A)

★ rs25532(C;T) info:3.02 **may be part of a haplotype associated with OCD**

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rs25532 is a SNP in a regulatory region upstream of the serotonin transporter SLC6A4 gene. The more common allele, rs25532(C), is the higher-expressing allele.

A study of patients with obsessive compulsive disorder (OCD) concluded that the major allele of rs25532 was a major contributor to the association of a corresponding haplotype with this disorder.

Gene related topics:

PSYCH(24.57) · depression(24.57) · suicide(6.88) · obsessive compulsive disorder(5.82) · IMMUNE(5.28) · irritable bowel syndrome(5.28) · personality traits(4.75) · anxiety disorder(4.50) · REPRODUCTION(4.00) · depressive disorder major bipolar disorder(4.00) · sexual dysfunction physiological(4.00) · personality disorders(3.78) · depressive disorder major(3.72) · affective disorder(3.12) · anxiety related traits(3.00) · decision making(3.00) · psychoses(3.00) · personality(2.60) · panic disorder(2.19) · autism(2.13) · mood disorders(2.02) · CHEMDEPENDENCY(2.00) · OTHER(2.00) · PHARMACOGENOMIC(2.00) · UNKNOWN(2.00) · alcohol abuse smoking behavior(2.00) · alcoholism attention deficit hyperactivity disorder(2.00) · alcoholism recurrence(2.00) · antidepressant adverse effects(2.00) · anxiety(2.00) · auditory evoked potential(2.00) · bipolar and unipolar disorder(2.00) · eating disorder(2.00) · hallucinations(2.00) · harm avoidance(2.00) · impulse control disorder(2.00) · manic depressive illness(2.00) · personality traits illegal drug use(2.00) · suicidal behavior(1.92) · alcoholism(1.92) · NEUROLOGICAL(1.80) · cocaine dependence(1.80) · major depression(1.80) · migraine migraine with aura(1.80) · neuroticism(1.80) · major depressive disorder(1.73) · migraine(1.73) · substance related disorders(1.67) · bulimia(1.47) · attention deficit hyperactivity disorder(1.16) · adhd attention deficit hyperactivity disorder(1.00) · aggressive behavior(1.00) · alcohol related disorders(1.00) · bipolar disorder depression(1.00) · conduct disorder(1.00) · eating disorders(1.00) · sudden infant death(1.00) · anorexia nervosa(0.93) · CARDIOVASCULAR(0.80) · hypertension pulmonary(0.80) · major psychoses(0.80) · bipolar disorder(0.79) · schizophrenia(0.69) · alcohol abuse(0.68) · bipolar affective disorder(0.67) · fibromyalgia(0.67) · sids sudden infant death syndrome(0.67) · smoking behavior(0.66) · mood disorder(0.60) · psychosis(0.60) · heroin abuse(0.53) · epilepsy temporal lobe(0.43) · sleep disorders(0.36) · cognitive function(0.33) · migraine with aura(0.33) · syncope vasovagal(0.33) · DEVELOPMENTAL(0.27) · mental retardation(0.27) · adhd(0.26) · METABOLIC(0.23) · weight gain(0.23) · smoking(0.21) · tourette syndrome(0.21) · alcohol(0.21) · migraine disorders(0.17) · cognitive ability(0.11) · chronic obstructive pulmonary disease(0.07) · hearing loss(0.07) · AGING(0.04) · longevity(0.04)

Relevant papers and external links:

PMID 18055562

PHARMGKB PA164920373

Evidence: PubMed ID:18055562This study found that the minor allele of the functional C > T single nucleotide polymorphism, rs25532, located less than 150 nucleotides centromeric of the serotonin transporter-linked polymorphic region indel (known as 5-HTTLPR) significantly decreased luciferase reporter gene expression levels by 15-80%, depending on 5-HTTLPR allele background and cell type.

dbSNP: rs25532 Genecard: SLC6A4 chromosome 17 SNPedia: rs25532(C;T)

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★ rs4624820(A;A) warning:2.99 26.38% **increased testicular cancer risk for men**

23andMe blog each rs4624820(A) increases the odds of testicular cancer by 1.37x

Established associations:

testicular carcinoma **you have 2 copies of the risk allele A**

(risk allele=A, odds ratio/beta 1.37 [1.19-1.58] with pval 3E-13, pubmedid=19483681; risk allele=A, odds ratio/beta 1.52 [1.38-1.67] with pval 3E-31, pubmedid=23666240; risk allele=G, odds ratio/beta 1.6393 [1.39-1.89] with pval 8E-10, pubmedid=23666239)

Gene related topics:

CANCER(5.00) · cancer(5.00) · testicular cancer(5.00)

Relevant papers and external links:

PMID 19483681
A genome-wide association study of testicular germ cell tumor.

PMID 23666240
Identification of nine new susceptibility loci for testicular cancer, including variants near dazl and prdm14.

PMID 23666239
Meta-analysis identifies four new loci associated with testicular germ cell tumor.

OMIM 273300

dbSNP: rs4624820 GWAS Catalog: rs4624820 Genecard: nan chromosome 5

SNPedia: rs4624820(A;A)

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★ rs7517847(G;T) info:2.96 45.85% **possibly reduced risk for Crohn's disease**

the association may be specific to Crohn's disease, as opposed to all types of IBD.

significant associations with rs1004819, rs7517847, and rs11209026. Having any CARD15 variant was associated with a significant risk for Crohn's disease ($P < 0.0001$).

rs7517847 (P=4.9x10(-9), OR 0.65, 0.56-0.75), is statistically independent of rs11209026.

Replicated reduced risk for Crohn's disease with the rs7517847(G) allele in a study of Italian patients, but not ulcerative colitis.

Each incidence of the G allele was associated with a decrease in Crohn's Disease in New Zealand populations studied.

Investigation of the IL23R gene in a Spanish rheumatoid arthritis cohort.

rs1004819 is the main disease-associated IL23R variant in German Crohn's disease patients: combined analysis of IL23R, CARD15, and OCTN1/2 variants.

Association analysis of IL-12B and IL-23R polymorphisms in myocardial infarction.

Fine mapping versus replication in whole-genome association studies.

CARD15 and IL23R influences Crohn's disease susceptibility but not disease phenotype in a Brazilian population.

IL23R: a susceptibility locus for celiac disease and multiple sclerosis?

IL23R and IL12B polymorphisms in Spanish IBD patients: no evidence of interaction.

IL23R in the Swedish, Finnish, Hungarian and Italian populations: association with IBD and psoriasis, and linkage to celiac disease.

No association between interleukin 23 receptor gene polymorphisms and systemic lupus erythematosus.

Parasites represent a major selective force for interleukin genes and shape the genetic predisposition to autoimmune conditions.

Variants of the IL23R gene are associated with ankylosing spondylitis but not with Sjogren syndrome in Hungarian population samples.

Interleukin-23 receptor gene variants in Hungarian systemic lupus erythematosus patients.

Polymorphisms of IL23R and Vogt-Koyanagi-Harada syndrome in a Chinese Han population.

Association of the interleukin-23 receptor gene variant rs11209026 with Crohn's disease in German children.

Evidence for STAT4 as a common autoimmune gene: rs7574865 is associated with colonic Crohn's disease and early disease onset.

An investigation of genome-wide studies reported susceptibility loci for ulcerative colitis shows limited replication in north Indians.

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Established associations:

omega-6 polyunsaturated fatty acid measurement **you have 1 copies of the risk allele T**

(risk allele=T, odds ratio/beta 0.19 unit increase with pval 4E-6, pubmedid=24823311)

Gene related topics:

IMMUNE(5.16) · inflammatory bowel diseases(5.16) · colitis ulcerative crohn disease(1.98) · arthritis psoriatic psoriasis(1.12) · crohn disease crohn s disease inflammatory bowel disease nos inflammatory bowel diseases(1.00) · celiac disease(1.00) · arthritis(1.00) · rheumatoid arthritis(1.00) · myocardial infarction(1.00) · multiple sclerosis(1.00) · systemic lupus erythematosus(1.00) · lupus erythematosus(1.00) · lupus(1.00) · crohn disease(0.96) · crohn s disease(0.64) · inflammatory bowel disease(0.54) · spondylitis ankylosing(0.44) · inflammatory bowel disease nos inflammatory bowel diseases(0.43) · colitis ulcerative(0.42) · chronic ulcerative colitis colitis ulcerative crohn disease crohn s disease inflammatory bowel disease nos inflammatory bowel diseases(0.40) · ulcerative colitis(0.24) · crohn disease crohn s disease(0.23) · psoriasis(0.20) · behcet syndrome(0.17) · crohn s disease ulcerative colitis(0.15) · ankylosing spondylitis(0.08) · lupus erythematosus systemic systemic lupus erythematosus(0.03)

Relevant papers and external links:

PMID 24823311
Genome-wide association study of plasma n6 polyunsaturated fatty acids within the cohorts for heart and aging research in genomic epidemiology consortium.

PMID 18047539

PMID 17508420

PMID 18698678

PMID 21253534

OMIM 612261

OMIM 607562

PHARMGKB PA162356596
Evidence: PubMed ID:17068223; Web Resource:External Link Results: A genome-wide association study identifies IL23R as an inflammatory bowel disease gene (Initial Sample Size: 547 cases, 548 controls; Replication Sample Size: 401 cases, 433 controls, 883 families, 1,119 affected offspring; Risk Allele: rs7517847-C).

PMID 17678723

PMID 17786191

PMID 17901940

PMID 17924341

PMID 18200510

PMID 18368064

PMID 18383521

PMID 19175939

PMID 19306001

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PMID 19468064
PMID 19522770
PMID 19757086
PMID 20116410
PMID 20192940
PMID 20454450
PMID 21304977

dbSNP: rs7517847 GWAS Catalog: rs7517847 Genecard: IL23R chromosome 1

SNPedia: rs7517847(G;T)

★ rs10830963(C;G) warning:2.96 38.50% **increased type-2 diabetes risk; higher gestational diabetes risk**

rs10830963 was associated with an increased risk of type-2 diabetes in our Han Chinese cohort (OR 1.16, 95% CI 1.03-1.31, p = 0.015). As previously described, the risk variant was also associated with increased fasting plasma glucose, showing an increase of 0.068 mmol/l (95% CI 0.036-0.100, p = 4 x 10(-5)) per risk allele

rs10830962, rs4753426, and rs10830963 were significantly associated with higher fasting plasma glucose concentrations and reduced OGTT- and IVGTT-induced insulin release. rs3781638 displayed significant association with lower fasting plasma glucose levels and increased OGTT-induced insulin release

A study totaling 19,000+ Europeans concluded that rs10830963 had the most influence of any MTNR1B gene SNP on the risk for type-2 diabetes. Specifically, the (G) allele increased the risk of isolated impaired fasting glycemia (OR=1.64, P=5.5x10(-11)) but not isolated impaired glucose tolerance.

900+ patients with gestational diabetes were studied, and the rs10830963(G) SNP was postulated to perhaps be a causal SNP for the condition, with an odds ratio of ~1.3 - 1.4.

Established associations:

body mass index **you have 1 copies of the risk allele G** (risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)
high density lipoprotein cholesterol measurement **you have 1 copies of the risk allele G** (risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

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triglyceride measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

body height **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

low density lipoprotein cholesterol measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

bone density **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

c-reactive protein measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

systolic blood pressure **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

total cholesterol measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

adiponectin measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

diastolic blood pressure **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

waist circumference **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

body weight **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

thyroid stimulating hormone measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

serum alanine aminotransferase measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

homocysteine measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

heart rate **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

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interleukin-6 measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

estradiol measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

icam-1 measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

aspartate aminotransferase measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

testosterone measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

ccl2 measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

birth weight **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

energy intake **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

fasting blood insulin measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

vitamin b12 measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

hormone measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

lean body mass **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

hip circumference **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

fatty acid measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

igfbp-3 measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

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maximal oxygen uptake measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

tumor necrosis factor-alpha measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

urinary metabolite measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

insulin sensitivity measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

blood urea nitrogen measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

homa-ir **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

ccl5 measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

igfbp-1 measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

igf-1 measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

amino acid measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

antioxidant measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

arm span **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

body composition measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

cortisol secretion measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

energy expenditure **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

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folic acid measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

leptin measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

gestational age **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

head circumference **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

metabolic rate measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

physical activity **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

sleep measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

transforming growth factor beta measurement **you have 1 copies of the risk allele G**

(risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661)

type ii diabetes mellitus **you have 1 copies of the risk allele G**

(risk allele=G, odds ratio/beta 1.11 [1.06-1.16] with pval 2E-7, pubmedid=24509480)

fasting blood glucose measurement **you have 1 copies of the risk allele G**

(risk allele=G, odds ratio/beta 0.07 [0.06-0.08] mmol/l increase with pval 3E-50, pubmedid=19060907; risk allele=G, GLU odds ratio/beta 0.05 mg/dL increase with pval 4E-8, pubmedid=23251661; risk allele=G, Fasting glucose odds ratio/beta 1.38 [1.01-1.75] mg/dL increase with pval 1E-12, pubmedid=22508271; risk allele=G complex/no impact summary; risk allele=G complex/no impact summary)

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Gene related topics:

glucose tolerance(1.00) · insulin(1.00) · METABOLIC(0.75) · fasting plasma glucose(0.75) · OTHER(0.64) · scoliosis(0.64) · bmi(0.25) · diabetes mellitus type ii diabetes mellitus type 2(0.20) · fasting glucose related traits(0.15) · glucose(0.06) · insulin resistance(0.04)

Relevant papers and external links:

PMID 24509480

Genome-wide trans-ancestry meta-analysis provides insight into the genetic architecture of type 2 diabetes susceptibility.

PMID 23251661
Novel genetic loci identified for the pathophysiology of childhood obesity in the hispanic population.

PMID 19060907
Variants in mtnr1b influence fasting glucose levels.

PMID 22508271
Fasting glucose gwas candidate region analysis across ethnic groups in the multiethnic study of atherosclerosis (mesa).

PMID 20081858
New genetic loci implicated in fasting glucose homeostasis and their impact on type 2 diabetes risk.

PMID 19241057

PMID 19088850

PMID 19324940

PMID 21658282

PHARMGKB PA165281934
Evidence: PubMed ID:20081858Phenotype 1: In a meta-analysis of 21 GWAS cohorts followed by analysis in additional individuals, this SNP was found to be associated with fasting glucose level. Study size: 112,844. Significance metric(s): p = 5.8 x 10(-175). Phenotype 2: In the same study, this SNP was found to be associated with HOMA-B (homeostasis model assessment of beta-cell function). Study size: 90,364. Significance metric(s): p = 2.7 x 10(-43). Study population/ethnicity: Non-diabetic Individuals of European descent. Type of association: CO;GN.

PHARMGKB PA164740301
Evidence: PubMed ID:19060907; Web Resource:External Link results: Variants in MTNR1B influence fasting glucose levels. (Initial Sample Size: 35,812 individuals; Replication Sample Size: NR); (Region: 11q21; Reported Gene(s): MTNR1B; Risk Allele: rs10830963-G); (p-value= 3E-50).This variant is associated with Fasting plasma glucose.

OMIM 613233

dbSNP: rs10830963 GWAS Catalog: rs10830963 Genecard: MTNR1B chromosome 11

SNPedia: rs10830963(C;G)

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★ **rs1004819(C;T)** warning:2.94 47.92% **1.5x risk of Crohn's disease**

SNP rs1004819, in the IL23R gene, is associated with increased risk for Crohn's disease in both Jewish and non-Jewish populations.

The same risk allele is also reported to increase the risk for developing ankylosing spondylitis, based on a large study of over 1,000 Caucasian patients. The odds ratio is 1.2 (p=8.8x10e-5).[PMID 17952073, PMID 18037607]

significant associations with rs1004819, rs7517847, and rs11209026. Having any CARD15 variant was associated with a significant risk for CD ($P < 0.0001$).

Investigation of the IL23R gene in a Spanish rheumatoid arthritis cohort.

rs1004819 is the main disease-associated IL23R variant in German Crohn's disease patients: combined analysis of IL23R, CARD15, and OCTN1/2 variants.

CARD15 and IL23R influences Crohn's disease susceptibility but not disease phenotype in a Brazilian population.

Genetic analysis of innate immunity in Crohn's disease and ulcerative colitis identifies two susceptibility loci harboring CARD9 and IL18RAP.

IL23R haplotypes provide a large population attributable risk for Crohn's disease.

Replication of interleukin 23 receptor and autophagy-related 16-like 1 association in adult- and pediatric-onset inflammatory bowel disease in Italy.

Lack of evidence for association of primary sclerosing cholangitis and primary biliary cirrhosis with risk alleles for Crohn's disease in Polish patients.

Association of polymorphisms in the Interleukin 23 receptor gene with osteonecrosis of femoral head in Korean population.

Ulcerative colitis-risk loci on chromosomes 1p36 and 12q15 found by genome-wide association study.

IL23R in the Swedish, Finnish, Hungarian and Italian populations: association with IBD and psoriasis, and linkage to celiac disease.

Genetic epistasis of IL23/IL17 pathway genes in Crohn's disease.

No association between interleukin 23 receptor gene polymorphisms and systemic lupus erythematosus.

Variants of the IL23R gene are associated with ankylosing spondylitis but not with Sjogren syndrome in Hungarian population samples.

Interleukin-23 receptor gene variants in Hungarian systemic lupus erythematosus patients.

Genome-wide association studies--a summary for the clinical gastroenterologist.

Association of interleukin 23 receptor polymorphisms with anti-topoisomerase-I positivity and pulmonary hypertension in systemic sclerosis.

Interaction of the major inflammatory bowel disease susceptibility alleles in Crohn's disease patients.

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The cannabinoid 1 receptor (CNR1) 1359 G/A polymorphism modulates susceptibility to ulcerative colitis and the phenotype in Crohn's disease.

NOD2/CARD15, ATG16L1 and IL23R gene polymorphisms and childhood-onset of Crohn's disease.

Evidence for STAT4 as a common autoimmune gene: rs7574865 is associated with colonic Crohn's disease and early disease onset.

Interleukin-23 receptor genetic polymorphisms and ankylosing spondylitis susceptibility: a meta-analysis.

An investigation of genome-wide studies reported susceptibility loci for ulcerative colitis shows limited replication in north Indians.

Gene related topics:

IMMUNE(5.16) · inflammatory bowel diseases(5.16) · colitis ulcerative crohn disease(1.98) · arthritis psoriatic psoriasis(1.12) · crohn disease crohn s disease inflammatory bowel disease nos inflammatory bowel diseases(1.00) · celiac disease(1.00) · arthritis(1.00) · cirrhosis(1.00) · rheumatoid arthritis(1.00) · primary biliary cirrhosis(1.00) · systemic sclerosis(1.00) · hypertension(1.00) · systemic lupus erythematosus(1.00) · lupus erythematosus(1.00) · lupus(1.00) · crohn disease(0.96) · crohn s disease(0.64) · inflammatory bowel disease(0.54) · spondylitis ankylosing(0.44) · inflammatory bowel disease nos inflammatory bowel diseases(0.43) · colitis ulcerative(0.42) · chronic ulcerative colitis colitis ulcerative crohn disease crohn s disease inflammatory bowel disease nos inflammatory bowel diseases(0.40) · ulcerative colitis(0.24) · crohn disease crohn s disease(0.23) · psoriasis(0.20) · behcet syndrome(0.17) · crohn s disease ulcerative colitis(0.15) · ankylosing spondylitis(0.08) · lupus erythematosus systemic systemic lupus erythematosus(0.03)

Relevant papers and external links:

PMID 17068223

PMID 18047539

OMIM 612261

OMIM 607562

PHARMGKB PA162356489

Evidence: PubMed ID:17804789; Web Resource:External Link results: Genome-wide association study for Crohn's disease in the Quebec Founder Population identifies multiple validated disease loci (Initial Sample Size: 382 trios; Replication Sample Size: 521 trios, 750 cases, 828 controls). This variant is associated with Crohn's disease. This variant is associated with Crohn's disease.

PMID 17678723

PMID 17786191

PMID 18200510

PMID 18439550

PMID 18470928

PMID 18698678

PMID 18715515

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PMID 18779654
PMID 19122664
PMID 19175939
PMID 19235914
PMID 19306001
PMID 19522770
PMID 19757086
PMID 19916168
PMID 19918037
PMID 20066736
PMID 20195480
PMID 20380008
PMID 20454450
PMID 21253733
PMID 21304977

dbSNP: rs1004819 Genecard: IL23R chromosome 1 SNPedia: rs1004819(C;T)

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★ rs11623713(A;G) warning:2.92 16.00% **; body mass index (you have the risk allele A)**

Established associations:

body mass index **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

triglyceride measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

high density lipoprotein cholesterol measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

body height **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

low density lipoprotein cholesterol measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

bone density **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

c-reactive protein measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

fasting blood glucose measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

systolic blood pressure **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

adiponectin measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

diastolic blood pressure **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

total cholesterol measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

waist circumference **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

body weight **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

thyroid stimulating hormone measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

serum alanine aminotransferase measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

interleukin-6 measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

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heart rate **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

homocysteine measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

testosterone measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

estradiol measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

icam-1 measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

ccl2 measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

aspartate aminotransferase measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

birth weight **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

energy intake **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

vitamin b12 measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

fasting blood insulin measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

hormone measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

lean body mass **you have 1 copies of the risk allele A**

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(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

igfbp-3 measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

insulin sensitivity measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

maximal oxygen uptake measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

hip circumference **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

fatty acid measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

tumor necrosis factor-alpha measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

urinary metabolite measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

blood urea nitrogen measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

homa-ir **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

arm span **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

antioxidant measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

amino acid measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

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igf-1 measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

igfbp-1 measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

ccl5 measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

body composition measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

transforming growth factor beta measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

sleep measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

physical activity **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

metabolic rate measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

leptin measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

head circumference **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

gestational age **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

folic acid measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

energy expenditure **you have 1 copies of the risk allele A**

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(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

cortisol secretion measurement **you have 1 copies of the risk allele A**

(risk allele=A, Free T3 odds ratio/beta 0.03 pg/mL increase with pval 8E-6, pubmedid=23251661)

Relevant papers and external links:

PMID 23251661
Novel genetic loci identified for the pathophysiology of childhood obesity in the hispanic population.

dbSNP: rs11623713 GWAS Catalog: rs11623713 Genecard: nan chromosome 14

SNPedia: rs11623713(A;G)

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★ **gs285** **info:2.92** **claimed to lose 2.5x as much weight on a low fat diet**

An interesting hypothesis, but not well validated, this patent is summarized in a simplified blog post and suggests a genotype to suggest diet and lifestyle changes which may be beneficial.

You are part of a group of 39% of all people.
Source: External Link and External Link

See also gs281, gs282, gs283, gs284

based on rs1799883 rs1801282 rs1042714 rs4994 and rs1042713

Gene related topics:

weight(5.00)

SNPedia: gs285

★ **rs17085007(C;T)** **warning:2.90** **29.97%** **1.35x increased risk for ulcerative colitis**

Established associations:

ulcerative colitis **you have 1 copies of the risk allele C**

(risk allele=C, odds ratio/beta 1.35 [1.21-1.51] with pval 7E-8, pubmedid=19915573; risk allele=C, odds ratio/beta 1.37 [1.22-1.55] with pval 3E-7, pubmedid=23511034; risk allele=C, odds ratio/beta 1.4 [1.21-1.63] with pval 8E-6, pubmedid=23511034; risk allele=C, odds ratio/beta 1.16 [1.10-1.21] with pval 1E-16, pubmedid=21297633)

inflammatory bowel disease **you have 1 copies of the risk allele C**

(risk allele=C, odds ratio/beta 1.106 [1.065-1.147] with pval 3E-19, pubmedid=23128233)

Relevant papers and external links:

PMID 19915573
A genome-wide association study identifies three new susceptibility loci for ulcerative colitis in the japanese population.

PMID 23511034
Genome-wide association study of ulcerative colitis in koreans suggests extensive overlapping of genetic susceptibility with caucasians.

PMID 21297633
Meta-analysis identifies 29 additional ulcerative colitis risk loci, increasing the number of confirmed associations to 47.

PMID 23128233
Host-microbe interactions have shaped the genetic architecture of inflammatory bowel disease.

OMIM 612796

dbSNP: rs17085007 GWAS Catalog: rs17085007 Genecard: nan chromosome 13

SNPedia: rs17085007(C;T)

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★ **rs2241880(C;C)** **warning:2.90** **15.68%** **2x-3x increased risk for Crohn's disease in Caucasians**

rs2241880, a SNP in the ATG16L1 gene encoding a threonine to alanine substitution ("T300A") in a protein known to be involved in the function of the epithelial cells lining the intestine, has been associated with Crohn's disease in several recent studies. [PMID 17200669, PMID 17435756]

In another recent (2007) report, rs2241880 is confirmed to be associated with both Crohn's disease and ileal disease, but additionally, the authors calculate risk for individuals who are homozygotes for this SNP plus 2 others (in the IBD5 and NOD2 genes). Individuals homozygous for the risk alleles for

all 3 of these SNPs are estimated to be at 20 fold higher risk (CI ~9-49) for Crohn's disease. From the largest most recent survey, the Crohn's disease-associated SNPs for IBD5 and NOD2 are, respectively, rs6596075 and rs17221417.

associated With Inflammatory Bowel Diseases but Not With Celiac Disease
rs11209026 had a protective effect for IBD in the case-control analysis (odds ratio [OR] 0.19, 95% confidence interval [CI] 0.10-0.37, P= 6.6E-09). Both CD (OR 0.14, CI 0.06-0.37, P= 3.9E-07) and UC (OR 0.33, CI 0.15-0.73, P= 1.4E-03) were associated with IL23R. rs2241880 was associated with CD susceptibility (OR 1.36, CI 1.12-1.66, P= 0.0017). The population-attributable risk of carrying allele G is 0.24 and is 0.19 for homozygosity for allele G in CD.

In another study, rs2241880 has been associated with Crohn's disease; the minor allele is somewhat protective in that it lessens the odds of acquiring the disease (odds ratio 0.74, CI: 0.65-0.84, p=3.7x10e-6).

Review of role of rs2241880 in Crohn's disease and possibly ulcerative colitis

Replicated increased risk for Crohn's disease with rs2241880(C) allele in a study of Italian patients, but saw no association to ulcerative colitis (UC).

strongly associated with ileal Crohn's disease (allelic P = 1.24 x 10(-6)). Children with GG genotype had a more than 3-fold elevated risk for disease as compared to the wildtype AA homozygotes (odds ratio [OR], 3.1; 95% confidence interval [CI], 1.93-4.94; P = 1.8 x 10(-6))

Meta-analysis of 24 studies performed, including 13,022 Crohn's disease cases and 17,532 controls. Confirmed increased risk for disease for carriers of (C) allele (1.9x or 1.4x, for homozygous and heterozgous genotypes), but only in Caucasians and not in Asians.

Study of 557 CD and 425 UC patients and 672 ethnically matched Spanish controls and a meta-analysis confirmed an association between rs2241880(C) and CD (p=6.5 x 10(-9), odds ratio =1.62).

A genome-wide association study identifies IL23R as an inflammatory bowel disease gene.

Novel Crohn disease locus identified by genome-wide association maps to a gene desert on 5p13.1 and modulates expression of PTGER4.

Confirmation of the role of ATG16L1 as a Crohn's disease susceptibility gene.

Systematic association mapping identifies NELL1 as a novel IBD disease gene.

IL23R R381Q and ATG16L1 T300A are strongly associated with Crohn's disease in a study of New Zealand Caucasians with inflammatory bowel disease.

Fine mapping versus replication in whole-genome association studies.

Autophagy gene ATG16L1 influences susceptibility and disease location but not childhood-onset in Crohn's disease in Northern Europe.

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CARD15 and IL23R influences Crohn's disease susceptibility but not disease phenotype in a Brazilian population.

[Correlation of the autophagosome gene ATG16L1 polymorphism and inflammatory bowel disease].

ATG16L1 and IL23 receptor (IL23R) genes are associated with disease susceptibility in Hungarian CD patients.

Lack of evidence for association of primary sclerosing cholangitis and primary biliary cirrhosis with risk alleles for Crohn's disease in Polish patients.

Gene variants influencing measures of inflammation or predisposing to autoimmune and inflammatory diseases are not associated with the risk of type 2 diabetes.

Searching for genotype-phenotype structure: using hierarchical log-linear models in Crohn disease.

Autophagy 16-like 1 rs2241880 G allele is associated with Crohn's disease in German children.

Lack of association of NKX2-3, IRGM, and ATG16L1 inflammatory bowel disease susceptibility variants with celiac disease.

Genome-wide association studies--a summary for the clinical gastroenterologist.

Interaction of the major inflammatory bowel disease susceptibility alleles in Crohn's disease patients.

NOD2, IL23R and ATG16L1 polymorphisms in Lithuanian patients with inflammatory bowel disease.

The cannabinoid 1 receptor (CNR1) 1359 G/A polymorphism modulates susceptibility to ulcerative colitis and the phenotype in Crohn's disease.

T300A polymorphism of ATG16L1 and susceptibility to inflammatory bowel diseases: a meta-analysis.

Is there a role for Crohn's disease-associated autophagy genes ATG16L1 and IRGM in formation of granulomas?

Evidence for STAT4 as a common autoimmune gene: rs7574865 is associated with colonic Crohn's disease and early disease onset.

ATG16L1 polymorphisms are associated with NOD2-induced hyperinflammation.

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Established associations:

crohn's disease **you have 2 copies of the risk allele C**

(risk allele=C, odds ratio/beta 1.32 [1.24-1.41] with pval 1E-12, pubmedid=22412388; risk allele=C, odds ratio/beta 1.45 [1.27-1.64] with pval 1E-13, pubmedid=17435756)

Gene related topics:

IMMUNE(1.05) · colitis ulcerative crohn disease(1.05) · celiac disease(1.00) · cirrhosis(1.00) · primary biliary cirrhosis(1.00) · ulcerative colitis(1.00) · type 2 diabetes(1.00) · inflammation(1.00) · crohn disease(0.81) · inflammatory bowel disease nos inflammatory bowel diseases(0.43) · inflammatory bowel disease(0.33) · inflammatory bowel diseases(0.16) · crohn s disease(0.13) · crohn disease crohn s disease(0.08) · crohn s disease ulcerative colitis(0.04)

Relevant papers and external links:

PMID 22412388
A genome-wide scan of ashkenazi jewish crohn's disease suggests novel susceptibility loci.

PMID 17435756
Genome-wide association study identifies new susceptibility loci for crohn disease and implicates autophagy in disease pathogenesis.

PMID 17484864

PMID 17554300

PMID 18047540

PMID 18162085

PMID 18366306

PMID 18698678

PMID 18985712

PMID 19337756

PMID 19491842

OMIM 611081

OMIM 266600

OMIM 610767

PHARMGKB PA162356605
Evidence: PubMed ID:17435756; Web Resource:External Link Results: Genome-wide association study identifies new susceptibility loci for Crohn disease and implicates autophagy in disease pathogenesis (Initial Sample Size: 946 cases, 977 controls; Replication Sample Size: 530 trios, 353 cases, 207 controls; Risk Allele: rs2241880-G).

OMIM 610767

PMID 17068223

PMID 17447842

PMID 17455206

PMID 17684544

PMID 17894849

PMID 17924341

PMID 18088053

PMID 18200510

PMID 18495612

PMID 18499543

PMID 18715515

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PMID 18853133
PMID 19185283
PMID 19659808
PMID 19683022
PMID 19916168
PMID 20066736
PMID 20082483
PMID 20195480
PMID 20222171
PMID 20395867
PMID 20454450
PMID 21673517

dbSNP: rs2241880 GWAS Catalog: rs2241880 Genecard: ATG16L1 chromosome 2

SNPedia: rs2241880(C;C)

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★ rs10468017(T;T) good:2.89 3.71%(rare) **associated with higher HDL cholesterol**

T allele is associated with 1.76mg/dl increase in HDL cholesterol (good cholesterol).

Established associations:

high density lipoprotein cholesterol measurement **you have 2 copies of the risk allele T**

(risk allele=T, HDL odds ratio/beta 0.104 [0.075-0.133] mmol/l increase with pval 3E-12, pubmedid=21943158; risk allele=T, odds ratio/beta 0.1 [0.06-0.14] s.d. increase with pval 8E-23, pubmedid=19060906)

triglyceride measurement **you have 2 copies of the risk allele T**

(risk allele=T, HDL odds ratio/beta 0.104 [0.075-0.133] mmol/l increase with pval 3E-12, pubmedid=21943158)

low density lipoprotein cholesterol measurement **you have 2 copies of the risk allele T**

(risk allele=T, HDL odds ratio/beta 0.104 [0.075-0.133] mmol/l increase with pval 3E-12, pubmedid=21943158)

c-reactive protein measurement **you have 2 copies of the risk allele T**

(risk allele=T, HDL odds ratio/beta 0.104 [0.075-0.133] mmol/l increase with pval 3E-12, pubmedid=21943158)

uric acid measurement **you have 2 copies of the risk allele T**

(risk allele=T, HDL odds ratio/beta 0.104 [0.075-0.133] mmol/l increase with pval 3E-12, pubmedid=21943158)

serum gamma-glutamyl transferase measurement **you have 2 copies of the risk allele T**

(risk allele=T, HDL odds ratio/beta 0.104 [0.075-0.133] mmol/l increase with pval 3E-12, pubmedid=21943158)

aspartate aminotransferase measurement **you have 2 copies of the risk allele T**

(risk allele=T, HDL odds ratio/beta 0.104 [0.075-0.133] mmol/l increase with pval 3E-12, pubmedid=21943158)

ferritin measurement **you have 2 copies of the risk allele T**

(risk allele=T, HDL odds ratio/beta 0.104 [0.075-0.133] mmol/l increase with pval 3E-12, pubmedid=21943158)

butyrylcholinesterase measurement **you have 2 copies of the risk allele T**

(risk allele=T, HDL odds ratio/beta 0.104 [0.075-0.133] mmol/l increase with pval 3E-12, pubmedid=21943158)

blood metabolite measurement **you have 2 copies of the risk allele T**

(risk allele=T, 1-palmitoylglycerophosphoethanolamine odds ratio/beta 0.019 [0.013-0.025] unit increase with pval 3E-12, pubmedid=24816252)

metabolic syndrome **you have 0 copies of the risk allele C**

(risk allele=C, HDLC-WC odds ratio/beta 0.16 [0.10-0.22] unit increase with pval 6E-8, pubmedid=21386085)

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Gene related topics:

cholesterol(5.00) · hdl cholesterol(5.00)

Relevant papers and external links:

PMID 21943158
Genetic variants in lpl, oasl and tomm40/apoe-c1-c2-c4 genes are associated with multiple cardiovascular-related traits.

PMID 19060906
Common variants at 30 loci contribute to polygenic dyslipidemia.

PMID 21386085
A bivariate genome-wide approach to metabolic syndrome: stampeed consortium.

PMID 24816252
An atlas of genetic influences on human blood metabolites.

PMID 18193043

PHARMGKB PA164740244

Evidence: PubMed ID:19060906; Web Resource:External Link results: Common variants at 30 loci contribute to polygenic dyslipidemia. (Initial Sample Size: 19,840 individuals; Replication Sample Size: Up to 20,623 individuals); (Region: 15q22.1; Reported Gene(s): LIPC; Risk Allele: rs10468017-T); (p-value= 8E-23).This variant is associated with HDL cholesterol.

dbSNP: rs10468017 GWAS Catalog: rs10468017 Genecard: nan chromosome 15

SNPedia: rs10468017(T;T)

★ gs101 good:2.88 **probably able to digest milk**

77% of Europeans with this are able to digest lactose and dairy products.

People without this are more likely to experience lactose intolerance.

Gene related topics:

lactose intolerance(1.00)

Relevant papers and external links:

PMID 11788828
PMID 15114531

SNPedia: gs101

★ gs159 good:2.88 **CYP1A2 fast metabolizer**

Your CYP1A2 fast metabolizer status means that you are less stimulated by caffeine.

Ciprofloxacin is also metabolized by CYP1A2, but is unclear if your genotype should influence its effect.

SNPedia: gs159

★ rs9272346(A;A) warning:2.74 **normal, but higher risk for type-1 diabetes**

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rs9272346 has been reported in a large study to be associated with type-1 diabetes.

The risk allele (oriented to the dbSNP entry) is (A); the odds ratio associated with heterozygotes is 5.49 (CI 4.83-6.24), and for homozygotes, 18.52 (CI 12.69-27.03).

Note that the (A) allele is the most common in various human populations; instead of labeling (A) as a risk allele, we consider the (G) allele protective.

Risk alleles for multiple sclerosis identified by a genomewide study.

Pharmacogenetics: data, concepts and tools to improve drug discovery and drug treatment.

Candidate causal regulatory effects by integration of expression QTLs with complex trait genetic associations.

A study of CNVs as trait-associated polymorphisms and as expression quantitative trait loci.

Established associations:

type i diabetes mellitus **you have 0 copies of the risk allele G**

(risk allele=G complex/no impact summary; risk allele=G, odds ratio/beta 5.49 [4.83-6.24] with pval 5E-134, pubmedid=17554300)

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Gene related topics:

IMMUNE(6.50) · diabetes type 1(6.50) · celiac disease(5.61) · diabetes mellitus insulin dependent diabetes mellitus type 1(2.59) · CARDIOVASCULAR(2.00) · hypertension stroke lacunar atherothrombotic brain infarction(2.00) · OTHER(1.00) · myasthenia gravis(0.69) · multiple sclerosis diabetes type 1(0.67) · multiple sclerosis optic neuritis(0.67) · urticaria(0.67) · UNKNOWN(0.58) · diabetes type 2 diabetes type 1(0.58) · myositis(0.50) · rheumatic fever(0.50) · NEUROLOGICAL(0.44) · guillain barre syndrome(0.44) · diabetes mellitus type 1(0.34) · autoimmune diseases(0.33) · CANCER(0.30) · cervical cancer(0.30) · antiphospholipid syndrome(0.27) · multiple sclerosis(0.26) · REPRODUCTION(0.26) · pregnancy loss recurrent(0.26) · graves disease graves disease(0.25) · lupus erythematosus(0.24) · alopecia areata(0.24) · METABOLIC(0.22) · cholangitis sclerosing(0.22) · INFECTION(0.22) · hepatitis c chronic(0.22) · abortion spontaneous(0.21) · rheumatic heart disease(0.21) · cirrhosis biliary primary(0.19) · nasopharyngeal cancer(0.19) · hepatitis b(0.18) · leprosy(0.18) · abdominal aortic aneurysm(0.17) · lupus erythematosus systemic(0.14) · arthritis(0.12) · vitiligo(0.10) · hepatitis b chronic(0.10) · diabetes mellitus type ii diabetes mellitus type 2(0.09) · h pylori infection(0.07) · graves disease(0.07) · cardiomyopathy(0.06) · helicobacter infections stomach neoplasms(0.06) · lymphoma(0.06) · psoriasis(0.06) · melanoma(0.06) · systemic lupus erythematosus(0.06) · graft versus host disease(0.05) · rheumatoid arthritis(0.04) · arthritis rheumatoid(0.03) · stomach cancer(0.03)

Relevant papers and external links:

PMID 18978792
Meta-analysis of genome-wide association study data identifies additional type 1 diabetes risk loci.

PMID 17554300

PHARMGKB PA164740758
Evidence: PubMed ID:18978792; Web Resource:External Link results: Meta-analysis of genome-wide association study data identifies additional type 1 diabetes risk loci. (Initial Sample Size: 3,561 cases, 4,646 controls; Replication Sample Size: 6,225 cases, 6,946 controls, 3,064 trios); (Region: 6p21.32; Reported Gene(s): HLA; Risk Allele: rs9272346-G); (p-value= 6E-129).This variant is associated with Type 1 diabetes.

PHARMGKB PA162355906
Evidence: PubMed ID:17554300A genome-wide association study in 2,000 individuals for each of 7 major diseases and a shared set of 3,000 controls found an association of this SNP with type 1 diabetes.

PMID 17660530
PMID 18224312
PMID 20369022
PMID 21304891

dbSNP: rs9272346 GWAS Catalog: rs9272346 Genecard: HLA-DQA1 chromosome 6

SNPedia: rs9272346(A;A)

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★ rs1344706(T;T) warning:2.70 48.37% **1.2x increased risk for schizophrenia**

rs1344706 is a SNP in the ZNF804A gene, and it is probably the first SNP to have been reported as achieving genome-wide significance for psychosis.

Reviews the literature, and concludes that rs1344706 is "robustly, if modestly" associated with increased risk for schizophrenia.

A genome-wide association study of schizophrenia (479 cases, 2,937 controls) followed by additional testing and a meta-analysis concluded that the strongest evidence for association was around ZNF804A (rs1344706; p = 1.61 x 10e-7) and that the finding was strengthened when the affected phenotype included bipolar disorder (p = 9.96 x 10e-9).

Executive function, neural circuitry, and genetic mechanisms in schizophrenia.

Apoptotic engulfment pathway and schizophrenia.

Persistence criteria for susceptibility genes for schizophrenia: a discussion from an evolutionary viewpoint.

Expanding the range of ZNF804A variants conferring risk of psychosis.

Effects of a genome-wide supported psychosis risk variant on neural activation during a theory-of-mind task.

Fine mapping of ZNF804A and genome-wide significant evidence for its involvement in schizophrenia and bipolar disorder.

New Genetic Findings in Schizophrenia: Is there Still Room for the Dopamine Hypothesis of Schizophrenia?

ZNF804A risk allele is associated with relatively intact gray matter volume in patients with schizophrenia.

The impact of a genome-wide supported psychosis variant in the ZNF804A gene on memory function in schizophrenia.

ZNF804A may be associated with executive control of attention.

Evaluation of risk loci for schizophrenia derived from genome-wide association studies in a German population.

Evidence of sex-modulated association of ZNF804A with schizophrenia.

Impact on schizotypal personality trait of a genome-wide supported psychosis variant of the ZNF804A gene.

ANK3, CACNA1C and ZNF804A gene variants in bipolar disorders and psychosis subphenotype.

Altered cortical network dynamics: a potential intermediate phenotype for schizophrenia and association with ZNF804A.

The schizophrenia risk gene ZNF804A influences the antipsychotic response of positive schizophrenia symptoms.

Association of the ZNF804A gene polymorphism rs1344706 with white matter density changes in Chinese schizophrenia.

Genome-wide supported psychosis risk variant in ZNF804A gene and impact on cortico-limbic WM integrity in schizophrenia.

ZNF804a regulates expression of the schizophrenia-associated genes PRSS16, COMT, PDE4B, and DRD2.

The effects of psychosis risk variants on brain connectivity: a review.

Association analysis of ZNF804A (zinc finger protein 804A) rs1344706 with therapeutic response to atypical antipsychotics in first-episode Chinese patients with schizophrenia.

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Established associations:

schizophrenia **you have 2 copies of the risk allele T**

(risk allele=T, odds ratio/beta 1.12 with pval 2E-7, pubmedid=18677311)

Gene related topics:

schizophrenia(5.00) · bipolar disorder(1.00) · personality(1.00) · memory(1.00) · brain(1.00) · PSYCH(0.27) · psychosis(0.27)

Relevant papers and external links:

PMID 20688871

PMID 18677311

OMIM 181500

OMIM 612361

PHARMGKB PA164856914
Evidence: PubMed ID:19407193Rs1344706, found in a separate GWAS to be associated with schizophrenia, was investigated here by Functional Magnetic Resonance Imaging. In 115 healthy German subjects, carriers of the "higher risk" allele(defined in this study as A) showed gene dosage-dependent alterations in functional coupling of dorsolateral prefrontal cortex across hemispheres and with hippocampus and also abnormal coupling of amygdala.

PHARMGKB PA162356547
Evidence: PubMed ID:18677311; Web Resource:External Link Results: Identification of loci associated with schizophrenia by genome-wide association and follow-up (Initial Sample Size: 479 cases, 2,937 controls; Replication Sample Size: 6,666 cases, 9,897 controls; Risk Allele: rs1344706-T). This variant is associated with Schizophrenia.

PMID 19693005

PMID 19721717

PMID 19911060

PMID 20048749

PMID 20231838

PMID 20368704

PMID 20485477

PMID 20934520

PMID 20957649

PMID 21040459

PMID 21302348

PMID 21349497

PMID 21457757

PMID 21767209

PMID 21810628

PMID 21892778

PMID 21911029

PMID 22328493

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PMID 22384243
PMID 22416237
PMID 22425527

dbSNP: rs1344706 GWAS Catalog: rs1344706 Genecard: ZNF804A chromosome 2

SNPedia: rs1344706(T;T)

★ rs806380(G;G) good:2.59 4.22%(rare) **uncommon. lowest odds of cannabis dependence**

This snp, located in intron 2 of the central cannabinoid receptor CNR1 gene, has been linked to cannabis dependence.

The (G) allele is reported to have a protective effect, and is a defining part of a haplotype that is associated with lower odds of developing cannabis dependence. The (A) allele is more common in all populations studied.

Variation in the human cannabinoid receptor CNR1 gene modulates gaze duration for happy faces. (G;G) is also associated with the longest gaze duration for happy faces.

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Gene related topics:

CHEMDEPENDENCY(0.67) · marijuana abuse(0.67)

Relevant papers and external links:

PMID 16917946
PMID 21714860

dbSNP: rs806380 Genecard: CNR1 chromosome 6 SNPedia: rs806380(G;G)

★ rs4129148(C;-) warning:2.57 **3x risk of schizophrenia.**

Located in the Pseudoautosomal region

rs4129148, located near the CSF2RA gene, has been reported in a whole genome association study to be associated with schizophrenia.

The risk allele (oriented to the dbSNP entry) is (G); the odds ratio associated with this allele is 3.23 (CI 2.04 - 5.15).

Given the results from the recent massive Psychiatric Genomics Consortium GWAS, it appears very unlikely
that any large effect common SNPs exist for schizophrenia. It is not completely clear, though, how the previously
replicated results for rs4129148 might be explained. It is currently predicted that 10,000 SNPs contribute to
schizophrenic illness.

The diagnosis for schizophrenia has now possibly shifted from asking: Schizophrenic? Yes or No to How much Schizophrenic?

Established associations:

schizophrenia **you have 1 copies of the risk allele C**

(risk allele=C, odds ratio/beta 3.23 [2.04-5.15] with pval 4E-7, pubmedid=17522711)

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Gene related topics:

schizophrenia(5.00)

Relevant papers and external links:

PMID 17522711

dbSNP: rs4129148 GWAS Catalog: rs4129148 Genecard: CSF2RA chromosome X

SNPedia: rs4129148(C;G)

★ gs281 warning:2.57 **part of the 88% of the population claimed not to maintain weight loss unless you perform high energy exercise**

An interesting hypothesis, but not well validated, this patent is summarized in a simplified blog post and suggests a genotype to suggest diet and lifestyle changes which may be beneficial.

Based on 2 snps
rs4994
rs1042713

Gene related topics:

weight loss(5.00) · weight(5.00)

Relevant papers and external links:

PMID 12917707

SNPedia: gs281

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★ rs6313(C;T) warning:2.56 49.31% **higher risk for RA**

rs6313, also known as T102C, is part of a 4-SNP haplotype in the serotonin 2A receptor gene HTR2A that has been associated with rheumatoid arthritis in a study of 1800 European patients. The risk allele is rs6313(C). The overall risk for the haplotype CTCC of SNPs rs6311-rs1328674-rs6313-rs6314 is 1.68 (CI: 1.20 - 2.34, p = 0.02).

Note: the orientation of rs1328674 in dbSNP is opposite that cited by this publication; therefore, with respect to dbSNP, the haplotype of risk as cited above is CACC rather than CTCC.

see also gs108 and gs226

Polymorphisms in the serotonin receptor gene HTR2A are associated with quantitative traits in panic disorder.

Gene related topics:

PSYCH(3.00) · anorexia nervosa(3.00) · anorexia nervosa bulimia(3.00) · PHARMACOGENOMIC(2.00) · impulsive behavior(2.00) · olanzapine(2.00) · response to clozapine(2.00) · schizophrenia affective disorder(2.00) · schizophrenia(1.64) · arthritis(1.00) · rheumatoid arthritis(1.00) · quantitative traits(1.00) · bulimia(0.94) · major depression(0.80) · suicidal behavior(0.69) · major depressive disorder(0.68) · depression(0.67) · obsessive compulsive disorder(0.58) · neuroleptic malignant syndrome(0.57) · schizophrenia tardive dyskinesia(0.57) · suicide(0.56) · personality disorders(0.50) · personality traits(0.47) · cognitive function(0.33) · depressive disorder major(0.29) · autism(0.28) · adhd(0.26) · mood disorders(0.23) · panic disorder(0.21) · adhd attention deficit hyperactivity disorder(0.21) · attention deficit hyperactivity disorder(0.21) · tourette syndrome(0.21) · bipolar disorder(0.20) · personality(0.14) · CHEMDEPENDENCY(0.13) · heroin abuse(0.13) · NEUROLOGICAL(0.11) · migraine(0.11) · tardive

dyskinesia(0.11) · smoking behavior(0.09) · CARDIOVASCULAR(0.05) · blood pressure determination(0.05) · alcohol abuse(0.04)

Relevant papers and external links:

PMID 18006541

PHARMGKB PA165109704
Evidence: PubMed ID:19494443Risk or phenotype-associated allele (s): T/T. Phenotype: Patients, with a diagnosis of probable Alzheimer's disease, carrying the TT genotype were the most delusional during the follow-up period of a study evaluating the association of HTR2A 102T/C polymorphism with psychotic symptom severity. Patients with delusion symptoms carrying the CT and TT genotypes were resistant to the treatment with antipsychotic drugs. Study size: 80. Study population/ethnicity: Caucasian

PMID 17440930

PHARMGKB PA164889040
Evidence: PubMed ID:19193342T allele is associated with olanzapine-induced weight gain

dbSNP: rs6313 Genecard: HTR2A chromosome 13 SNPedia: rs6313(C;T)

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★ rs53576(G;G) good:2.50 37.29% **Optimistic and empathetic; handle stress well**

The one in four subjects who inherited a variation in this allele called G/G were significantly better at accurately reading the emotions of others by observing their faces than were the remaining three-quarters of subjects, who had inherited either a pair of A's or an A and a G from their parents at this site. Compared to the three-fourths with A/A or A/G variations, the G/G individuals were also less likely to startle when blasted by a loud noise, or to become stressed at the prospect of such a noise. And by their own reports, the G/G subjects were mellower and more attuned to other people than were the A/As or A/Gs. news

Effect on Parental Sensitivity

Controlling for maternal education, depression and marital discord, OXTR [F (1,152)=4.32, P=0.04, partial n2=0.03] gene was significantly associated with maternal sensitivity. Mothers with OXTR AA or AG genotypes were less sensitive than mothers with the GG genotype. The genetic difference accounts for 3% influence on variation in sensitive parenting.
[Bakermans-Kranenburg MJ, van Ijzendoorn MH. Oxytocin receptor (OXTR) and serotonin transporter (5-HTT) genes associated with observed parenting. Soc Cogn Affect Neurosci. 2008 Jun;3(2):128-34.]

In another study, heart rate responses of 40 healthy females without children were measured during the presentation of three episodes of infant cry sounds. Participants with the presumably more efficient variant of the oxytonergic system gene (OXTR GG) had more pronounced physiological

reactivity to repeated cry sounds, except when they showed more symptoms of depression. [Riem MM, Pieper S, Out D, Bakermans-Kranenburg MJ, van Ijzendoorn MH. Oxytocin receptor gene and depressive symptoms associated with physiological reactivity to infant crying. Soc Cogn Affect Neurosci. 2010 Apr 16.]

optimism

Yet another study () found that people with the G;G genotype were better able to discern the emotional state of others and to handle stress, compared to those who carry the A-allele. The study encompassed 192 participants of both sexes and mixed ethnicities. The study subjects underwent a number of tests to determine their level of empathy and stress reactivity. They found that G;G individuals performed significantly better on the behavioral measure of empathy and were 22.7% less likely to make a mistake on the "Reading the Mind in the Eyes Test" (RMET) (a behavioural measure of empathic accuracy) than A;A/A;G individuals (P = 0.005). Similarly, G;G individuals reported higher levels of dispositional empathy than A;A/A;G individuals: mean (SE) = 3.69 (0.06) and 3.53 (0.04) for G;G and A;A/A;G, respectively (P = 0.025), and were less affected by stress (as measured by their heart rates): mean (SE) = 72.1 (0.54) and 78.4 (1.19) for G;G and A;A/A;G, respectively. rs53576 is a silent G to A change in the oxytocin receptor (OXTR) gene. Studies have demonstrated that individuals with the G allele are more empathetic, feel less lonely, employ more sensitive parenting techniques, and have lower rates of autism (discussed in)

The blog Not Exactly Rocket Science discusses that Americans with (G;G) tend to be more sensitive parents, more empathetic and less lonely than those with an ‘A’. In a Korean population people with (G;G) were less likely to seek support from their peers.

Culture, distress, and oxytocin receptor polymorphism (OXTR) interact to influence emotional support seeking.

Oxytocin receptor genetic variation relates to empathy and stress reactivity in humans. In brief, people with the G;G genotype were better able to discern the emotional state of others than those who carried the A-allele (blog summary found here). The study encompassed 192 participants of both sexes and mixed ethnicities. The study subjects underwent a number of tests to determine their level of empathy and stress reactivity. They found that G;G individuals performed significantly better on the behavioral measure of empathy and were 22.7% less likely to make a mistake on the "Reading the Mind in the Eyes Test" (RMET) (a behavioural measure of empathic accuracy) than A;A/A;G individuals (P = 0.005). Similarly, G;G individuals reported higher levels of dispositional empathy than A;A/A;G individuals: mean (SE) = 3.69 (0.06) and 3.53 (0.04) for G;G and A;A/A;G, respectively (P = 0.025), and were less affected by stress (as measured by their heart rates): mean (SE) = 72.1 (0.54) and 78.4 (1.19) for G;G and A;A/A;G, respectively.

Associations between the oxytocin receptor gene (OXTR) and affect, loneliness and intelligence in normal subjects.

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This study examined the association between the OXTR gene and parenting. A total of 176 mothers of toddlers were included in the study. After controlling for marital discord, depression, and education status, rs53576 was found to significantly correlate with parenting, with the G;G genotype being associated with a significantly more sensitive parenting style than A;A or A;G genotypes. However, they concluded that the major factor influencing parenting was the maternal education level.

Forty-five female subjects (17 twin pairs and 11 twins without their sibling) were genotyped and tested for their ability to hear and understand conversations in noisy environments. Participants with the G;G genotype reported less difficulty hearing and understanding (difficulty hearing total score [Mean (SD)], 9.2 (4.2)) than participants with A;A/A;G genotypes (13.9 (3.8); $p < 0.001$). Even after adjusting for age, the relationship between genotype and difficulty hearing scores was still significant ($p = 0.003$).

Two hundred three healthy German male university students (mean age 23.2 y, SD 2.9 y) were subjected to a mock interview and their saliva cortisol levels were tested before and at a number of time points after the interview. Half of the participants were told to bring a female friend as social support, whereas the other half came alone to the interview. The authors found that subjects carrying the G allele (G;G or A;G genotypes) had significantly lower cortisol responses to stress when they had social support ($P < 0.01$). Conversely, there were no differences in cortisol levels in subjects with the A;A genotype receiving or not receiving social support ($P = 0.46$). Moreover, there was a trend ($P = 0.08$) for people with the A;A genotype to display higher levels of cortisol throughout the session than G carriers (no differences between the genotypes were observed at baseline).

23andMe blog discusses the snp.

blogs.scientificamerican discusses the snp.

In this study, 345 healthy subjects were examined by multimodal neuroimaging techniques to identify structural and functional alterations in OXTR risk allele carriers and their link to temperament. The authors found that there was a significant association between the different rs53576 genotypes and grey matter volume in the hypothalamus, with the hypothalamus volume being significantly larger in G;G carriers than in A;A carriers ($P = 0.012$). Moreover, they investigated the relationship between rs53576 and reward dependence (RD) using the Tridimensional Personality Questionnaire, and found that the G;G, G;A, and A;A genotypes were associated with the highest, intermediate, and lowest RD values, respectively (G;G vs. A;A, $p=0.02$; A;G vs. A;A, $p=0.07$). Lastly, they also demonstrated that rs53576 is associated with amygdala activation, with the A;A genotype being associated with a significant reduction in activation compared to the G;G genotype ($p=0.036$); and with the functional relationship between the amygdala and the hypothalamus, with A;A carrier having a significantly increased functional correlation ($p=0.036$). The authors concluded that there is likely a neural mechanism linking both structural and neural signalling alterations in the oxytocinergic system to individual differences in emotional reactivity and prosocial temperament.

This study found that there is no association between rs53576 variants and the risk of autism in Caucasian children and adolescents.

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Followup:

Gene related topics:

stress(5.00) · personality(1.00) · OTHER(1.00) · temperament(1.00) · heart rate(1.00) · depression(1.00) · intelligence(1.00) · PSYCH(0.28) · autism(0.28)

Relevant papers and external links:

PMID 19934046
PMID 20724662
PMID 20724662
PMID 19934046
PMID 19376182
PMID 19015103

dbSNP: rs53576 Genecard: OXTR chromosome 3 SNPedia: rs53576(G;G)

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★ rs4988235(C;T) good:2.47 27.06% **likely to be able to digest milk as an adult**

Approximately 80% of individuals with this genotype are able to digest milk (lactose) as adults.
Also known as "C/T(-13910)", and located in the MCM6 gene but with influence on the lactase LCT gene, rs4988235 is one of two SNPs that is associated with the primary haplotype associated with hypolactasia, more commonly known as lactose intolerance in European Caucasian populations. ,

In these populations, the rs4988235(T) allele is both the more common allele and the one associated with lactase persistence; individuals who are rs4988235(C;C) are likely to be lactose intolerant.

In populations of sub-Saharan Africans, though, the rs4988235(T) allele is so rare that it's unlikely to be predictive of lactase persistence, and other SNPs are predictive instead. [PMID 15106124, PMID 17159977]

See also OMIM 601806.0001

Measuring European population stratification with microarray genotype data.

Adult-type hypolactasia is not a predisposing factor for the early functional and structural changes of atherosclerosis: the Cardiovascular Risk in Young Finns Study.

Genetic polymorphisms in PTPN22, PADI-4, and CTLA-4 and risk for rheumatoid arthritis in two longitudinal cohort studies: evidence of gene-environment interactions with heavy cigarette smoking.

Heterogeneity in gene loci associated with type 2 diabetes on human chromosome 20q13.1.

Genetic testing for adult-type hypolactasia in Italian families.

Lactase persistence-related genetic variant: population substructure and health outcomes.

Gender differences in genetic risk profiles for cardiovascular disease.

Genetic lactase non-persistence, consumption of milk products and intakes of milk nutrients in Finns from childhood to young adulthood.

Geographical structure and differential natural selection among North European populations.

Natriuretic peptide system gene variants are associated with ventricular dysfunction after coronary artery bypass grafting.

A non-synonymous variant in ADH1B is strongly associated with prenatal alcohol use in a European sample of pregnant women.

Polymorphism in the oxytocin promoter region in patients with lactase non-persistence is not related to symptoms.

European lactase persistence genotype shows evidence of association with increase in body mass index.

Variation in the 4q25 chromosomal locus predicts atrial fibrillation after coronary artery bypass graft surgery.

Haplotype allelic classes for detecting ongoing positive selection.

Usefulness of Mendelian randomization in observational epidemiology.

Adult-type hypolactasia and lactose malabsorption in Poland.

Association of the LCT-13910C>T polymorphism with obesity and its modulation by dairy products in a Mediterranean population.

Lactase persistence genotypes and malaria susceptibility in Fulani of Mali.

The lactase persistence -13910C>T polymorphism shows indication of association with abdominal obesity among Portuguese children.

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Established associations:

body mass index **you have 1 copies of the risk allele T**

(risk allele=T, EA odds ratio/beta 0.016 [0.0091-0.0225] kg/m2 increase with pval 5E-6, pubmedid=25673413; risk allele=T, odds ratio/beta 0.016 [0.0094-0.0224] kg/m2 increase with pval 2E-6, pubmedid=25673413)

Gene related topics:

lactase persistence(1.00) · cardiovascular risk(1.00) · arthritis(1.00) · cardiovascular disease(1.00) · atherosclerosis(1.00) · CARDIOVASCULAR(1.00) · rheumatoid arthritis(1.00) · atrial fibrillation(1.00) · body mass(1.00) · hypolactasia(1.00) · malaria(1.00) · OTHER(1.00) · cardiovascular(1.00) · alcohol(1.00) · body mass index(1.00) · obesity(1.00) · type 2 diabetes(1.00) · smoking(1.00) · METABOLIC(0.19) · lactose intolerance(0.19)

Relevant papers and external links:

PMID 25673413
Genetic studies of body mass index yield new insights for obesity biology.

PMID 11788828

PMID 15114531

PMID 17436249

PMID 18194137

PMID 18462498

PMID 18602983

PMID 18605960

PMID 18797476

PMID 18974842

PMID 19138442

PMID 19265028

PMID 19326473

PMID 19687126

PMID 19943975

PMID 20015952

PMID 20031626

PMID 20109229

PMID 20616999

PMID 21152447

PMID 21193851

PMID 21235777

PMID 23252911

PHARMGKB PA165363227
Evidence: PubMed ID:11788828Risk or phenotype-associated allele: C .Phenotype: The C allele of this SNP completely associated with lactase non-persistence in nine extended Finnish pedigrees and in 236/236 individuals from four different populations . Study size: 145 family members/9 familes; 236 individuals . Study population/ethnicity: families: Finnish; individuals: Finnish(196), Italian(9), German(9),South Korean(22) .Type of association: CO;GN.

PHARMGKB PA165363255

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Evidence: PubMed ID:15114531Phenotype: This SNP, for which the T allele previously has been shown to be tightly associated with lactase persistance, is part of a haplotype that extends for > 1 Mb and includes rs182549(the A allele is associated with lactase persistance). The haplotype appears to be under strong recent positive selection in European Americans. In European Americans, African Americans and East Asians, the alleles associated with lactase persistance were found to occur at frequencies roughly matching the rates of lactase persistance. Study population/ethnicity: European American, African American, East Asian, Scandinavian. Type of association: CO;GN.

OMIM 601806

dbSNP: rs4988235 GWAS Catalog: rs4988235 Genecard: MCM6 chromosome 2

SNPedia: rs4988235(C;T)

★ rs1800497(C;T) warning:2.46 43.92% **A1/A2: Bad at avoidance of errors. 0.5x lower OCD risk, 0.87x lower Tardive Diskinesia risk, higher ADHD risk. More Alcohol Dependence. Lower risk of Postoperative Nausea. Increased obesity. Bupropion is not effective.**

The DRD2 TaqIA A1/A2 version causes less dopamine receptors. Doesn't learn from mistakes well. Reduced response to errors and increased addictive behavior

Men may have half the risk of Obsessive Compulsive Disorder but higher risk of ADHD. Women have lower Persistence. 0.87x reduced risk of Tardive Diskinesia when taking dopamine receptor antagonists. Slightly increased risk of alcoholism and smoking addiction. Slower recovery from traumatic brain injury. Lower risk of postoperative nausea within 6 hours of surgery. Bupropion (Wellbutrin, Budeprion, Prexaton, Elontril, Aplenzin, Zyban, Voxra) doesn't help to quit smoking. Increased obesity due to decreased pleasure response to food.

Single marker analysis the TaqIA rs1800497 and TaqIB rs1079597 variants were associated with heroin dependence.

DRD2/ANKK1 TaqIA polymorphism is not associated with early infant temperament. There were no differences in the temperament style distribution between the T-carrier and non-T carrier groups. There were also no statistically significant differences between the two groups in the score of the nine temperament dimensions.

Extrapyramidal adverse effects are associated with increased drug occupancy of the dopamine 2 receptors DRD2. The A1 allele of the DRD2/ANKK1, rs1800497, is associated with decreased striatal DRD2 density. Results strongly suggested that A1+ variants of the DRD2/ANKK1 Taq1A allele do confer an associated risk for akathisia in patients who were treated with second-generation antipsychotics SGAs, and these variants may explain inconsistencies found across prior studies, when comparing FGAs and SGAs.

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DRD2/ANKK1 Taq1A (rs 1800497 C>T) genotypes are associated with susceptibility to second generation antipsychotic-induced akathisia.

Multivariate analysis of dopaminergic gene variants as risk factors of heroin dependence.

[DRD2/ANKK1 Taq IA polymorphism and early infant temperament].
rs1800497, a SNP also known as the TaqIA (or Taq1A) polymorphism of the dopamine D2 receptor DRD2 gene (even though it is actually located over 10,000bp downstream of the gene), gives rise to the DRD2*A1 allele. This allele (rs1800497(T)) is associated with a reduced number of dopamine binding sites in the brain .

Reduced response to errors and increased addictive behavior

It has been postulated to play a role in alcoholism, smoking, and certain neuropsychiatric disorders.

The reduced number of dopamine binding sites may play a role in nicotine addiction by causing an "understimulated" state that can be relieved by smoking (and/or use of other drugs).

A wide variety of reports have been published over more than ten years either linking rs1800497 to aspects of nicotine use and smoking cessation success, or finding no such association. A meta-analysis of 41 such studies published in 2004 concluded that overall the association of rs1800497 with such phenomena was statistically weak. This same group recently (2009) published a study showing no association between rs1800497 and improved response to nicotine replacement therapy (NRT), contrary to their previous study.

More recently, a relatively large study of over 700 patients attempting to kick their smoking habit using the drug bupropion determined that smokers homozygous for the A2/A2 genotype were more successful than A1/A2 or A1/A1 individuals. The A2/A2 smokers were more than three times as likely, relative to placebo, to be abstinent at end of treatment (35.2% vs. 15.1%; odds ratio = 3.25, CI: 2.00-5.28) and at 6 months of follow-up (26.7% vs. 12.2%; odds ratio = 2.81, CI: 1.66-4.77), whereas not so much by 12 months (16.3% vs. 10.7%; OR = 1.70, CI: 0.95-3.05). Basically, A1/A2 and A1/A1 genotype smokers didn't gain anything from using bupropion versus placebo; bupropion only helped A2/A2 genotypes stop smoking.

In a study of individuals in the Polyp Prevention Trial with any, multiple (>/=2) or advanced colorectal adenoma recurrence after 4 years, compared to those without adenoma recurrence, rs1800497 (T;T) individuals were significantly associated with all advanced adenoma recurrence (odds ratio 2.40, CI: 1.11-5.20). The authors speculate this increased risk of adenoma recurrence (and an association with colorectal cancer) may be related to SNP-associated differences in alcohol and fat intake.

A 2014 preliminary study links an individual's ANKK1 rs1800497 genotype as likely to experience greater positive subjective effects following cocaine exposure, including greater 'high' and 'like', and these individuals may have increased vulnerability to continue using cocaine or they may be at greater risk to relapse during periods of abstinence. The study states that replication of these findings is necessary to confirm these findings.

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SNPs rs6276, rs6277, and rs1800497) in the human DRD2 gene are associated with decreased D2R expression and function, as well as high blood pressure. Data supports the hypothesis that D2R function has protective effects in human renal proximal tubule cells hRPTCs and suggest that carriers of these SNPs may be prone to chronic renal disease and high blood pressure.

SNPs, rs1800497 and rs2283265, located near and within the dopamine receptor 2 DRD2 gene, respectively, were significantly associated with improvements during working memory training ($p < .003$ and $p < .0004$, respectively).

A small (54 patient) study of patients with traumatic brain injury concluded that carriers of rs1800497 (A) alleles recover slower as assessed by memory and attention tests.

23andMe blog diminished pleasure response from food

GENETICALLY DETERMINED DIFFERENCES IN LEARNING FROM ERRORS

Obsessive Compulsive Disorder

Avoidance of Errors

Postoperative Nausea and Vomiting

A variant in ANKK1 modulates acute subjective effects of cocaine: a preliminary study.

Living in the moment: Effects of time perspective and emotional valence of episodic thinking on delay discounting.

Single-nucleotide polymorphisms of the dopamine d2 receptor increase inflammation and fibrosis in human renal proximal tubule cells.

Polymorphisms in the dopamine receptor 2 gene region influence improvements during working memory training in children and adolescents.

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Gene related topics:

adhd(5.00) · alcohol(5.00) · alcohol dependence(5.00) · obesity(5.00) · obsessive compulsive disorder(1.00) · memory(1.00) · blood pressure(1.00) · CANCER(1.00) · brain(1.00) · postoperative nausea and vomiting(1.00) · alcoholism(1.00) · cancer(1.00) · RENAL(1.00) · colorectal cancer(1.00) · OTHER(1.00) · nicotine(1.00) · smoking cessation(1.00) · heroin dependence(1.00) · temperament(1.00) · smoking(1.00) · inflammation(1.00)

Relevant papers and external links:

PMID 18063800
PMID 18058343
PMID 23840506
PMID 20199723
PMID 23118020

PMID 23118020

PMID 23840506

PMID 20199723

PMID 9672901

PMID 18063800

PMID 8873216

PMID 15370155

PMID 19273465

PMID 18058343

PMID 19065655

PMID 24528631

PMID 24379187

PMID 24001007

PMID 18698520

OMIM 608774

PHARMGKB PA165291720

Evidence: PubMed ID:1969501Risk or phenotype-associated genotype: T. Phenotype: The A1 allele is associated with 24 (69%) of 35 alcoholics, but it associated with only 7 (20%) of 35 nonalcoholics. The absence of the A1 allele is associated with 28 (80%) of 35 nonalcoholics and with only 11 (31%) of 35 alcoholics. Study size: 35 alcoholics and 35 nonalcoholics. Study population: 46 whites; 24 blacks

PHARMGKB PA161145174

Evidence: Web Resource:External Link with 12-month smoking cessation outcomes following treatment with a combination of bupropion and behavioral counseling in women.

PHARMGKB PA162171873

Evidence: PubMed ID:18058343This variant (A2/A2 genotype) is associated with improved response to bupropion efficacy for smoking cessation.

PHARMGKB PA162356250

Evidence: PubMed ID:18855532This SNP is significant predictor of treatment response to risperidone in first-episode schizophrenia.

PHARMGKB PA162370416

Evidence: PubMed ID:18927395In blood oxygen level-dependent functional magnetic resonance imaging studies, obese college-aged women and adolescent girls showed a blunted striatal response to chocolate milkshake tasting (vs. tasting of a tasteless solution) when compared to lean women and girls, and the genotype at this SNP affected the extent of this response. This SNP is known as the TaqIA allele of DRD2 even though it is more than 10 KB downstream from the gene boundary.

PHARMGKB PA165108050

Evidence: PubMed ID:19339912Risk or phenotype-associated allele: T. Phenotype: Carriers of one or two copies of the T allele of this variant (also known as Taq1A A1) were at higher risk of developing hyperprolactinemia than those with two copies of the C allele (also known as Taq1A A2). This variant is a SNP located about 9.5 kb downstream of the DRD2 coding region, and results in a Glu (C allele)

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to Lys (T allele) amino acid substitution in the ANKK1 protein. Study size: 90. Study population/ethnicity: 7-17-year-old patients chronically treated with risperidone; non-Hispanic Caucasians. Significance metric(s): OR = 3.1; p < 0.05. Type of association: CO.

PHARMGKB PA165291714
Evidence: PubMed ID:18086475Risk or phenotype-associated genotype: T/T. Phenotype: Patients with one or two A1 alleles (T/T) had a greater risk of significant side effects, particularly if they were male. Study size: 116. Study population: Caucasians.

PHARMGKB PA165291715
Evidence: PubMed ID:17767146Risk or phenotype-associated genotype: C/C. Phenotype: A meta-analysis showed that compared to tardive dyskinesia (TD)-negative patients, TD-positive patients had a higher A2 allele frequency (P = 0.003), with an effect-size of 1.30 (95% CI: 1.09-1.55), and higher A2/A2 genotype frequency (P = 0.001), with an effect-size of 1.50 (95% CI: 1.17-1.92). Study size: 1256 schizophrenia patients.

PHARMGKB PA165291718
Evidence: PubMed ID:19273465Phenotype: This study found no association between the Taq1A polymorphism and response to nicotine replacement therapy.

PHARMGKB PA165291719
Evidence: PubMed ID:15492764Risk or phenotype-associated genotype: T. Phenotype: Compared to women who carry both A2 (CC) alleles, women with at least one A1 (T) allele were more likely to report having stopped taking bupropion due to medication side effects (odds ratio (OR)=1.91, 95% confidence interval (CI)=1.01-3.60; P<0.04) and at 12 months were somewhat more likely to report smoking (OR=0.76, 95% CI=0.56-1.03; P<0.076). Significant associations or trends were not observed in men. Study size: 451 participants. Study population: Caucasian.

PHARMGKB PA165291721
Evidence: PubMed ID:17989061Risk or phenotype-associated genotype: T. Phenotype: This meta-analysis included 44 studies with 9,382 participants. For all studies combined, when we assumed the dominant model of gene action (A1A1 + A1A2 vs. A2A2), a small but significant association of alcohol dependency with being homozygote or heterozygote for the A1 allele was detected. The odds ratio was 1.38 (95 percent confidence interval (CI): 1.20, 1.58) when random effects were used. Study size: 5,273 cases and 3,995 controls.

PHARMGKB PA165291618
Evidence: PubMed ID:18563706Risk or phenotype-associated allele: T (A1). Phenotype: In a smoking cessation study the TaqIA SNP was significantly associated with being abstinent at 1 year (P = 0.01). Participants who carried at least one minor allele (A1) were less likely to quit compared to A2A2 homozygous (Odds Ratio: 0.47, 95% CI: 0.24-0.94). Study size: 881. metric(s): OR =0.47, 95% CI: 0.24-0.94.

PHARMGKB PA165291713
Evidence: PubMed ID:15286066Risk or phenotype-associated allele: T. Phenotype: In the combined medication group patients with the A1 (T) allele had 40% higher prolactin levels than patients without this allele. Study size: 144 schizophrenic patients. Study population: White patients.

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PHARMGKB PA165291717
Evidence: PubMed ID:15077009Risk associated genotype: C/C. Phenotype: In a randomized controlled trial investigating the short-term effectiveness of the nicotine patch, after 1 week of the trial the patch was more effective for smokers with CT/TT genotype (OR 2.80, 95% CI 1.70-4.61) than with CC (OR 1.41, 0.94-2.12; P for difference in ORs 0.04). Study size: 1532.

PHARMGKB PA165291679
Evidence: PubMed ID:20194480Phenotype: A meta-analysis, including eight individual studies, was not able to detect an association between clinical response to antipsychotics and the Taq1A variant. Study size: meta-analysis included 8 individual studies, 748 patients total.

PHARMGKB PA165291688
Evidence: PubMed ID:19603545Phenotype: In this case-control study the polymorphisms -141C Ins/Del (rs1799732); C957T (rs6277); A1385G (rs6276); and TaqIA (rs1800497) were genotyped. The haplotypes I-C-G-A2 and I-C-A-A1 occurred with a higher frequency in alcoholics [P=0.026, odds ratio (OR): 1.340; P=0.010, OR: 1.521, respectively]. Study size: 360 alcoholics and 368 controls. Study population:Caucasian individuals of German origin.

PHARMGKB PA165291677
Evidence: PubMed ID:18926547Phenotype: Compared with patients who had the A2/A2 (C/C) genotype, the positive and negative syndrome scale (PANSS) positive scores of A1/A1 (T/T) patients were 3.71 lower (p = 0.01) and of A1/A2 patients were 1.36 lower (p = 0.03) after 4-week aripiprazole treatment. These results suggest that A1 carriers are associated with superior therapeutic response on positive symptoms. Study size: 128 schizophrenic patients. Study population: Chinese.

PHARMGKB PA165291712
Evidence: PubMed ID:10823405Risk or phenotype-associated allele: T. Phenotype: In schizophrenic patients treated with nemonapride, the delta prolactin at 1 and 3 weeks was significantly (P<0.05) higher in female patients with the A1 allele than in males with or with no A1 allele. The delta prolactin at 3 weeks was also significantly (P<0.05) higher in the female patients with the A1 allele than in those with no A1 allele. Study size: 25 schizophrenic inpatients. Study population: Japanese.

PHARMGKB PA165291716
Evidence: PubMed ID:18180754Risk or phenotype-associated genotype: C/C. Phenotype: This meta-analysis suggests multiple genetic influences on tardive dyskinesia, including the Taq1A SNP. For Taq1A using the A1 allele as reference category, the study showed a risk-increasing effect for the A2 variant (OR=1.30, 95% CI: 1.03-1.65, P=0.026), and for A2-A2 homozygotes using A1-A1 as reference category (OR=1.80, 95% CI: 1.03-3.15, P=0.037).

PHARMGKB PA165374659
Evidence: PubMed ID:20714340Risk or phenotype-associated allele: T. Phenotype: The T allele was associated with weight gain of at least 7% in schizophrenia patients treatment with clozapine or olanzapine. Study size: 206. Study population/ethnicity: European american; German; Schizophrenia. Significance metric(s): OR = 2.18 (CI: 1.00-4.75). Type of association: PD.

OMIM 608774
PMID 24528631
PMID 24512061

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PMID 24379187
PMID 24001007

dbSNP: rs1800497 Genecard: ANKK1 chromosome 11 SNPedia: rs1800497(C;T)

★ rs1169300(A;A) warning:2.45 10.16% **~2x increased lung cancer risk**

rs1169300 is a SNP that was originally associated with C-reactive protein levels, and given other findings that suggest C-reactive protein levels may be predictive of cancer risk, was thought to be linked to cancer risk.

A large study pooling data from 3 Finnish studies totaling over 18,000 individuals concluded that while this SNP is not likely to be causative (relative to cancer), it and one other CRP SNP (rs2464196) are associated with increased risk for lung cancer. The odds ratio for minor alleles of either SNP was about 1.5x and 2x for heterozygotes and homozygotes, respectively. One other CRP SNP (rs1892534) was associated with increased overall cancer risk. CRP SNPs were not associated with colorectal, prostate or breast cancer risk.

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Gene related topics:

cancer(5.00) · lung cancer(5.00) · UNKNOWN(2.00) · diabetes mellitus type 1 diabetes mellitus type 2(2.00) · breast cancer(1.00) · OTHER(1.00) · METABOLIC(0.64) · gamma glutamyltransferase(0.64) · diabetes mellitus type ii diabetes mellitus type 2(0.57) · IMMUNE(0.36) · inflammation(0.36) · diabetes type 2(0.28) · CARDIOVASCULAR(0.16) · c reactive protein(0.16) · diabetes mellitus type 2(0.09) · diabetes mellitus(0.07) · CANCER(0.03) · pancreatic neoplasms(0.03)

Relevant papers and external links:

PMID 20727736

dbSNP: rs1169300 Genecard: HNF1A chromosome 12 SNPedia: rs1169300(A;A)

★ rs12970134(A;A) info:2.41 4.30%(rare) **adult waist 1.8cm larger on average**

A study of 14,000 Indian Asian or Caucasian adults indicates that rs12970134(A) alleles are associated with obesity. The average increase in waist circumference is 0.9cm BMI units per risk allele. see also

The common obesity variant near MC4R gene is associated with higher intakes of total energy and dietary fat, weight change and diabetes risk in women.

Variants near MC4R are associated with obesity and influence obesity-related quantitative traits in a population of middle-aged people: studies of 14,940 Danes.

Investigation of the locus near MC4R with childhood obesity in Americans of European and African ancestry.

The role of obesity-associated loci identified in genome-wide association studies in the determination of pediatric BMI.

Large effects on body mass index and insulin resistance of fat mass and obesity associated gene (FTO) variants in patients with polycystic ovary syndrome (PCOS).

From monogenic to polygenic obesity: recent advances.

A genome-wide association study of the metabolic syndrome in Indian Asian men.

Obesity and diabetes genes are associated with being born small for gestational age: results from the Auckland Birthweight Collaborative study.

FTO and MC4R gene variants are associated with obesity in polycystic ovary syndrome.

Common variants near MC4R: exploring gender effects in overweight and obese children and adolescents participating in a lifestyle intervention.

A variant near the melanocortin-4 receptor gene regulates postprandial lipid metabolism in a healthy Caucasian population.

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Established associations:

high density lipoprotein cholesterol measurement **you have 2 copies of the risk allele A**

(risk allele=A, waist circumference odds ratio/beta 0.88 [0.59-1.17] cm increase with pval 2E-9, pubmedid=18454146)

triglyceride measurement **you have 2 copies of the risk allele A**

(risk allele=A, waist circumference odds ratio/beta 0.88 [0.59-1.17] cm increase with pval 2E-9, pubmedid=18454146)

waist circumference **you have 2 copies of the risk allele A**

(risk allele=A, waist circumference odds ratio/beta 0.88 [0.59-1.17] cm increase with pval 2E-9, pubmedid=18454146)

body weight **you have 2 copies of the risk allele A**

(risk allele=A, odds ratio/beta 4.66 [3.41-5.91] % SD with pval 5E-13, pubmedid=19079260)

body mass index **you have 2 copies of the risk allele A**

(risk allele=A, odds ratio/beta 4.38 [3.16-5.60] % SD with pval 1E-12, pubmedid=19079260)

type ii diabetes mellitus **you have 2 copies of the risk allele A**

(risk allele=A, odds ratio/beta 1.08 [1.03-1.12] with pval 3E-8, pubmedid=24509480)

Gene related topics:

METABOLIC(2.73) · obesity(2.73) · PSYCH(2.00) · binge eating(2.00) · polycystic ovary syndrome(1.00) · metabolism(1.00) · insulin resistance(1.00) · body mass(1.00) · bmi(1.00) · metabolic syndrome(1.00) · weight(1.00) · quantitative traits(1.00) · body mass index(1.00) · insulin(1.00) · waist circumference(1.00) · lipid metabolism(1.00) · body weight obesity(0.71) · obesity morbid(0.33) · diabetes mellitus type 2 obesity(0.31) · diabetes mellitus type ii diabetes mellitus type 2(0.09)

Relevant papers and external links:

PMID 24509480
Genome-wide trans-ancestry meta-analysis provides insight into the genetic architecture of type 2 diabetes susceptibility.

PMID 19079260
Genome-wide association yields new sequence variants at seven loci that associate with measures of obesity.

PMID 18454146

OMIM 601665

OMIM 155541

PHARMGKB PA164740168
Evidence: PubMed ID:19079260; Web Resource:External Link results: Genome-wide association yields new sequence variants at seven loci that associate with measures of obesity. (Initial Sample Size: 80,969 individuals; Replication Sample Size: 11,036 individuals); (Region: 18q21.32; Reported Gene(s): MC4R; Risk Allele: rs12970134-A); (p-value= 0.0000000000005).This variant is associated with Weight.

PHARMGKB PA162356414
Evidence: PubMed ID:18454146; Web Resource:External Link Results: Common genetic variation near MC4R is associated with waist circumference and insulin resistance (Initial Sample Size: 2,684 Asian Indian men; Replication Sample Size: 11,955 Asian Indian and European individuals; Risk Allele: rs12970134-A). This variant is associated with Waist circumference and related phenotypes.

PHARMGKB PA164740146
Evidence: PubMed ID:19079260; Web Resource:External Link results: Genome-wide association yields new sequence variants at seven loci that associate with measures of obesity. (Initial Sample Size: 80,969 individuals; Replication Sample Size: 11,036 individuals); (Region: 18q21.32; Reported Gene(s): MC4R; Risk Allele: rs12970134-A); (p-value= 0.000000000001).This variant is associated with Body mass index.

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PMID 18697794
PMID 19073769
PMID 19265794
PMID 19478790
PMID 20092643
PMID 20127379
PMID 20694148
PMID 20712903
PMID 21283731
PMID 21372613
PMID 21736789

dbSNP: rs12970134 GWAS Catalog: rs12970134 Genecard: nan chromosome 18

SNPedia: rs12970134(A;A)

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★ rs4474514(A;A) warning:2.38 39.21% **>3x increased testicular cancer risk for men**

found in 65% of caucasians but linked 3x higher risk of testicular cancer than the (G;G) form. Testicular germ cell tumor risk l("TGCT"; i.e. testicular cancer risk) was increased 3x per copy of the major (A) allele at rs3782179 and rs4474514 (OR = 3.08, CI: 2.29–4.13; OR = 3.07, CI = 2.29–4.13, respectively), based on a genome-wide scan of 277 primarily Caucasian TGCT cases and 919 controls. Men who have two copies of the common version of the c-KIT ligand KITLG gene have a 4.5-fold higher risk of testicular cancer than men who have two copies of the less common or minor version of the gene.

SNPs from the same gene were also implicated in testicular cancer risk in a study of 730 cases and 1,435 controls from the UK and replicating associations in a further 571 cases and 1,806 controls.

A second independent locus within DMRT1 is associated with testicular germ cell tumor susceptibility.

Associations between variants in KITLG, SPRY4, BAK1, and DMRT1 and pediatric germ cell tumors.

Established associations:

testicular carcinoma **you have 2 copies of the risk allele A**

(risk allele=A, odds ratio/beta 3.07 [2.29-4.13] with pval 6E-15, pubmedid=19483682)

Gene related topics:

cancer(5.00) · testicular cancer(5.00) · CANCER(2.00) · neoplasms germ cell and embryonal testicular neoplasms(2.00) · testicular neoplasms(1.29) · testicular germ cell tumor(1.00) · IMMUNE(0.80) · ige(0.80)

Relevant papers and external links:

PMID 19483682

PMID 19483681

OMIM 273300

PMID 21551455

PMID 22072546

dbSNP: rs4474514 GWAS Catalog: rs4474514 Genecard: KITLG chromosome 12

SNPedia: rs4474514(A;A)

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★ rs9536314(T;T) info:2.38 75.69% **most common in all populations**

but having one copy of the rarer (C) allele is linked to increased executive function

Variation in longevity gene KLOTHO is associated with greater cortical volumes

Gene related topics:

METABOLIC(2.00) · age related skeletal disorders(2.00) · AGING(0.04) · longevity(0.04)

Relevant papers and external links:

dbSNP: rs9536314 Genecard: KL chromosome 13 SNPedia: rs9536314(T;T)

★ rs10994336(C;T) warning:2.38 21.60% **1.45x increased odds of developing bipolar disorder**

23andMe blog rs10994336 or (rs4948418) Each T at this SNP increased the odds of developing bipolar disorder by 1.45 times compared to the CC genotype.

This is one of the SNPs reported by NutraHacker.

Established associations:

bipolar disorder **you have 1 copies of the risk allele T**

(risk allele=T, odds ratio/beta 1.45 with pval 9E-9, pubmedid=18711365)

Gene related topics:

METABOLIC(0.14) · cholesterol ldl(0.14) · PSYCH(0.10) · bipolar disorder(0.10) · CARDIOVASCULAR(0.06) · arteries(0.06)

Relevant papers and external links:

PMID 18711365
Collaborative genome-wide association analysis supports a role for ank3 and cacna1c in bipolar disorder.

OMIM 125480

OMIM 612357

PHARMGKB PA162356412
Evidence: PubMed ID:18711365; Web Resource:External Link Results: Collaborative genome-wide association analysis supports a role for ANK3 and CACNA1C in bipolar disorder (Initial Sample Size: 1,098 cases, 1,267 controls; Replication Sample Size: 4,387 cases, 6,209 controls; Risk Allele: rs10994336-T). This variant is associated with Bipolar disorder.

PHARMGKB PA162355588
Evidence: PubMed ID:18711365A genome-wide association analysis study in 4,387 cases and 6,209 controls identified a strong association of this variant in the ANK3 gene with bipolar disorder.

dbSNP: rs10994336 GWAS Catalog: rs10994336 Genecard: ANK3 chromosome 10

SNPedia: rs10994336(C;T)

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★ **rs17070145(C;T)** **good:2.38** **49.92%** **increased memory performance**

In a Swiss cohort: "Carriers of KIBRA rs17070145 T allele had 24% better free recall performance 5 min after word presentation (P = 0.000004) and 19% better free recall performance 24 hours after word presentation (P = 0.0008) than did noncarriers."
In a US cohort: "T allele carriers had significantly better memory scores than non-carriers in the Buschke's Selective Reminding Test (SRT). Performance on another episodic memory task, the Rey

Auditory Verbal Learning Test (AVLT), was also significantly different between allele groups."
In another Swiss cohort, with a visual episodic memory task: "T allele carriers also performed significantly better than did noncarriers in this population."
"Functional magnetic resonance imaging detected KIBRA allele-dependent differences in hippocampal activations during memory retrieval."

Replication in a healthy elderly cohort.

Age-dependent association of KIBRA genetic variation and Alzheimer's disease risk.

Genetic correlates of brain aging on MRI and cognitive test measures: a genome-wide association and linkage analysis in the Framingham Study.

KIBRA genetic polymorphism influences episodic memory in later life, but does not increase the risk of mild cognitive impairment.

Failure to replicate effect of Kibra on human memory in two large cohorts of European origin.

Evidence for an association between KIBRA and late-onset Alzheimer's disease.

Association of KIBRA and memory.

Caspase-1 genetic variation is not associated with Alzheimer's disease risk.

KIBRA: A New Gateway to Learning and Memory?

Impact of common KIBRA allele on human cognitive functions.

The role of memory-related gene polymorphisms, KIBRA and CLSTN2, on replicate memory assessment in the elderly.

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Gene related topics:

memory(5.00) · aging(1.00) · cognitive impairment(1.00) · brain(1.00) · AGING(1.00) · NEUROLOGICAL(0.22) · memory disorders(0.22)

Relevant papers and external links:

PMID 17053149
PMID 17353070
OMIM 610533
OMIM 610533
PMID 17707552
PMID 17903297
PMID 18194457
PMID 18205171
PMID 18789830

PMID 19397951
PMID 20184726
PMID 20552044
PMID 21346737
PMID 21643791

dbSNP: rs17070145 Genecard: WWC1 chromosome 5 SNPedia: rs17070145(C;T)

★ rs1012053(A;A) warning:2.34 59.41% **Normal (higher) risk of Bipolar Disorder.**

This is the most common version of this SNP, so most people have this. But it is suspected of having a higher risk of Bipolar Disorder compared to the other rare versions of this SNP.
Bipolar disorder failed to replicate in

Established associations:

bipolar disorder **you have 2 copies of the risk allele A**

(risk allele=A, odds ratio/beta 1.59 [1.35-1.87] with pval 2E-8, pubmedid=17486107)

Gene related topics:

OTHER(1.00) · PSYCH(0.10) · bipolar disorder(0.10)

Relevant papers and external links:

PMID 17486107
A genome-wide association study implicates diacylglycerol kinase eta (dgkh) and several other genes in the etiology of bipolar disorder.

PHARMGKB PA162356620
Evidence: PubMed ID:17486107; Web Resource:External Link Results: A genome-wide association study implicates diacylglycerol kinase eta (DGKH) and several other genes in the etiology of bipolar disorder (Initial Sample Size: 461 cases, 563 controls; Replication Sample Size: 772 cases, 876 controls; Risk Allele: rs1012053-A).

dbSNP: rs1012053 GWAS Catalog: rs1012053 Genecard: DGKH chromosome 13

SNPedia: rs1012053(A;A)

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★ rs4911414(G;T) warning:2.27 32.27% **2-4x higher risk of sun sensitivity if part of risk haplotype**

rs4911414 is a SNP near the ASIP (agouti signaling protein) gene on chromosome 20.

This SNP is one of a tightly-linked pair that increases the likelihood of an individual being prone to sun sensitivity, in other words, freckles and sunburn, based on a study of 6,000+ Caucasians (5,000+ Icelanders + 1,000+ Dutch). The odds ratios, presumably on a dominant basis, and at least in the largest population (Icelanders) for "freckles and burns vs. no freckles and tans" for the haplotype pair rs1015362(G) - rs4911414(T) is 3.91 (CI: 2.54-6.03) for males and 2.42 (CI: 1.52-3.86) for females, with an overall p=0.051.

Based on a study by the same authors of 4,000+ skin cancer patients, this haplotype was seen to confer significant increased risk for cutaneous malignant melanoma (odds ratio 1.45, p = 1.2 x 10-9) as well as basal cell carcinoma (odds ratio 1.33, p = 1.2 x 10-6).

Melanoma susceptibility variants on chromosome 20q11.22 are associated with pigmentary traits and the risk of nonmelanoma skin cancer.

Web-based, participant-driven studies yield novel genetic associations for common traits.

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Established associations:

sunburn **you have 1 copies of the risk allele T**

(risk allele=T, odds ratio/beta 2.56 [2.06-3.18] with pval 6E-37, pubmedid=18488028)

freckles **you have 1 copies of the risk allele T**

(risk allele=T, odds ratio/beta 1.95 [1.65-2.32] with pval 8E-29, pubmedid=18488028; risk allele=T, odds ratio/beta 2.56 [2.06-3.18] with pval 6E-37, pubmedid=18488028)

skin sensitivity to sun **you have 1 copies of the risk allele T**

(risk allele=T, odds ratio/beta 1.76 [1.49-2.08] with pval 2E-24, pubmedid=18488028)

hair color **you have 1 copies of the risk allele T**

(risk allele=T, odds ratio/beta 1.76 [1.34-2.31] with pval 3E-9, pubmedid=18488028)

suntan **you have 1 copies of the risk allele G**

(risk allele=G, odds ratio/beta 0.07 [0.050-0.090] unit decrease with pval 4E-9, pubmedid=23548203)

Gene related topics:

malignant melanoma(1.00) · melanoma(1.00) · CANCER(1.00) · cancer(1.00) · OTHER(1.00)

Relevant papers and external links:

PMID 23548203
Genome-wide association studies identify several new loci associated with pigmentation traits and skin cancer risk in european americans.

PMID 18488028

PMID 18488027

OMIM 612263

OMIM 611742

OMIM 600201

PHARMGKB PA162356581
Evidence: PubMed ID:18488028; Web Resource:External Link Results: Two newly identified genetic determinants of pigmentation in Europeans (Initial Sample Size: 5,130 individuals; Replication Sample Size: 3,330 individuals; Risk Allele: rs1015362-G + rs4911414-T). This variant is associated with Burning and freckling.

PHARMGKB PA162355809
Evidence: PubMed ID:18488027This variant is associated with the risk of cutaneous melanoma and basal cell carcinoma. It is also associated blue vs green eyes and skin sensitivity to sun.

PHARMGKB PA162356583
Evidence: PubMed ID:18488028; Web Resource:External Link Results: Two newly identified genetic determinants of pigmentation in Europeans (Initial Sample Size: 5,130 individuals; Replication Sample Size: 3,330 individuals; Risk Allele: rs1015362-G + rs4911414-T). This variant is associated with Skin sensitivity to sun; freckles and red vs. non-red hair.

PMID 19995372

PMID 20585627

dbSNP: rs4911414 GWAS Catalog: rs4911414 Genecard: nan chromosome 20

SNPedia: rs4911414(G;T)

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★ rs10784502(C;T) good:2.25 44.95% **Slightly higher intracranial volume**

associated with intracranial volume (12q14.3; N = 15,782; P = 1.12 × 10−12)

External Link

(C;C) has higher cranial capacity (about 2 teaspoons) and over 2% higher IQ.

(C;T) is half this positive effect.

Intelligence

Established associations:

brain measurement **you have 1 copies of the risk allele C**

(risk allele=C, Intracranial volume odds ratio/beta 9006.71 [6525.78-11487.64] mm3 increase with pval 1E-12, pubmedid=22504417)

Gene related topics:

intelligence(1.00) · DEVELOPMENTAL(0.31) · primary tooth development(0.31) · height(0.12)

Relevant papers and external links:

PMID 22504417

dbSNP: rs10784502 GWAS Catalog: rs10784502 Genecard: HMGA2 chromosome 12

SNPedia: rs10784502(C;T)

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★ **rs1801394(G;G)** **warning:2.25** **13.27%** **1.4x higher risk for meningiomas**

This common variant (HapMap allele frequency of 31.3%) in a protein involved in folate (B9) and cobalamin (B12) metabolism and is often reported as "MTRR I22M" (for the Ile to Met substitution at amino acid 22). Mothers homozygous for this variant were found in a few studies to have an increased risk for Down syndrome babies (risk of 0.4%, average risk in population is 0.25%), but other studies (including in Italian, Irish, French, and Indian-Gujarati women) found no increased risk. Notably, age plays a far larger role in the rate of Down syndrome (risk is 4.5% for a mother 45-years-of-age), and, it is unknown how this variant may combine with the effect of age. There are conflicting reports associating this variant with incidence of neural tube defects, possibly when combined with MTHFR A222V.

rs1801394, also known as A66G or Ile22Met, is a SNP in the methionine synthase MTRR gene. This gene encodes one of the two enzymes involved in the production of methionine (the other is

MTR). The protein encoded by an rs1801394 allele has a lower affinity for MTR () and is inconsistently associated with homocysteine level, although it is a risk factor for neural tube defects () and Down syndrome () in conditions of higher homocysteine.

Based on a study of British patients with primary brain tumors, (1,005 glioma cases and 631 meningioma cases), rs1801394(G;G) individuals were at higher risk for meningioma (odds ratio 1.41, CI: 1.02-1.94). In general, genotypes associated with increased 5,10-methylenetetrahydrofolate levels are associated with elevated risk for these types of brain cancer.

Neural tube defects and folate pathway genes: family-based association tests of gene-gene and gene-environment interactions.

Correlating observed odds ratios from lung cancer case-control studies to SNP functional scores predicted by bioinformatic tools.

Dietary vitamin B6 intake and the risk of colorectal cancer.

Folate and one-carbon metabolism gene polymorphisms and their associations with oral facial clefts.

BRCA1 promoter methylation is associated with increased mortality among women with breast cancer.

B-vitamin intake, one-carbon metabolism, and survival in a population-based study of women with breast cancer.

Vitamins B2, B6, and B12 and risk of new colorectal adenomas in a randomized trial of aspirin use and folic acid supplementation.

Prevalence in the United States of selected candidate gene variants: Third National Health and Nutrition Examination Survey, 1991-1994.

Low-penetrance alleles predisposing to sporadic colorectal cancers: a French case-controlled genetic association study.

No association of single nucleotide polymorphisms in one-carbon metabolism genes with prostate cancer risk.

Global DNA hypomethylation (LINE-1) in the normal colon and lifestyle characteristics and dietary and genetic factors.

One-carbon metabolism and breast cancer: an epidemiological perspective.

Lack of association between genetic polymorphisms in enzymes associated with folate metabolism and unexplained reduced sperm counts.

Association of folate-pathway gene polymorphisms with the risk of prostate cancer: a population-based nested case-control study, systematic review, and meta-analysis.

Polymorphisms in methionine synthase, methionine synthase reductase and serine hydroxymethyltransferase, folate and alcohol intake, and colon cancer risk.

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Associations of folate, vitamin B12, homocysteine, and folate-pathway polymorphisms with prostate-specific antigen velocity in men with localized prostate cancer.

Folate and choline metabolism gene variants and development of uterine cervical carcinoma.

Polymorphic variants of genes involved in homocysteine metabolism in celiac disease.

Folate and choline metabolism gene variants in relation to ovarian cancer risk in the Polish population.

Gene related topics:

DEVELOPMENTAL(2.20) · down syndrome(2.20) · CARDIOVASCULAR(1.50) · aorta(1.50) · ovarian cancer(1.00) · celiac disease(1.00) · metabolism(1.00) · survival(1.00) · prostate cancer(1.00) · brain(1.00) · UNKNOWN(1.00) · cancer(1.00) · breast cancer(1.00) · glioma(1.00) · colorectal cancer(1.00) · OTHER(1.00) · alcohol(1.00) · colorectal adenomas(1.00) · lung cancer(1.00) · mortality(1.00) · brain cancer(1.00) · METABOLIC(0.68) · homocysteine(0.68) · congenital abnormalities(0.67) · neural tube defects(0.55) · congenital heart defects heart defects congenital(0.33) · hyperhomocysteinemia(0.25) · PHARMACOGENOMIC(0.22) · methotrexate toxicity(0.22) · CANCER(0.15) · precursor cell lymphoblastic leukemia lymphoma(0.15) · spinal dysraphism(0.15) · VISION(0.13) · choroidal neovascularization macular degeneration(0.13) · IMMUNE(0.09) · kidney transplant complications(0.09) · thromboembolism venous(0.05) · HEMATOLOGICAL(0.05) · cholesterol(0.05) · erythrocytes(0.05) · multiple myeloma(0.05) · REPRODUCTION(0.04) · infertility male(0.04) · cleft lip cleft palate(0.04) · leukemia(0.02)

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Relevant papers and external links:

PMID 12416982

PMID 10444342

PMID 10930360

PMID 18483342

PHARMGKB PA165223225
Evidence: PubMed ID:17024475Risk or phenotype-associated allele: G Phenotype: Maternal MTRR 66GG genotype was associated with an overall increase in Spina bifida risk. Study size: 413 Study population/ethnicity: Mothers with a child affected by Spina Bifida and control women; Netherlands Significance metric(s): OR= 2.1 (95% CI 1.3-3.3) Type of association: CO

OMIM 602568

PMID 17035141

PMID 18191955

PMID 18199722

PMID 18203168

PMID 18521744

PMID 18708404

PMID 18708408

PMID 18936436

PMID 18992148

PMID 19064578
PMID 19336559
PMID 19376481
PMID 19657388
PMID 19706844
PMID 19776626
PMID 20852008
PMID 21349258
PMID 21688148
PMID 22183302

dbSNP: rs1801394 Genecard: MTRR chromosome 5 SNPedia: rs1801394(G;G)

★ rs2802292(G;T) good:2.25 49.81% **One copy of a longevity gene. Slightly increased lifespan.**

This is a well known longevity gene, FOXO3, although only one of your copies is the good version. It seems to improve insulin sensitivity, and decrease oxidative stress, among other things.
A study of 5 genes in 3,741 Japanese men, some of whom lived significantly longer than average (centarians), concluded that the FOXO3 gene (also known as FOXO3A) harbors several SNPs linked to longevity, including this SNP, rs2802292, as well as rs2764264 and rs13217795.
The odds ratio for rs2802292(G;G) vs (T;T) carriers was 2.75 (p = 0.00009; adjusted p = 0.00135).
It is not known yet if this correlation is specific to Japanese males.

This is discussed extensively at geneticfuture as a longevity snp.
(T) shorter lifespan
(G) longer lifespan

Quoted from the geneticfuture article:
"No-one should consider this association to be definitive until it's been replicated in independent cohorts.
Secondly, the effects of this variant are likely to differ between groups. According to the HapMap database the frequency of the "die early" (T) version of the variant varies substantially between populations: it's around 76% in Japanese, slightly more common (81%) in Chinese, less common (58%) in Europeans, and quite rare (17%) in West Africans. Given that the Japanese have the highest life expectancy of any of these populations, this is a counter-intuitive result, which reinforces the fact that there are many genes and other factors influencing variation in longevity."

23andMe blog longevity rs2802288(A) increased the odds of living to 100 by 1.5x. rs2802288 is located very close to rs2802292 which was previously reported to have a similar effect.

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A study of males from the Southern Italian Centenarian Study also replicated the association between rs2802288, a proxy of rs2802292, and longevity, with an odds ratio of 1.51 (CI: 1.15-1.98, p = 0.003).

Gene related topics:

insulin sensitivity(1.00) · stress(1.00) · OTHER(1.00) · insulin(1.00) · oxidative stress(1.00) · AGING(0.15) · longevity(0.15)

Relevant papers and external links:

PMID 18765803

PMID 19415983

OMIM 602681

OMIM 606460

dbSNP: rs2802292 Genecard: FOXO3 chromosome 6 SNPedia: rs2802292(G;T)

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★ rs363050(A;G) info:2.21 50.00% **The (A;G) genotype averages 2.8 PIQ points higher than the (G;G), but averages 2.84 PIQ points lower than (A;A) genotype**

The (A;G) genotype averages 2.8 PIQ points higher than the (G;G), but averages 2.84 PIQ points lower than (A;A) genotype

SNPs in the SNAP25 gene were initially linked to intelligence but have failed to replicate .

While now suspect, the original work can be found in .

The (A;A) genotype averaged 2.84 PIQ points higher than the (A;G) genotype, which averages 2.8 PIQ points higher than the (G;G) genotype (p=0.0002). Variance in this SNP may account for 3.4% of the phenotypic variance in PIQ.

rs363050 is in close linkage with rs353016, with r2 = 0.98, such that the rs363050(A) allele typically is linked to the rs353016(T) allele. For SNPs rs363039 - rs363043 - rs353016, respectively, the haplotype C-T-T is most associated with higher intelligence in children, whereas the haplotype C-C-T is most associated in adults (SNPs oriented as in dbSNP).

Measures of Intelligence

Gene related topics:

intelligence(1.00) · PSYCH(0.29) · adhd attention deficit hyperactivity disorder(0.29) · attention deficit

hyperactivity disorder(0.29) · adhd(0.26)

Relevant papers and external links:

PMID 17160701
PMID 19418213
PMID 16801949
PMID 16801949
PMID 17908175

dbSNP: rs363050 Genecard: SNAP25 chromosome 20 SNPedia: rs363050(A;G)

★ rs1892534(G;G) good:2.19 21.72% **Slightly higher C-Reactive Protein levels. Possibly slightly lower cancer risk.**

This causes a median hsCRP Level of 2.09 mg/L in women.

Strangely this was associated with a slightly lower overall cancer risk (in Finnish people). rs1892534 is a SNP that was originally associated with C-reactive protein levels, and given other findings that suggest C-reactive protein levels may be predictive of cancer risk, was thought to be linked to cancer risk.

A large study pooling data from 3 Finnish studies totaling over 18,000 individuals concluded that while the rs1892534(A) allele was not likely to be causative (relative to cancer), it is associated with increased risk for cancer overall. The odds ratio for 1 or 2 such alleles was 1.05 (CI: 0.90 - 1.23) and 1.2 (CI: 1.01 - 1.42) overall. Two other CRP SNPs (rs1169300 and rs2464196) were specifically associated with increased lung cancer risk. CRP SNPs were not associated with colorectal, prostate or breast cancer risk.

Established associations:

c-reactive protein measurement **you have 0 copies of the risk allele A**

(risk allele=A, odds ratio/beta 0.17 mg/dl decrease with pval 7E-21, pubmedid=18439548)

Gene related topics:

CANCER(5.00) · cancer(5.00) · breast cancer(1.00) · OTHER(1.00) · lung cancer(1.00) · IMMUNE(0.51) · inflammation(0.51) · CARDIOVASCULAR(0.03) · c reactive protein(0.03)

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Relevant papers and external links:

PMID 18439548
PMID 21647738
PMID 20727736
PMID 20727736
PHARMGKB PA162356792
Evidence: PubMed ID:18439548; Web Resource:External Link Results: Loci Related to Metabolic-Syndrome Pathways Including LEPR, HNF1A, IL6R, and GCKR Associate with Plasma C-Reactive Protein: The Women's Genome Health Study (Initial Sample Size: 6,345 women; Replication Sample Size: NR; Risk Allele: rs1892534-A). This variant is associated with C-reactive protein levels.

PHARMGKB PA161614204
Evidence: PubMed ID:18439548In a GWAS of apparantly healthy women, seven loci were found to be significantly associated with C-Reactive protein levels. Nine SNPs were significant in LEPR, and this was the lead SNP.

dbSNP: rs1892534 GWAS Catalog: rs1892534 Genecard: LEPROT chromosome 1

SNPedia: rs1892534(G;G)

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★ rs3131296(G;G) warning:2.10 **1.4x increased risk for schizophrenia**

23andMe blog schizophrenia
rs3131296(C) increased the odds of schizophrenia by 1.19x
rs12807809(T) increased the odds of schizophrenia by 1.15x
rs9960767(C) increased the odds of schizophrenia by 1.23 times.

A study of ~2,500 Han Chinese patients with schizophrenia did find rs3131296 to be significantly associated in this population.

Established associations:

schizophrenia **you have 2 copies of the risk allele G**
(risk allele=G, odds ratio/beta 1.19 with pval 2E-10, pubmedid=19571808)

Gene related topics:

PSYCH(0.38) · schizophrenia(0.38) · IMMUNE(0.34) · diabetes mellitus type 1(0.34) · scleroderma systemic(0.15) · lupus erythematosus systemic(0.14) · VISION(0.12) · macular degeneration(0.12) · CARDIOVASCULAR(0.04) · heart rate(0.04)

Relevant papers and external links:

PMID 19571808
Common variants conferring risk of schizophrenia.

PMID 20673877

dbSNP: rs3131296 GWAS Catalog: rs3131296 Genecard: NOTCH4 chromosome 6

SNPedia: rs3131296(G;G)

★ rs6323(G;-) info:2.09 14.07% **Increased monoamine oxidase A activity**

rs6323 (R297R / Arg297Arg) is a SNP in the MAOA (monoamine oxidase A) gene. Monoamine oxidase A degrades serotonin, dopamine, epineprine, and norepinephrine. The G allele encodes for the higher activity form of the enzyme.

Subjects with major depressive disorder with the highest activity form of the enzyme (G or G/G) had a significantly lower magnitude of placebo response.

Gene related topics:

PSYCH(3.00) · antisocial personality disorder(3.00) · bp major depressive(2.00) · personality disorders(1.12) · aggressive behavior(1.00) · mood disorder(0.60) · UNKNOWN(0.56) · sudden infant death(0.56) · CHEMDEPENDENCY(0.51) · smoking behavior(0.51) · personality traits(0.47) · suicide(0.32) · panic disorder(0.31) · depression(0.30) · personality(0.25) · major depressive disorder(0.24) · alcoholism(0.24) · bipolar disorder(0.24) · adhd attention deficit hyperactivity disorder(0.21) · attention deficit hyperactivity disorder(0.21) · smoking(0.21) · alcohol abuse(0.17) · adhd(0.15) · NEUROLOGICAL(0.13) · restless legs syndrome(0.13) · autism(0.12) · parkinson s disease(0.12) · migraine(0.11) · depressive disorder major(0.10) · mood disorders(0.10) · obsessive compulsive disorder(0.06) · dementia(0.04)

Relevant papers and external links:

PMID 19593178

dbSNP: rs6323 Genecard: MAOA chromosome X SNPedia: rs6323(G;G)

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★ rs27072(C;T) good:2.07 32.60% **Lower risk of alcohol withdrawal seizures, and perhaps lower odds of ADHD.**

Alcoholics with this are less likely to have seizures during alcohol withdrawal.

rs27072, a SNP in the dopamine transporter SLC6A3 gene, has been associated with more severe symptoms upon alcohol withdrawal, such as seizures, in a study of 250 Caucasian alcohol-dependent patients. Two haplotypes appear to be tagged by this SNP and a neighbor, rs27048. The rs27072 polymorphism has been shown to be significantly associated with ADHD in clinical samples of Canadian children aged 6 to 16 [Feng et al., 2005] and the significant association was also reported by Ouellet-Morin et al. in a Canadian twin study, 2008 (Am. J. of Medical Genetics Part B, 147B:1442-1449 (2008).

The interaction between the dopamine transporter gene and age at onset in relation to tobacco and alcohol use among 19-year-olds.

Association of promoter variants of human dopamine transporter gene with schizophrenia in Han Chinese.

Dopamine Transporter Gene Variant Affecting Expression in Human Brain is Associated with Bipolar Disorder.

Sequence variation in the 3'-untranslated region of the dopamine transporter gene and attention-deficit hyperactivity disorder (ADHD).

Association of the dopamine transporter gene and ADHD symptoms in a Canadian population-based sample of same-age twins.

Candidate gene studies of ADHD: a meta-analytic review.

Association between polymorphism of the dopamine transporter gene and early smoking onset: an interaction risk on nicotine dependence.

Gene and gene by sex associations with initial sensitivity to nicotine in nonsmokers.

Financial and psychological risk attitudes associated with two single nucleotide polymorphisms in the nicotine receptor (CHRNA4) gene.

Haplotypes of dopamine and serotonin transporter genes are associated with antisocial personality disorder in alcoholics.

Pharacogenetic effects of dopamine transporter gene polymorphisms on response to chlorpromazine and clozapine and on extrapyramidal syndrome in schizophrenia.

The role of dopamine transporter (SLC6A3) and dopamine D2 receptor/ankyrin repeat and kinase domain containing 1 (DRD2/ANKK1) gene polymorphisms in personality traits.

Alcohol Dependence

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Gene related topics:

PSYCH(9.95) · attention deficit hyperactivity disorder(9.95) · seizures(5.00) · adhd attention deficit hyperactivity disorder(3.41) · schizophrenia methamphetamine abuse(2.00) · CHEMDEPENDENCY(1.25) · smoking behavior(1.25) · perceptual disorders(1.00) · bipolar disorder(1.00) · brain(1.00) · antisocial personality disorder(1.00) · nicotine(1.00) · alcohol dependence(1.00) · nicotine dependence(1.00) · personality(0.98) · cocaine related disorders(0.67) · psychosis(0.60) · delirium(0.57) · impulsivity(0.57) · tourette syndrome(0.47) · adhd(0.40) · NEUROLOGICAL(0.40) · parkinson s disease(0.40) · REPRODUCTION(0.36) · prenatal exposure delayed effects(0.36) · schizophrenia(0.27) · substance related disorders(0.27) · heroin dependence(0.25) · bulimia(0.24) · alcohol(0.21) · dyslexia(0.19) · alcohol abuse(0.17) · alcoholism(0.16) · personality traits(0.12) · migraine disorders(0.08) · depression(0.07) · obsessive compulsive disorder(0.06) · autism(0.05) · smoking(0.05) · PHARMACOGENOMIC(0.02)

Relevant papers and external links:

PMID 18070248

OMIM 143465

OMIM 126455

PMID 19740369

PMID 19879111

PMID 21525861

PMID 16082693

PMID 18165969

PMID 19506906

PMID 14685824

PMID 18690117

PMID 19693267

PMID 20505557

PMID 20580759

PMID 21354244

dbSNP: rs27072 Genecard: SLC6A3 chromosome 5 SNPedia: rs27072(C;T)

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★ rs460000(C;C) info:2.06 33.60% **Better response to amphetamine**

Individuals with the C/C genotype at rs460000 (N = 83) reported approximately twofold higher ratings of stimulation and euphoria relative to the A/A+A/C (N = 69) genotype group, at both the 10 and 20 mg doses.

[PMID 20091113]

Gene related topics:

PSYCH(9.95) · attention deficit hyperactivity disorder(9.95) · adhd attention deficit hyperactivity disorder(3.41) · schizophrenia methamphetamine abuse(2.00) · CHEMDEPENDENCY(1.25) · smoking behavior(1.25) · perceptual disorders(1.00) · personality(0.98) · cocaine related disorders(0.67) · psychosis(0.60) · delirium(0.57) · impulsivity(0.57) · tourette syndrome(0.47) · adhd(0.40) · NEUROLOGICAL(0.40) · parkinson s disease(0.40) · REPRODUCTION(0.36) · prenatal exposure delayed effects(0.36) · schizophrenia(0.27) · substance related disorders(0.27) · heroin dependence(0.25) · bulimia(0.24) · alcohol(0.21) · dyslexia(0.19) · alcohol abuse(0.17) · alcoholism(0.16) · personality traits(0.12) · migraine disorders(0.08) · depression(0.07) · obsessive compulsive disorder(0.06) · autism(0.05) · smoking(0.05) · PHARMACOGENOMIC(0.02)

Relevant papers and external links:

dbSNP: rs460000 Genecard: SLC6A3 chromosome 5 SNPedia: rs460000(C;C)

★ rs2395185(G;G) good:2.05 **1.33x increased risk of Ulcerative Colitis, but much lower Type 1 Diabetes risk.**

This raises the risk of Ulcerative Colitis (according to 23andMe), but also greatly lowers the risk of Type 1 Diabetes. It may also affect other autoimmune diseases.

A genome-wide association study using DNA samples from 1,052 individuals with ulcerative colitis and preexisting data from 2,571 controls, all of European ancestry, concluded that this SNP and several others were associated with altered risk for the disease.

Established associations:

lung carcinoma **you have 0 copies of the risk allele T**

(risk allele=T, odds ratio/beta 1.17 [1.11-1.23] with pval 1E-8, pubmedid=23143601)

ulcerative colitis **you have 2 copies of the risk allele G**

(risk allele=G, odds ratio/beta 1.92 [1.68-2.19] with pval 5E-22, pubmedid=19915573)

Gene related topics:

ulcerative colitis(5.00) · type 1 diabetes(5.00) · autoimmune diseases(1.00) · OTHER(1.00)

Relevant papers and external links:

PMID 19837788

PMID 19915573

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A genome-wide association study identifies three new susceptibility loci for ulcerative colitis in the japanese population.

PMID 23143601
Genome-wide association analysis identifies new lung cancer susceptibility loci in never-smoking women in asia.

PMID 19122664

PHARMGKB PA164740081
Evidence: PubMed ID:19122664; Web Resource:External Link results: Ulcerative colitis-risk loci on chromosomes 1p36 and 12q15 found by genome-wide association study. (Initial Sample Size: 1,022 cases, 2,503 controls; Replication Sample Size: 1,387 cases, 1,115 controls); (Region: 6p21.32; Reported Gene(s): BTNL2, HLA-DRA, HLA-DRB5, HLA-DRB1, HLA-DQA1, HLA-DQB1; Risk Allele: rs2395185-?); (p-value= 0.0000000000000001).This variant is associated with Ulcerative colitis.

OMIM 604519

dbSNP: rs2395185 GWAS Catalog: rs2395185 SNPedia: rs2395185(G;G)

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★ rs10761659(A;A) warning:2.04 27.10% **1.5x risk of Crohn's disease**

rs10761659 has been reported in a large study to be associated with Crohn's disease.

The risk allele (oriented to the dbSNP entry) is (G); the odds ratio associated with heterozygotes is 1.23 (CI 1.05-1.45), and for homozygotes, 1.55 (CI 1.3-1.84).

Established associations:

crohn's disease **you have 0 copies of the risk allele G**

(risk allele=G, odds ratio/beta 1.28 with pval 5E-6, pubmedid=22936669; risk allele=G, odds ratio/beta 1.23 [1.18-1.29] with pval 4E-22, pubmedid=21102463; risk allele=G, odds ratio/beta 1.23 [1.05-1.45] with pval 2E-6, pubmedid=17554300)

inflammatory bowel disease **you have 0 copies of the risk allele G**

(risk allele=G, odds ratio/beta 1.166 [1.134-1.20] with pval 6E-46, pubmedid=23128233)

Relevant papers and external links:

PMID 22936669

A genome-wide association study on a southern european population identifies a new crohn's disease susceptibility locus at rbx1-ep300.

PMID 21102463
Genome-wide meta-analysis increases to 71 the number of confirmed crohn's disease susceptibility loci.

PMID 23128233
Host-microbe interactions have shaped the genetic architecture of inflammatory bowel disease.

PMID 17554300
OMIM 612255

dbSNP: rs10761659 GWAS Catalog: rs10761659 Genecard: nan chromosome 10

SNPedia: rs10761659(A;A)

★ rs655601(A;A) warning:2.01 51.13% **; body mass index (you have the risk allele A)**

HDL cholesterol levels being the quantitative trait associated with in

Established associations:

body mass index **you have 2 copies of the risk allele A**
(risk allele=A, HDL-C odds ratio/beta 0.23 mg/dL increase with pval 5E-6, pubmedid=19197348)

triglyceride measurement **you have 2 copies of the risk allele A**
(risk allele=A, HDL-C odds ratio/beta 0.23 mg/dL increase with pval 5E-6, pubmedid=19197348)

high density lipoprotein cholesterol measurement **you have 2 copies of the risk allele A**
(risk allele=A, HDL-C odds ratio/beta 0.23 mg/dL increase with pval 5E-6, pubmedid=19197348)

body height **you have 2 copies of the risk allele A**
(risk allele=A, HDL-C odds ratio/beta 0.23 mg/dL increase with pval 5E-6, pubmedid=19197348)

low density lipoprotein cholesterol measurement **you have 2 copies of the risk allele A**
(risk allele=A, HDL-C odds ratio/beta 0.23 mg/dL increase with pval 5E-6, pubmedid=19197348)

c-reactive protein measurement **you have 2 copies of the risk allele A**
(risk allele=A, HDL-C odds ratio/beta 0.23 mg/dL increase with pval 5E-6, pubmedid=19197348)

systolic blood pressure **you have 2 copies of the risk allele A**
(risk allele=A, HDL-C odds ratio/beta 0.23 mg/dL increase with pval 5E-6, pubmedid=19197348)

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diastolic blood pressure **you have 2 copies of the risk allele A**

(risk allele=A, HDL-C odds ratio/beta 0.23 mg/dL increase with pval 5E-6, pubmedid=19197348)

total cholesterol measurement **you have 2 copies of the risk allele A**

(risk allele=A, HDL-C odds ratio/beta 0.23 mg/dL increase with pval 5E-6, pubmedid=19197348)

waist circumference **you have 2 copies of the risk allele A**

(risk allele=A, HDL-C odds ratio/beta 0.23 mg/dL increase with pval 5E-6, pubmedid=19197348)

body weight **you have 2 copies of the risk allele A**

(risk allele=A, HDL-C odds ratio/beta 0.23 mg/dL increase with pval 5E-6, pubmedid=19197348)

thyroid stimulating hormone measurement **you have 2 copies of the risk allele A**

(risk allele=A, HDL-C odds ratio/beta 0.23 mg/dL increase with pval 5E-6, pubmedid=19197348)

body fat distribution **you have 2 copies of the risk allele A**

(risk allele=A, HDL-C odds ratio/beta 0.23 mg/dL increase with pval 5E-6, pubmedid=19197348)

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Gene related topics:

cholesterol(1.00) · hdl cholesterol(1.00)

Relevant papers and external links:

PMID 19197348

dbSNP: rs655601 GWAS Catalog: rs655601 Genecard: MGAT1 chromosome 5

SNPedia: rs655601(A;A)

★ **rs7294919(T;T)** **info:2.01** **62.12%** **Average hippocampal volume**

rs7294919, showed a particularly strong link to a reduced hippocampus volume, suggesting that this gene is very important to hippocampus development or health. No associations for brain volume, but they did discover that intracranial volume was significantly associated with two loci: rs4273712, a known height locus on chromosome 6q22, and rs9915547, tagging the inversion on chromosome 17q21. The SNP is located between two genes, HRK and FBXW8 , but evidence suggests that it

influences the expression level of a gene 3' to FBXW8, TESC . Each copy of the T allele was associated with a 107.8 mm3 decrease in hippocampal volume . In European populations, the effect allele (T) is found at frequency of 0.898 . The minor allele (C) is found at a frequency of 0.102.

Background

The hippocampus is a critical brain structure involved in learning and memory. In particular, it is associated with the ability to form long-term memories of facts and events . This is in contrast to short-term and working memory, which have been shown to be independent of the hippocampus . Hippocampal size decreases with age and is diminished in several disorders including Alzheimer's Disease , Major Depressive Disorder , Post-traumatic Stress Disorder , and Schizophrenia . Moreover, the size of the structure is heritable, with estimates of heritability ranging from 40-70% .

Studies

Two major studies were conducted which found an association between rs7294919 and hippocampal volume. They were published in the April 15th, 2012 issue of Nature Genetics . Though the P-values and regression slopes differ, both studies, together comprising tens of thousands of individuals, agree that the T allele is negatively associated with hippocampal volume.

The first study uses the CHARGE consortium (Cohorts for Heart and Aging Research in Genomic Epidemiology) as its discovery cohort . CHARGE comprises 8 sub-cohorts representing 9,232 people with an average age of 67.1 years. Most of the sub-cohorts were from Europe and hippocampal volume was determined using MRI. Second stage verification/replication was performed on two cohorts comprising 2,318 subjects. This study found that rs7294919 was associated with hippocampal volume with a meta-P value (combining the discovery and replication cohorts) of 2.9 x 10-11 and a regression slope (β) of -107.8 mm3 hippocampal volume. The authors provide some speculation about the SNP's mechanism of action by reviewing the neighboring two genes. HRK is involved in apoptosis of neurons and is thought to play a role in ischemia-induced apoptosis. FBXW8 targets an E3 ubiquitin ligase to protein aggregates and has been shown to be involved in hippocampal neuron dendrite growth. Potential pitfalls of this study include the older age of the participants, the mixture of both computerized and manual hippocampus tracing in MRIs, and the predominantly European ethnic makeup of the cohorts.

The second study's discovery cohort comprised 17 European-ancestry cohorts representing 5,775 healthy people with a mean age of 34.8 years . An additional 2,020 people with various neuropsychiatric disorders were also included to determine whether the SNPs had disease-specific effects. To verify/replicate their findings, they used several cohorts comprising both European ancestry and non-European populations (Yoruba and Los Angeles Mexican).

The study found that rs7294919 was associated with hippocampal volume with a combined P value of 1.99 x 10-7 with a regression slope of -42.74 mm3. The authors also examined the association of the SNP with IQ. While no association was found with full-scale IQ, a small association was found between having the C (minor) allele and an increase in verbal IQ (P = 0.043, β = 0.126). Using data from existing databases , rs7294919 (really rs4767492, a SNP used to impute rs7294919) was associated with expression of tescalin (TESC), a gene which lies 3' to FBXW8 in the brain. The T

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allele (associated with lower hippocampal volume) was associated with higher expression levels of TESC. TESC itself is expressed during brain development and is thought to regulate differentiation and proliferation. However, it is still not clear how increased TESC levels could be responsible for a decrease in hippocampal volume without additional studies.

References/Links

External Link

External Link

External Link

External Link

Established associations:

hippocampal volume **you have 2 copies of the risk allele T**

(risk allele=T, odds ratio/beta 107.8 [76.05-139.55] mm3 decrease with pval 3E-11, pubmedid=22504421)

brain measurement **you have 0 copies of the risk allele C**

(risk allele=C, Mean bilateral hippocampal volume odds ratio/beta 47.58 [36.04-59.12] mm3 increase with pval 7E-16, pubmedid=22504417)

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Gene related topics:

aging(1.00) · height(1.00) · memory(1.00) · stress(1.00) · brain(1.00) · schizophrenia(1.00) · hippocampus(1.00) · AGING(1.00) · major depressive disorder(1.00)

Relevant papers and external links:

PMID 22504417
Identification of common variants associated with human hippocampal and intracranial volumes.

PMID 22504421
Common variants at 12q14 and 12q24 are associated with hippocampal volume.

dbSNP: rs7294919 GWAS Catalog: rs7294919 Genecard: nan chromosome 12

SNPedia: rs7294919(T;T)

★ rs1815739(C;C) good:2.00 35.91% **Better performing muscles. Likely sprinter.**

This genotype indicates better performing muscles, particularly for sprinting and power sports. Fast-twitch muscle fibers are able to produce alpha-actinin-3. Professional sprinters usually have this, although it is less common for endurance athletes.
This SNP, in the ACTN3 gene, encodes a premature stop codon in a muscle protein called alpha-actinin-3. The polymorphism alters position 577 of the alpha-actinin-3 protein.

In the (C;C) genotype is often called RR. The truncated (T;T) is often called XX.

According to (T;T) is under-represented in elite strength athletes, consistent with previous reports indicating that alpha-actinin-3 deficiency appears to impair muscle performance. However in 2016 failed to replicate casting some doubt.

The most common nucleotide at this position, (C), encodes an arginine (amino acid code R), the alternative T allele encodes a stop codon (X). Hence, the SNP is referred to as R577X, with homozygotes being either RR or XX and heterozygotes being RX. XX individuals completely lack the expression of alpha-actinin-3.

One of the earliest report studying a relatively small number of Australian elite (i.e. ~Olympic) athletes found that, at least in females, the R allele (ie rs1815739(C)) is associated with sprinters, while the X allele (rs1815739(T)) is associated with endurance athletes. No female or Olympic-level sprinters were XX homozygotes (rs1815739(T;T)). The association tended the same way but was statistically weaker in males. . There have been several subsequent studies, but few with large sample sizes and thus few with much statistical power. An example of a typical study: no increase in endurance ability was associated with the X allele in elite male cyclists.

An extensive blog post from one of the original authors of this research.

This is one of the SNPs reported by NutraHacker.

Distribution and effects of nonsense polymorphisms in human genes.

Is there an optimum endurance polygenic profile?

ACTN3 R577X polymorphism and Israeli top-level athletes.

Is the interaction between HIF1A P582S and ACTN3 R577X determinant for power/sprint performance?

ACTN3 R577X polymorphism does not influence explosive leg muscle power in elite volleyball players.

Web-based, participant-driven studies yield novel genetic associations for common traits.

ACTN3 R577X and other polymorphisms are not associated with elite endurance athlete status in the Genathlete study.

Association between the ACTN3 R577X polymorphism and female endurance athletes in China.

Are 'endurance' alleles 'survival' alleles? Insights from the ACTN3 R577X polymorphism.

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ACTN3 genotype, athletic status, and life course physical capability: meta-analysis of the published literature and findings from nine studies.

ACTN3 R577X polymorphism and performance phenotypes in young Chinese male soldiers.

Category:Visual arts

Gene related topics:

survival(1.00) · OTHER(1.00) · NORMALVARIATION(0.67) · muscle testing(0.67) · endurance performance(0.36)

Relevant papers and external links:

PMID 18043716
PMID 18043716
PMID 26824906
PMID 12879365
PMID 16612741
OMIM 102574
PMID 18852891
PMID 19237423
PMID 19544227
PMID 20005538
PMID 20561285
PMID 20585627
PMID 20845221
PMID 20936592
PMID 21407828
PMID 21542061
PMID 22224919

dbSNP: rs1815739 Genecard: ACTN3 chromosome 11 SNPedia: rs1815739(C;C)

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★ rs4680(G;G) good:1.99 39.79% **(warrior) multiple associations, see details**

You have the VAL/VAL version of the snp discussed in this news article. It is able to perform better in a test where the optimal strategy changes.

Placebo is less effective for you

External Link
rs4680 (Val158Met) is a well studied SNP in the COMT gene. 23andMe blog summarizes them as
rs4680(A) = Worrier. Met, more exploratory, lower COMT enzymatic activity, therefore higher dopamine levels; lower pain threshold, enhanced vulnerability to stress, yet also more efficient at processing information under most conditions
rs4680(G) = Warrior. Val, less exploratory, higher COMT enzymatic activity, therefore lower dopamine levels; higher pain threshold, better stress resiliency, albeit with a modest reduction in executive cognition performance under most conditions

Roughly speaking, the predominant wisdom (known colloquially as the warrior/worrier hypothesis; summary at) posits that people with Val alleles have increased COMT activity and lower prefrontal extracellular dopamine compared with those with the Met substitution. Val158 alleles may be associated with an advantage in the processing of aversive stimuli (warrior strategy), while Met158 alleles may be associated with an advantage in memory and attention tasks (worrier strategy). Under conditions of increased dopamine release (eg, stress), individuals with Val158 alleles may have improved dopaminergic transmission and better performance, while individuals with Met158 alleles may have less efficient neurotransmission and worse performance. Some evidence suggests that Val158 alleles are associated with schizophrenia, while Met158 alleles are associated with anxiety.

Specific studies include:

rs4680, a functional Val/Met polymorphism, showed modest association with Irish familial schizophrenia. Haplotype A-G-A for SNPs rs737865-rs4680-rs165599 was preferentially transmitted to the affected subjects.

A study of 400 individuals reported that an increase in plasma total homocysteine (tHcy) of 10.4% (CI: 0.01-0.21, p=0.03) for associated with rs4680(A;A) homozygotes compared with rs4680(G;G) subjects. The (A;A) genotype was also more common, but statistically not that significantly, in venous thrombosis patients (OR 1.61, CI: 0.97-2.65], p=0.06) compared to control subjects.

And from :
"Adolescent cannabis use was associated with increased risk of schizophreniform disorder in adulthood among Val/Val (i.e. rs4680(G;G)) individuals (OR = 10.9, 95% CI: 2.2â€“54.1) and, to a lesser extent, among Met/Val individuals (rs4680(A;G)); OR = 2.5, 95% CI: .78 â€“ 8.2), but not among Met/Met (rs4680(A;A)) individuals (OR = 1.1, 95% CI: .21â€“5.4)."

Also potentially associated with schizophrenia

As part of a haplotype with rs4633, the (G) allele is also associated with endometrial cancer

In a study of 2 populations of breast cancer patients (2,000+ patients), increased risk was associated with rs4680(G;G) genotypes in both the Ontario [odds ratio 2.22, CI: 1.49-3.28] and Finland [OR 1.73, CI: 1.08-2.78] populations.

23andMe blog The A version of rs4680 appears to boost working memory and cognitive function compared to G â€” but it also hampers emotional control.

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A study of 330 cocaine-dependent individuals, all of African descent, concluded that there was a slight (odds ratio 1.44, CI: 1.12-1.86, p = 0.014) association between the rs4680(A) allele, in other words the Met encoding allele, and cocaine dependence.

A study of the antidepressant paroxetine found better response in Met/Met homozygotes, worse effects in Val/Val homozygotes and intermediate effects in heterozygotes. The effect became significant at the third week of treatment. Paroxetine daily dose was proportional to baseline severity, but did not influence outcome.

g2b2mh (A;A) subjects deploy more attentional focus when they realize they have made an error.

A meta-analysis of neuroimaging studies found a significant association between the COMT genotype and prefrontal activation; strong and opposing effects were found for executive cognition paradigms (favoring Met allele carriers) and emotional paradigms (favoring Val).

gray matter volume and interacts with rs2097603 related to extracellular dopamine

rs6275/DRD2 and rs4680/COMT useful in predicting disease risk among schizophrenia patients

rs4680(G;G) carriers deprived of sleep respond quite well to 2x 100mg modafinil in terms of improved vigor and well-being, and maintained baseline performance with respect to executive functioning, whereas rs4680(A;A) individuals barely responded to the drug at all.

Met158 allele carriers had "a more focused response...during a working memory task." "The met158 allele seems to be beneficial during the performance of working memory and attention-related tasks, whereas the val158 allele appears to be advantageous during the processing of aversive emotional stimuli."

"Increased limbic and prefrontal activation elicited by unpleasant stimuli in subjects with more met158 alleles might contribute to the observed lower emotional resilience against negative mood states."

In response to pain Met/Met allele carriers showed greater "sensory and affective ratings of pain and a more negative internal affective state. Opposite effects were observed in val158 homozygotes. The COMT val158met polymorphism thus influences the human experience of pain and may underlie interindividual differences in the adaptation and responses to pain and other stressful stimuli."

Reports that ease of entering hypnosis correlates with number of rs4680(G) alleles.

see gs226 for a report related to impulsiveness

External Link and blog discussion of data about rs4680 thinking about nothing in an fmri.

Breast Cancer Risk Modifiers

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Established associations:

blood metabolite measurement **you have 0 copies of the risk allele A**

(risk allele=A, X-01911 odds ratio/beta 0.044 [0.032-0.056] unit decrease with pval 1E-13, pubmedid=24816252; risk allele=A, X-11593--O-methylascorbate odds ratio/beta 0.049 [0.045-0.053] unit decrease with pval 5E-178, pubmedid=24816252)

Gene related topics:

UNKNOWN(6.00) · digeorge syndrome pharyngeal pouch syndrome(6.00) · PSYCH(5.72) · schizophrenia(5.72) · CANCER(3.00) · neoplasms pain(3.00) · fibromyalgia(2.67) · NEUROLOGICAL(2.00) · mamographic density(2.00) · pain temporomandibular joint disorders(2.00) · psychosis(1.67) · obsessive compulsive disorder(1.31) · CHEMDEPENDENCY(1.07) · substance related disorders(1.07) · panic disorder(1.03) · adhd attention deficit hyperactivity disorder(1.00) · eating disorders(1.00) · sleep(1.00) · cognitive function(1.00) · anxiety(1.00) · homocysteine(1.00) · memory(1.00) · stress(1.00) · cancer(1.00) · thrombosis(1.00) · venous thrombosis(1.00) · cocaine dependence(1.00) · pain(0.94) · breast cancer(0.83) · marijuana abuse(0.67) · mood disorders(0.62) · parkinson s disease(0.62) · DEVELOPMENTAL(0.60) · leiomyoma uterine neoplasms(0.60) · mental retardation(0.60) · schizophrenia bipolar disorder(0.60) · OTHER(0.57) · sleep deprivation(0.57) · bipolar disorder(0.50) · depression(0.50) · fractures bone(0.50) · personality(0.38) · mammographic density(0.36) · personality traits(0.33) · heroin abuse(0.30) · alcoholism(0.29) · adhd(0.26) · cognitive ability(0.25) · heroin dependence(0.25) · bulimia(0.24) · endometrial cancer(0.23) · smoking(0.21) · anorexia nervosa(0.15) · suicide(0.14) · liver cancer(0.13) · tobacco use disorder(0.11) · narcolepsy(0.11) · METABOLIC(0.10) · weight gain(0.10) · smoking behavior(0.09) · endometrial neoplasms(0.07) · attention deficit hyperactivity disorder(0.05) · IMMUNE(0.05) · vitiligo(0.05) · autism(0.03) · REPRODUCTION(0.02) · endometriosis(0.02)

Relevant papers and external links:

PMID 24816252
An atlas of genetic influences on human blood metabolites.

PMID 17008817

PMID 18064318

PMID 15866551

PMID 18194538

PMID 18704099

PMID 18989660

PMID 19417742

PMID 19071221

PMID 19207030

PMID 19037200

PMID 16878403

PMID 15673663

PMID 12595695

PMID 20509070

OMIM 116790

OMIM 104300

PHARMGKB PA162630417

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Evidence: PubMed ID:19207030This variant (rs4680/COMT) along with rs6275/DRD2 are associated with risk for schizophrenia in a study consisting of 254 patients and 225 controls of southern indian origin.

PHARMGKB PA161145211
Evidence: Web Resource:External Link variant is responsible for low (Met) and high (Val) activity alleles that have been studied in relation to schizophrenia and other psychiatric/neurological disorders and in relation to breast cancer.

PHARMGKB PA161748325
Evidence: PubMed ID:17156920In an study at 207 patients treated with morphine, carriers of COMT Val/Val and Val/Met genotype required 63% and 23%, respectively, higher morphine dose compared to carriers of Met/Met genotype (p=0.02).

PHARMGKB PA164944057
Evidence: PubMed ID:19365560The minor allele A of this 472G>A variant produces a valine to methionine substitution, resulting in a less thermostable COMT enzyme that exhibits a 3-fold reduction in activity.

PHARMGKB PA164944062
Evidence: PubMed ID:15537663On the basis of subjects' pain responsiveness, haplotypes involving rs6269 (A/G), rs4633 (C/T), rs4818 (C/G), and rs4680 (G/A) were designated as low (low pain sensitivity (LPS) haplotype; GCGG), average (average pain sensitivity (APS) haplotype; ATCA), or high (high pain sensitivity (HPS) haplotype; ACCG) pain sensitive.

dbSNP: rs4680 GWAS Catalog: rs4680 Genecard: COMT chromosome 22

SNPedia: rs4680(G;G)

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★ rs1801133(C;C) good:1.98 56.94% **Common genotype: normal homocysteine levels**

rs1801133 is a SNP that is relatively common and has been studied for (relatively) a long time. Also known as C677T, Ala222Val, and A222V, it encodes a variant in the MTHFR gene, which encodes an enzyme involved in folate metabolism.

Homozygous rs1801133(T;T) individuals have ~30% of the expected MTHFR enzyme activity, and rs1801133(C;T) heterozygotes have ~65% activity, compared to the most common genotype, rs1801133(C;C).

This reduced activity (i.e. this SNP) has been linked at least once to each of the following disorders (though not necessarily reproducibly):

autism
cancer, including
gastric cancer
lung cancer
head and neck cancer
renal cancer
cleft lip and cleft palate
coronary artery disease
dementia
depression
hyperhomocysteinemia
migraine
neural tube defects
pre-eclampsia (gestational hypertension)
schizophrenia
thrombosis
down syndrome

PubMed lists over 2,300 studies linking Rs1801133 to a long list of disorders in various populations across the world, of which only some are mentioned above. Considering the central role of MTHFR in folate metabolism and in control of homocysteine levels this is not surprising. A nice summary of the pathological significance of elevated homocysteine levels could be found in this article.

With regard to gastric cancer, a meta-analysis combining 16 studies and ~2,700 patients concluded that the increased risk (odds ratio) associated with rs1801133 (T;T) and (C;T) genotypes, was 1.52 (CI 1.31-1.77) and 1.17 (CI 0.99-1.39), respectively, compared to the (C;C) genotype. Roughly the same risks were seen for Caucasians and Asians. Smoking and having low folate levels (presumably from diets low in fruits and veggies) increased the risk for (T;T) individuals from ~1.5x to ~2x, whereas having high folate levels almost reduced the risk for (T;T) genotypes pretty much down to the (C;C) level, ie. the average risk.

Another meta-analysis of three studies on rs1801133 stratified according to dietary folate intake showed an increased risk for individuals with low folate intake (OR=1.37, CI: 0.92-2.06 for head and neck cancer and OR=1.28, CI: 0.97-1.68 for lung) versus high folate intake (OR=0.85, CI: 0.63-1.16 for head and neck cancer, and OR=0.94, CI: 0.79-1.12 for lung).

rs1801131 and rs1801133 have been linked to increased risk for several types of brain cancer. The highest risk of meningioma was associated with heterozygosity for both MTHFR SNPs (odds ratio 2.11, CI: 1.42-3.12, p=0.002). The corresponding odds ratio for glioma was 1.23 (CI: 0.91-1.66, p=0.02. In general, risks were increased with genotypes associated with reduced MTHFR activity.

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Based on a study of 25,000 Caucasian women followed for 11 years (on average), the rs1801133 (T;T) genotype individual was less likely to have migraine with aura (odds ratio 0.79, CI: 0.65-0.9 6, p = 0.02) and did not have increased risk for cardiovascular disease. However, if a (T;T) genotype did have migraine with aura, then the risk for cardiovascular disease was increased 3.66 fold (CI: 1.69-7.90, p = 0.001). This was apparently driven by a 4x increased risk for ischemic stroke (multivariable-adjusted relative risk 4.19, CI: 1.38-12.74, p = 0.01).

A study of 677 patients with end-stage renal disease (ESRD) concluded that the adjusted hazard ratio for mortality in all patients was 2.27 (CI: 1.07 - 4.84, p = 0.03) for rs1801133(T;T) homozygotes, in other words, they died at about twice the rate of the other 2 genotypes over the time course of this study.

In a case-control study of 152 cases (men) and 304 age-matched healthy controls conducted in one geographical area of Iran, evidence was seen that the MTHFR polymorphisms might contribute to increased clear cell renal cell carcinoma (CCRCC) risk in men. After controlling for confounding factors, a significant increase in CCRCC risk was found among carriers of the 677CT genotype compared with those with the 677CC genotype (odds ratio 2.21, 95% confidence interval 1.31-3.76), with a significant trend (P=0.014). Statistically significant odds ratios were also found in patients homozygous for MTHFR C677T, who have a 1.58-fold higher risk of developing CCRCC (95% confidence interval=1.21-2.44; P=0.024). Compared with the MTHFR 677CC genotype, the odds ratio (95% confidence interval) for the MTHFR 677TT genotype was 6.18 (95% confidence interval=4.75-8.34) for stage IV cancer and 4.68 (95% confidence interval=2.72-6.54) for grade 3 CCRCC (both P=0.0001). Clin Oncol (R Coll Radiol). 2012 May;24(4):269-81. doi: 10.1016/j.clon.2011.03.005. Epub 2011 Apr 13.

A 2-year follow-up study of 122 newly diagnosed patients with acute lymphoblastic leukemia (ALL) found that carriers of a rs1801133(T) allele were at increased risk for hepatic toxicity from methotrexate treatment. Hepatic toxicity was increased ~2x and ~5x for heterozygous and homozygous rs1801133(T) genotypes, respectively (p=0.028). If a carrier of a rs1801133(T) allele was also a carrier of a rs70991108 deletion allele, the risk for hepatic toxicity was even higher (odds ratio 6.8, p=0.018).

Note: Another SNP in dbSNP, rs59514310, represents the same location as rs1801133.

blog The MTHFR gene polymorphism is associated with lean body mass but not fat body mass

rs1801133(T;T) post-menopausal women said to be at 2x higher risk for osteoporosis.

MTHFR 677C→T polymorphism is more prevalent among mothers of children with Down syndrome than among control mothers, with an odds ratio of 1.91

Gene-wide association study between the methylenetetrahydrofolate reductase gene (MTHFR) and schizophrenia in the Japanese population, with an updated meta-analysis on currently available data.

rs1801133 shows no consistent association with schizophrenia overall in 12 studies totaling 3,000+ patients

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Established associations:

homocysteine measurement **you have 0 copies of the risk allele T**

(risk allele=T, odds ratio/beta 0.1583 [0.14-0.17] unit increase with pval 4E-104, pubmedid=23824729; risk allele=T, WGHS odds ratio/beta 0.05 unit increase in loghomocysteine with pval 8E-35, pubmedid=20031578)

Gene related topics:

METABOLIC(14.69) · hyperhomocysteinemia(14.69) · DEVELOPMENTAL(14.49) · neural tube defects(14.49) · down syndrome(11.36) · CARDIOVASCULAR(10.00) · coronary artery disease hyperhomocysteinemia(10.00) · folic acid deficiency(7.00) · homocysteine(6.92) · UNKNOWN(5.00) · chronic renal failure hyperhomocysteinemia kidney failure chronic(5.00) · coronary disease coronary heart disease hyperhomocysteinemia(5.00) · CANCER(4.00) · adenoma colorectal neoplasms neoplasm recurrence local(4.00) · hyperhomocysteinemia retinal vein occlusion(4.00) · spinal dysraphism(3.85) · colorectal cancer(3.74) · arteriosclerosis(3.57) · apoplexy stroke(3.12) · apoplexy brain ischemia hyperhomocysteinemia stroke(3.00) · cardiovascular diseases hyperhomocysteinemia(3.00) · congenital heart defects heart defects congenital(3.00) · diabetes mellitus type ii diabetes mellitus type 2 hyperhomocysteinemia(3.00) · hearing loss sudden(3.00) · hyperhomocysteinemia postoperative complications(3.00) · hyperhomocystinemia(3.00) · vascular diseases(3.00) · cardiovascular diseases(2.93) · apoplexy brain ischemia stroke(2.61) · REPRODUCTION(2.61) · pregnancy loss(2.61) · abortion spontaneous(2.58) · leukemia lymphocytic acute l1 precursor cell lymphoblastic leukemia lymphoma(2.58) · pregnancy complications(2.40) · precursor cell lymphoblastic leukemia lymphoma(2.16) · colonic neoplasms(2.13) · IMMUNE(2.00) · NEUROLOGICAL(2.00) · VISION(2.00) · abortion habitual hyperhomocysteinemia(2.00) · abortion habitual pregnancy complications hematologic thrombophilia(2.00) · abruptio placentae(2.00) · activated protein c resistance hyperhomocysteinemia thrombophilia venous thrombosis(2.00) · acute lymphocytic leukemia hyperhomocysteinemia precursor cell lymphoblastic leukemia lymphoma(2.00) · atherosclerosis thrombosis(2.00) · cardiovascular diseases chronic renal failure hyperhomocysteinemia kidney failure chronic(2.00) · cerebrovascular disease ischemic(2.00) · chronic ulcerative colitis colitis ulcerative folic acid deficiency hyperhomocysteinemia(2.00) · cleft lip cleft palate syndrome(2.00) · epilepsy hyperhomocysteinemia(2.00) · glaucoma angle closure glaucoma open angle(2.00) · heart defects congenital(2.00) · hyperhomocysteinemia hyperlipidemias(2.00) · hyperhomocysteinemia hypertension(2.00) · hyperhomocysteinemia pre eclampsia(2.00) · retinal artery occlusion retinal vein occlusion(2.00) · hyperhomocysteinemia venous thrombosis(1.80) · venous thrombosis(1.78) · abortion habitual thrombophilia(1.71) · stomach neoplasms(1.71) · HEMATOLOGICAL(1.58) · thrombophilia(1.58) · thrombosis(1.44) · PHARMACOGENOMIC(1.39) · methotrexate toxicity(1.39) · thromboembolism venous(1.35) · anemia sickle cell sickle cell anemia(1.29) · cleft lip cleft palate(1.29) · adenocarcinoma stomach neoplasms(1.25) · acute lymphocytic leukemia precursor cell lymphoblastic leukemia lymphoma(1.25) · diabetes mellitus type ii diabetes mellitus type 2 diabetic nephropathies diabetic nephropathy(1.12) · coronary disease coronary heart disease(1.10) · pre eclampsia(1.05) · coronary artery disease(1.04) · antiphospholipid syndrome thrombophilia thrombosis(1.00) · brain ischemia(1.00) · brain neoplasms glioma meningeal neoplasms meningioma(1.00) · carcinoma squamous cell skin neoplasms squamous cell carcinoma(1.00) · cervical intraepithelial neoplasia cervical neoplasm papillomavirus infections uterine cervical neoplasms(1.00) · migraine with aura migraine without aura(1.00) · thrombophilia and vascular disease(1.00) · gastric cancer(1.00) · metabolism(1.00) · renal cancer(1.00) · brain(1.00) · body mass(1.00) · cancer(1.00) · glioma(1.00) · cleft lip(1.00) · enzyme activity(1.00) · lung cancer(1.00) · mortality(1.00) · brain cancer(1.00) · smoking(1.00) · head and neck cancer(1.00) · breast cancer(0.93) · migraine disorders(0.92) · thrombosis deep vein(0.86) · RENAL(0.84) · diabetic nephropathy(0.84) · pregnancy loss recurrent(0.83) · pre eclampsia thrombophilia(0.82) · esophageal cancer stomach cancer(0.80) · abortion habitual(0.75) · preeclampsia(0.73) · arthritis rheumatoid rheumatoid arthritis(0.71) · PSYCH(0.67) · behcet syndrome venous thrombosis(0.67) · brain ischemia stroke thrombophilia(0.67) · cerebrovascular disorders(0.67) · congenital abnormalities(0.67) · dementia vascular(0.67) · diabetic nephropathies diabetic nephropathy(0.67) · aortic aneurysm abdominal(0.59) · insulin resistance polycystic ovarian syndrome polycystic ovary syndrome(0.57) · heart anomalies congenital(0.53) · leukemia(0.52) · atherosclerosis(0.51) · hypertension(0.51) · cardiovascular risk(0.50) · cerebrovascular

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disease(0.50) · colonic neoplasms rectal neoplasms(0.50) · thrombosis venous thrombosis(0.50) · chronic renal failure kidney failure chronic(0.49) · esophageal cancer(0.44) · fatty liver(0.44) · adenoma colorectal neoplasms(0.44) · migraine(0.43) · inflammatory bowel disease nos inflammatory bowel diseases(0.43) · OTHER(0.40) · chromosome aberrations dna damage(0.40) · chronic progressive chorea huntington disease(0.40) · infertility male(0.38) · venous thromboembolism(0.36) · alzheimer s disease(0.36) · intima media thickness(0.33) · migraine with aura(0.33) · brain ischemia stroke(0.31) · infertility female(0.31) · stroke ischemic(0.30) · schizophrenia(0.30) · multiple myeloma(0.29) · atherosclerosis coronary(0.29) · schizophrenia bipolar disorder(0.27) · colorectal neoplasms(0.26) · body weight obesity(0.26) · cardiovascular disease(0.25) · stroke(0.24) · bladder neoplasm urinary bladder neoplasms(0.24) · stomach cancer(0.23) · carcinoma squamous cell esophageal neoplasms oesophageal neoplasm squamous cell carcinoma(0.23) · diabetes type 2(0.23) · arthritis rheumatoid(0.23) · myocardial ischemia(0.21) · CHEMDEPENDENCY(0.21) · alcohol(0.21) · bladder cancer(0.20) · kidney transplant complications(0.19) · neoplasms(0.19) · myocardial infarction(0.17) · arthritis juvenile rheumatoid chronic childhood arthritis(0.17) · myocardial infarct(0.16) · cerebral infarction(0.15) · kidney failure chronic(0.14) · lymphoma(0.14) · kidney diseases(0.14) · choroidal neovascularization macular degeneration(0.13) · cleft lip with cleft palate cleft lip without cleft palate(0.12) · restenosis(0.12) · type 2 diabetes(0.12) · carcinoma hepatocellular lcc liver cell carcinoma liver neoplasms(0.11) · INFECTION(0.11) · helicobacter infections(0.11) · depression(0.11) · leukemia lymphocytic chronic b cell(0.11) · carotid artery diseases(0.10) · cerebral palsy(0.09) · AGING(0.08) · longevity(0.08) · cardiovascular(0.08) · carcinoma squamous cell head and neck neoplasms squamous cell carcinoma(0.08) · polycystic ovary syndrome(0.08) · graft vs host disease(0.06) · dna damage(0.06) · autism(0.05) · atrial fibrillation(0.05) · lymphoma non hodgkin(0.05) · NORMALVARIATION(0.04) · normal variation(0.04) · crohn s disease ulcerative colitis(0.04) · dementia(0.04) · osteoporosis(0.04) · coronary disease(0.02) · cervical cancer(0.02)

Relevant papers and external links:

PMID 23824729
Common genetic loci influencing plasma homocysteine concentrations and their effect on risk of coronary artery disease.

PMID 20031578
Novel associations of cps1, mut, nox4, and dpep1 with plasma homocysteine in a healthy population: a genome-wide evaluation of 13 974 participants in the women's genome health study.

PMID 18162478

PMID 18789576

PMID 18483342

PMID 18672474

PMID 19272686

PMID 21489764

PMID 19648163

PHARMGKB PA165110386
Evidence: PubMed ID:18580170Risk or phenotype-associated allele(s): T/T. Phenotype:Patients with a homozygous MTHFR 677C>T mutation (n = 25) developed higher plasma homocysteine concentrations (median [interquartile range], 14.9 [10.0-26.4] microm) than wild-type or heterozygous patients (9.3 [7.5-15.5] microm; n = 115). The change in homocysteine after nitrous oxide anesthesia was tripled in homozygous patients compared with wild-type (5.6 microm [+60%] vs. 1.8 microm [+22%]). Study size: 140. Study population/ethnicity: healthy patients undergoing elective surgery. Type of association: GN.

OMIM 607093

PHARMGKB PA161145165
Evidence: Web Resource:External Link studied, associated with multiple phenotypes.

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PHARMGKB PA162316602
Evidence: PubMed ID:17488658This variant is associated with methotrexated-induced mucositis, thrombocytopenia and hepatic toxicity

PHARMGKB PA162316601
Evidence: PubMed ID:18381794This variant is associated with methotrexated-induced alopecia in African Americans with rheumatoid arthritis.

PHARMGKB PA164920416
Evidence: PubMed ID:19581920This variant protein has reduced catalytic activity and thermolability, and is thus associated with elevated homocysteine levels under conditions of impaired folate status.

PHARMGKB PA164918288
Evidence: PubMed ID:19303062The T allele of this SNP was found to be associated with higher blood concentration of homocysteine in Italians (Combined studies of Tuscan Italians (inCHIANTI:N =1175 and Progetto Nutrizione study:N = 687) and Sardinians (SardiNIA: N=1115)) but not in the BLSA cohort (from Baltimore-Washington DC). The study authors inferred that this was related to U.S. food fortification and hence higher folate status.

PHARMGKB PA164918205
Evidence: PubMed ID:15643524In a case control study of Asian gastric cancer (n=633), individuals with 6 variant alleles of three MTHFR common variants (i.e. C677T, A1298C and G1793A) were at increased risk for gastric cardia adenocarcinoma (OR =4.64, 95% CI =1.34-16.01) compared with those having 0-2 variants.

PHARMGKB PA165106866
Evidence: PubMed ID:12915598In 53 newly diagnosed patients with childhood acute lymphoblastic leukemia who were treated with two courses of high-dose methotrexate (MTX), no association was found between MTX-induced increases in plasma or cerebrospinal fluid homocysteine levels or MTX-induced toxicity (seizures or thrombosis) based upon the MTHFR 677C>T or RFC (SLC19A1) 80G>A genotypes.

PHARMGKB PA165106864
Evidence: PubMed ID:11418485In patients with chronic myelogenous leukemia undergoing bone marrow transplantation and taking methotrexate, carriers of the MTHFR 677TT genotype versus CT and CC genotypes showed significantly greater post-allograft drug-induced toxicity in the form of oral mucositis (p=0.046), and a non-significant trend toward slower platelet recovery.

PHARMGKB PA165108052
Evidence: PubMed ID:17976958Risk or phenotype-associated allele: T. Phenotype: 53% of T allele carriers met metabolic syndrome criteria, compared with 23% in the CC genotype group. Study size: 58. Study population/ethnicity: Individuals with schizophrenia receiving atypical antipsychotics (AAPs) for 12 or more months. Over 85% of the subjects were Caucasian. Significance metric(s): OR = 3.7; p = 0.02. Type of association: CO.

PHARMGKB PA165108053
Evidence: PubMed ID:17976958Risk or phenotype-associated allele: T. Phenotype: Both waist circumference and the MTHFR genotype were significant factors associated with insulin resistance. Study size: 58. Study population/ethnicity: Individuals with schizophrenia receiving atypical

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antipsychotics (AAPs) for 12 or more months. Over 85% of the subjects were Caucasian. Significance metric(s): p < 0.0001. Type of association: CO.

PHARMGKB PA165106826
Evidence: PubMed ID:11710708MTHFR rs1801133, 667CT or 667TT genotypes were associated with an increased risk of methotrexate treatment discontinuation due to adverse events (relative risk 2.01), mostly as a result of increased risk of elevated levels of liver enzyme alanine aminotransferase (relative risk 2.38) in rheumatoid arthritis patients.

PHARMGKB PA165106865
Evidence: PubMed ID:12453860In a retrospective analysis of 61 Italian patients experiencing methotrexate toxicity during treatment for acute lymphoblastic leukemia or acute promyelocytic leukemia, carriers of the MTHFR 677TT genotype (60%) showed significantly greater drug-induced toxicity (p=0.03) compared to CC and CT genotypes.

PHARMGKB PA165109580
Evidence: PubMed ID:11710708Risk or phenotype-associated allele: CT and TT genotypes. Phenotype: The 677 CT or TT genotypes were associated with greater incidence of discontinuation of methorexate treatment because of adverse events, mainly due to elevation of liver enzymes. Study size: 236. Study population/ethnicity: Patients who started methorexate treatment with (n = 157) or without (n = 79) folic or folinic acid supplementation for rheumatoid arthritis. Significance metric(s): RR = 2.01 Type of association: CO, GN.

PHARMGKB PA165282249
Evidence: PubMed ID:19005482Risk or phenotype-associated allele: T. Phenotype: MTHFR:677T genotype in the transplant recipient was associated with acute graft vs host disease. Study size:107 . Study population/ethnicity: Donors and patients with leukemia after HLA-identical hematopoietic stem cell transplantation, France. Significance metric(s): p = 0.03. Type of association: CO

PHARMGKB PA165106616
Evidence: PubMed ID:16572443Given methotrexate and folic acid therapy, patients with the MTHFR 1298AA / 677CC diplotype showed greater clinical improvement for early rheumatoid arthritis.

PHARMGKB PA165106621
Evidence: PubMed ID:19016697In 330 patients who completed 3 months methotrexate treatment for psoriasis, no significant genotypic associations were found between clinical outcome (e.g. efficacy, toxicity) and 50 SNPs in pathway genes for methotrexate metabolism (ATIC, FPGS, GGH, MTHFR), including 47 common (>5% minor allele frequency) haplotype-tagging SNPs (r(2) > 0.8) plus 3 additional SNPs.

PMID 10930360

PMID 20692813

PMID 21302350

OMIM 607093

dbSNP: rs1801133 GWAS Catalog: rs1801133 Genecard: MTHFR chromosome 1

SNPedia: rs1801133(C;C)

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★ rs838133(C;C) info:1.95 59.56% **Lower odds preferring sweet snack**

“Genetic associations with traits in 23andMe customers.” 23andMe White Paper.

Salty or sweet - 23andMe blog entry

Established associations:

energy intake **you have 0 copies of the risk allele T**

(risk allele=T, Protein - model 2 odds ratio/beta 0.11 [0.071-0.149] percent decrease with pval 8E-9, pubmedid=23372041)

homocysteine measurement **you have 0 copies of the risk allele T**

(risk allele=T, odds ratio/beta 0.0422 [0.028-0.056] unit increase with pval 7E-9, pubmedid=23824729)

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Relevant papers and external links:

PMID 23824729
Common genetic loci influencing plasma homocysteine concentrations and their effect on risk of coronary artery disease.

PMID 23372041
Novel locus including fgf21 is associated with dietary macronutrient intake.

dbSNP: rs838133 GWAS Catalog: rs838133 Genecard: FUT1 chromosome 19

SNPedia: rs838133(C;C)

★ rs1468271(A;A) good:1.93 97.38%

Physiogenomic analysis of weight loss induced by dietary carbohydrate restriction.

Exploring genetic variations that may be associated with the direct effects of some antipsychotics on lipid levels.

Physiogenomic comparison of human fat loss in response to diets restrictive of carbohydrate or fat.

Physiogenomic comparison of edema and BMI in patients receiving rosiglitazone or pioglitazone.

Neuropeptide Y gene polymorphisms confer risk of early-onset atherosclerosis.

Gene related topics:

atherosclerosis(1.00) · bmi(1.00) · weight loss(1.00) · lipid levels(1.00) · weight(1.00) · CHEMDEPENDENCY(0.22) · alcohol dependence(0.22) · alcohol abuse(0.17) · CANCER(0.05) · lymphoma non hodgkin(0.05) · CARDIOVASCULAR(0.03) · c reactive protein(0.03)

Relevant papers and external links:

PMID 16700901

PMID 18031993

PMID 18254975

PMID 18996102

PMID 19119412

dbSNP: rs1468271 Genecard: NPY chromosome 7 SNPedia: rs1468271(A;A)

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★ rs1495741(A;G) good:1.89 45.80% **NAT2 intermediate metabolizer (predicted)**

see discussion at rs1495741

With 99% sensitivity and 95% specificity, at least in Caucasian populations, rs1495741 is reported to classify the NAT2 phenotype of an individual; it therefore is a candidate to replace the 7-SNP panel used in genosets such as gs138, gs139, and gs140. The rs1495741(A;A), (A;G) and (G;G) genotypes predicted slow, intermediate, and rapid NAT2 acetylation phenotypes, respectively.

Established associations:

triglyceride measurement **you have 1 copies of the risk allele G**

(risk allele=G, odds ratio/beta 0.04 mg/dL increase with pval 3E-12, pubmedid=24097068; risk allele=G, odds ratio/beta 2.97 [2.15-3.79] mg/dL increase with pval 4E-14, pubmedid=20686565)

total cholesterol measurement **you have 1 copies of the risk allele G**

(risk allele=G, odds ratio/beta 0.032 unit increase with pval 3E-8, pubmedid=24097068; risk allele=G, odds ratio/beta 1.07 [0.66-1.48] mg/dL increase with pval 2E-9, pubmedid=20686565)

blood metabolite measurement **you have 1 copies of the risk allele A**

(risk allele=A, 1-methylurate odds ratio/beta 0.057 [0.047-0.067] unit increase with pval 1E-27, pubmedid=24816252)

bladder carcinoma **you have 1 copies of the risk allele A**

(risk allele=A, odds ratio/beta 1.14 [1.09-1.18] with pval 2E-10, pubmedid=24163127)

Relevant papers and external links:

PMID 24097068
Discovery and refinement of loci associated with lipid levels.

PMID 20686565
Biological, clinical and population relevance of 95 loci for blood lipids.

PMID 24163127
Genome-wide association study identifies multiple loci associated with bladder cancer risk.

PMID 24816252
An atlas of genetic influences on human blood metabolites.

PMID 20739907

dbSNP: rs1495741 GWAS Catalog: rs1495741 Genecard: nan chromosome 8

SNPedia: rs1495741(A;G)

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★ **rs2710102(C;T)** **info:1.88** **48.43%** **None**

rs2710102, a common SNP in the CNTNAP2 gene, was found to be significantly associated ($p<0.028$) with a delayed onset of speech, as measured by the age at which a child speaks their first words, in children with autism. This effect is primarily seen in males, perhaps correlated with the 4-5x overrepresentation of males with autism compared with females.

The confirmatory Stage 2 study was performed on 304 independent parent-child trios. However, the risk allele and the degree to which speech is delayed per genotype is unclear as published and awaits clarification by the authors.

Speech development rs4431523, rs17236239 and significant associations (with P values from 0.01 to 5.0x10–5) between nonsense-word repetition and nine intronic SNPs (rs851715, rs10246256, rs2710102, rs759178, rs1922892, rs2538991, rs17236239, rs2538976, and rs2710117)

Gene related topics:

PSYCH(0.67) · mental disorders(0.67) · OTHER(0.12) · breath tests(0.12) · autism(0.09) · CARDIOVASCULAR(0.03) · echocardiography(0.03) · METABOLIC(0.03) · diabetes mellitus(0.03)

Relevant papers and external links:

PMID 18179893

PMID 18179893

PMID 18987363

OMIM 604569

PHARMGKB PA162168113
Evidence: PubMed ID:18179893This variant in CNTNAP2 is associated with delay of speech in autism spectrum disorder samples and appeared to be most pronounced in males.

OMIM 604569

dbSNP: rs2710102 Genecard: CNTNAP2 chromosome 7 SNPedia: rs2710102(C;T)

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★ rs851715(A;G) info:1.84 35.60% **None**

Speech development rs4431523, rs17236239 and significant associations (with P values from 0.01 to 5.0x10–5) between nonsense-word repetition and nine intronic SNPs (rs851715, rs10246256, rs2710102, rs759178, rs1922892, rs2538991, rs17236239, rs2538976, and rs2710117)

Gene related topics:

PSYCH(0.67) · mental disorders(0.67) · OTHER(0.12) · breath tests(0.12) · autism(0.09) · CARDIOVASCULAR(0.03) · echocardiography(0.03) · METABOLIC(0.03) · diabetes mellitus(0.03)

Relevant papers and external links:

PMID 18987363

dbSNP: rs851715 Genecard: CNTNAP2 chromosome 7 SNPedia: rs851715(A;G)

★ rs1858830(C;C) info:1.80 29.43% **2x risk of autism**

The C allele appears to increase risk of autism by an odds ratio of perhaps 1.8x
rs1858830, located in promoter of the MET gene, has been linked to a 2x increase in the risk of autism based on a study of ~700 families.

From OMIM 164860:

"In case-control analysis, the relative risk for autism was 2.27 for the CC genotype and 1.67 for the GC genotype compared to the GG genotype."

The association of rs1858830 in the MET gene with autism failed to replicate in 325 multiplex families and 10 trios of the International Molecular Genetic Study of Autism Consortium (IMGSAC), although another MET SNP did associate with autism (rs38845)

Disruption of cerebral cortex MET signaling in autism spectrum disorder.

Distinct genetic risk based on association of MET in families with co-occurring autism and gastrointestinal conditions.

Association of genetic variation in the MET proto-oncogene with schizophrenia and general cognitive ability.

Further evidence for the role of MET in autism susceptibility.

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Gene related topics:

schizophrenia(1.00) · cognitive ability(1.00) · PSYCH(0.12) · autism(0.12)

Relevant papers and external links:

PMID 17053076
PMID 19002214
OMIM 611015
OMIM 164860
PMID 17696172
PMID 19255034
PMID 20080979
PMID 20615438

dbSNP: rs1858830 Genecard: MET chromosome 7 SNPedia: rs1858830(C;C)

★ rs3819331(C;T) info:1.80 28.92% **increased risk of autism**

Increased risk of autism.
Examination of tetrahydrobiopterin pathway genes in autism.

Examination of association to autism of common genetic variation in genes related to dopamine.

Gene related topics:

autism(5.00)

Relevant papers and external links:

PMID 19674121
PMID 19360691

dbSNP: rs3819331 Genecard: PTS chromosome 11 SNPedia: rs3819331(C;T)

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★ rs309375(G;G) good:1.80 2.89%(rare) **Smaller mosquito bites**

External Link mosquito bite size

Relevant papers and external links:

dbSNP: rs309375 Genecard: nan chromosome 4 SNPedia: rs309375(G;G)

★ rs2007153(G;G) info:1.74 37.48% **increased risk of schizophrenia in limited study**

Associated with schizophrenia in a study of Croatians. Due to the limited number and diversity of the studied population, more study is needed to confirm any link. The G allele was associated with an increased risk of schizophrenia.

Gene related topics:

schizophrenia(5.00) · PSYCH(2.00) · UNKNOWN(2.00) · dopamine beta hydroxylase activity(2.00) · interpersonal sensitivity paranoid ideation psychoticism(2.00) · perceptual disorders(1.00) ·

NEUROLOGICAL(0.33) · migraine with aura(0.33) · attention deficit hyperactivity disorder(0.21) · CHEMDEPENDENCY(0.16) · smoking behavior(0.16) · adhd attention deficit hyperactivity disorder(0.15) · personality(0.14) · migraine disorders(0.08) · adhd(0.06) · smoking(0.05) · autism(0.03)

Relevant papers and external links:

PMID 19673036

dbSNP: rs2007153 Genecard: DBH chromosome 9 SNPedia: rs2007153(G;G)

★ rs762551(A;A) info:1.68 39.66% **Fast Caffeine Metabolizer.**

Unlike the majority of people, caffeine is broken down faster in your liver, so it has less effect on you. Supposedly this decreases heart attack risk, although other studies show caffeine is generally good for the heart. Caffeine will be less effective at preventing Breast Cancer, Alzheimer's Disease, and Parkinson's disease. Caffeine will not make your breasts smaller.
rs762551, also known as -164A>C or -163C>A, is a SNP encoding the CYP1A2*1F allele of the CYP1A2 gene. For historic reasons, the rs762551(C) allele is considered the wild-type, even though it is the rarer allele in most populations. The rs762551(A) allele is the "fast metabolizer" allele known as CYP1A2*1F; the (C) allele is by comparison a slower metabolizer of certain substrates (including caffeine).

In terms of genotypes, only rs762551(A;A) individuals are considered fast metabolizers. Individuals who are rs762551(A;C) heterozygotes or rs762551(C;C) homozygotes are both considered slow metabolizers.

The CYP1A2 gene encodes a member of the cytochrome p450 family of proteins, which metabolize nutrients and drugs. One well known substrate of CYP1A2 is caffeine; individuals who are carry one or more CYP1A2*1C alleles are "slow" caffeine metabolizers, whereas carriers of the variant CYP1A2*1F are "fast" caffeine metabolizers. The same amount of caffeine will therefore tend to have more stimulating effect on CYP1A2 slow metabolizers than on CYP1A2 fast metabolizers.

A study of healthy premenopausal non-hormone using women concluded that drinkers of 3 or more cups of coffee per day tended to have lower breast volume (smaller breasts), but only if they had at least one rs762551(C) allele (p(interaction)=0.02), which was said to be consistent with reports that coffee protects only C-allele carriers against breast cancer.

Another study by this same group looked at coffee consumption as related to breast cancer. Among 458 such patients (age 25-99 years), rs762551(A;A) women (about 1/2 of the total study) who drank 2 or more cups of coffee per day tended to have a later age at diagnosis compared with low

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coffee consumption (59.8 versus 52.6 years, p = 0.0004). These patients were also more likely to have ER- tumors than patients with low consumption (14.7% versus 0%, p = 0.018). Coffee consumption had no associations in carriers of a rs762551(C) allele.

An independent study of 411 BRCA1 mutation carriers (170 cases and 241 controls) looked at the association between breast cancer, coffee consumption before age 35, and CYP1A2 genotype. While CYP1A2 genotype did not affect breast cancer risk, women with at least one rs762551(C) allele who consumed coffee had a 64% reduction in breast cancer risk, compared with women who never consumed coffee (odds ratio 0.36, CI: 0.18-0.73). No such protective effect was seen in rs762551 (A;A) women.

A study of 2,000 Costa Ricans who survived a first heart attack observed a general trend between coffee consumption and increased risk among carriers of rs762551(C) alleles, i.e. more coffee led to increased nonfatal heart attack risk. No association was seen for rs762551(A;A) individuals.

Gene related topics:

HEMATOLOGICAL(2.00) · UNKNOWN(2.00) · porphyria(2.00) · urinary mutagenicity(2.00) · METABOLIC(1.80) · cholesterol hdl cholesterol ldl(1.80) · cyp1a2 activity(1.80) · cancer(1.00) · breast cancer(1.00) · OTHER(1.00) · PHARMACOGENOMIC(0.36) · dyskinesia drug induced(0.36) · CANCER(0.30) · liver cancer(0.30) · NEUROLOGICAL(0.24) · tardive dyskinesia(0.24) · colorectal cancer(0.22) · CARDIOVASCULAR(0.10) · pulmonary disease chronic obstructive(0.10) · endometrial cancer(0.10) · bladder cancer(0.09) · pancreatic cancer(0.08) · adenoma colorectal neoplasms(0.07) · endometrial neoplasms(0.07) · REPRODUCTION(0.04) · pregnancy loss recurrent(0.04)

Relevant papers and external links:

PMID 18813311

PMID 18398030

PMID 17507615

PMID 16522833

PHARMGKB PA165109602
Evidence: PubMed ID:19636338Risk or phenotype-associated allele: A. Phenotype: CYP1A2*1F/*1F genotype was associated with reduced dose-/body weight-normalized olanzapine serum concentrations compared to homo- and heterozygote carriers of CYP1A2*1A. Study size: 124. Study population/ethnicity: Caucasian; Patients with psychiatric disorders.Type of association: PD.

PHARMGKB PA161145040
Evidence: PubMed ID:10233211; PubMed ID:12445035Very common SNP, high inducibility.

PHARMGKB PA162191314
Evidence: PubMed ID:18496682A study in 105 patients with rheumatoid arthritis found that patients with the CYP1A2*1F CC genotype had a 9.7-fold higher risk for overall leflunomide-induced toxicity than the carriers of CYP1A2*1F A allele.

PHARMGKB PA162263496

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Evidence: PubMed ID:16522833Carriers of the C allele of this SNP(*1F) are slow metabolizers of caffeine. In a study of Costa Rican Hispanic Americans, the risk of nonfatal Myocardial Infarction increased with coffee intake for *1F carriers.

dbSNP: rs762551 Genecard: CYP1A2 chromosome 15 SNPedia: rs762551(A;A)

★ rs6277(T;T) good:1.66 5.95% **normal schizophrenia risk, learns NoGo faster**

According to 23andMe blog this genotype is better for a particular learning task.
rs6277 (957C>T, Pro319Pro) is one of several SNPs in the dopamine receptor DRD2 gene.

In a study of 300+ Russian patients with schizophrenia, the rs6277(C) allele was associated with higher risk. From a pooled meta-study (total of 4 other reports plus this one) the allelic odds ratio is 1.42 (CI: 1.26-1.61, p<0.00005), and the genotypic odds ratio for the (C;C) genotype was 1.6 (CI: 1.32-1.95, p<0.00005).

Note that rs6275 and rs6277 are only 18bp apart, hence their very tight linkage (r2=1).

gs278 reflects a gene-gene interaction between this SNP and a NR3A SNP, rs10989591, in which older adults with a particular combination of genotypes at these two (independent) loci are reported to exhibit better episodic memory.

C allele associated significantly (p = 0.021) with post-traumatic stress disorder in a sample of (assumedly Australian) 127 war veterans w/ 228 controls

23andMe blog people with two As at rs6277(T;T) were better at NoGo learning.

[pharmgkb] T allele associated with decrease in mRNA levels

Gene related topics:

CHEMDEPENDENCY(6.04) · alcoholism(6.04) · alcoholism substance related disorders(4.00) · PSYCH(2.30) · schizophrenia(2.30) · METABOLIC(2.00) · NEUROLOGICAL(2.00) · hyperprolactinaemia hyperprolactinemia(2.00) · nervous system diseases(2.00) · substance withdrawal syndrome(2.00) · substance withdrawal syndrome tobacco use disorder(2.00) · tic combined vocal and multiple motor tourette syndrome(2.00) · smoking behavior(1.74) · UNKNOWN(1.29) · neuroleptic malignant syndrome(1.29) · heroin dependence(1.00) · methamphetamine abuse(1.00) · memory(1.00) · stress(1.00) · OTHER(1.00) · tardive dyskinesia(0.95) · heroin abuse(0.83) · cocaine dependence(0.80) · cocaine related disorders(0.67) · delirium(0.57) · impulsivity(0.57) · alcohol abuse(0.52) · tobacco use disorder(0.44) · adhd(0.40) · mood disorders(0.40) · parkinson s disease(0.40) · smoking(0.33) · alcohol(0.32) · mood disorder(0.27) · substance related disorders(0.27) · CARDIOVASCULAR(0.25) · atrial natriuretic factor(0.25) · alcohol dependence(0.22) · personality traits(0.21) · tourette syndrome(0.21) · dyslexia(0.19) · migraine disorders(0.17) · adhd attention deficit hyperactivity disorder(0.15) · personality(0.14) · personality disorders(0.12) · depressive disorder major(0.10) · bipolar disorder(0.07) · attention deficit hyperactivity disorder(0.05) · autism(0.03)

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Relevant papers and external links:

PMID 18255274

PMID 18833581

PHARMGKB PA165374657
Evidence: PubMed ID:20714340Risk or phenotype-associated allele: CC. Phenotype: The CC genotype was associated with weight gain of at least 7% in schizophrenia patients treatment with clozapine or olanzapine. Study size: 206. Study population/ethnicity: European american; African American; German; Schizophrenia. Significance metric(s): OR = 3.37 (CI = 1.00-11.36). Type of association: PD.

PHARMGKB PA161145172
Evidence: Web Resource:External Link allele led to a decrease in mRNA levels, C/C genotype was reported to be associated with schizophrenia.

PHARMGKB PA165291683
Evidence: PubMed ID:18687376Phenotype: The 957CC carriers were more frequently non-responders to methadone treatment (OR=2.4; p=0.02). The 957CC carriers were 25% (95% CI = 15-36%) in the non-responder group (n = 73) whereas they were only 12% (95% CI = 8-18%) in the responder group (n = 165; $\chi 2 = 5.9$; p = 0.015). No significant difference was observed for the TaqI A genotype frequency in responder and non-responder groups (p = 0.9). Study size: 238 patients. Study population: Caucasian.

PHARMGKB PA165291681
Evidence: PubMed ID:15567074The T allele led to a decrease in mRNA levels and stability whereas the C allele was not found to be associated with any mRNA changes, which led to a relative increase in the expression of DRD2 in carriers of the C allele.

PHARMGKB PA165291682
Evidence: PubMed ID:19582781Results of an in vivo study with [11C]raclopride indicated that this variant increased binding potential by decreasing DRD2 KD (C/C>C/T>T/T), while Bmax was not significantly altered.

PHARMGKB PA165291689
Evidence: PubMed ID:16973280Risk or phenotype-associated allele: C. Phenotype: A greater proportion of patients with schizophrenia than healthy controls were C-allele carriers. Study size: 188 schizophrenia patients and 384 healthy controls. Study population: Finnish. Metric(s): OR =1.5, 95% confidence interval (CI) 1.0-2.3, p=0.05.

PHARMGKB PA165291685
Evidence: PubMed ID:19603545Phenotype: In this case-control study the polymorphisms -141C Ins/Del (rs1799732); C957T (rs6277); A1385G (rs6276); and TaqIA (rs1800497) were genotyped. The haplotypes I-C-G-A2 and I-C-A-A1 occurred with a higher frequency in alcoholics [P=0.026, odds ratio (OR): 1.340; P=0.010, OR: 1.521, respectively]. Study size: 360 alcoholics and 368 controls. Study population:Caucasian individuals of German origin.

PHARMGKB PA165291676

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Evidence: PubMed ID:18926547Phenotype: Patients with C/C genotype were associated with poor aripiprazole response for excitement symptoms (measured on the positive and negative syndrome scale, PANSS - excitement subscale) when compared with T/T patients. Study size: 128 schizophrenic patients. Study population: Chinese.

dbSNP: rs6277 Genecard: DRD2 chromosome 11 SNPedia: rs6277(T;T)

★ rs7171755(A;G) info:1.65 49.86% **very slight decrease in cortical thickness and IQ; see full text**

rs7171755 is a SNP near the synaptic cell adhesion glycoprotein-encoding NPTN gene.

A study of ~1,500 adolescents determined that the rs7171755(A) allele is associated with a slightly thinner left hemisphere cortex and also a slight decrease in verbal and nonverbal intelligence (IQ). The differences were really quite slight; each copy of the risk allele decreased cortical thickness by 0.7%, and decreased verbal and nonverbal IQ by 1.8 and 1.4 points, respectively. This study awaits independent replication. Also, note that eight SNPs in the NPTN locus were reported as jointly showing significant association with cortical thickness in addition to rs7171755: rs7176637, rs11854138, rs8028749, rs12185108, rs1564492, rs899981, rs7178269 and rs4075802.

Gene related topics:

intelligence(1.00)

Relevant papers and external links:

dbSNP: rs7171755 Genecard: REC114 chromosome 15 SNPedia: rs7171755(A;G)

★ rs13266634(C;C) info:1.63 55.47% **increased risk for type-2 diabetes**

rs13266634 is a SNP in the zinc transporter protein member 8 SLC30A8 gene that has primarily been associated with type-2 diabetes in several studies. This SNP is also known as the Arg325Trp or R325W variant; the (C) allele encodes the arginine (R), and the (T) allele encodes the tryptophan (W).

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significantly associated p = 0.0073; in 1,630 Japanese subjects with type-2 diabetes and in 1,064 controls

The major alleles of the SLC30A8 SNP rs13266634 and the HHEX SNP rs7923837 associate with reduced insulin secretion, but not with insulin resistance.

46% of European non-diabetic offspring of type-2 diabetes patients are rs13266634(C;C) homozygotes; they are diabetes-prone and characterised by a 19% decrease in first-phase insulin release following an intravenous glucose load.

Note: this SNP, rs13266634, is not represented on the Affymetrix 5.0 chip. Since it is also in an area of high recombination, it also lacks a proxy on the Affy chip and thus could not have been detected in the large genome-wide type-2 diabetes study performed by the Wellcome Trust Consortium.

1,638 type-2 diabetes patients and 1,858 controls
rs13266634 in SLC30A8 (OR 1.20, 95% CI: 1.09-1.33, p = 3.9 x 10(-4)).

rs13266634(C) allele is associated with younger age of onset of type-1 diabetes

This SNP was confirmed to be associated with type-2 diabetes in a study of 500+ Japanese patients plus pooled meta-analysis with 6 previous association studies (also of Japanese).

spitotoon associated with blood sugar levels (glycated hemoglobin levels) rs13266634(C)

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Established associations:

type ii diabetes mellitus **you have 2 copies of the risk allele C**

(risk allele=C, odds ratio/beta 1.16 [1.10-1.22] with pval 8E-8, pubmedid=19734900; risk allele=C, odds ratio/beta 1.12 [1.07-1.17] with pval 5E-7, pubmedid=23945395; risk allele=C, odds ratio/beta 1.22 [1.16-1.28] with pval 2E-14, pubmedid=19401414; risk allele=C, DGI+FUSION+WTCCC odds ratio/beta 1.12 [1.07-1.16] with pval 5E-8, pubmedid=17463249; risk allele=C, odds ratio/beta 1.15 [1.08-1.22] with pval 3E-6, pubmedid=17460697; risk allele=C, odds ratio/beta 1.18 [0.69-1.67] with pval 6E-8, pubmedid=17293876; risk allele=C, DGI+FUSION+WTCCC odds ratio/beta 1.12 [1.07-1.16] with pval 5E-8, pubmedid=17463246; risk allele=C, DGI+FUSION+WTCCC odds ratio/beta 1.12 [1.07-1.16] with pval 5E-8, pubmedid=17463248)

autoantibody measurement **you have 2 copies of the risk allele C**

(risk allele=C, odds ratio/beta 1.16 [1.10-1.22] with pval 8E-8, pubmedid=19734900)

a1c measurement **you have 0 copies of the risk allele A**

(risk allele=A, odds ratio/beta 0.05 unit decrease with pval 2E-7, pubmedid=24647736; risk allele=A, odds ratio/beta 0.02 % decrease with pval 5E-8, pubmedid=19096518)

Gene related topics:

glucose(1.00) · insulin resistance(1.00) · insulin secretion(1.00) · insulin(1.00) · METABOLIC(0.91) · type 2 diabetes(0.91) · diabetes type 2(0.87) · diabetes mellitus type 2(0.56) · UNKNOWN(0.09) · diabetes type 2 diabetes type 1(0.09) · HEMATOLOGICAL(0.08) · erythrocyte count(0.08) · hemoglobins(0.07) · diabetes mellitus(0.03)

Relevant papers and external links:

PMID 19734900
Genetic variant near irs1 is associated with type 2 diabetes, insulin resistance and hyperinsulinemia.

PMID 23945395
Genome-wide association study identifies three novel loci for type 2 diabetes.

PMID 19401414
Confirmation of multiple risk loci and genetic impacts by a genome-wide association study of type 2 diabetes in the japanese population.

PMID 17463249
Replication of genome-wide association signals in uk samples reveals risk loci for type 2 diabetes.

PMID 17460697
A variant in cdkal1 influences insulin response and risk of type 2 diabetes.

PMID 17293876
A genome-wide association study identifies novel risk loci for type 2 diabetes.

PMID 17463246
Genome-wide association analysis identifies loci for type 2 diabetes and triglyceride levels.

PMID 17463248
A genome-wide association study of type 2 diabetes in finns detects multiple susceptibility variants.

PMID 24647736
Multiple nonglycemic genomic loci are newly associated with blood level of glycated hemoglobin in east asians.

PMID 19096518
Novel association of hk1 with glycated hemoglobin in a non-diabetic population: a genome-wide evaluation of 14,618 participants in the women's genome health study.

PMID 18162508

PMID 17786204

PMID 18324385

PMID 17554300

PMID 18437351

PMID 18548167

PMID 19033397

OMIM 611145

PHARMGKB PA164740300
Evidence: PubMed ID:19056611; Web Resource:External Link results: Adiposity-related heterogeneity in patterns of type 2 diabetes susceptibility observed in genome wide association data. (Initial Sample Size: 1,924 cases, 2,938 controls; Replication Sample Size: 3,757 cases, 5,346 controls); (Region:

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8q24.11; Reported Gene(s): SLC30A8; Risk Allele: rs13266634-?); (p-value= 0.000007).This variant is associated with Type 2 diabetes.

PHARMGKB PA161749011
Evidence: PubMed ID:17786204; PubMed ID:18162508; PubMed ID:18264689; PubMed ID:18324385This variant has been reported to be significantly associated with type 2 diabetes.

PHARMGKB PA162191339
Evidence: PubMed ID:17463249rs13266634 is associated with susceptibility to Type 2 Diabetes. The association has been noted in two case-control studies of UK subjects as well as in two other large case-control studies.

PHARMGKB PA162191360
Evidence: PubMed ID:17463248In a large Finnish case-control GWAS, rs13266634 was found to be associated with susceptibility to Type 2 Diabetes.

PHARMGKB PA162356600
Evidence: PubMed ID:17293876; Web Resource:External Link Results: A genome-wide association study identifies novel risk loci for type 2 diabetes (Initial Sample Size: 1,380 cases, 1,323 controls; Replication Sample Size: 2,617 cases, 2,894 controls; Risk Allele: rs13266634-C).

OMIM 611145

dbSNP: rs13266634 GWAS Catalog: rs13266634 Genecard: SLC30A8 chromosome 8

SNPedia: rs13266634(C;C)

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★ rs4343(A;A) info:1.61 41.37% **usually the ACE Alu insertion; better endurance vs strength;**

A and G alleles of rs4343 are strongly associated marking insertion and deletion alleles of ACE I/D respectively. For those looking for the ACE Alu Insertion/Deletion, this is a very good substitute for people searching for ACE Del16. G would mean deletion.

A haplotype of rs4311, rs4343, rs699 increases risk of diabetic nephropathy 4x.

Established associations:

angiotensin converting enzyme activity measurement **you have 0 copies of the risk allele G**

(risk allele=G, odds ratio/beta 16.2 % variance with pval 3E-25, pubmedid=20066004)

blood metabolite measurement **you have 2 copies of the risk allele A**

(risk allele=A, aspartylphenylalanine/X-14450--phenylalanylleucine odds ratio/beta 0.059 [0.049-0.069] unit decrease with pval 1E-37, pubmedid=24816252)

serum metabolite measurement **you have 2 copies of the risk allele A**

(risk allele=A, [H]HWESASLLR[OH] odds ratio/beta 0.35 unit increase with pval 1E-18, pubmedid=24625756; risk allele=G, Threonylphenylalanine odds ratio/beta 0.21 unit increase with pval 8E-14, pubmedid=24625756; risk allele=G, Aspartylphenylalanine odds ratio/beta 0.22 unit increase with pval 9E-25, pubmedid=24625756)

Gene related topics:

CARDIOVASCULAR(22.41) · hypertension(22.41) · hypertrophy left ventricular left ventricular hypertrophy(9.33) · left ventricular hypertrophy(4.17) · coronary disease coronary heart disease myocardial infarction(4.00) · coronary restenosis(4.00) · UNKNOWN(3.00) · altitude sickness pulmonary edema(3.00) · heart failure hypertension(3.00) · cardiovascular(2.88) · diabetic nephropathies diabetic nephropathy(2.67) · METABOLIC(2.63) · diabetes type 2(2.63) · apoplexy stroke(2.46) · OTHER(2.27) · altitude sickness(2.27) · chronic renal failure glomerulonephritis iga iga glomerulonephritides kidney failure chronic(2.27) · IMMUNE(2.00) · RENAL(2.00) · adult respiratory distress syndrome respiratory distress syndrome adult(2.00) · albuminuria hypertension(2.00) · aortic valve stenosis hypertrophy left ventricular left ventricular hypertrophy(2.00) · calcinosis coronary artery disease(2.00) · cardiomyopathy hypertrophic hypertrophic cardiomyopathy hypertrophy left ventricular left ventricular hypertrophy(2.00) · chronic renal failure hypertension kidney failure chronic polycystic kidney autosomal dominant(2.00) · coronary disease coronary heart disease dilatation pathologic(2.00) · coronary disease coronary heart disease hypertension(2.00) · coronary disease coronary heart disease recurrence(2.00) · diabetes mellitus type 1 hypoglycemia(2.00) · elite athletes(2.00) · fetal growth retardation intrauterine growth retardation(2.00) · glomerulonephritis(2.00) · heart failure systolic(2.00) · hypertension kidney failure kidney failure(2.00) · hypertension myocardial infarction(2.00) · hypertension myocardial ischemia(2.00) · insulin sensitivity(2.00) · left ventricular mass(2.00) · myocardial infarction ventricular dysfunction left(2.00) · myocardial infarction ventricular remodeling(2.00) · proteinuria(2.00) · vesico ureteral reflux vesicoureteral reflux(2.00) · myocardial infarction(1.98) · diabetic nephropathy(1.89) · REPRODUCTION(1.87) · pre eclampsia(1.87) · glycogen storage disease type v(1.80) · pneumonia(1.80) · polycystic kidney disease(1.60) · coronary disease coronary heart disease(1.54) · coronary atherosclerosis(1.50) · hypertension hypertrophy left ventricular left ventricular hypertrophy(1.47) · chronic renal failure kidney failure chronic(1.37) · aortic aneurysm abdominal(1.33) · cerebral infarction(1.33) · glomerulonephritis iga iga glomerulonephritides(1.33) · peripheral vascular diseases(1.32) · hypertension left ventricular hypertrophy(1.29) · sarcoidosis pulmonary(1.29) · erectile dysfunction(1.14) · diabetes mellitus type ii diabetes mellitus type 2 diabetic nephropathies diabetic nephropathy(1.12) · lupus(1.04) · NEUROLOGICAL(1.00) · alzheimer s disease vascular dementia(1.00) · cardiomyopathies(1.00) · idiopathic dilated cardiomyopathy(1.00) · iga nephropathy(1.00) · renal insufficiency(1.00) · alzheimer s disease(0.96) · myocardial ischemia(0.84) · NORMALVARIATION(0.82) · endurance performance(0.82) · nephropathy(0.82) · apoplexy brain ischemia stroke(0.81) · heart failure(0.77) · atherosclerosis(0.67) · PSYCH(0.67) · cerebral infarct(0.67) · dementia vascular(0.67) · heart disease(0.67) · muscle testing(0.67) · vesicoureteral reflux(0.67) · essential hypertension(0.64) · atrial fibrillation(0.60) · cardiovascular diseases(0.58) · heart failure diastolic(0.57) · insulin resistance polycystic ovarian syndrome polycystic ovary syndrome(0.57) · kidney failure chronic(0.56) · blood pressure arterial(0.54) · type 2 diabetes(0.52) · nephrotic syndrome(0.52) · retinopathy diabetic(0.52) · coronary artery disease(0.51) · arterial stiffness(0.50) · kidney disease(0.50) · sarcoidosis(0.47) · coronary artery disease myocardial infarction(0.46) · glomerulonephritis iga(0.45) · nephropathy diabetic(0.45) · AGING(0.44) · aging(0.44) · brain ischemia(0.44) · VISION(0.40) · diabetes mellitus type 2 diabetic nephropathies(0.40) · glaucoma glaucoma primary open angle(0.40) · myotonic dystrophy(0.36) · coronary heart disease(0.36) · hypertension pregnancy induced(0.33) · intima media thickness(0.33) · migraine with aura(0.33) · syncope vasovagal(0.33) · insulin resistance(0.32) · kidney diseases(0.31) · brain ischemia stroke(0.31) · obesity overweight(0.31) · migraine disorders(0.30) · systemic sclerosis(0.29) · chronic obstructive pulmonary disease(0.28) · bronchopulmonary dysplasia(0.27) · pregnancy complications(0.27) · restenosis(0.26) · metabolic syndrome(0.26) · bmi(0.25) · cardiovascular disease(0.25) · cardiomyopathy hypertrophic hypertrophic cardiomyopathy(0.24) · nephropathy in other diseases(0.19) · abortion habitual(0.19) · pulmonary disease chronic obstructive(0.18) · atherosclerosis coronary(0.18) · stroke(0.16) · asthma(0.15) · INFECTION(0.14) · copd(0.14) · sepsis systemic infection(0.14) · stroke ischemic(0.11) · migraine(0.11) · thrombosis(0.11) · vitiligo(0.10) · preeclampsia(0.10) · myocardial infarct(0.10) · weight gain(0.10) · diabetes mellitus type 2(0.09) · systemic lupus erythematosus(0.09) · dementia(0.09) · behcet s disease(0.08) · lupus erythematosus systemic systemic lupus erythematosus(0.07) · cardiomyopathy(0.06) · arthritis(0.06) · diabetes mellitus insulin dependent

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diabetes mellitus type 1(0.05) · atopy(0.05) · longevity(0.04) · hypercholesterolemia(0.04) · suicide(0.04) · dna damage(0.03) · coronary disease(0.02) · CANCER(0.02) · stomach neoplasms(0.02) · endometriosis(0.02)

Relevant papers and external links:

PMID 20066004
A genome-wide association study identifies new loci for ace activity: potential implications for response to ace inhibitor.

PMID 24816252
An atlas of genetic influences on human blood metabolites.

PMID 24625756
Genetic determinants influencing human serum metabolome among african americans.

PMID 19108684

PHARMGKB PA165107172
Evidence: PubMed ID:18057531While the ACE:I/D has dbSNP identifiers rs1799752, rs4340, rs13447447 and rs4646994, measurement of a single base at these positions does not give an informative genotype. In Europeans, rs4343 is in complete LD with the ACE:I/D, with the A and G alleles of rs4343 marking the insertion and deletion alleles of ACE:I/D respectively.

PHARMGKB PA161795939
Evidence: PubMed ID:16642441In an inbred Israeli Arab community, rs4343 was shown to be significantly associated with risk of developing Alzheimer Disease, especially when analyzed in a combined haplotype with rs4351.

dbSNP: rs4343 GWAS Catalog: rs4343 Genecard: ACE chromosome 17

SNPedia: rs4343(A;A)

★ rs909525(A;-) good:1.59 32.29% **Probably MAOA 4 or 5 repeats: not Warrior Gene.**

People with this SNP usually also have either the 4 or 5 repeat (non-Warrior) version of MAOA which makes them less aggressive and less anti-social. If you are male, you inherited this from your mother. Do not confuse this with the completely different warrior/worrier gene.
This is the best proxy for the number of repeats of the MAOA warrior gene. In 105 samples (69 males, 36 females) from the Stanley foundation brain collection, it was always A in people with the 4 or 5 repeat non-Warrior version and always G in people with the 3 repeat Warrior version. The combination rs909525(A) AND rs6323(G) AND rs3027399(G) indicate specifically that it is the 5 repeat version. The 2 repeat version and the 3.5 repeat version weren't mentioned.

mentioned as potentially affecting white matter volume, sample size tiny

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Gene related topics:

PSYCH(3.00) · antisocial personality disorder(3.00) · bp major depressive(2.00) · personality disorders(1.12) · aggressive behavior(1.00) · brain(1.00) · mood disorder(0.60) · UNKNOWN(0.56) · sudden infant death(0.56) · CHEMDEPENDENCY(0.51) · smoking behavior(0.51) · personality traits(0.47) · suicide(0.32) · panic disorder(0.31) · depression(0.30) · personality(0.25) · major depressive disorder(0.24) · alcoholism(0.24) · bipolar disorder(0.24) · adhd attention deficit hyperactivity disorder(0.21) · attention deficit hyperactivity disorder(0.21) · smoking(0.21) · alcohol abuse(0.17) · adhd(0.15) · NEUROLOGICAL(0.13) · restless legs syndrome(0.13) · autism(0.12) · parkinson s disease(0.12) · migraine(0.11) · depressive disorder major(0.10) · mood disorders(0.10) · obsessive compulsive disorder(0.06) · dementia(0.04)

Relevant papers and external links:

PMID 16893905

PMID 19028548

dbSNP: rs909525 Genecard: MAOA chromosome X SNPedia: rs909525(A;A)

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★ rs6265(G;G) good:1.55 63.80% **common/normal**

Val/val (normal) variant of the Val66Met snp in the BDNF gene that codes for brain-derived neurotrophic factor. Met/met is associated with susceptibility to neuropsychiatric disorders. rs6265, also known as Val66Met, is a SNP in brain-derived neurotrophic factor BDNF gene. The more common G allele encodes the Val, while the A allele encodes Met.

Met/A allele associated with introversion

The A allele may also be protective against depression when subjected to repeated defeat g2b2mh blog

On a driving-based motor learning task subjects with this genotype showed greater error during short-term learning and poorer retention over 4 days, relative to subjects without the polymorphism. The presence of this BDNF polymorphism is associated with differences in brain motor system function, altered short-term plasticity, and greater error in short-term motor learning.

Wired discusses the research as does 23andMe blog.

Alzheimer's disease risk for non ApoE4 carriers is affected by the heterozygous form of rs6265, as well as the diplotypes of rs6265, rs11030104, and rs2049045.

Depressed patients who had (GA or AA) appeared to show a significantly increased risk of suicidal behaviour

In 2 studies of British women, rs6265(A;A) women had a lower body mass index than rs6265(G) allele carriers, with the difference being -0.57-0.911 kg/m(2), depending on the study.

Established associations:

smoking behavior **you have 2 copies of the risk allele G**

(risk allele=G, smoking initiation odds ratio/beta 1.06 [1.04-1.08] with pval 2E-8, pubmedid=20418890)

body mass index **you have 2 copies of the risk allele G**

(risk allele=G, odds ratio/beta 4.58 [3.07-6.09] % SD with pval 5E-10, pubmedid=19079260)

body weight **you have 2 copies of the risk allele G**

(risk allele=G, odds ratio/beta 4 [2.47-5.53] % SD with pval 2E-7, pubmedid=19079260)

Gene related topics:

PSYCH(2.33) · depression(2.33) · NEUROLOGICAL(2.00) · affective psychoses(2.00) · alzheimer disease amnesia(2.00) · bipolar disorder(1.66) · CHEMDEPENDENCY(1.00) · amphetamine related disorders(1.00) · brain(1.00) · body mass(1.00) · body mass index(1.00) · schizophrenia(0.94) · major depressive disorder(0.68) · alzheimer s disease(0.59) · METABOLIC(0.56) · weight(0.56) · personality(0.55) · obsessive compulsive disorder(0.40) · UNKNOWN(0.36) · dyskinesia drug induced(0.36) · cognitive function(0.33) · parkinsons disease(0.29) · DEVELOPMENTAL(0.27) · IMMUNE(0.27) · dermatitis atopic(0.27) · mental retardation(0.27) · huntington s disease(0.26) · adhd attention deficit hyperactivity disorder(0.21) · anorexia nervosa(0.15) · autism(0.12) · body weight obesity(0.11) · cognitive ability(0.11) · panic disorder(0.08) · migraine disorders(0.08) · attention deficit hyperactivity disorder(0.05) · personality traits(0.05) · smoking(0.05) · epilepsy(0.04) · smoking behavior(0.04) · suicide(0.04)

Relevant papers and external links:

PMID 19079260
Genome-wide association yields new sequence variants at seven loci that associate with measures of obesity.

PMID 20418890
Genome-wide meta-analyses identify multiple loci associated with smoking behavior.

PMID 20042999

PMID 17956738

PMID 19745020

PMID 17293537

PMID 18600033

PMID 19209189

OMIM 113505

PHARMGKB PA164857051

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Evidence: PubMed ID:19414708This nonsynonymous variant (G/A) in an exon in BDNF gene was associated with major depressive disorder in a study of 264 controls and 272 Mexican Americans with major depressive disorder. (OR= 1.66; p value=0.009; risk allele=G)

PHARMGKB PA162364070
Evidence: PubMed ID:18319075In DNA from frontal-cortex brain tissue of a group of Schizophrenia, Bipolar Disorder and Control individuals, this SNP was found to be associated with methylation at CpG sites surrounding the gene. CC genotype was associated with a higher level of methylation.

PHARMGKB PA164740154
Evidence: PubMed ID:19079260; Web Resource:External Link results: Genome-wide association yields new sequence variants at seven loci that associate with measures of obesity. (Initial Sample Size: 80,969 individuals; Replication Sample Size: 11,036 individuals); (Region: 11p14.1; Reported Gene(s): BDNF; Risk Allele: rs6265-G); (p-value= 0.0000000005).This variant is associated with Body mass index.

PHARMGKB PA164740174
Evidence: PubMed ID:19079260; Web Resource:External Link results: Genome-wide association yields new sequence variants at seven loci that associate with measures of obesity. (Initial Sample Size: 80,969 individuals; Replication Sample Size: 11,036 individuals); (Region: 11p14.1; Reported Gene(s): BDNF; Risk Allele: rs6265-G); (p-value= 0.0000002).This variant is associated with Weight.

OMIM 113505

dbSNP: rs6265 GWAS Catalog: rs6265 Genecard: BDNF chromosome 11

SNPedia: rs6265(G;G)

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★ rs2201841(C;T) info:1.55 47.81% **1.5x increased risk for Crohn's disease; 2x increased risk for Graves' disease**

SNP rs2201841, in the IL23R gene, is associated with increased risk for Crohn's disease in both Jewish and non-Jewish populations.

Another study found that the "A allele" and "AA genotype" were significantly overrepresented in Graves' disease patients with Graves ophthalmopathy, based on 216 North American patients. The odds ratios reported were 2.04 for the allele (p=1x10-4), and for the so-called "AA" genotype (presumably rs2201841(T;T) when correctly oriented to the dbSNP orientation), 2.4 (p=1x10-4).

23andMe blog psoriasis
Europeans
rs2201841(G) 1.13x risk

Established associations:

ulcerative colitis **you have 1 copies of the risk allele T**

(risk allele=T, odds ratio/beta 1.27 with pval 1E-13, pubmedid=20228799)

psoriasis **you have 1 copies of the risk allele C**

(risk allele=C, odds ratio/beta 1.13 with pval 3E-8, pubmedid=19169254)

Gene related topics:

IMMUNE(5.16) · inflammatory bowel diseases(5.16) · colitis ulcerative crohn disease(1.98) · arthritis psoriatic psoriasis(1.12) · crohn disease crohn s disease inflammatory bowel disease nos inflammatory bowel diseases(1.00) · graves ophthalmopathy(1.00) · crohn disease(0.96) · crohn s disease(0.64) · inflammatory bowel disease(0.54) · spondylitis ankylosing(0.44) · inflammatory bowel disease nos inflammatory bowel diseases(0.43) · colitis ulcerative(0.42) · chronic ulcerative colitis colitis ulcerative crohn disease crohn s disease inflammatory bowel disease nos inflammatory bowel diseases(0.40) · ulcerative colitis(0.24) · crohn disease crohn s disease(0.23) · psoriasis(0.20) · behcet syndrome(0.17) · crohn s disease ulcerative colitis(0.15) · ankylosing spondylitis(0.08) · lupus erythematosus systemic systemic lupus erythematosus(0.03)

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Relevant papers and external links:

PMID 20228799
Genome-wide association identifies multiple ulcerative colitis susceptibility loci.

PMID 19169254
Genome-wide scan reveals association of psoriasis with il-23 and nf-kappab pathways.

PMID 17068223

PMID 18073300

OMIM 605606

PHARMGKB PA164740040
Evidence: PubMed ID:19169254; Web Resource:External Link results: Genome-wide scan reveals association of psoriasis with IL-23 and NF-kappaB pathways. (Initial Sample Size: 1,359 cases, 1,400 controls; Replication Sample Size: 5,048 cases, 5,041 controls); (Region: 1p31.3; Reported Gene(s): IL23R; Risk Allele: rs2201841-G); (p-value= 0.00000003).This variant is associated with Psoriasis.

PHARMGKB PA162356491
Evidence: PubMed ID:17804789; Web Resource:External Link results: Genome-wide association study for Crohn's disease in the Quebec Founder Population identifies multiple validated disease loci (Initial Sample Size: 382 trios; Replication Sample Size: 521 trios, 750 cases, 828 controls). This variant is associated with Crohn's disease. This variant is associated with Crohn's disease.

dbSNP: rs2201841 GWAS Catalog: rs2201841 Genecard: IL23R chromosome 1

SNPedia: rs2201841(C;T)

★ rs7221412(A;G) info:1.52 42.83% **Intermediate riser. Wakes up at a time between the AA and GG genotypes.**

Research also suggests, then you are also more likely to die on a morning, shortly before 11am.

External Link

A common polymorphism near PER1 and the timing of human behavioral rhythms. Study showed that it had influence on wake-up times, as well as time of day that death occurs. Suggests that common (but unidentified) polymorphisms may be associated with a preference for daytime or nighttime activity.

rs7221412 appears to sit in a haplotype block and is flanked by rs9914077 and rs2585408

Relevant papers and external links:

PMID 23034908

dbSNP: rs7221412 Genecard: nan chromosome 17 SNPedia: rs7221412(A;G)

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★ rs3087243(A;A) info:1.51 13.62% **increased risk for auto-immune diseases**

The rs3087243 SNP is also known in the literature as the CT60 G>A or the +6230G>A polymorphism, and it is located in the CTLA4 gene.

In Asian (Japanese) populations, the presence of an rs3087243(G) allele represents a 1.3 fold increased risk of autoimmune thyroid disease, and for those with autoimmune thyroid disease, a 1.5 fold increased risk of type-1 diabetes. However, in individuals without autoimmune thyroid disease, no association was seen between this SNP and type-1 diabetes. The authors speculate that earlier studies may have reported associations between this SNP and type-1 diabetes that were actually primarily based on the association with autoimmune thyroid disease.

This same SNP, rs3087243, has also been implicated as a (minor) risk factor for developing rheumatoid arthritis (RA). A study of 2,000+ European RA patients led to a calculated odds ratio of 1.13 (CI: 1.03 - 1.24) for the rs3087243(G) risk allele.

In a study of 395 Spanish patients with lupus, rs3087243(G) allele carriers were calculated to have an odds ratio of 1.71 (CI: 1.18-2.49, p=0.003, p(corr) = 0.006).

In a different study involving recipients of liver transplants, although also with Spanish patients, the rs3087243(G) allele was significantly associated with acute rejection (odds ratio 1.49, p(corr)=0.038). Patients who lacked this allele had the lowest risk of acute rejection development. Allograft survival data did not show statistical differences between genotypes.

rs2292399 and rs2903692 both significantly associated with type 1 diabetes odds ratio 1.37 and 1.28. A joint analysis revealed that rs3087243, rs2292399, and rs2903692, but not INS rs689, were significant risk factors for the cooccurrence of AITD

Established associations:

rheumatoid arthritis **you have 0 copies of the risk allele G**

(risk allele=G, EA odds ratio/beta 1.15 [1.12-1.18] with pval 4E-22, pubmedid=24390342; risk allele=G, odds ratio/beta 1.14 [1.11-1.17] with pval 3E-25, pubmedid=24390342; risk allele=G, odds ratio/beta 1.15 [1.10-1.20] with pval 1E-8, pubmedid=20453842)

autoantibody measurement **you have 2 copies of the risk allele A**

(risk allele=A, T1D odds ratio/beta 1.2 with pval 2E-17, pubmedid=21829393)

type i diabetes mellitus **you have 2 copies of the risk allele A**

(risk allele=A complex/no impact summary; risk allele=A, T1D odds ratio/beta 1.2 with pval 2E-17, pubmedid=21829393)

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Gene related topics:

IMMUNE(7.75) · graves disease(7.75) · diabetes type 1(5.56) · UNKNOWN(3.00) · wegener s granulomatosis(3.00) · autoimmune thyroid disease(2.67) · OTHER(2.00) · asthma atopy specific ige total serum ige(2.00) · autoimmune hypothyroidism(2.00) · diabetes type 1 thyroid disease autoimmune(2.00) · grave s disease(2.00) · graves disease graves ophthalmopathy(2.00) · oral submucous fibrosis(2.00) · thyroiditis hashimoto s(2.00) · VISION(1.60) · graves ophthalmopathy(1.60) · addison s disease(1.29) · survival(1.00) · sclerosis systemic(0.96) · multiple sclerosis(0.92) · rheumatoid arthritis(0.89) · ige levels(0.80) · cirrhosis biliary primary(0.76) · METABOLIC(0.67) · biliary cirrhosis liver cirrhosis biliary(0.67) · systemic lupus erythematosus(0.59) · lupus erythematosus(0.54) · chronic ulcerative colitis colitis ulcerative(0.47) · myasthenia gravis(0.44) · liver transplant(0.40) · diabetes mellitus type 1(0.27) · celiac disease(0.27) · HEMATOLOGICAL(0.25) · hemophilia a(0.25) · arthritis rheumatoid(0.23) · arthritis rheumatoid rheumatoid arthritis(0.23) · diabetes mellitus insulin dependent diabetes mellitus type 1(0.21) · lupus erythematosus systemic systemic lupus erythematosus(0.19) · RENAL(0.18) · kidney transplant(0.18) · lupus(0.17) · liver cirrhosis biliary(0.15) · ulcerative colitis(0.15) · crohn s disease ulcerative colitis(0.15) · CANCER(0.14) · lymphoma(0.14) · pemphigus(0.14) · asthma(0.13) · arthritis(0.12) · sjogren s syndrome(0.12) · graves disease graves disease(0.11) · behcet syndrome(0.09) · ankylosing spondylitis(0.08) · behcet s disease(0.08) · type 1 diabetes(0.06) · cervical cancer(0.05) · graft versus host disease(0.05) · colitis ulcerative(0.05) · multiple myeloma(0.05) · vitiligo(0.05) · lymphoma non hodgkin(0.05) · INFECTION(0.04) · hepatitis b chronic(0.04) · REPRODUCTION(0.04) · pregnancy loss recurrent(0.04) · hepatitis c(0.04) · sarcoidosis(0.04) · melanoma(0.03)

Relevant papers and external links:

PMID 24390342
Genetics of rheumatoid arthritis contributes to biology and drug discovery.

PMID 20453842
Genome-wide association study meta-analysis identifies seven new rheumatoid arthritis risk loci.

PMID 18978792
Meta-analysis of genome-wide association study data identifies additional type 1 diabetes risk loci.

PMID 21829393
Genome-wide association analysis of autoantibody positivity in type 1 diabetes cases.

PMID 16352685

PMID 16380915

PMID 15248219

PMID 18047932

PMID 18940880

OMIM 123890

PHARMGKB PA164740759
Evidence: PubMed ID:18978792; Web Resource:External Link results: Meta-analysis of genome-wide association study data identifies additional type 1 diabetes risk loci. (Initial Sample Size: 3,561 cases, 4,646 controls; Replication Sample Size: 6,225 cases, 6,946 controls, 3,064 trios); (Region: 2q33.2; Reported Gene(s): CTLA4; Risk Allele: rs3087243-A); (p-value= 0.00000000008).This variant is associated with Type 1 diabetes.

OMIM 123890

dbSNP: rs3087243 GWAS Catalog: rs3087243 Genecard: CTLA4 chromosome 2

SNPedia: rs3087243(A;A)

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★ rs140701(A;G) info:1.48 50.00% **Increased risk for anxiety disorders**

A study of patients with panic disorder or social anxiety disorder, both moderately heritable anxiety disorders, concluded that the rs140701(A) allele was associated with increased risk for both disorders. This SNP was actually part of a tightly linked haplotype A-A-G (for SNPs rs3794808, rs140701 and rs4583306, respectively) that had 1.7x increased risk for panic disorder (CI: 1.2-2.3).

Gene related topics:

PSYCH(24.57) · depression(24.57) · suicide(6.88) · obsessive compulsive disorder(5.82) · IMMUNE(5.28) · irritable bowel syndrome(5.28) · personality traits(4.75) · anxiety disorder(4.50) · REPRODUCTION(4.00) · depressive disorder major bipolar disorder(4.00) · sexual dysfunction

physiological(4.00) · personality disorders(3.78) · depressive disorder major(3.72) · affective disorder(3.12) · anxiety related traits(3.00) · decision making(3.00) · psychoses(3.00) · personality(2.60) · panic disorder(2.19) · autism(2.13) · mood disorders(2.02) · CHEMDEPENDENCY(2.00) · OTHER(2.00) · PHARMACOGENOMIC(2.00) · UNKNOWN(2.00) · alcohol abuse smoking behavior(2.00) · alcoholism attention deficit hyperactivity disorder(2.00) · alcoholism recurrence(2.00) · antidepressant adverse effects(2.00) · anxiety(2.00) · auditory evoked potential(2.00) · bipolar and unipolar disorder(2.00) · eating disorder(2.00) · hallucinations(2.00) · harm avoidance(2.00) · impulse control disorder(2.00) · manic depressive illness(2.00) · personality traits illegal drug use(2.00) · suicidal behavior(1.92) · alcoholism(1.92) · NEUROLOGICAL(1.80) · cocaine dependence(1.80) · major depression(1.80) · migraine migraine with aura(1.80) · neuroticism(1.80) · major depressive disorder(1.73) · migraine(1.73) · substance related disorders(1.67) · bulimia(1.47) · attention deficit hyperactivity disorder(1.16) · adhd attention deficit hyperactivity disorder(1.00) · aggressive behavior(1.00) · alcohol related disorders(1.00) · bipolar disorder depression(1.00) · conduct disorder(1.00) · eating disorders(1.00) · sudden infant death(1.00) · anorexia nervosa(0.93) · CARDIOVASCULAR(0.80) · hypertension pulmonary(0.80) · major psychoses(0.80) · bipolar disorder(0.79) · schizophrenia(0.69) · alcohol abuse(0.68) · bipolar affective disorder(0.67) · fibromyalgia(0.67) · sids sudden infant death syndrome(0.67) · smoking behavior(0.66) · mood disorder(0.60) · psychosis(0.60) · heroin abuse(0.53) · epilepsy temporal lobe(0.43) · sleep disorders(0.36) · cognitive function(0.33) · migraine with aura(0.33) · syncope vasovagal(0.33) · DEVELOPMENTAL(0.27) · mental retardation(0.27) · adhd(0.26) · METABOLIC(0.23) · weight gain(0.23) · smoking(0.21) · tourette syndrome(0.21) · alcohol(0.21) · migraine disorders(0.17) · cognitive ability(0.11) · chronic obstructive pulmonary disease(0.07) · hearing loss(0.07) · AGING(0.04) · longevity(0.04)

Relevant papers and external links:

PMID 18663369

dbSNP: rs140701 Genecard: SLC6A4 chromosome 17 SNPedia: rs140701(A;G)

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★ **rs2538976(A;G)** **info:1.46** **49.80%** **None**

Speech development rs4431523, rs17236239 and significant associations (with P values from 0.01 to 5.0x10–5) between nonsense-word repetition and nine intronic SNPs (rs851715, rs10246256, rs2710102, rs759178, rs1922892, rs2538991, rs17236239, rs2538976, and rs2710117)

Gene related topics:

PSYCH(0.67) · mental disorders(0.67) · OTHER(0.12) · breath tests(0.12) · autism(0.09) · CARDIOVASCULAR(0.03) · echocardiography(0.03) · METABOLIC(0.03) · diabetes mellitus(0.03)

Relevant papers and external links:

PMID 18987363

dbSNP: rs2538976 Genecard: CNTNAP2 chromosome 7 SNPedia: rs2538976(A;G)

★ rs10889677(A;C) info:1.46 47.69% **1.5x increased risk for certain autoimmune diseases; 2x increased risk for Graves disease**

SNP rs10889677, in the IL23R gene, is associated with increased risk for Crohn's disease in both Jewish and non-Jewish populations.

The same risk allele for this SNP has been associated with increased risk for ankylosing spondylitis in a large study of over 1,000 Caucasian patients. The odds ratio is 1.3 (p=1.3x10e-6).[PMID 17952073, PMID 18037607]

In a study of 216 North American patients with Graves' disease, the C allele of rs10889677 was 2.03x overrepresented (p=1.3x10â€“4), and the homozygous rs10889677(C;C) genotype was also overrepresented (2.36x; p=1.4x10-4) in Graves ophthalmopathy patients.

A genome-wide association study using DNA samples from 1,052 individuals with ulcerative colitis and preexisting data from 2,571 controls, all of European ancestry, concluded that this SNP and several others were associated with altered risk for the disease.

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Established associations:

ulcerative colitis **you have 1 copies of the risk allele A**

(risk allele=A, odds ratio/beta 1.29 with pval 1E-8, pubmedid=19122664)

Gene related topics:

IMMUNE(5.16) · inflammatory bowel diseases(5.16) · autoimmune diseases(5.00) · graves disease(5.00) · colitis ulcerative crohn disease(1.98) · arthritis psoriatic psoriasis(1.12) · crohn disease crohn s disease inflammatory bowel disease nos inflammatory bowel diseases(1.00) · graves ophthalmopathy(1.00) · crohn disease(0.96) · crohn s disease(0.64) · inflammatory bowel disease(0.54) · spondylitis ankylosing(0.44) · inflammatory bowel disease nos inflammatory bowel diseases(0.43) · colitis ulcerative(0.42) · chronic ulcerative colitis colitis ulcerative crohn disease crohn s disease inflammatory bowel disease nos inflammatory bowel diseases(0.40) · ulcerative colitis(0.24) · crohn disease crohn s disease(0.23) · psoriasis(0.20) · behcet syndrome(0.17) · crohn s disease ulcerative colitis(0.15) · ankylosing spondylitis(0.08) · lupus erythematosus systemic systemic lupus erythematosus(0.03)

Relevant papers and external links:

PMID 17068223

PMID 18073300

PMID 19122664

PHARMGKB PA164740079

Evidence: PubMed ID:19122664; Web Resource:External Link results: Ulcerative colitis-risk loci on chromosomes 1p36 and 12q15 found by genome-wide association study. (Initial Sample Size: 1,022 cases, 2,503 controls; Replication Sample Size: 1,387 cases, 1,115 controls); (Region: 1p31.3; Reported Gene(s): IL23R; Risk Allele: rs10889677-A); (p-value= 0.00000001).This variant is associated with Ulcerative colitis.

dbSNP: rs10889677 GWAS Catalog: rs10889677 Genecard: IL23R chromosome 1

SNPedia: rs10889677(A;C)

★ rs763361(C;T) info:1.45 49.81% **Slightly increased risk for multiple autoimmune diseases, such as type-1 diabetes**

Higher risk of type-1 diabetes.

rs763361, also known as Gly307Ser, is a SNP in the immune response CD226 gene. The rs763361(T) allele (in dbSNP orientation) encodes the Ser.

Based on multiple studies (by one group) now totaling over 2,000 patients, the rs763361(T) allele is associated with increased risk for multiple autoimmune diseases, including type-1 diabetes (p = 3 x 10e-9), multiple sclerosis (p = 4 x 10e-4), and possibly rheumatoid arthritis (p = 0.017). Based on tag SNP analysis, the authors conclude that this SNP could be the causal variant.

Tested in a large multiple sclerosis data set consisting of 2369 trio families, 5737 cases and 10 296 unrelated controls, SNP rs763361 was associated with disease risk (p = 5.4 x 10e-8)

Established associations:

autoantibody measurement **you have 1 copies of the risk allele C**

(risk allele=C, T1D odds ratio/beta 1.12 with pval 1E-9, pubmedid=21829393)

type i diabetes mellitus **you have 0 copies of the risk allele A**

(risk allele=A, odds ratio/beta 1.16 [1.10-1.22] with pval 1E-8, pubmedid=17554260; risk allele=C, T1D odds ratio/beta 1.12 with pval 1E-9, pubmedid=21829393)

Gene related topics:

autoimmune diseases(5.00) · arthritis(1.00) · rheumatoid arthritis(1.00) · multiple sclerosis(1.00) · IMMUNE(1.00)

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Relevant papers and external links:

PMID 17554260
Robust associations of four new chromosome regions from genome-wide analyses of type 1 diabetes.

PMID 21829393
Genome-wide association analysis of autoantibody positivity in type 1 diabetes cases.

PMID 18971939

PMID 18987646

PHARMGKB PA162356634
Evidence: PubMed ID:17554260; Web Resource:External Link Results: Robust associations of four new chromosome regions from genome-wide analyses of type 1 diabetes (Initial Sample Size: 1,963 cases, 2,938 controls; Replication Sample Size: 4,000 cases, 5,000 controls, 2,997 trios; Risk Allele: rs763361-A).

dbSNP: rs763361 GWAS Catalog: rs763361 Genecard: CD226 chromosome 18

SNPedia: rs763361(C;T)

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★ **rs10210302(T;T)** **info:1.44** **15.36%** **1.8x risk**

rs10210302 has been reported in a large study to be associated with Crohn's disease.

The risk allele (oriented to the dbSNP entry) is (T); the odds ratio associated with heterozygotes is 1.19 (CI 1.01-1.41), and for homozygotes, 1.85 (CI 1.56-2.21).

Established associations:

crohn's disease **you have 2 copies of the risk allele T**
(risk allele=T, odds ratio/beta 1.19 [1.01-1.41] with pval 5E-14, pubmedid=17554300)

Gene related topics:

IMMUNE(1.05) · colitis ulcerative crohn disease(1.05) · crohn disease(0.81) · inflammatory bowel disease nos inflammatory bowel diseases(0.43) · inflammatory bowel disease(0.33) · inflammatory bowel diseases(0.16) · crohn s disease(0.13) · crohn disease crohn s disease(0.08) · crohn s disease ulcerative colitis(0.04)

Relevant papers and external links:

PMID 17554300
OMIM 611081

dbSNP: rs10210302 GWAS Catalog: rs10210302 Genecard: ATG16L1 chromosome 2

SNPedia: rs10210302(T;T)

★ rs1061170(T;T) good:1.40 53.79% **lower risk for AMD, generally longer live than (C) allele carriers**

rs1061170 is a SNP in the complement factor H CFH gene; it is also known as Tyr402His or p.Y402H. The rs1061170(T) allele encodes the more common Tyr (Y), while the generally rarer rs1061170(C) encodes the His (H).

This SNP has been associated primarily with age related macular degeneration, and to a lesser extent, with longevity.

This research paper shows that CFH Y402H is the relevant mutation.

A haplotype of rs1061170 rs3753394 rs800292 rs1329428 (TGTC) was found to confer a significantly increased likelihood of exudative AMD.

rs1061170 (aka Y402H 1277 T>C) was not associated with exudative AMD.

CFH variations appear to contribute to ARMD in Caucasians, but not in Japanese

CFH variant rs1061170 Y402H is strongly associated with both dry and wet AMD

rs1061170(C) alleles are significantly associated with increased susceptibility to early AMD in Taiwan Chinese populations.

Some SNPs in other genes (like C7 and MBL2) may protect individuals with one or two of the rs1061170(C) risk alleles.

An analysis of the joint effects of rs1061170, rs11200638 and rs10490924 on AMD

In a 4 year study of longevity of 491 nonagenarians in the Finnish Vitality 90+ study, risk factor-adjusted mortality was significantly higher among the rs1061170(C) carriers compared to non-carriers (odds ratio 1.78, CI 1.19-2.67, p = 0.005), and the survival curves of these carriers and non-carriers deviated significantly (p = 0.016). In other words, in this study, rs1061170(T;T) individuals generally lived longer than (C) allele carriers.

age related macular degeneration

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Established associations:

age-related macular degeneration **you have 0 copies of the risk allele C**

(risk allele=C, odds ratio/beta 2.41 with pval 1E-261, pubmedid=21665990)

Gene related topics:

VISION(25.76) · macular degeneration(25.76) · choroidal neovascularization macular degeneration(4.65) · METABOLIC(2.00) · age related maculopathy(2.00) · hemolytic uremic syndrome(2.00) · macular degeneration age related(2.00) · survival(1.00) · OTHER(1.00) · longevity(1.00) · mortality(1.00) · age related macular degeneration(0.75) · UNKNOWN(0.50) · haemolytic uraemic syndrome hemolytic uremic syndrome(0.50) · choroid diseases macular degeneration peripheral vascular diseases(0.36) · CARDIOVASCULAR(0.03) · myocardial infarct(0.03)

Relevant papers and external links:

PMID 21665990
Common variants near frk/col10a1 and vegfa are associated with advanced age-related macular degeneration.

PMID 16849663

PMID 17167412

PMID 16710702

PMID 17306880

PMID 18784628

PMID 17266113

PMID 18682806

PMID 19000922

PMID 15870199

OMIM 134370

PHARMGKB PA161889381
Evidence: PubMed ID:17053109In confirmation of other studies, the C allele of rs1061170 was found to be significantly associated with risk of Age-related Macular degeneration in a Caucasion (White) case-control study.

dbSNP: rs1061170 GWAS Catalog: rs1061170 Genecard: CFH chromosome 1

SNPedia: rs1061170(T;T)

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★ rs4988234(G;G) good:1.37 99.96% **common in complete genomics**

Some news linked it to lactose intolerance:
Screening of variants for lactase persistence/non-persistence in populations from South Africa and Ghana.
Association of the European lactase persistence variant (LCT-13910 C>T polymorphism) with obesity in the Canary Islands.
Several different lactase persistence associated alleles and high diversity of the lactase gene in the admixed Brazilian population.
It is however a typo: “There was a typographical error in the rs number in the Abstract and Introduction. The correct rs number is rs4988235.”

Gene related topics:

lactase persistence(1.00) · obesity(1.00) · METABOLIC(0.19) · lactose intolerance(0.19)

Relevant papers and external links:

PMID 19575818
PMID 22937140
PMID 23029545

dbSNP: rs4988234 Genecard: MCM6 chromosome 2 SNPedia: rs4988234(G;G)

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★ rs17391694(C;C) info:1.35 94.29% **; crohn's disease (you have the risk allele C)**

Established associations:

body height **you have 0 copies of the risk allele T**

(risk allele=T, odds ratio/beta 0.042 unit increase with pval 2E-11, pubmedid=20881960; risk allele=T, odds ratio/beta 0.043 [0.033-0.053] unit increase with pval 4E-16, pubmedid=25282103)

crohn's disease **you have 2 copies of the risk allele C**

(risk allele=C, odds ratio/beta 1.134 [1.077-1.194] with pval 3E-9, pubmedid=23128233)

Relevant papers and external links:

PMID 20881960
Hundreds of variants clustered in genomic loci and biological pathways affect human height.

PMID 25282103
Defining the role of common variation in the genomic and biological architecture of adult human height.

PMID 23128233
Host-microbe interactions have shaped the genetic architecture of inflammatory bowel disease.

dbSNP: rs17391694 GWAS Catalog: rs17391694 Genecard: nan chromosome 1

SNPedia: rs17391694(C;C)

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★ **rs1799978(A;A)** **good:1.30** **77.61%**

Association between ADORA2A and DRD2 polymorphisms and caffeine-induced anxiety.

Physiogenomic analysis of localized FMRI brain activity in schizophrenia.

Evaluation of genetic variability in the dopamine receptor D2 in relation to behavioral inhibition and impulsivity/sensation seeking: an exploratory study with d-amphetamine in healthy participants.

Pharmacogenetics and antipsychotics: therapeutic efficacy and side effects prediction.

Gene related topics:

CHEMDEPENDENCY(6.04) · alcoholism(6.04) · alcoholism substance related disorders(4.00) · PSYCH(2.30) · schizophrenia(2.30) · METABOLIC(2.00) · NEUROLOGICAL(2.00) · hyperprolactinaemia hyperprolactinemia(2.00) · nervous system diseases(2.00) · substance withdrawal syndrome(2.00) · substance withdrawal syndrome tobacco use disorder(2.00) · tic combined vocal and multiple motor tourette syndrome(2.00) · smoking behavior(1.74) · UNKNOWN(1.29) · neuroleptic malignant syndrome(1.29) · heroin dependence(1.00) · methamphetamine abuse(1.00) · anxiety(1.00) · brain(1.00) · tardive dyskinesia(0.95) · heroin abuse(0.83) · cocaine dependence(0.80) · cocaine related disorders(0.67) · delirium(0.57) · impulsivity(0.57) · alcohol abuse(0.52) · tobacco use disorder(0.44) · adhd(0.40) · mood disorders(0.40) · parkinson s disease(0.40) · smoking(0.33) · alcohol(0.32) · mood disorder(0.27) · substance related disorders(0.27) · CARDIOVASCULAR(0.25) · atrial natriuretic factor(0.25) · alcohol dependence(0.22) · personality traits(0.21) · tourette syndrome(0.21) · dyslexia(0.19) · migraine disorders(0.17) · adhd attention deficit hyperactivity disorder(0.15) · personality(0.14) · personality disorders(0.12) · depressive disorder major(0.10) · bipolar disorder(0.07) · attention deficit hyperactivity disorder(0.05) · autism(0.03)

Relevant papers and external links:

PHARMGKB PA165108051
Evidence: PubMed ID:19339912Risk or phenotype-associated allele: A. Phenotype: Having the AA genotype of this variant was associated with a protective effect from increased prolactin concentration. Study size: 90. Study population/ethnicity: 7-17-year-old patients chronically treated with risperidone; non-Hispanic Caucasians. Significance metric(s): p = 0.002. Type of association: CO.

PHARMGKB PA162356251
Evidence: PubMed ID:18855532This SNP is significant predictor of treatment response to risperidone in first-episode schizophrenia.

PMID 18305461

PMID 18330705

PMID 19968402

PMID 21162693

dbSNP: rs1799978 Genecard: DRD2 chromosome 11 SNPedia: rs1799978(A;A)

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★ **rs17300539(G;G)** **good:1.30** **94.72%** **normal**

rs17300539, also known as -11391 G/A, is a SNP in the promoter of the adiponectin ADIPOQ gene.

A small study of 180 Spanish patients with obesity concluded that rs17300539(G;G) individuals are at increased risk of insulin resistance and MetS complications compared to carriers of an (A) allele. The rs17300539(A) allele confered protection from weight regain, particularly at 32-60 weeks after the low-calorie dietary intervention, when improvement in GG subjects had disappeared

adiponectin levels and risk of diabetes rs17300539 P(n) = 2.6 x 10(-8) and rs822387 (P(n) = 3.8 x 10(-5)

Gene related topics:

METABOLIC(4.00) · adiponectin levels(4.00) · CARDIOVASCULAR(2.00) · atherosclerosis coronary diabetes type 2(2.00) · adiponectin(1.44) · UNKNOWN(1.00) · chronic renal failure diabetes mellitus insulin dependent diabetes mellitus type 1 diabetic nephropathies diabetic nephropathy kidney failure chronic(1.00) · weight(1.00) · insulin(1.00) · insulin resistance obesity(0.69) · diabetes type 2(0.55) · obesity(0.53) · metabolic syndrome(0.51) · obesity morbid(0.33) · diabetes mellitus type 2 obesity(0.31) · insulin resistance(0.22) · type 2 diabetes(0.14) · polycystic ovary syndrome(0.08) · RENAL(0.06) · kidney failure chronic(0.06) · diabetes mellitus type 2(0.05)

Relevant papers and external links:

PMID 18949681

PMID 18776141
OMIM 612556

dbSNP: rs17300539 Genecard: ADIPOQ chromosome 3 SNPedia: rs17300539(G;G)

★ rs9939609(T;T) good:1.30 43.55% **lower risk of obesity and Type-2 diabetes**

rs9939609 is a SNP in the fat mass and obesity associated FTO gene, aka the "Fat Gene" . The original paper describing it is here .

2015 research suggests rs1421085 may be the causal snp.

The researchers identified 10 different FTO SNPs in the first intron of the gene that associated with both BMI and type-2 diabetes. Because they all correlated with each other, they chose to examine one of the SNPs (rs9939609) in detail.

rs9939609 has also been reported in a large study to be associated with type-2 diabetes.
The risk allele (oriented to the dbSNP entry) is (A); the odds ratio associated with heterozygotes is 1.34 (CI 1.17-1.52), and for homozygotes, 1.55 (CI 1.3-1.84).

In a different study, this SNP was found in a linkage block in the FTO gene with rs1121980; see rs1121980 for the association of this block with early onset obesity since it showed the strongest association.

The increases in body mass index associated with rs9939609(A) carriers appears to begin at a young age and are maintained throughout adulthood, according to a study of 5,600+ Utah families.

This SNP did not show any association with obesity, type-2 diabetes or any other related traits in a study of 3,210 Chinese subjects. Furthermore, the minor allele frequency was much lower in Chinese populations compared to populations of European descent.

Note: The three FTO SNPs, rs1421085, rs17817449, and rs9939609, are in strong linkage disequilibrium (pairwise r2>0.97), and there are two primary haplotypes, C-G-A (42.0 %) and T-T-T (55.5 %).

1,638 type 2 diabetes patients and 1,858 controls
rs9939609 in FTO (OR 1.14, 95% CI: 1.04-1.25, p = 0.006)

A study of 3,000+ UK children indicated that (A;A) genotypes were less satieted (i.e. had greater appetite) than other genotypes, indicating this risk allele for obesity is likely to influence appetite.

This SNP is also associated with (severe) obesity in a study of 927 Japanese patients.

This SNP may contribute to variation in plasma C-reactive protein (CRP) levels independently of obesity; average CRP levels may be 12-14% higher per rs9939609(A) allele.

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This SNP is also associated with adult obesity in Mexicans.

A longitudinal study of Finnish children concluded that the effect of rs9939609 on BMI only becomes evident after age 7, and that the FTO gene is not directly associated with energy intake or physical activity.

Independent of fatness, the A-allele of the FTO SNP appears to increase mortality of a magnitude similar to smoking, but without a particular underlying disease pattern barring an increase in the risk of diseases of the nervous system.

Each copy of the risk-allele A at rs9939609 was significantly associated with 0.45 kg/m(2) increase in BMI (95% confidence interval (CI): 0.16-0.74; P = 0.004) and 0.97 cm increase in waist circumference (95% CI: 0.21-0.65; P = 0.0002). Similar results were observed for rs8050136. may confer a modest susceptibility to obesity in an ethnicity-specific manner, but may be unlikely to contribute to a clinically significant diabetes risk.

Established associations:

type ii diabetes mellitus **you have 0 copies of the risk allele A**

(risk allele=A, Obese odds ratio/beta 1.25 [1.19-1.30] with pval 1E-20, pubmedid=22693455; risk allele=A, odds ratio/beta 1.34 [1.17-1.52] with pval 2E-7, pubmedid=17554300)

body mass index **you have 0 copies of the risk allele A**

(risk allele=A, BMI odds ratio/beta 0.24 [0.14-0.32] kg/m2 increase with pval 2E-7, pubmedid=19396169; risk allele=A, odds ratio/beta 0.33 [0.27-0.39] kg/m2 increase with pval 4E-51, pubmedid=19079261; risk allele=A, odds ratio/beta 0.36 kg/m2 per copy in adults with pval 2E-20, pubmedid=17434869)

bone density **you have 0 copies of the risk allele A**

(risk allele=A, BMI odds ratio/beta 0.24 [0.14-0.32] kg/m2 increase with pval 2E-7, pubmedid=19396169)

systolic blood pressure **you have 0 copies of the risk allele A**

(risk allele=A, BMI odds ratio/beta 0.24 [0.14-0.32] kg/m2 increase with pval 2E-7, pubmedid=19396169)

diastolic blood pressure **you have 0 copies of the risk allele A**

(risk allele=A, BMI odds ratio/beta 0.24 [0.14-0.32] kg/m2 increase with pval 2E-7, pubmedid=19396169)

waist-hip ratio **you have 0 copies of the risk allele A**

(risk allele=A, BMI odds ratio/beta 0.24 [0.14-0.32] kg/m2 increase with pval 2E-7, pubmedid=19396169)

heart rate **you have 0 copies of the risk allele A**

(risk allele=A, BMI odds ratio/beta 0.24 [0.14-0.32] kg/m2 increase with pval 2E-7, pubmedid=19396169)

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age at menarche **you have 0 copies of the risk allele A**

(risk allele=A, odds ratio/beta 2.1 [1.32-2.88] week decrease with pval 3E-8, pubmedid=21102462)

Gene related topics:

METABOLIC(4.07) · obesity(4.07) · diabetes mellitus type 2 obesity(3.77) · diabetes mellitus obesity(3.00) · obesity related traits(3.00) · obesity morbid(1.33) · obesity type 2 diabetes(1.29) · body mass(1.00) · OTHER(1.00) · waist circumference(1.00) · mortality(1.00) · smoking(1.00) · bmi(0.56) · body weight obesity(0.46) · adiposity(0.40) · diabetes type 2(0.36) · body mass index(0.34) · obesity overweight(0.31) · weight(0.25) · type 2 diabetes(0.21) · diabetes mellitus type 2(0.20) · metabolic syndrome(0.09) · UNKNOWN(0.09) · diabetes type 2 diabetes type 1(0.09) · CARDIOVASCULAR(0.08) · cell adhesion molecules(0.08) · metabolic syndrome x(0.08)

Relevant papers and external links:

PMID 22693455
Stratifying type 2 diabetes cases by bmi identifies genetic risk variants in lama1 and enrichment for risk variants in lean compared to obese cases.

PMID 19396169
A large-scale genome-wide association study of asian populations uncovers genetic factors influencing eight quantitative traits.

PMID 19079261
Six new loci associated with body mass index highlight a neuronal influence on body weight regulation.

PMID 21102462
Thirty new loci for age at menarche identified by a meta-analysis of genome-wide association studies.

PMID 17434869

PMID 17554300

PMID 18159244

PMID 18239580

PMID 17959933

PMID 18437351

PMID 18583465

PMID 18379722

PMID 18948966

PMID 18719664

PMID 19158205

PMID 19214238

PMID 18787525

OMIM 612460

OMIM 125853

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PHARMGKB PA165110735
Evidence: PubMed ID:17942823Risk or phenotype-associated allele: rs9939609 A allele. Phenotype: In studies of 3,856 type 2 diabetic case subjects and 4,861 normal glucose-tolerant control subjects, the minor A-allele of rs9939609 associated with type 2 diabetes (OR = 1.13, 95% CI 1.06-1.20, p = 9 × 10−5), which was abolished when adjusting for BMI (OR = 1.06, CI = 0.97-1.16, p = 0.2). The FTO rs9939609 genotype was influenced by the habitual level of physical activity in a population-based study of 5,722 treatment-naive Danish. Physical inactivity was associated with a BMI increase of 1.95 ± 0.33 kg/m2 in FTO rs9939609 AA genotype carriers, whereas no major effect of sedentary lifestyle was found comparing TT and TA genotypes. Among 17,162 middle-aged Danes, the A-allele associated with overweight (OR = 1.19, CI = 1.13-1.24, p = 1 x 10(-12)) and obesity (OR = 1.27, CI = 1.20-1.34, p = 2 x 10(-16)). Body weight, waist circumference, fat mass, and fasting serum leptin levels were significantly elevated in A-allele carriers. An interaction between the FTO rs9939609 genotype and physical activity (p = 0.007) was found, where physically inactive homozygous risk A-allele carriers had an 1.95 +/- 0.3 kg/m(2) increase in BMI relative to TT genotype carriers. Interaction between the FTO rs9939609 genotype and physical activity (P = 0.007) was observed, where physically inactive homozygous risk A-allele carriers had a 1.95 ± 0.3 kg/m2 increase in BMI compared with TT genotype carriers. Study size: Subsets of 17,508. Study population/ethnicity: Anglo-Danish-Dutch. Significance metric(s): OR = (1.06-1.27). Type of association: GN; CO; PD.

PHARMGKB PA162168074
Evidence: PubMed ID:17434869The A allele of rs9939609 is reproducibly associated with BMI and obesity in European whites from childhood on.

PHARMGKB PA162168246
Evidence: PubMed ID:18583465Children homozygous for the A allele of this intronic SNP exhibited reduced Satiety Responsiveness, assessed using the Child Eating Behavior Questionnaire (CEBQ), relative to AT heterozygotes and TT homozygotes. There was no statistically significant association between genotype at this SNP and scores for Enjoyment of Food, also assessed using the CEBQ.

PHARMGKB PA161660933
Evidence: PubMed ID:17434869SNP in the FTO gene region is strongly associated with type 2 diabetes. This association was replicated by analyzing the SNP in a further 3757 type 2 diabetes cases and 5346 controls.

PHARMGKB PA162355880
Evidence: PubMed ID:18853134In a large study of 16,143 non-diabetic individuals this SNP in the FTO gene predicted the development of at least two components of the metabolic syndrome.

PHARMGKB PA162356662
Evidence: PubMed ID:17554300; Web Resource:External Link Results: Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls (Initial Sample Size: 1,924 cases, 2,938 controls; Replication Sample Size: (see Zeggini 2007); Risk Allele: rs9939609-A). This variant is associated with type 2 diabetes.

PHARMGKB PA162356604

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Evidence: PubMed ID:17434869; Web Resource:External Link Results: A common variant in the FTO gene is associated with body mass index and predisposes to childhood and adult obesity (Initial Sample Size: 10,657 adults; Replication Sample Size: 19,424 adults, 10,172 children; Risk Allele: rs9939609-A).

PHARMGKB PA164740175
Evidence: PubMed ID:19079261; Web Resource:External Link results: Six new loci associated with body mass index highlight a neuronal influence on body weight regulation. (Initial Sample Size: 32,387 individuals; Replication Sample Size: 59,092 individuals); (Region: 16q12.2; Reported Gene(s): FTO; Risk Allele: rs9939609-A); (p-value= 4E-51).This variant is associated with Body mass index.

PHARMGKB PA165110734
Evidence: PubMed ID:19687793Risk or phenotype-associated allele: rs9939609 A allele. Phenotype: Among participants enrolled in a 10-week dietary intervention study, the study drop-out rate was higher among rs9939609 A-allele carriers on a high-fat/low carbohydrate (HF) versus low-fat/high carbohydrate (LF) diet (AT genotype p = 0.002; AT/AA combined genotype p = 0.003). Individuals carrying the rs9939609 TT gentoype had smaller decrease in resting energy expenditure on the LF versus HF diet (p = 0.0055, n = 387), and the largest reduction in measures of insulin release (p = 0.0083, n = 612) and insulin resistance (p = 0.047, n = 612). Individuals with rs9939609 TT genotype were more likely to have been randomized to HF (55.2%) than LF diet (44.8%) (p = 0.027), and carriers of the heterozygous AT genotype were distributed evenly. Study size: Subsets of 764. Study population/ethnicity: Obese women and men recruited in a 10-week European multi-center dietary intervention study from May 2001 until September 2002. Significance metric(s): p < 0.05. Type of association: GN; CO; PD.

OMIM 155541

dbSNP: rs9939609 GWAS Catalog: rs9939609 Genecard: FTO chromosome 16

SNPedia: rs9939609(T;T)

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★ **rs3803300(G;G)** **info:1.27** **45.04%**

G allele associated with schizophrenia

among 120 (likely Japanese) first-episode neuroleptic-naive schizophrenics treated with risperidone genotyped for 30 variants in misc. dopamine and serotonin (receptors and otherwise) two SNPs in DRD2 (rs1799989 and rs1800497) and two SNPs in AKT1 (rs3803300 and rs2494732) were significant predictors of treatment response to risperidone

The ancestral allele is G in the dbSNP database.

Gene related topics:

schizophrenia(1.00)

Relevant papers and external links:

PMID 17915974

PMID 18855532

PHARMGKB PA162356248
Evidence: PubMed ID:18855532This SNP is significant predictor of treatment response to risperidone in first-episode schizophrenia.

dbSNP: rs3803300 Genecard: ZBTB42 chromosome 14 SNPedia: rs3803300(G;G)

★ rs167771(A;A) info:1.19 34.65% **Possibly more "Insistence on Sameness" on ADI-R test. Possibly increased FTND nicotine dependence.**

Possibly more "Insistence on Sameness" on ADI-R test.

Possibly increased FTND nicotine dependence.

rs167771 was significantly associated with autism spectrum disorder in a study of 144 patients.

G allele associated with increased extra-pyramidal symptom risk as a result of risperidone treatment in a study of 321 psychiatric inpatients (81 presenting with EPS and 189 without)

Gene related topics:

nicotine(5.00) · nicotine dependence(5.00) · NEUROLOGICAL(3.79) · tardive dyskinesia(3.79) · essential tremor(2.88) · PSYCH(2.37) · schizophrenia(2.37) · CHEMDEPENDENCY(2.00) · UNKNOWN(2.00) · akathisia drug induced(2.00) · cocaine abuse(2.00) · autism(1.00) · cocaine related disorders(0.67) · schizophrenia tardive dyskinesia(0.57) · tourette syndrome(0.47) · alcohol dependence(0.22) · parkinson s disease(0.14) · personality disorders(0.12) · bipolar disorder(0.10) · obsessive compulsive disorder(0.06) · adhd attention deficit hyperactivity disorder(0.05) · personality traits(0.05) · alcohol abuse(0.04) · smoking behavior(0.04)

Relevant papers and external links:

PMID 19058789

PMID 19506579

PHARMGKB PA165109604
Evidence: PubMed ID:19506579Risk or phenotype-associated allele: G. Phenotype: The G variant of rs167771 was associated with increased risk for extrapyramidal symptoms in psychiatric patients receiving risperidone. Study size: 132. Study population/ethnicity: Patients with Schizophrenia or

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Bipolar disorder receiving antipsychotics; Caucasians; Spain; Catalonia. Significance metric(s): OR = 3.3; p = 0.00013. Type of association: PD.

dbSNP: rs167771 Genecard: DRD3 chromosome 3 SNPedia: rs167771(A;A)

★ rs4570625(G;G) info:1.18 41.93% **maybe: higher scores on anxiety-related personality traits; greater placebo response**

One study of (only) 25 people shows this genotype was a significant predictor of clinical placebo response, being associated with greater improvement in anxiety symptoms. A criticism of this study has also been published, and it has not been replicated since the original publication years ago.

This SNP has been linked to several psychiatric and/or behavioral phenomena, including:
higher scores on anxiety-related personality traits
obsessive compulsive disorder
panic disorder with a possible gender-dependent effect
rs4570625, also known as G-703T, is a SNP in the tryptophan hydroxylase 2 TPH2 gene. This SNP has been linked to several psychiatric and/or behavioural phenomena, including:

rs4570625 and rs4565946 linked to the pathogenesis of early-onset obsessive compulsive disorder

rs4570625 and rs11178997 in TPH2's regulatory region display preferential transmission

rs4570625(G;G) homozygosity was a significant predictor of clinical placebo response, being associated with greater improvement in anxiety symptoms.

g2b2mh is rs4570625(G;G) is a significant predictor of clinical placebo response? maybe not

panic disorder with a possible gender-dependent effect

Transmission disequilibrium of polymorphic variants in the tryptophan hydroxylase-2 gene in attention-deficit/hyperactivity disorder.

Association between a polymorphism in the promoter region of the TPH2 gene and the personality trait of harm avoidance.

Tryptophan hydroxylase-2 gene variation influences personality traits and disorders related to emotional dysregulation.

Case-control and family-based association studies of candidate genes in autistic disorder and its endophenotypes: TPH2 and GLO1.

Characterization of a functional promoter polymorphism of the human tryptophan hydroxylase 2 gene in serotonergic raphe neurons.

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Genetic variation of serotonin function and cognitive control.

Tryptophan hydroxylase 2 gene and alcohol use among college students.

Resequencing of serotonin-related genes and association of tagging SNPs to citalopram response.

TPH2 gene variants and anxiety during alcohol detoxification outcome.

Epistasis between IL1A, IL1B, TNF, HTR2A, 5-HTTLPR and TPH2 variations does not impact alcohol dependence disorder features.

Alternative splicing and extensive RNA editing of human TPH2 transcripts.

Association study of tryptophan hydroxylase-2 gene in schizophrenia and its clinical features in Chinese Han population.

Pharmacogenetics of antidepressant response.

Serotonin and early cognitive development: variation in the tryptophan hydroxylase 2 gene is associated with visual attention in 7-month-old infants.

Emotion appraisal and the tryptophan hydroxylase 2 (TPH2) gene.

Common variants in the TPH2 promoter confer susceptibility to paranoid schizophrenia.

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Gene related topics:

anxiety(5.00) · personality(5.00) · OTHER(1.00) · chronic fatigue syndrome(1.00) · citalopram(1.00) · obsessive compulsive disorder(1.00) · harm avoidance(1.00) · alcohol(1.00) · alcohol dependence(1.00) · schizophrenia(1.00) · PSYCH(0.56) · suicide(0.56) · METABOLIC(0.45) · waist circumference(0.45) · autism(0.17) · adhd attention deficit hyperactivity disorder(0.15) · depression(0.15) · adhd(0.15) · panic disorder(0.08) · NEUROLOGICAL(0.08) · migraine disorders(0.08) · attention deficit hyperactivity disorder(0.05) · personality traits(0.05) · depressive disorder major(0.05)

Relevant papers and external links:

PMID 19052197
PMID 16146581
PMID 19052197
PMID 19132526
OMIM 143465
OMIM 607478
OMIM 613003
PMID 16116490
PMID 17176491
PMID 17176492
PMID 17346350

PMID 17568567
PMID 17892388
PMID 18782386
PMID 19077664
PMID 19361870
PMID 19742166
PMID 20126463
PMID 20938755
PMID 21172166
PMID 21418063
PMID 22322887
PMID 22392150

dbSNP: rs4570625 Genecard: TPH2 chromosome 12 SNPedia: rs4570625(G;G)

★ rs11805303(C;T) info:1.18 47.11% **1.4x risk of Crohn's disease**

rs11805303 has been reported in a large study to be associated with Crohn's disease.

The risk allele (oriented to the dbSNP entry) is (T); the odds ratio associated with heterozygotes is 1.39 (CI 1.22-1.58), and for homozygotes, 1.86 (CI 1.54-2.24).

Established associations:

crohn's disease **you have 1 copies of the risk allele T**

(risk allele=T, odds ratio/beta 1.39 [1.22-1.58] with pval 6E-12, pubmedid=17554300)

Gene related topics:

IMMUNE(5.16) · inflammatory bowel diseases(5.16) · colitis ulcerative crohn disease(1.98) · arthritis psoriatic psoriasis(1.12) · crohn disease crohn s disease inflammatory bowel disease nos inflammatory bowel diseases(1.00) · crohn disease(0.96) · crohn s disease(0.64) · inflammatory bowel disease(0.54) · spondylitis ankylosing(0.44) · inflammatory bowel disease nos inflammatory bowel diseases(0.43) · colitis ulcerative(0.42) · chronic ulcerative colitis colitis ulcerative crohn disease crohn s disease inflammatory bowel disease nos inflammatory bowel diseases(0.40) · ulcerative colitis(0.24) · crohn disease crohn s disease(0.23) · psoriasis(0.20) · behcet syndrome(0.17) · crohn s disease ulcerative colitis(0.15) · ankylosing spondylitis(0.08) · lupus erythematosus systemic systemic lupus erythematosus(0.03)

Relevant papers and external links:

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PMID 17554300
OMIM 612261
OMIM 607562

dbSNP: rs11805303 GWAS Catalog: rs11805303 Genecard: IL23R chromosome 1

SNPedia: rs11805303(C;T)

★ rs2060793(G;G) info:1.17 43.53% **Higher serum levels of vitamin D**

Levels of 25-hydroxyvitamin D in familial longevity: the Leiden Longevity Study. Concluded that familial longevity was associated with a lower vitamin D levels and a lower frequency of the allelic variation in the CYP2R1 gene.

Genome-wide association study of circulating vitamin D levels. This SNP is part of CYP2R1, which encodes a key C-25 hydroxylase that converts vitamin D3 to an active vitamin D receptor ligand. This SNP, along with rs2282679, rs3829251, and rs6599638 accounted for 2.8% of the variance in circulating vitamin D levels.

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Established associations:

vitamin d measurement **you have 0 copies of the risk allele A**

(risk allele=A, odds ratio/beta 0.25 [0.15-0.35] unit increase with pval 3E-17, pubmedid=20418485)

Gene related topics:

vitamin d(5.00) · longevity(1.00)

Relevant papers and external links:

PMID 23128285
PMID 20418485

dbSNP: rs2060793 GWAS Catalog: rs2060793 Genecard: CYP2R1 chromosome 11

SNPedia: rs2060793(G;G)

★ rs6908425(C;C) info:1.17 61.11% **1.95x increased risk of developing Crohn's disease**

rs6908425 has been reported in a large study to be associated with Crohn's disease.

The risk allele (oriented to the dbSNP entry) is (C); the odds ratio associated with heterozygotes is 1.63 (CI 1.38-2.25), and for homozygotes, 1.95 (CI 1.43-2.67).

Established associations:

crohn's disease **you have 2 copies of the risk allele C**

(risk allele=C, odds ratio/beta 1.17 [1.11-1.23] with pval 1E-8, pubmedid=21102463; risk allele=C, odds ratio/beta 1.21 with pval 9E-10, pubmedid=18587394)

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Gene related topics:

METABOLIC(1.00) · diabetes mellitus type 2 insulin resistance(1.00) · type 2 diabetes(0.91) · diabetes mellitus type 2(0.81) · diabetes type 2(0.78) · UNKNOWN(0.09) · diabetes type 2 diabetes type 1(0.09) · diabetes mellitus(0.03) · IMMUNE(0.03) · crohn disease(0.03)

Relevant papers and external links:

PMID 21102463
Genome-wide meta-analysis increases to 71 the number of confirmed crohn's disease susceptibility loci.

PMID 18587394
Genome-wide association defines more than 30 distinct susceptibility loci for crohn's disease.

PMID 17554300

OMIM 177900

dbSNP: rs6908425 GWAS Catalog: rs6908425 Genecard: CDKAL1 chromosome 6

SNPedia: rs6908425(C;C)

★ rs25531(A;A) info:1.12 74.38% **short form of 5-HTTLPR. lower levels of serotonin, slightly less happy, benefits from more support**

23andMe reports that this genotype is not reliable on their platform. 23andMe users should ignore this result.

This genotype is associated with lower levels of serotonin and appears to have a cognitive effect related to emotions, but the effect is slight and no one in SNPedia has yet found a good description. However the (G;G) genotype generally seems to have more positive associations, while this (A;A) is more negative. Examples of the differences include biased attention for negative word stimuli and gaze bias for emotional information.

More reluctant to endorse utilitarian actions resulting in foreseen harm to an innocent individual

The genotype of this SNP is not meaningful on 23andme v3.
External Link
According to David Hinds of 23andMe on community forums, "nearly everyone (99.97%) is getting called as CC, and there is no clear heterozygote cluster" ... "the genotype calls for rs25531 on our platform are not meaningful." Looking at OpenSNP frequency data, this seems to be universal for direct to consumer genotyping services at this stage.

The minor allele (G) of rs25531 is almost always in phase with the long (L) allele of the 5-HTTLPR, which is the most commonly studied variation in this region. The (L) long allele may be associated with lower levels of serotonin.

The article and video from "The Atlantic" suggests that children with the short allele (A), may need and benefit greatly from more maternal support
External Link

In contrast to earlier reports linking rs25531, a SNP in the serotonin transporter SLC6A4 gene, to obsessive compulsive disorder (OCD), a March 2007 publication finds no significant association studying "the largest sample size of OCD patients (N=347) and controls (N=749) ever investigated".

A separate study of 75 patients suffering mood disorders after traumatic brain injury (TBI) concluded that there was no evidence of association between rs25531 and depression post-TBI.

A small study (62 patients and 62 matched controls) investigated this SNP in relation to blushing. (Blushing is considered to be one of the prime pathophysiological markers of social anxiety disorder.) The less active 5-HTTLPR genotypes were nominally significantly associated with increased blushing propensity in patients with social anxiety disorder, and association remained significant even when statistically controlled for he influence of depression.

A study of 94 depressed patients who attempted suicide compared to 94 without psychiatric disorders found no evidence of an association with rs25531.

A study of 97 "normal" individuals concluded that rs25531(G;G) individuals selectively process positive affective material alongside selective avoidance of negative affective material, in a manner absent among individuals carrying a rs25531(A) allele. To put it another way, if this conclusion drawn from a very small sample of people can be replicated, rs25531(G;G) individuals may have a tendency to be optimists and to be more resilient to stress.

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A study published in 2011 of 2,500+ individuals followed from 1995 - 2008 as part of the US National Longitudinal Study of Adolescent Health found that twice as many respondents with two 5-HTTLPR long variants said they were very satisfied with life compared with carriers of two short versions. Conversely, 26% of those carrying two short alleles said they were dissatisfied with life, compared with 20% of people carrying two long variants. This blog post investigates the statistics.

A study of 135 patients with major depressive disorder (mean age 31.1+/-11.6 years, 68% females) treated with escitalopram 10-20 mg/day for 12 weeks concluded that patients carrying the rs25531 (A) allele may be at increased risk for side effects (e.g. headaches), but they say no associations with efficacy.

Protocol for genotyping rs25531

Gene related topics:

PSYCH(24.57) · depression(24.57) · suicide(6.88) · obsessive compulsive disorder(5.82) · IMMUNE(5.28) · irritable bowel syndrome(5.28) · personality traits(4.75) · anxiety disorder(4.50) · REPRODUCTION(4.00) · depressive disorder major bipolar disorder(4.00) · sexual dysfunction physiological(4.00) · personality disorders(3.78) · depressive disorder major(3.72) · affective disorder(3.12) · anxiety related traits(3.00) · decision making(3.00) · psychoses(3.00) · personality(2.60) · panic disorder(2.19) · autism(2.13) · mood disorders(2.02) · CHEMDEPENDENCY(2.00) · OTHER(2.00) · PHARMACOGENOMIC(2.00) · UNKNOWN(2.00) · alcohol abuse smoking behavior(2.00) · alcoholism attention deficit hyperactivity disorder(2.00) · alcoholism recurrence(2.00) · antidepressant adverse effects(2.00) · anxiety(2.00) · auditory evoked potential(2.00) · bipolar and unipolar disorder(2.00) · eating disorder(2.00) · hallucinations(2.00) · harm avoidance(2.00) · impulse control disorder(2.00) · manic depressive illness(2.00) · personality traits illegal drug use(2.00) · suicidal behavior(1.92) · alcoholism(1.92) · NEUROLOGICAL(1.80) · cocaine dependence(1.80) · major depression(1.80) · migraine migraine with aura(1.80) · neuroticism(1.80) · major depressive disorder(1.73) · migraine(1.73) · substance related disorders(1.67) · bulimia(1.47) · attention deficit hyperactivity disorder(1.16) · adhd attention deficit hyperactivity disorder(1.00) · aggressive behavior(1.00) · alcohol related disorders(1.00) · bipolar disorder depression(1.00) · conduct disorder(1.00) · eating disorders(1.00) · sudden infant death(1.00) · stress(1.00) · brain(1.00) · psychiatric disorders(1.00) · anorexia nervosa(0.93) · CARDIOVASCULAR(0.80) · hypertension pulmonary(0.80) · major psychoses(0.80) · bipolar disorder(0.79) · schizophrenia(0.69) · alcohol abuse(0.68) · bipolar affective disorder(0.67) · fibromyalgia(0.67) · sids sudden infant death syndrome(0.67) · smoking behavior(0.66) · mood disorder(0.60) · psychosis(0.60) · heroin abuse(0.53) · epilepsy temporal lobe(0.43) · sleep disorders(0.36) · cognitive function(0.33) · migraine with aura(0.33) · syncope vasovagal(0.33) · DEVELOPMENTAL(0.27) · mental retardation(0.27) · adhd(0.26) · METABOLIC(0.23) · weight gain(0.23) · smoking(0.21) · tourette syndrome(0.21) · alcohol(0.21) · migraine disorders(0.17) · cognitive ability(0.11) · chronic obstructive pulmonary disease(0.07) · hearing loss(0.07) · AGING(0.04) · longevity(0.04)

Relevant papers and external links:

PMID 8929413

PMID 17375136

PMID 18465388

PMID 18629430

PMID 19103261

PMID 19272758

PHARMGKB PA164920378

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Evidence: PubMed ID:15993855This vatiant in the SLC6A4 gene, located upstream of the HTTLPR polymorphism, showed evidence of an association with treatment response, and biochemical experiments showed this polymorphism altered binding of nuclear extracts to a consensus sequence for the activator protein 2 transcription factor, which is believed to be a critical factor in regulating neural gene expression in mammals.

PHARMGKB PA162008701
Evidence: PubMed ID:18618621This promoter SNP is not associated with depression remission frequency upon treatment with citalopram in white Hispanics, white non-Hispanics, or Blacks.

dbSNP: rs25531 Genecard: SLC6A4 chromosome 17 SNPedia: rs25531(A;A)

★ rs10883365(A;G) info:1.03 49.44% **1.2x increased risk for developing Crohn's disease**

rs10883365 has been reported in a large study to be associated with Crohn's disease.

The risk allele (oriented to the dbSNP entry) is (G); the odds ratio associated with heterozygotes is 1.2 (CI 1.03-1.39), and for homozygotes, 1.62 (CI 1.37-1.92).

The association between rs10883365 and Crohn's disease was replicated in a Japanese population.

Pharmacogenetics: data, concepts and tools to improve drug discovery and drug treatment.

Genetic determinants of ulcerative colitis include the ECM1 locus and five loci implicated in Crohn's disease.

Worldwide population differentiation at disease-associated SNPs.

Gene variants influencing measures of inflammation or predisposing to autoimmune and inflammatory diseases are not associated with the risk of type 2 diabetes.

Unbiased estimation of odds ratios: combining genomewide association scans with replication studies.

Lack of association of NKX2-3, IRGM, and ATG16L1 inflammatory bowel disease susceptibility variants with celiac disease.

NKX2-3 and IRGM variants are associated with disease susceptibility to IBD in Eastern European patients.

Established associations:

crohn's disease **you have 1 copies of the risk allele G**

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(risk allele=G, odds ratio/beta 1.2 [1.03-1.39] with pval 6E-8, pubmedid=17554300)

Gene related topics:

celiac disease(1.00) · ulcerative colitis(1.00) · inflammatory bowel disease(1.00) · type 2 diabetes(1.00) · inflammation(1.00)

Relevant papers and external links:

PMID 17554300

PMID 18936107

OMIM 612288

PHARMGKB PA162356575
Evidence: PubMed ID:17554261; Web Resource:External Link results: Sequence variants in the autophagy gene IRGM and multiple other replicating loci contribute to Crohn's disease susceptibility (Initial Sample Size: 1,748 cases, 2,938 controls; Replication Sample Size: 1,182 cases, 2,024 controls). This variant is associated with Crohn's disease.

PHARMGKB PA162356636
Evidence: PubMed ID:17554300; Web Resource:External Link Results: Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls (Initial Sample Size: 1,748 cases, 2,938 controls; Replication Sample Size: (see Parkes 2007); Risk Allele: rs10883365-G). This variant is associated with crohn's disease.

PMID 18224312

PMID 18438406

PMID 18533027

PMID 18853133

PMID 19140132

PMID 19683022

PMID 21049557

dbSNP: rs10883365 GWAS Catalog: rs10883365

Genecard: LINC01475 chromosome 10 SNPedia: rs10883365(A;G)

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★ rs174548(C;G) info:1.00 44.03% **intermediate phosphatidylcholine values**

FADS1 codes for an enzyme involved in fatty acid unsaturation and the FADS1 gene SNP rs174548 seems to explain as much as ten percent of the variance in the metabolism of some

glycerophospholipids. news

A study of metabolite concentrations in the blood of 284 adult males from Southern Germany determined that several SNPs were associated either directly with a metabolite range or with a ratio of metabolites. The rarer rs174548(G) allele was associated with decreased phosphatidylcholine values.

295px|thumb|left|

Established associations:

albumin:globulin ratio measurement **you have 1 copies of the risk allele G**

(risk allele=G, ALB/GLB odds ratio/beta 0.017 [0.011-0.023] unit increase with pval 4E-8, pubmedid=23303382)

triglyceride measurement **you have 1 copies of the risk allele G**

(risk allele=G, odds ratio/beta 0.03 [0.02-0.04] unit increase with pval 5E-14, pubmedid=20864672)

high density lipoprotein cholesterol measurement **you have 1 copies of the risk allele G**

(risk allele=G, odds ratio/beta 0.01 [0.007-0.015] unit decrease with pval 1E-12, pubmedid=20864672)

blood metabolite measurement **you have 1 copies of the risk allele C**

(risk allele=C, arachidonate 20:4n6 odds ratio/beta 0.049 [0.043-0.055] unit increase with pval 1E-84, pubmedid=24816252; risk allele=C, arachidonate 20:4n6/dihomo-linolenate 20:3n3 or n6 odds ratio/beta 0.071 [0.067-0.075] unit increase with pval 2E-361, pubmedid=24816252)

cis/trans-18:2 fatty acid measurement **you have 1 copies of the risk allele C**

(risk allele=C, Cis/trans-18:2, EA odds ratio/beta 0.0035 [0.0027-0.0043] unit decrease with pval 5E-15, pubmedid=25646338)

total trans-18:1 fatty acid measurement **you have 1 copies of the risk allele C**

(risk allele=C, Cis/trans-18:2, EA odds ratio/beta 0.0035 [0.0027-0.0043] unit decrease with pval 5E-15, pubmedid=25646338)

trans fatty acid measurement **you have 1 copies of the risk allele C**

(risk allele=C, Cis/trans-18:2, EA odds ratio/beta 0.0035 [0.0027-0.0043] unit decrease with pval 5E-15, pubmedid=25646338)

trans-16:1n-7 fatty acid measurement **you have 1 copies of the risk allele C**

(risk allele=C, Cis/trans-18:2, EA odds ratio/beta 0.0035 [0.0027-0.0043] unit decrease with pval 5E-15, pubmedid=25646338)

trans/cis-18:2 fatty acid measurement **you have 1 copies of the risk allele C**

(risk allele=C, Cis/trans-18:2, EA odds ratio/beta 0.0035 [0.0027-0.0043] unit decrease with pval 5E-15, pubmedid=25646338)

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trans/trans-18:2 fatty acid measurement **you have 1 copies of the risk allele C**

(risk allele=C, Cis/trans-18:2, EA odds ratio/beta 0.0035 [0.0027-0.0043] unit decrease with pval 5E-15, pubmedid=25646338)

Gene related topics:

metabolism(1.00) · METABOLIC(0.57) · serum metabolites(0.57) · phospholipids(0.16) · fasting glucose related traits(0.15) · lipoproteins ldl(0.06)

Relevant papers and external links:

PMID 20864672
Genetic variants influencing circulating lipid levels and risk of coronary artery disease.

PMID 24816252
An atlas of genetic influences on human blood metabolites.

PMID 25646338
Genetic loci associated with circulating phospholipid trans fatty acids: a meta-analysis of genome-wide association studies from the charge consortium.

PMID 23303382
Genome-wide association study of serum albumin:globulin ratio in korean populations.

PHARMGKB PA164740318
Evidence: PubMed ID:19043545; Web Resource:External Link results: Genetics Meets Metabolomics: A Genome-Wide Association Study of Metabolite Profiles in Human Serum. (Initial Sample Size: 284 males; Replication Sample Size: NR); (Region: 11q12.2; Reported Gene(s): FADS1; Risk Allele: rs174548-?); (p-value= 0.00000005).This variant is associated with Serum metabolites.

dbSNP: rs174548 GWAS Catalog: rs174548 Genecard: FADS1 chromosome 11

SNPedia: rs174548(C;G)

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★ **rs6601764(C;T)** **info:0.97** **44.66%** **1.16x increased risk of developing Crohn's disease**

rs6601764 has been reported in a large study to be associated with Crohn's disease.

The risk allele (oriented to the dbSNP entry) is (C); the odds ratio associated with heterozygotes is 1.16 (CI 1.01-1.33), and for homozygotes, 1.52 (CI 1.28-1.80).

Established associations:

crohn's disease **you have 1 copies of the risk allele C**

(risk allele=C, odds ratio/beta 1.16 [1.01-1.33] with pval 9E-6, pubmedid=17554300)

Relevant papers and external links:

PMID 17554300

dbSNP: rs6601764 GWAS Catalog: rs6601764 Genecard: nan chromosome 10

SNPedia: rs6601764(C;T)

★ **rs13316193(C;T)** **info:0.95** **48.05%**

The oxytocin receptor (OXTR) contributes to prosocial fund allocations in the dictator game and the social value orientations task.

Evidence that genetic variation in the oxytocin receptor (OXTR) gene influences social cognition in ADHD.

The association between oxytocin receptor gene (OXTR) polymorphisms and affective temperaments, as measured by TEMPS-A.

Gene related topics:

adhd(1.00) · PSYCH(0.28) · autism(0.28)

Relevant papers and external links:

PMID 19461999
PMID 20347913
PMID 20488544

dbSNP: rs13316193 Genecard: OXTR chromosome 3 SNPedia: rs13316193(C;T)

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★ rs7807268(C;C) info:0.89 23.66% **1.4x risk for Crohn's disease**

rs7807268 has been reported in a large study to be associated with Crohn's disease.

The risk allele (oriented to the dbSNP entry) is (G); the odds ratio associated with heterozygotes is 1.38 (CI 1.20-1.60), and for homozygotes, 1.47 (CI 1.24-1.74).

Note: there is a slight amount of ambiguity over the orientation of this SNP information relative to the dbSNP entry.

Established associations:

crohn's disease **you have 0 copies of the risk allele G**
(risk allele=G, odds ratio/beta 1.38 [1.20-1.60] with pval 4E-6, pubmedid=17554300)

Relevant papers and external links:

PMID 17554300

dbSNP: rs7807268 GWAS Catalog: rs7807268 Genecard: nan chromosome 7

SNPedia: rs7807268(C;C)

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★ rs1800795(G;G) info:0.83 73.76% **more IL6; certain risks, see details**

rs1800795 is a SNP in the promoter of the interleukin-6 IL6 gene, affecting the levels made of this important cytokine. In the literature, it is almost universally referred to as the IL6 "-174" polymorphism. It tends to be quite polymorphic in Caucasians, but Asian and African populations are almost monomorphic (for the (G) allele).

The rs1800795(G) allele, generally associated with higher levels of IL6, has been associated with increased risk in the following studies:

The rs1800795(G) allele was significantly associated with type-2 diabetes (odds ratio 1.51, CI: 1.11-2.07, p=0.0096) in a study of 700 elderly Caucasians.[PMID 15472205]

Following a kidney transplant, patients with rs1800795(G;G) genotypes have a higher risk of new-onset diabetes and higher C-reactive protein levels) compared to the (C;C) genotype. Since this was more pronounced in overweight patients, the authors suggest that diabetes-inducing drug

administration should be limited in overweight patients who are rs1800795(G;G) following renal transplantation.[PMID 16837641]

The development of bronchiolitis obliterans syndrome (BOS) after lung transplantation is more likely and happens earlier for rs1800795(G) carriers.[PMID 12451269]

Among HIV+ men, the lifetime risk of developing Kaposi sarcoma is higher for rs1800795(G;G) homozygotes compared to (C;C) homozygotes.[PMID 11001912]

Patients with Hodgkin's lymphoma who are rs1800795(G;G) were less likely to be successfully treated, with odds ratios for failure of 1.75 (CI: 1.04-2.92, p=0.03).[PMID 17496310]

A study of 139 elderly males with acute coronary syndrome (ACS) indicated that rs1800795(G;G) genotypes were at 3.89 fold (CI: 1.71-8.86, p=0.001) higher risk of dying within one year of their ACS event than (C;G) or (C;C) genotypes.[PMID 16098388]

Although the odds of having a stroke weren't different, among patients (in this case, under 60 years of age) who did have a stroke those with a rs1800795(G;G) genotype had more severe disability after 1 week (odds ratio 3.2, CI: 1.5-6.6, p=0.002).[PMID 14512079]

Among Chinese patients with hypertension, the rs1800795(G) allele is more common, and (G;G) genotypes had significantly higher plasma PAI-1 activity than (C;C) genotypes.[PMID 15831362]

In 168 Brazilian patients, rs1800795(G) allele frequency was higher in gastric cancer than in patients with chronic gastritis. [PMID 17560462]

Although the odds of having Crohn's disease aren't affected, among 153 children with it, those with the rs1800795(G;G) genotype were more growth-retarded at diagnosis than (C;G) or (C;C) genotypes. (G;G) patients also had higher circulating levels of C-reactive protein (CRP).[PMID 16150725]

rs1800795(G) carriers don't increase their high-density lipoprotein (HDL) levels after 24 weeks of aerobic exercise as much as rs1800795(C) carriers.[PMID 15904871]

The rs1800795(G;G) genotype was more frequent in Australian infants with sudden infant death syndrome (58%) than in control subjects (38%, p=0.02).[PMID 17055359] This was not seen in a Norwegian population.[PMID 17509454]

Women with endometriosis who are rs1800795(G) carriers appear to be more likely to develop chocolate cysts, and (G) allele carriers may be at slightly higher risk of developing endometriosis. [PMID 12517591]

The rs1800795(G) allele is more frequent in women with hyperandrogenism than in controls.[PMID 11889177]
rs1800795 is a SNP in the promoter of the interleukin-6 IL6 gene, affecting the levels made of this

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important cytokine. In the literature, it is almost universally referred to as the IL6 "-174" polymorphism. It tends to be quite polymorphic in Caucasians, but Asian and African populations are almost monomorphic (for the (G) allele).

It was first described in 1998, when it was shown that the rs1800795(C) allele produces less IL6 than the (G) allele, which supported the hypothesis that a protective genotype against systemic onset juvenile rheumatoid arthritis would be rs1800795(C;C), and indeed, few juvenile RA patients had that genotype.

Studies on rs1800795 now include potential associations with heart disease, Kaposi's sarcoma, type-2 diabetes, stroke, obesity, Hodgkin's lymphoma, sudden infant death syndrome, cancer (including breast cancer, gastric cancer, prostate cancer), hypertension, periodontitis, and complications arising after organ transplantations or grafts. It is worth noting that some of the associations seen for rs1800795 alleles are not for the risk of developing a given disorder; instead, they relate to how a disorder might progress or respond to various treatments. Summaries of several major associations are available at OMIM.

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A study of 139 elderly males with acute coronary syndrome (ACS) indicated that rs1800795(G;G) genotypes were at 3.89 fold (CI: 1.71-8.86, p=0.001) higher risk of dying within one year of their ACS event than (C;G) or (C;C) genotypes.

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Women with endometriosis who are rs1800795(G) carriers appear to be more likely to develop chocolate cysts, and (G) allele carriers may be at slightly higher risk of developing endometriosis.

The rs1800795(G) allele is more frequent in women with hyperandrogenism than in controls.

Children with the rs1800795(G;G) genotype are more likely to have recurrent acute otitis, based on a study of 348 patients, with an odds ratio of 1.64 (p=0.02).

(G) allele is increased risk ((C) allele is decreased risk) for type-2 diabetes; this SNP is one of 4 relatively common SNPs reported to represent risk for type-2 diabetes in the DESIR prospective study of 3,877 Caucasian participants. Under an additive model, the odds ratio for rs1800795(C) carriers is 0.79 (CI: 0.65-0.96, p=0.01). For the 4 SNPs, each risk allele increased type-2 diabetes risk by 1.34x (p=2x10e-6), with an odds ratio of 2.48 (CI: 1.59-3.86) for carriers of 4 or more compared to those with one or none (risk alleles).

The rs1800795(C) allele, generally associated with lower levels of IL6, has been associated with increased risk in these studies:

rs1800795 (C;C) and (C;G) Caucasians who are excessively heavy (body mass index ~33 +/- 5kg/m2) are at increased risk (odds ratio 5.2, p=0.003) for developing obesity-related metabolic disorders such as hypertension, atherogenic dyslipidemia, and insulin resistance.

Among 571 patients with type-2 diabetes, the rs1800795(C) allele is correlated with higher body mass index, but it showed no correlation among non-diabetics.

A study of 238 Caucasians with type-2 diabetes concluded that rs1800795(C) carriers have an insulin resistance "IL-6-sensitive" phenotype and perhaps strategies to manage insulin resistance should be different between (C) carriers vs. (G;G) patients.

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In patients with end-stage renal disease (ESRD) being treated by hemodialysis, rs1800795(C) allele carriers had higher diastolic blood pressure (p=0.008) and a higher left ventricular mass index (p=0.026) than (G;G) homozygotes. Among diabetic patients in dialysis, the prevalence of left ventricular hypertrophy in (C) allele carriers was 87.5% vs. 36.3% among (G;G) genotypes (p= 0.02).

Among recipient of a kidney transplant, rs1800795(C) allele carriers had worse three-year graft survival (71/104 = 68.3%; p=0.0059) with a 3.7-fold increased relative risk of graft loss compared to rs1800795(G;G) genotypes (48/54 = 88.9%). The authors suggest that determining the rs1800795 genotype may offer a new method for identifying patients at increased risk of allograft loss.

Among prostate cancer patients, the rs1800795(C) allele is associated with more aggressive cancer and higher prostate-specific antigen levels compared to rs1800795(G;G) homozygotes.

Although the rs1800795(C) allele was not associated with a higher frequency of heart attacks, it did have an association with inflammation and infarcts detectable only by MRI, suggesting that rs1800795(C) may chronically predispose an individual to develop atherosclerosis.

The combination of carrying at least one rs1800795(C) and one rs1205(T), a SNP in the C-reactive protein gene, led to higher risk of a stroke after cardiac surgery (odds ratio 3.3, CI: 1.4-8.1, p=0.0023) compared to individuals who were rs1800795(G;G) and rs1205(C;C).

The rs1800795(C) allele was associated with higher postoperative IL6 levels and a less favorable clinical outcome following coronary revascularization surgery.

The rs1800795(C;C) genotype was significantly higher in the group of 122 adult periodontitis patients than in controls (odds ratio 1.896, CI: 1.1-3.2, p=0.0283).

The degree of periodontitis was more severe in rs1800795(C) carriers than (G) carriers.

The rs1800795(C) allele was over-represented in patients with Alzheimer's disease compared to controls and the (C;C) genotype was associated with higher risk in women.

The rs1800795(C) allele was higher in non-survivors compared to (nonagenarian) survivors in a case/control study of ~100 pairs of matched Finnish individuals; to put it another way, the rs1800795(G) allele may be associated with increased longevity.

rs1800797, rs1800796 and rs1800795 have been shown to affect both the transcription and secretion of IL-6, to symptomatic distal interphalangeal osteoarthritis based on 535 women. the G alleles of two promoter single-nucleotide polymorphisms (SNP) G-597A and G-174C were more common among the subjects with symptomatic DIP OA than among those with no disease (p-values corrected for multiple testing 0.020 and 0.024). Also, the carriage of at least one G allele in these positions increased the risk of disease (p=0.006 and, p=0.008, respectively). Carrying a haplotype with the G allele in all three promoter SNPs increased the risk of symptomatic DIP OA more than four-fold (OR 4.45, p=0.001). Carriage of the G-G diplotype indicated an increased risk of both symmetrical DIP OA (OR 1.52 95% CI 1.01 to 2.28) and symptomatic DIP OA (OR 3.67 95% CI 1.50 to 9.00)

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Suggests that "healthy elderly individuals may have a proinflammatory profile playing as a downregulating factor for inducible Hsp70, particularly if -174 G/C-negative" and that therefore, a nutraceutical intervention (fermented papaya preparation 9 g/day) might be of benefit.

rs1800795 coronary artery disease 190 affected Asian Indian sibling pairs

Gene related topics:

METABOLIC(8.00) · bone mineralization(8.00) · UNKNOWN(3.00) · inflammation myocardial infarction(3.00) · CARDIOVASCULAR(2.00) · IMMUNE(2.00) · arterial occlusive diseases peripheral vascular diseases(2.00) · critical illness multiple organ failure systemic inflam response synd systemic inflammatory response syndrome(2.00) · endotoxemia(2.00) · giant cell arteritis polymyalgia rheumatica temporal arteritis(2.00) · gingival overgrowth(2.00) · inflamatory bowel disease(2.00) · juvenile rheumatoid arthritis(2.00) · REPRODUCTION(1.80) · cardiovascular diseases inflammation(1.80) · obstetric labor premature(1.80) · RENAL(1.00) · kidney failure(1.00) · pneumoconiosis(1.00) · gastric cancer(1.00) · survival(1.00) · lymphoma(1.00) · left ventricular hypertrophy(1.00) · insulin resistance(1.00) · prostate cancer(1.00) · lipoprotein(1.00) · left ventricular mass(1.00) · stroke(1.00) · blood pressure(1.00) · body mass(1.00) · cancer(1.00) · breast cancer(1.00) · hypertension(1.00) · diastolic blood pressure(1.00) · hiv(1.00) · heart disease(1.00) · osteoarthritis(1.00) · coronary artery disease(1.00) · body mass index(1.00) · insulin(1.00) · gastritis(1.00) · inflammation(0.91) · stomatitis aphthous(0.80) · preterm delivery(0.76) · INFECTION(0.72) · sepsis(0.72) · periodontitis(0.72) · kawasaki disease(0.67) · respiratory syncytial virus bronchiolitis(0.67) · coronary disease coronary heart disease(0.58) · CANCER(0.58) · multiple myeloma(0.58) · coronary artery disease inflammation(0.57) · otitis media recurrence(0.57) · kidney transplant complications(0.53) · kidney transplant(0.50) · renal allograft outcome(0.50) · AGING(0.46) · longevity(0.46) · allograft outcome(0.40) · bone marrow transplantation(0.40) · bronchiolitis obliterans syndrome(0.40) · graft vs host disease(0.40) · NEUROLOGICAL(0.38) · alzheimer s disease(0.38) · type 2 diabetes(0.37) · juvenile arthritis(0.36) · brucellosis(0.36) · bone mineral density(0.35) · asthma eczema allergic disease(0.33) · sepsis systemic infection(0.32) · chronic periodontitis(0.31) · metabolic syndrome x(0.31) · premature birth(0.31) · pulmonary fibrosis(0.27) · periodontal disease(0.26) · graves disease graves disease(0.25) · hodgkin disease(0.25) · kidney failure chronic(0.25) · sudden infant death(0.25) · myocardial infarction(0.25) · intracranial aneurysm(0.22) · peripheral vascular diseases(0.21) · coronary heart disease(0.20) · respiratory syncytial virus(0.20) · pulmonary disease chronic obstructive(0.18) · pregnancy loss recurrent(0.16) · bone density(0.16) · obesity(0.16) · multiple sclerosis(0.15) · aortic aneurysm abdominal(0.15) · cardiovascular diseases(0.14) · pancreatitis chronic(0.14) · skin cancer non melanoma(0.14) · acute coronary syndrome(0.14) · rheumatoid arthritis(0.13) · heart disease ischemic(0.13) · apoplexy brain ischemia stroke(0.13) · arthritis(0.12) · irritable bowel syndrome(0.12) · osteoporosis postmenopausal(0.12) · graft versus host disease(0.12) · pre eclampsia(0.12) · hepatitis b(0.10) · PSYCH(0.10) · mood disorders(0.10) · atherosclerosis(0.09) · NORMALVARIATION(0.09) · normal variation(0.09) · sarcoidosis(0.09) · OTHER(0.09) · cerebral palsy(0.09) · endometriosis(0.08) · ankylosing spondylitis(0.08) · hepatitis c chronic(0.08) · celiac disease(0.07) · crohn disease(0.06) · myocardial infarct(0.06) · systemic lupus erythematosus(0.06) · coronary disease(0.05) · chronic renal failure kidney failure chronic(0.05) · diabetes mellitus insulin dependent diabetes mellitus type 1(0.05) · stroke ischemic(0.05) · atopy(0.05) · hepatitis b chronic(0.04) · hepatitis c(0.04) · arthritis rheumatoid(0.03) · diabetes mellitus(0.03) · lupus erythematosus systemic systemic lupus erythematosus(0.03) · esophageal cancer(0.03) · melanoma(0.03) · cervical cancer(0.02) · cystic fibrosis(0.02) · neuroblastoma(0.02)

Relevant papers and external links:

PMID 9769329

PMID 15472205

PMID 16837641

PMID 12451269

PMID 11001912

PMID 17496310

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PMID 16098388
PMID 14512079
PMID 15831362
PMID 17560462
PMID 16150725
PMID 15904871
PMID 17055359
PMID 17509454
PMID 12517591
PMID 11889177
PMID 17908769
PMID 17977958
PMID 17998015
PMID 15172007
PMID 16140413
PMID 12846758
PMID 12371985
PMID 16006970
PMID 12482836
PMID 16051899
PMID 16183563
PMID 17209781
PMID 17286759
PMID 12928051
PMID 15664628
PMID 18257935
PMID 18056967
PMID 18449426
OMIM 147620

dbSNP: rs1800795 Genecard: IL6 chromosome 7 SNPedia: rs1800795(G;G)

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★ **rs4504469(C;T)** **info:0.82** **32.70%** **1.5x risk**

Rs4504469, a nonsynonymous SNP in exon 4 of the KIAA0319 gene, is in a region that crops up in several independent studies as likely to associated with dyslexia. The risk allele in the Caucasian populations studied is (T).

The odds ratio in a study of case:control study of ~400 Caucasians associated with rs4504469(T) is 1.51 (CI: 1.17â€“1.95, p = 0.002).

A 77-kilobase region of chromosome 6p22.2 is associated with dyslexia in families from the United Kingdom and from the United States.

Strong genetic evidence of DCDC2 as a susceptibility gene for dyslexia.

A common variant associated with dyslexia reduces expression of the KIAA0319 gene.

Convergent genetic linkage and associations to language, speech and reading measures in families of probands with Specific Language Impairment.

Gene related topics:

NEUROLOGICAL(1.19) · dyslexia(1.19)

Relevant papers and external links:

PMID 15717286
OMIM 609269
OMIM 600202
PMID 15514892
PMID 16385449
PMID 19325871
PMID 19997522

dbSNP: rs4504469 Genecard: KIAA0319 chromosome 6 SNPedia: rs4504469(C;T)

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★ rs5751876(C;C) good:0.79 19.56% **None**

Variation at this SNP, rs5751876, also referred to as 1976C>T or 1976T>C, appears to influence how an individual responds to moderate levels (150mg) of caffeine.

T/T carriers displaying significantly increased subjective levels of anxiety after consumption.

However a PharmGKB variant annotation suggests it is (C;C) genotypes who are more sensitive to caffeine.

rs5751876 is not associated with how much coffee one drinks, though, nor with risk for Parkinson's disease, based on a study of 1,200 subjects.

"rs2298383 shows functional potential based on in silico analyses and might therefore represent the true underlying causal variant"

Gene related topics:

PSYCH(2.00) · caffeine induced anxiety(2.00) · anxiety(1.00) · panic disorder(0.21)

Relevant papers and external links:

PMID 12825092
PMID 18305461
PMID 18759349

dbSNP: rs5751876 Genecard: ADORA2A chromosome 22 SNPedia: rs5751876(C;C)

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★ rs17388568(A;G) info:0.78 24.16% **1.3x risk of type-1 diabetes**

rs17388568 has been reported in a large study to be associated with type-1 diabetes.

The risk allele (oriented to the dbSNP entry) is (A); the odds ratio associated with heterozygotes is 1.26 (CI 1.11-1.42), and for homozygotes, 1.58 (CI 1.27-1.95).

In an expanded follow-up study of >6,000 controls and 6,000 patients, the heterozygote odds ratio for this SNP was recalculated to be 1.11 (CI 1.05â€“1.18).

Established associations:

autoantibody measurement **you have 1 copies of the risk allele A**

(risk allele=A, T1D odds ratio/beta 1.1 with pval 6E-6, pubmedid=21829393)

type i diabetes mellitus **you have 1 copies of the risk allele A**

(risk allele=A, odds ratio/beta 1.26 [1.11-1.42] with pval 3E-6, pubmedid=17554300; risk allele=A, T1D odds ratio/beta 1.1 with pval 6E-6, pubmedid=21829393)

ulcerative colitis **you have 1 copies of the risk allele A**

(risk allele=A, odds ratio/beta 1.12 [1.07-1.17] with pval 9E-7, pubmedid=21297633)

allergy **you have 1 copies of the risk allele A**

(risk allele=A, odds ratio/beta 0.0727 [0.047-0.099] unit increase with pval 4E-8, pubmedid=23817569)

Relevant papers and external links:

PMID 21297633
Meta-analysis identifies 29 additional ulcerative colitis risk loci, increasing the number of confirmed associations to 47.

PMID 21829393
Genome-wide association analysis of autoantibody positivity in type 1 diabetes cases.

PMID 23817569
A genome-wide association meta-analysis of self-reported allergy identifies shared and allergy-specific susceptibility loci.

PMID 17554300

PMID 17554260

OMIM 612622

OMIM 222100

PHARMGKB PA162356644
Evidence: PubMed ID:17554300; Web Resource:External Link Results: Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls (Initial Sample Size: 1,963 cases, 2,938 controls; Replication Sample Size: (see Todd 2007); Risk Allele: rs17388568-A). This variant is associated with type 1 diabetes.

dbSNP: rs17388568 GWAS Catalog: rs17388568 Genecard: ADAD1 chromosome 4

SNPedia: rs17388568(A;G)

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★ rs182549(C;T) good:0.75 27.33% **Can digest milk.**

Also known as "G/A(-22018)" and located in the MCM6 but with influence on the lactase LCT gene, rs182549 is one of two SNPs that is associated with the primary haplotype associated with hypolactasia, more commonly known as lactose intolerance in European Caucasian populations. [PMID 11788828, PMID 15114531]

In these populations, the rs182549(C) allele (as named in accordance with dbSNP) is both the more common allele and the one associated with lactose intolerance.

In populations of sub-Saharan Africans, though, the rs182549(C) allele is unlikely to be predictive of lactose intolerance, and other SNPs are predictive instead. [PMID 15106124, PMID 17159977]

See also OMIM 601806.0002

Gender differences in genetic risk profiles for cardiovascular disease.

Natriuretic peptide system gene variants are associated with ventricular dysfunction after coronary artery bypass grafting.

Ancestry analysis in the 11-M Madrid bomb attack investigation.

Variation in the 4q25 chromosomal locus predicts atrial fibrillation after coronary artery bypass graft surgery.

Gene related topics:

cardiovascular disease(1.00) · CARDIOVASCULAR(1.00) · atrial fibrillation(1.00) · hypolactasia(1.00) · OTHER(1.00) · cardiovascular(1.00) · METABOLIC(0.19) · lactose intolerance(0.19)

Relevant papers and external links:

PHARMGKB PA165363256
Evidence: PubMed ID:15114531Phenotype: This SNP is part of a haplotype that extends for > 1 Mb and includes rs4988235. the the A allele of rs182549 and the T allele of rs4988235 are associated with lactase persistance. The haplotype appears to be under strong recent positive selection in Northern- European derived populations. In European Americans, African Americans and East Asians, the alleles associated with lactase persistance were found to occur at frequencies roughly matching the rates of lactase persistance. Study population/ethnicity: European American, African American, East Asian, Scandinavian. Type of association: CO;GN.

OMIM 601806
PMID 18974842
PMID 19326473
PMID 19668368
PMID 20031626

dbSNP: rs182549 Genecard: MCM6 chromosome 2 SNPedia: rs182549(C;T)

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★ rs4994(T;T) good:0.70 78.32% **normal**

rs4994, also known as Trp64Arg or as the Arg64 variant, is a SNP in the ADRB3 gene. The rs4994 (C) allele encodes the variant arginine.

In a study of 600+ women initially referred for coronary angiography and then followed for 6 years, during which time 115 experienced a heart problem, rs4994(C) individuals were calculated to be at higher risk (adjusted hazard ratio 2.10, CI: 1.05-4.24). This risk was due to subtle increases in risk for all of the individual endpoints, and was only seen in women without obstructive coronary artery disease.

Response to Diet and Exercise

Gene related topics:

METABOLIC(2.00) · insulin resistance metabolic syndrome x(2.00) · obesity(1.84) · obesity weight loss(1.29) · hypertension obesity weight gain(1.00) · coronary artery disease(1.00) · angiography(1.00) · insulin resistance obesity(0.69) · hypertension obesity(0.57) · obesity morbid(0.33) · insulin resistance(0.32) · body mass(0.24) · diabetes type 2(0.21) · metabolic syndrome x(0.17) · CANCER(0.10) · endometrial cancer(0.10) · weight gain(0.10) · metabolic syndrome(0.09)

Relevant papers and external links:

PMID 18331634

OMIM 109691

PHARMGKB PA164889041
Evidence: PubMed ID:19193342Arg in position 64 associated with olanzapine-induced weight gain

dbSNP: rs4994 Genecard: ADRB3 chromosome 8 SNPedia: rs4994(T;T)

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★ rs1535(A;G) good:0.69 43.66% **4+ IQ points for breastfeeding**

4+ IQ points for breastfeeding with the AA or AG genotypes

Breastfeeding and IQ

Established associations:

metabolic syndrome **you have 1 copies of the risk allele G**

(risk allele=G, HDL odds ratio/beta 0.03 [0.02-0.04] mmol/l decrease with pval 4E-7, pubmedid=20694148)

response to statin **you have 1 copies of the risk allele G**

(risk allele=G complex/no impact summary)

cis/trans-18:2 fatty acid measurement **you have 1 copies of the risk allele A**

(risk allele=A, Cis/trans-18:2, EA odds ratio/beta 0.0032 [0.0024-0.004] unit decrease with pval 5E-14, pubmedid=25646338)

total trans-18:1 fatty acid measurement **you have 1 copies of the risk allele A**

(risk allele=A, Cis/trans-18:2, EA odds ratio/beta 0.0032 [0.0024-0.004] unit decrease with pval 5E-14, pubmedid=25646338)

trans fatty acid measurement **you have 1 copies of the risk allele A**

(risk allele=A, Cis/trans-18:2, EA odds ratio/beta 0.0032 [0.0024-0.004] unit decrease with pval 5E-14, pubmedid=25646338)

trans-16:1n-7 fatty acid measurement **you have 1 copies of the risk allele A**

(risk allele=A, Cis/trans-18:2, EA odds ratio/beta 0.0032 [0.0024-0.004] unit decrease with pval 5E-14, pubmedid=25646338)

trans/cis-18:2 fatty acid measurement **you have 1 copies of the risk allele A**

(risk allele=A, Cis/trans-18:2, EA odds ratio/beta 0.0032 [0.0024-0.004] unit decrease with pval 5E-14, pubmedid=25646338)

trans/trans-18:2 fatty acid measurement **you have 1 copies of the risk allele A**

(risk allele=A, Cis/trans-18:2, EA odds ratio/beta 0.0032 [0.0024-0.004] unit decrease with pval 5E-14, pubmedid=25646338)

phospholipid measurement **you have 1 copies of the risk allele A**

(risk allele=A, ALA odds ratio/beta 0.02 % decrease with pval 3E-63, pubmedid=21829377; risk allele=A, DPA odds ratio/beta 0.07 % increase with pval 3E-152, pubmedid=21829377)

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Gene related topics:

METABOLIC(0.36) · phospholipids(0.36) · hdl cholesterol(0.08) · lipoproteins ldl(0.06) · ldl cholesterol(0.06) · PSYCH(0.02) · adhd attention deficit hyperactivity disorder(0.02)

Relevant papers and external links:

PMID 20339536
Genome-wide association of lipid-lowering response to statins in combined study populations.

PMID 20694148
A genome-wide association study of the metabolic syndrome in indian asian men.

PMID 21829377
Genetic loci associated with plasma phospholipid n-3 fatty acids: a meta-analysis of genome-wide association studies from the charge consortium.

PMID 25646338

Genetic loci associated with circulating phospholipid trans fatty acids: a meta-analysis of genome-wide association studies from the charge consortium.

PMID 17984066

dbSNP: rs1535 GWAS Catalog: rs1535 Genecard: FADS2 chromosome 11

SNPedia: rs1535(A;G)

★ rs11827962(C;C) info:0.66 96.44% **; schizophrenia (you have the risk allele C)**

Established associations:

schizophrenia **you have 2 copies of the risk allele C**

(risk allele=C, 5 degree of freedom test odds ratio/beta 1.044 [0.94-1.15] with pval 6E-6, pubmedid=23453885)

bipolar disorder **you have 2 copies of the risk allele C**

(risk allele=C, 5 degree of freedom test odds ratio/beta 1.044 [0.94-1.15] with pval 6E-6, pubmedid=23453885)

unipolar depression **you have 2 copies of the risk allele C**

(risk allele=C, 5 degree of freedom test odds ratio/beta 1.044 [0.94-1.15] with pval 6E-6, pubmedid=23453885)

attention deficit hyperactivity disorder **you have 2 copies of the risk allele C**

(risk allele=C, 5 degree of freedom test odds ratio/beta 1.044 [0.94-1.15] with pval 6E-6, pubmedid=23453885)

autism spectrum disorder **you have 2 copies of the risk allele C**

(risk allele=C, 5 degree of freedom test odds ratio/beta 1.044 [0.94-1.15] with pval 6E-6, pubmedid=23453885)

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Relevant papers and external links:

PMID 23453885

Identification of risk loci with shared effects on five major psychiatric disorders: a genome-wide analysis.

dbSNP: rs11827962 GWAS Catalog: rs11827962 Genecard: nan chromosome 11

SNPedia: rs11827962(C;C)

★ rs6596075(C;C) info:0.59 55.00% **2x risk of Crohn's disease**

rs6596075 has been reported in a large study to be associated with Crohn's disease.

The risk allele (oriented to the dbSNP entry) is (C); the odds ratio associated with heterozygotes is 1.55 (CI 1.00-2.39), and for homozygotes, 2.06 (CI 1.35-3.14).

Established associations:

crohn's disease **you have 2 copies of the risk allele C**

(risk allele=C, odds ratio/beta 1.55 [1.00-2.39] with pval 3E-6, pubmedid=17554300)

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Gene related topics:

IMMUNE(0.59) · colitis ulcerative crohn disease(0.59) · crohn s disease ulcerative colitis(0.09) · crohn disease(0.03) · crohn s disease(0.02)

Relevant papers and external links:

PMID 17554300

OMIM 606348

dbSNP: rs6596075 GWAS Catalog: rs6596075 Genecard: nan chromosome 5

SNPedia: rs6596075(C;C)

★ rs3024505(C;C) info:0.37 83.49% **; type i diabetes mellitus (you have the risk allele C)**

Established associations:

ulcerative colitis **you have 0 copies of the risk allele T**

(risk allele=T, odds ratio/beta 1.23 with pval 1E-8, pubmedid=20228799; risk allele=T, odds ratio/beta 1.46 [1.31-1.62] with pval 1E-12, pubmedid=18836448; risk allele=A, odds ratio/beta 1.25 [1.19-1.32] with pval 6E-17, pubmedid=21297633)

crohn's disease **you have 0 copies of the risk allele T**

(risk allele=T, odds ratio/beta 1.12 [1.07-1.17] with pval 2E-14, pubmedid=21102463)

type i diabetes mellitus **you have 2 copies of the risk allele C**

(risk allele=C, T1D odds ratio/beta 1.2 with pval 5E-10, pubmedid=21829393)

autoantibody measurement **you have 2 copies of the risk allele C**

(risk allele=C, T1D odds ratio/beta 1.2 with pval 5E-10, pubmedid=21829393)

inflammatory bowel disease **you have 0 copies of the risk allele A**

(risk allele=A, odds ratio/beta 1.208 [1.163-1.254] with pval 7E-42, pubmedid=23128233)

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Gene related topics:

CANCER(2.00) · IMMUNE(2.00) · OTHER(2.00) · RENAL(2.00) · UNKNOWN(2.00) · acute and chronic kidney transplant outcome(2.00) · cachexia stomach neoplasms(2.00) · epstein barr virus infection(2.00) · fibrosis hepatitis c(2.00) · focal segmental glomsclerosis glomerulonephritis iga glomerulosclerosis focal segmental iga glomerulonephritides(2.00) · gastric carcinoma(2.00) · gingivitis(2.00) · INFECTION(1.80) · parvovirus b19 infection(1.80) · chronic ulcerative colitis colitis ulcerative crohn disease crohn s disease inflammatory bowel disease nos inflammatory bowel diseases(1.60) · lichen planus oral(1.00) · liver disease alcoholic(1.00) · tuberculosis tuberculosis pulmonary(1.00) · sepsis systemic infection(0.89) · graft versus host disease(0.83) · lupus erythematosus systemic systemic lupus erythematosus(0.74) · sjogren s syndrome(0.74) · eosinophilia(0.67) · brucellosis(0.64) · dermatitis atopic eczema allergic(0.60) · heart transplant complications(0.57) · helicobacter infections stomach neoplasms(0.56) · sudden infant death(0.56) · hepatitis b(0.56) · kidney diseases(0.55) · kidney transplant complications(0.53) · tuberculosis pulmonary(0.51) · kidney transplant(0.50) · renal allograft outcome(0.50) · melanoma skin neoplasms(0.49) · tuberculosis(0.47) · AGING(0.46) · longevity(0.46) · NEUROLOGICAL(0.44) · guillain barre syndrome(0.44) · kidney cancer(0.44) · scleroderma systemic systemic scleroderma(0.44) · hepatitis c chronic(0.43) · systemic lupus erythematosus(0.42) · allograft outcome(0.40) · bone marrow transplantation(0.40) · bronchiolitis obliterans syndrome(0.40) · lymphoma large b cell diffuse(0.40) · graft vs host disease(0.40) · hepatitis b chronic(0.39) · lupus(0.38) · juvenile arthritis(0.36) · asthma eczema allergic disease(0.33) · sepsis(0.32) · carcinoma squamous cell mouth neoplasms(0.31) · dengue hemorrhagic fever(0.31) · periodontitis(0.30) · stomach neoplasms(0.29) · meningococcal disease(0.29) · irritable bowel syndrome(0.28) · arthritis rheumatoid rheumatoid arthritis(0.28) · acquired immunodeficiency syndrome(0.27) · hepatitis c(0.25) · bronchiolitis viral respiratory syncytial virus infections(0.25) · helicobacter infections(0.24) · ulcerative colitis(0.24) · malaria falciparum(0.23) · asthma(0.22) · mucocutaneous lymph node syndrome(0.22) · arthritis psoriatic psoriatic arthropathy(0.21) · chronic ulcerative colitis colitis ulcerative(0.21) · REPRODUCTION(0.21) · pre eclampsia(0.21) · respiratory syncytial virus(0.20) · inflammatory bowel disease nos inflammatory bowel diseases(0.19) · colitis ulcerative(0.19) · lymphoma non hodgkin(0.18) · endometriosis(0.18) · CARDIOVASCULAR(0.18) · cardiovascular(0.18) · leprosy(0.18) · multiple sclerosis(0.18) · behcet syndrome(0.17) · arthritis juvenile rheumatoid chronic childhood arthritis(0.17) · h pylori infection(0.17) · stomach cancer(0.16) · aortic aneurysm abdominal(0.15) · pancreatitis chronic(0.14) · kidney failure chronic(0.14) · pemphigus(0.14) · skin cancer non melanoma(0.14) · liver cancer(0.13) · celiac disease(0.13) · chronic renal failure kidney failure chronic(0.12) · psoriasis(0.12) · periodontal disease(0.12) · graves disease graves disease(0.11) · hiv(0.11) · multiple myeloma(0.11) · leukemia lymphocytic chronic b cell(0.11) · lymphoma non hodgkin lymphoma non hodgkin s(0.11) · PSYCH(0.10) · mood disorders(0.10) · rheumatoid arthritis(0.10) · pregnancy loss recurrent(0.09) · NORMALVARIATION(0.09) · normal variation(0.09) · sarcoidosis(0.09) · abortion habitual(0.08) ·

premature birth(0.08) · chronic obstructive pulmonary disease copd(0.07) · hiv infections x human immunodeficiency virus disease(0.07) · pancreatitis(0.07) · atherosclerosis(0.07) · lymphoma(0.06) · lupus erythematosus(0.06) · inflammation(0.06) · arthritis(0.06) · cystic fibrosis(0.05) · vitiligo(0.05) · hiv infections(0.05) · atopy(0.05) · preeclampsia(0.05) · lupus erythematosus systemic(0.04) · pregnancy loss(0.04) · crohn disease crohn s disease(0.04) · crohn disease(0.03) · inflammatory bowel disease(0.03) · melanoma(0.03) · cervical cancer(0.02) · diabetes mellitus insulin dependent diabetes mellitus type 1(0.02) · crohn s disease(0.02)

Relevant papers and external links:

PMID 21102463
Genome-wide meta-analysis increases to 71 the number of confirmed crohn's disease susceptibility loci.

PMID 20228799
Genome-wide association identifies multiple ulcerative colitis susceptibility loci.

PMID 18836448
Sequence variants in il10, arpc2 and multiple other loci contribute to ulcerative colitis susceptibility.

PMID 21297633
Meta-analysis identifies 29 additional ulcerative colitis risk loci, increasing the number of confirmed associations to 47.

PMID 21829393
Genome-wide association analysis of autoantibody positivity in type 1 diabetes cases.

PMID 23128233
Host-microbe interactions have shaped the genetic architecture of inflammatory bowel disease.

OMIM 612381

PHARMGKB PA164740884
Evidence: PubMed ID:18836448; Web Resource:External Link variants in IL10, ARPC2 and multiple other loci contribute to ulcerative colitis susceptibility. (Initial Sample Size: 1,167 cases, 777 controls; Replication Sample Size: 1,855 cases, 3,091 controls); (Region: 1q32.1; Reported Gene: IL10; Risk Allele: rs3024505-T) This variant is associated with Ulcerative colitis.

PHARMGKB PA162355781
Evidence: PubMed ID:18836448A genome-wide association study in 1,167 individuals with ulcerative colitis and 777 healthy controls found significant association of this variant immediately flanking the IL10 gene and ulcerative colitis.

PHARMGKB PA164740842
Evidence: PubMed ID:18836448; Web Resource:External Link results: Sequence variants in IL10, ARPC2 and multiple other loci contribute to ulcerative colitis susceptibility. (Initial Sample Size: 1,167 cases, 777 controls; Replication Sample Size: 1,855 cases, 3,091 controls); (Region: 1q32.1; Reported Gene(s): IL10; Risk Allele: rs3024505-T); (p-value= 9.99999999999999E-13).This variant is associated with Ulcerative colitis.

OMIM 222100

dbSNP: rs3024505 GWAS Catalog: rs3024505 Genecard: nan chromosome 1

SNPedia: rs3024505(C;C)

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★ rs2710117(A;T) info:0.34 46.86% **None**

Speech development rs4431523, rs17236239 and significant associations (with P values from 0.01 to 5.0x10–5) between nonsense-word repetition and nine intronic SNPs (rs851715, rs10246256, rs2710102, rs759178, rs1922892, rs2538991, rs17236239, rs2538976, and rs2710117)

Gene related topics:

PSYCH(0.67) · mental disorders(0.67) · OTHER(0.12) · breath tests(0.12) · autism(0.09) · CARDIOVASCULAR(0.03) · echocardiography(0.03) · METABOLIC(0.03) · diabetes mellitus(0.03)

Relevant papers and external links:

PMID 18987363

dbSNP: rs2710117 Genecard: CNTNAP2 chromosome 7 SNPedia: rs2710117(A;T)

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★ rs165599(A;G) info:0.25 49.98%

anxiety-related personality traits, ADHD, schizophrenia

part of a three marker haplotype rs737865-rs4680-rs165599

epistasis between SNPs in COMT (rs2097603, Val158Met (rs4680), rs165599) and polymorphisms in other schizophrenia susceptibility genes

popular in pubmed

is associated with bipolar disorder and influences prefrontal aspects of verbal memory in bipolar patients and healthy controls.

Gene related topics:

UNKNOWN(6.00) · digeorge syndrome pharyngeal pouch syndrome(6.00) · PSYCH(5.72) · schizophrenia(5.72) · CANCER(3.00) · neoplasms pain(3.00) · fibromyalgia(2.67) ·

NEUROLOGICAL(2.00) · mamographic density(2.00) · pain temporomandibular joint disorders(2.00) · psychosis(1.67) · obsessive compulsive disorder(1.31) · CHEMDEPENDENCY(1.07) · substance related disorders(1.07) · panic disorder(1.03) · adhd attention deficit hyperactivity disorder(1.00) · eating disorders(1.00) · anxiety(1.00) · memory(1.00) · pain(0.94) · breast cancer(0.83) · marijuana abuse(0.67) · mood disorders(0.62) · parkinson s disease(0.62) · DEVELOPMENTAL(0.60) · leiomyoma uterine neoplasms(0.60) · mental retardation(0.60) · schizophrenia bipolar disorder(0.60) · OTHER(0.57) · sleep deprivation(0.57) · bipolar disorder(0.50) · depression(0.50) · fractures bone(0.50) · personality(0.38) · mammographic density(0.36) · personality traits(0.33) · heroin abuse(0.30) · alcoholism(0.29) · adhd(0.26) · cognitive ability(0.25) · heroin dependence(0.25) · bulimia(0.24) · endometrial cancer(0.23) · smoking(0.21) · anorexia nervosa(0.15) · suicide(0.14) · liver cancer(0.13) · tobacco use disorder(0.11) · narcolepsy(0.11) · METABOLIC(0.10) · weight gain(0.10) · smoking behavior(0.09) · endometrial neoplasms(0.07) · attention deficit hyperactivity disorder(0.05) · IMMUNE(0.05) · vitiligo(0.05) · autism(0.03) · REPRODUCTION(0.02) · endometriosis(0.02)

Relevant papers and external links:

PMID 17547583

OMIM 104300

OMIM 116790

PHARMGKB PA164888988
Evidence: PubMed ID:19451915In a study of 78 African American and 65 white patients diagnosed with schizophrenia or schizoaffective disorder, this SNP in the COMT gene was found to have significant associations with response to risperidone over 2-12 weeks in both African-American and white patients.

PHARMGKB PA162168065
Evidence: PubMed ID:16876132This variant may predict a favorable outcome for bupropion treatment for smoking cessation

dbSNP: rs165599 Genecard: COMT chromosome 22 SNPedia: rs165599(A;G)

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★ **rs35753505(T;T)** **info:0.22** **48.12%**

Rs35753505 (SNP8NRG221533) is located within the 5'-flanking region of the NRG1 (Neuregulin 1) gene.

g2b2mh blog rs35753505(C;C) correlates with frontal brain having to working harder during a working memory task.

personality traits - C allele carriers associated with higher (NEO-FFI) scores of neuroticism and lower scores of conscientiousness in a sample of 523 healthy individuals

The results suggest that subjects with risk alleles show hyperactivations in areas associated with elaborate episodic memory encoding and retrieval strategies.

Gene related topics:

personality traits(1.00) · personality(1.00) · neuroticism(1.00) · memory(1.00) · brain(1.00)

Relevant papers and external links:

PMID 18455369
PMID 20036336

dbSNP: rs35753505 Genecard: nan chromosome 8 SNPedia: rs35753505(T;T)

★ rs174570(C;C) info:0.18 59.56% **; a1c measurement (you have the risk allele C)**

per the 23andMe blog, the minor allele of this SNP (T) was associated with decreased cholesterol and increased triglycerides

SNPRarer alleleLDLHDLTG rs6544713 T + rs2650000 A + rs471364 C - rs1800961 T - rs7679 C - + rs2967605 T - rs2409722 T - rs10903129 A - - - rs6756629 A - + - rs12670798 C + + + rs7395662 A - + + rs174570 T - - + rs2271293 A - + - rs2624265 C + rs2167079 T + rs9891572 T + rs4844614 T + rs5031002 G +

rs7115739 and rs174570 External Link Greenlandic Inuit show genetic signatures of diet and climate adaptation

Established associations:

trans-16:1n-7 fatty acid measurement **you have 0 copies of the risk allele T**

(risk allele=T, Cis/trans-18:2, EA odds ratio/beta 0.0032 [0.002-0.0044] unit increase with pval 1E-7, pubmedid=25646338)

trans/trans-18:2 fatty acid measurement **you have 0 copies of the risk allele T**

(risk allele=T, Cis/trans-18:2, EA odds ratio/beta 0.0032 [0.002-0.0044] unit increase with pval 1E-7, pubmedid=25646338)

trans/cis-18:2 fatty acid measurement **you have 0 copies of the risk allele T**

(risk allele=T, Cis/trans-18:2, EA odds ratio/beta 0.0032 [0.002-0.0044] unit increase with pval 1E-7, pubmedid=25646338)

trans fatty acid measurement **you have 0 copies of the risk allele T**

(risk allele=T, Cis/trans-18:2, EA odds ratio/beta 0.0032 [0.002-0.0044] unit increase with pval 1E-7, pubmedid=25646338)

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total trans-18:1 fatty acid measurement **you have 0 copies of the risk allele T**

(risk allele=T, Cis/trans-18:2, EA odds ratio/beta 0.0032 [0.002-0.0044] unit increase with pval 1E-7, pubmedid=25646338)

cis/trans-18:2 fatty acid measurement **you have 0 copies of the risk allele T**

(risk allele=T, Cis/trans-18:2, EA odds ratio/beta 0.0032 [0.002-0.0044] unit increase with pval 1E-7, pubmedid=25646338)

low density lipoprotein cholesterol measurement **you have 0 copies of the risk allele G**

(risk allele=G, odds ratio/beta 0.11 s.d. increase with pval 4E-13, pubmedid=19060911)

total cholesterol measurement **you have 0 copies of the risk allele G**

(risk allele=G, odds ratio/beta 0.09 s.d. increase with pval 2E-10, pubmedid=19060911)

high density lipoprotein cholesterol measurement **you have 0 copies of the risk allele G**

(risk allele=G, odds ratio/beta 0.06 s.d. increase with pval 4E-6, pubmedid=19060911)

a1c measurement **you have 2 copies of the risk allele C**

(risk allele=C, odds ratio/beta 0.04 [0.020-0.060] unit increase with pval 2E-7, pubmedid=24647736)

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Gene related topics:

cholesterol(1.00) · triglycerides(1.00) · METABOLIC(0.36) · phospholipids(0.36) · hdl cholesterol(0.08) · lipoproteins ldl(0.06) · ldl cholesterol(0.06) · PSYCH(0.02) · adhd attention deficit hyperactivity disorder(0.02)

Relevant papers and external links:

PMID 19060911
Loci influencing lipid levels and coronary heart disease risk in 16 european population cohorts.

PMID 24647736
Multiple nonglycemic genomic loci are newly associated with blood level of glycated hemoglobin in east asians.

PMID 25646338
Genetic loci associated with circulating phospholipid trans fatty acids: a meta-analysis of genome-wide association studies from the charge consortium.

PHARMGKB PA164740210
Evidence: PubMed ID:19060911; Web Resource:External Link results: Loci influencing lipid levels and coronary heart disease risk in 16 European population cohorts. (Initial Sample Size: 21,412 individuals; Replication Sample Size: NR); (Region: 11q12.2; Reported Gene(s): FADS2, FADS3; Risk Allele: rs174570-G); (p-value= 0.000004).This variant is associated with HDL cholesterol.

PHARMGKB PA164740199

Evidence: PubMed ID:19060911; Web Resource:External Link results: Loci influencing lipid levels and coronary heart disease risk in 16 European population cohorts. (Initial Sample Size: 22,562 individuals; Replication Sample Size: NR); (Region: 11q12.2; Reported Gene(s): FADS2, FADS3; Risk Allele: rs174570-G); (p-value= 0.0000000002).This variant is associated with Cholesterol, total.

PHARMGKB PA164740223
Evidence: PubMed ID:19060911; Web Resource:External Link results: Loci influencing lipid levels and coronary heart disease risk in 16 European population cohorts. (Initial Sample Size: 17,797 individuals; Replication Sample Size: NR); (Region: 11q12.2; Reported Gene(s): FADS2, FADS3; Risk Allele: rs174570-G); (p-value= 0.0000000000004).This variant is associated with LDL cholesterol.

dbSNP: rs174570 GWAS Catalog: rs174570 Genecard: FADS2 chromosome 11

SNPedia: rs174570(C;C)

★ **rs2802288(A;G)** **info:0.18** **49.81%**

23andMe blog longevity rs2802288(A) increased the odds of living to 100 by 1.5x. rs2802288 is located very close to rs2802292 which was previously reported to have a similar effect.

Gene related topics:

AGING(0.15) · longevity(0.15)

Relevant papers and external links:

dbSNP: rs2802288 Genecard: FOXO3 chromosome 6 SNPedia: rs2802288(A;G)

★ **rs5746059(A;G)** **info:0.17** **23.70%** **slightly higher fat mass**

rs5746059 is a SNP in the tumor necrosis factor-alpha receptor 2 TNFRSF1B gene.

A study of ~1,800 subjects from 400 Caucasian families concluded that carriers of a rs5746059(A) allele had a 4.6% higher fat mass and a 2.5% increased percentage fat mass compared to noncarriers; the results were significant after multiple testing correction. This same correlation was

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also seen in 700 independent Chinese Han adults and 1,000 additional Caucasians. This SNP therefore appears to be associated with risk for obesity.

Gene related topics:

obesity(1.00) · IMMUNE(0.33) · rheumatoid arthritis(0.33) · systemic lupus erythematosus(0.28) · arthritis(0.22) · METABOLIC(0.08) · bone density(0.08) · REPRODUCTION(0.08) · premature birth(0.08) · arthritis rheumatoid rheumatoid arthritis(0.07) · arthritis rheumatoid(0.06) · crohn s disease(0.05) · crohn s disease ulcerative colitis(0.04) · CARDIOVASCULAR(0.04) · coronary disease coronary heart disease(0.04) · endometriosis(0.02)

Relevant papers and external links:

PMID 18685868

dbSNP: rs5746059 Genecard: TNFRSF1B chromosome 1 SNPedia: rs5746059(A;G)

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★ rs4320932(A;A) info:0.14 53.09% **normal**

rs4320932 is a SNP in the IF2 insulin-like growth factor 2 gene.

A study totaling ~5,000 non-Hispanic white patients with ovarian cancer found a 13% decreased risk of ovarian cancer per copy of the rs4320932(G) allele carried (CI: 0.81-0.93, P-trend=7.4 × 10e-5).

Associated with schizophrenia in a study of Croatians. Due to the limited number (~100 patients) and diversity of the studied population, more study is needed to confirm any link. The G allele was associated with an increased risk of schizophrenia with $P < 0.001$.

Small size study with 212 cases linked the G allele earlier found protective against ovarian cancer increasing relapse by 3.05 (95% confidence interval CI: 1.47–6.37, P-trend 2*10e-4) and mortality 3.28 (95% CI: 1.64–6.57, P-trend 6*10e-5).

Association of Tagging Single Nucleotide Polymorphisms on 8 Candidate Genes in Dopaminergic Pathway with Schizophrenia in Croatian Population

Gene related topics:

ovarian cancer(1.00) · CANCER(1.00) · cancer(1.00) · schizophrenia(1.00) · insulin(1.00) · mortality(1.00) · NORMALVARIATION(0.67) · muscle testing(0.67) · METABOLIC(0.57) · insulin obesity(0.57) · body mass(0.24) · UNKNOWN(0.16) · birth weight(0.16) · AGING(0.08) · longevity(0.08) · IMMUNE(0.04) · diabetes mellitus type 1(0.04)

Relevant papers and external links:

PMID 21422097
PMID 19673036
PMID 23677070
PMID 19673036

dbSNP: rs4320932 Genecard: IGF2 chromosome 11 SNPedia: rs4320932(A;A)

★ rs1801282(C;C) good:0.08 86.44% **normal fat metabolism; common in clinvar**

rs1801282, also known as Pro12Ala, is a common SNP in the peroxisome proliferator-activated receptor PPARG gene. The more common (C) allele (in dbSNP orientation) encodes the 'Pro' amino acid at this SNP position.

rs1801282 has been reported to be associated with metabolic syndrome, but other studies have not been able to replicate any strong or significant effect.

The association between type-2 diabetes and SNP rs1801282 is mentioned as being replicated in

rs1801282 and risk of cognitive decline in elders? Maybe with diabetes.

A ten-year study of 679 male patients with symptomatic coronary artery disease found that rs1801282 (G) carriers had a much lower (10 year) cardiovascular risk, with the hazard ratio estimated to be 0.10 (CI: 0.01-0.70, p = 0.02) for ischemic heart disease and 0.24 for vascular death (CI: 0.08-0.74, p = 0.013) per copy of the allele.

23andMe blog carriers could be informed that they really need to watch out for high fat in their diets

Response to Diet and Exercise

Established associations:

type ii diabetes mellitus **you have 2 copies of the risk allele C**

(risk allele=C, DGI+FUSION+WTCCC odds ratio/beta 1.14 [1.08-1.20] with pval 2E-6, pubmedid=17463249; risk allele=C, odds ratio/beta 1.16 [1.10-1.23] with pval 6E-10, pubmedid=24509480; risk allele=C, DGI+FUSION+WTCCC odds ratio/beta 1.14 [1.08-1.20] with pval 2E-6, pubmedid=17463248; risk allele=C, DGI+FUSION+WTCCC odds ratio/beta 1.14 [1.08-1.20] with pval 2E-6, pubmedid=17463246)

Gene related topics:

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metabolism(5.00) · METABOLIC(2.56) · type 2 diabetes(2.56) · diabetes mellitus diabetes mellitus type ii diabetes mellitus type 2(2.00) · diabetes mellitus type ii diabetes mellitus type 2 hyperinsulinism insulin resistance(2.00) · diabetes mellitus type ii diabetes mellitus type 2 obesity(2.00) · insulin resistance obesity(1.92) · diabetes mellitus type ii diabetes mellitus type 2 insulin resistance(1.78) · obesity(1.75) · diabetes type 2(1.72) · metabolic syndrome(1.50) · REPRODUCTION(1.44) · polycystic ovarian syndrome polycystic ovary syndrome(1.44) · insulin resistance polycystic ovarian syndrome polycystic ovary syndrome(1.29) · UNKNOWN(1.00) · fatty liver(1.00) · weight loss(1.00) · cardiovascular risk(1.00) · OTHER(1.00) · cardiovascular(1.00) · heart disease(1.00) · coronary artery disease(1.00) · diabetes mellitus type ii diabetes mellitus type 2(0.82) · CARDIOVASCULAR(0.80) · cardiovascular diseases inflammation(0.80) · insulin obesity(0.57) · obesity type 2 diabetes(0.57) · insulin resistance(0.32) · bmi(0.25) · peripheral vascular diseases(0.21) · birth weight(0.16) · acute coronary syndrome(0.14) · body weight obesity(0.11) · diabetes type 2 diabetes type 1(0.09) · osteoporosis(0.08) · metabolic syndrome x(0.08) · polycystic ovary syndrome(0.08) · atherosclerosis(0.07) · diabetes mellitus type 2(0.05) · bone mineral density(0.04) · AGING(0.04) · longevity(0.04) · insulin(0.03) · myocardial infarct(0.03) · endometriosis(0.02)

Relevant papers and external links:

PMID 17463249
Replication of genome-wide association signals in uk samples reveals risk loci for type 2 diabetes.

PMID 24509480
Genome-wide trans-ancestry meta-analysis provides insight into the genetic architecture of type 2 diabetes susceptibility.

PMID 17463248
A genome-wide association study of type 2 diabetes in finns detects multiple susceptibility variants.

PMID 17463246
Genome-wide association analysis identifies loci for type 2 diabetes and triglyceride levels.

PMID 18959602

PMID 17554300

PMID 18280041

PMID 19228871

OMIM 601487

PHARMGKB PA165111428
Evidence: PubMed ID:16783862Risk or phenotype-associated allele: G. Phenotype: For the Pro12Ala (rs1801282) SNP in the PPARG gene, an association was noted for the minor allele between psoriatic arthritis cases and controls (9.0% vs 13.8%, respectively; p = 0.017, OR 0.62, 95% CI 0.45-0.93). Study size: X. Study population/ethnicity: Caucasian

PHARMGKB PA162191393
Evidence: PubMed ID:17463248In a large Finnish case-control GWAS, rs1801282 was found to be associated with susceptibility to Type 2 Diabetes.

OMIM 601487

dbSNP: rs1801282 GWAS Catalog: rs1801282 Genecard: PPARG chromosome 3

SNPedia: rs1801282(C;C)

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★ rs601338(A;G) info:0.08 43.64% **susceptible to Norovirus infections**

rs601338 is found on chromosome 19 in the alpha(1,2)-fucosyltransferase FUT2 gene. The wild-type rs601338(G) encodes the "secretor" (Se) allele, while rs601338(A) encodes the "non-secretor" (se) allele.

A study of 115 Swedish adults concluded that rs601338(A;A) homozygotes have genetic immunity to infection by the Norwalk norovirus, a major (and contagious) cause of acute gastroenteritis worldwide among adults. This illness is also known as "cruise ship gastroenteritis."

Being a non-secretor may have other consequences, such as greater susceptibility to infection by influenza viruses and by some types of bacteria. 23andMe discusses these topics.

In some non-Caucasian populations, a different SNP is responsible for non-secretor phenotypes. For example, although the Se allele is absent in Japanese, 15% are non-secretors based on being homozygous for the non-secretor "sej" allele of SNP rs1047781.

It also appears to affect the composition of the microbiome , with secretors apparently having higher levels of Bifidobacteria.

External Link

External Link

Novel association of ABO histo-blood group antigen with soluble ICAM-1: results of a genome-wide association study of 6,578 women.

Common variants of FUT2 are associated with plasma vitamin B12 levels.

Histo-blood group gene polymorphisms as potential genetic modifiers of infection and cystic fibrosis lung disease severity.

Development of a fingerprinting panel using medically relevant polymorphisms.

Potential etiologic and functional implications of genome-wide association loci for human diseases and traits.

Common genetic variation and the control of HIV-1 in humans.

Population based allele frequencies of disease associated polymorphisms in the Personalized Medicine Research Project.

FUT2 nonsecretor status links type 1 diabetes susceptibility and resistance to infection.

Norovirus Resistance

Established associations:

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blood metabolite measurement **you have 1 copies of the risk allele A**

(risk allele=A, ADSGEGDFXAEGGGVR/ADpSGEGDFXAEGGGVR odds ratio/beta 0.055 [0.043-0.067] unit decrease with pval 2E-20, pubmedid=24816252; risk allele=A, ADpSGEGDFXAEGGGVR odds ratio/beta 0.041 [0.029-0.053] unit increase with pval 3E-11, pubmedid=24816252)

Gene related topics:

INFECTION(3.00) · METABOLIC(3.00) · norovirus infection(3.00) · vitamin b 12(3.00) · cystic fibrosis(1.00) · OTHER(1.00) · hiv(1.00) · type 1 diabetes(1.00) · helicobacter pylori infection(0.50) · h pylori infection(0.30) · IMMUNE(0.04) · crohn disease crohn s disease(0.04) · crohn disease(0.03)

Relevant papers and external links:

PMID 24816252
An atlas of genetic influences on human blood metabolites.

PMID 12692541

PMID 16306606

PMID 8621666

OMIM 612542

OMIM 182100

PHARMGKB PA162355649
Evidence: PubMed ID:7876235Results from in vitro expression assays indicate that this polymorphism, which creates the translation termination codon, inactivates this allele. Approximately 20% of randomly-selected individuals were found to be homozygous for this enzyme-inactivating nonsense allele, in correspondence to the frequency of the non-secretor phenotype in most human populations.

PMID 18604267

PMID 18776911

PMID 19169360

PMID 19379518

PMID 19474294

PMID 20041166

PMID 20565774

PMID 22025780

dbSNP: rs601338 GWAS Catalog: rs601338 Genecard: FUT2 chromosome 19

SNPedia: rs601338(A;G)

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★ rs2943641(C;C) info:0.01 56.37% **increased risk for type 2 diabetes**

identified a significant association of rs2943641 near IRS1 locus, with type 2 diabetes in the Japanese population.

Prioritizing genes for follow-up from genome wide association studies using information on gene expression in tissues relevant for type 2 diabetes mellitus.

The type 2 diabetes and insulin-resistance locus near IRS1 is a determinant of HDL cholesterol and triglycerides levels among diabetic subjects.

Insulin receptor substrate 1 gene variation modifies insulin resistance response to weight-loss diets in a 2-year randomized trial: the Preventing Overweight Using Novel Dietary Strategies (POUNDS LOST) trial.

Established associations:

type ii diabetes mellitus **you have 2 copies of the risk allele C**

(risk allele=C, odds ratio/beta 1.19 [1.13-1.25] with pval 9E-12, pubmedid=19734900)

autoantibody measurement **you have 2 copies of the risk allele C**

(risk allele=C, odds ratio/beta 1.19 [1.13-1.25] with pval 9E-12, pubmedid=19734900)

Gene related topics:

type 2 diabetes(5.00) · cholesterol(1.00) · insulin resistance(1.00) · hdl cholesterol(1.00) · diabetes mellitus(1.00) · weight(1.00) · triglycerides(1.00) · insulin(1.00)

Relevant papers and external links:

PMID 19734900
Genetic variant near irs1 is associated with type 2 diabetes, insulin resistance and hyperinsulinemia.

OMIM 147545

PMID 20043853

PMID 21353221

PMID 21747052

dbSNP: rs2943641 GWAS Catalog: rs2943641 Genecard: nan chromosome 2

SNPedia: rs2943641(C;C)

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★ rs174537(G;T) info:0.00 42.23% **common; higher LDL-C and total cholesterol**

rs174537 is a SNP near the FADS1 gene on chromosome 11. The FADS1 gene is one of 3 fatty acid desaturases in this region; higher polyunsaturated fatty acids (PUFA) levels are generally correlated with lower risk for heart disease.

A study of 1,075 participants in the InCHIANTI study on aging concluded that rs174537(T;T) individuals had lower arachidonic acid (AA), as well as lower levels of all other polyunsaturates measured (excepting linoleic acid, which followed the opposite pattern, and alpha-linoleic acid), compared to (G;G) homozgyotes (p = 6 x 10e-46), and that this one SNP accounted for almost 19% of the variance in AA levels. Individuals carrying at least one rs174537(G) allele had higher low-density lipoprotein (LDL-C) and total cholesterol levels. This effect of rs174537 was confirmed in an independent sample of 1,076 participants in the GOLDN study.

Genome-wide association analysis of metabolic traits in a birth cohort from a founder population.

Genetic determinants of circulating sphingolipid concentrations in European populations.

FADS genetic variants and omega-6 polyunsaturated fatty acid metabolism in a homogeneous island population.

Genetic variation in lipid desaturases and its impact on the development of human disease.

FADS gene polymorphisms in Koreans: association with omega6 polyunsaturated fatty acids in serum phospholipids, lipid peroxides, and coronary artery disease.

Genetics and genomics of human ageing.

Genetic variants of the FADS gene cluster and ELOVL gene family, colostrums LC-PUFA levels, breastfeeding, and child cognition.

Established associations:

trans/trans-18:2 fatty acid measurement **you have 1 copies of the risk allele T** (risk allele=T, Cis/trans-18:2, EA odds ratio/beta 0.0031 [0.0023-0.0039] unit increase with pval 8E-14, pubmedid=25646338)
trans/cis-18:2 fatty acid measurement **you have 1 copies of the risk allele T** (risk allele=T, Cis/trans-18:2, EA odds ratio/beta 0.0031 [0.0023-0.0039] unit increase with pval 8E-14, pubmedid=25646338)
cis/trans-18:2 fatty acid measurement **you have 1 copies of the risk allele T**

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(risk allele=T, Cis/trans-18:2, EA odds ratio/beta 0.0031 [0.0023-0.0039] unit increase with pval 8E-14, pubmedid=25646338)

total trans-18:1 fatty acid measurement **you have 1 copies of the risk allele T**

(risk allele=T, Cis/trans-18:2, EA odds ratio/beta 0.0031 [0.0023-0.0039] unit increase with pval 8E-14, pubmedid=25646338)

trans fatty acid measurement **you have 1 copies of the risk allele T**

(risk allele=T, Cis/trans-18:2, EA odds ratio/beta 0.0031 [0.0023-0.0039] unit increase with pval 8E-14, pubmedid=25646338)

trans-16:1n-7 fatty acid measurement **you have 1 copies of the risk allele T**

(risk allele=T, Cis/trans-18:2, EA odds ratio/beta 0.0031 [0.0023-0.0039] unit increase with pval 8E-14, pubmedid=25646338)

colorectal cancer **you have 1 copies of the risk allele G**

(risk allele=G, East Asian odds ratio/beta 1.16 [1.12-1.19] with pval 9E-21, pubmedid=24836286)

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Gene related topics:

cholesterol(5.00) · aging(1.00) · metabolism(1.00) · lipoprotein(1.00) · phospholipids(1.00) · OTHER(1.00) · heart disease(1.00) · METABOLIC(1.00) · coronary artery disease(1.00) · AGING(1.00)

Relevant papers and external links:

PMID 24836286
Large-scale genetic study in east asians identifies six new loci associated with colorectal cancer risk.

PMID 25646338
Genetic loci associated with circulating phospholipid trans fatty acids: a meta-analysis of genome-wide association studies from the charge consortium.

PMID 19148276

OMIM 612795

PMID 19060910

PMID 19798445

PMID 20562440

PMID 20565855

PMID 21040914

PMID 21115529

PMID 21383846

dbSNP: rs174537 GWAS Catalog: rs174537 Genecard: MYRF chromosome 11

SNPedia: rs174537(G;T)

★ rs6280(T;T) info:0.00 26.38% **normal**

rs6280, also known as Ser9Gly, is a SNP in the dopamine receptor D3 DRD3 gene. The rs6280 (C) allele encodes a glycine, and the (T) allele encodes a serine (in dbSNP orientation).

In a study of 88 patients being treated for schizophrenia with olanzapine, those who were rs6280 (C;C) homozygotes had greater positive symptom remission (endpoint rating of minimal or none on all PANSS clinical response positive items, 39.1%), as compared with (C;T) or (T;T) genotypes (13.8%; p = 0.033).

Ser9Gly has been implicated in executive function in some studies, but the results are conflicting. Gly/Gly carriers showed significantly (p = 0.002) poorer performance than Ser/Ser carriers on executive functioning tasks in a somewhat small Caucasian sample (84 patients with first-episode psychosis and 85 controls).
Gly/Ser heterozygotes had 23% more preservative errors on the WCST compared to Ser/Ser homozygotes in a small (216) healthy Han Chinese sample (p = 0.009). Differences between homozygotes were not statistically significant.
No association between WCST scores and Ser9Gly was found in 138 schizophrenic patients.

Associated in a family association study and pooled sample of 2,037 with nicotine dependence in Americans of European descent.

Preliminary evidence for an association between a dopamine D3 receptor gene variant and obsessive-compulsive personality disorder in patients with major depression. DRD3 polymorphism for an individual with Gly/Gly (C;C) genotype is 2.4 (P = 0.017) times more likely to be diagnosed with OCPD. Male gender was also found to be a significant predictor of OCPD diagnosis (OR = 2.82, P = 0.001). DRD3 may contribute to the development of OCPD. This association was tested using two independent groups of individuals with a history of depression, from a clinical sample (n = 149) and a family study (n = 213).

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Gene related topics:

NEUROLOGICAL(3.79) · tardive dyskinesia(3.79) · essential tremor(2.88) · PSYCH(2.37) · schizophrenia(2.37) · CHEMDEPENDENCY(2.00) · UNKNOWN(2.00) · akathisia drug induced(2.00) · cocaine abuse(2.00) · personality(1.00) · olanzapine(1.00) · nicotine(1.00) · major depression(1.00) · psychosis(1.00) · depression(1.00) · nicotine dependence(1.00) · cocaine related disorders(0.67) · schizophrenia tardive dyskinesia(0.57) · tourette syndrome(0.47) · alcohol dependence(0.22) · parkinson s disease(0.14) · personality disorders(0.12) · bipolar disorder(0.10) · obsessive compulsive disorder(0.06) · adhd attention deficit hyperactivity disorder(0.05) · personality traits(0.05) · alcohol abuse(0.04) · smoking behavior(0.04)

Relevant papers and external links:

PMID 18320559

PMID 18351593

PMID 2186374

PMID 15785860

PMID 18348205

PMID 16583407

PHARMGKB PA165111549
Evidence: PubMed ID:20029384Risk or phenotype-associated allele: Phenotype: This meta-analysis showed no difference in clozipine response between the Ser and Gly allele. Study size: 758 Study population/ethnicity: Patients with Schizophrenia Significance metric(s): OR = 0.82, 95% confidence interval (CI): 0.65-1.04; p = 0.1 Type of association: PD

PHARMGKB PA162316713
Evidence: PubMed ID:18320559A study on 88 olanzapine-treated patients with schizophrenia found that the Gly/gly genotype was significantly associated with greater positive symptom improvement.

PHARMGKB PA164807597
Evidence: PubMed ID:19396436This variant in the DRD3 gene was significantly associated with the therapeutic efficacy of pramipexole in Chinese patients with Parkinson's disease in a study of 30 patients.

PHARMGKB PA165111370
Evidence: PubMed ID:19997080Risk or phenotype-associated allele: C Phenotype: Carriers of the C variant (Gly) of DRD3:Ser9Gly had greater reductions in Autism Treatment Evaluation Checklist (ATEC) scores, indicating improved symptoms and response to risperidone, than TT homozygotes. Study size: 45 Study population/ethnicity: Children with Autism receiving risperidone Significance metric(s): p = 0.012 Type of association: PD

PHARMGKB PA165110867
Evidence: PubMed ID:16809426Risk or phenotype-associated allele: C. Phenotype: A study comparing 276 patients with essential tremor and 184 normal controls confirmed the association of the Gly-9 variant in the DRD-3 gene with risk and age-at-onset of essential tremor. Study size: 276. Study population/ethnicity: French.

OMIM 126451

dbSNP: rs6280 Genecard: DRD3 chromosome 3 SNPedia: rs6280(T;T)

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★ **rs1042713(G;G)** **info:0.00** **27.50%** **normal**

rs1042713, also known as Arg16, G16R, and 16Arg>Gly, is a SNP in the adrenergic, beta-2-, surface receptor ADRB2 gene. The rs1042713(G) allele encodes the G/glycine form that is the more common one in most populations, while rs1042713(A) encodes the R/arginine residue at this position of the ADRB2 protein.

A study of 1,182 young Scottish asthma patients (age, 3-22 years) concluded that rs1042713(A) alleles are significantly associated with "exacerbations" of their condition, regardless of treatment regimens (odds ratio 1.30, CI: 1.09-1.55, p=0.003). This didn't hold true for patients using inhalers less than once per day; and it was particularly true for those receiving daily inhaled long- or short-acting beta(2)-agonist treatment (OR, 1.64, CI: 1.22-2.20, p=.001). This may therefore indicate that regular inhalant use of albuterol (found in Ventolin) or long-acting agonists, such as salmeterol found in Advair, may be counter-productive in young asthma patients carrying one or especially two rs1042713(A) alleles.

Gene related topics:

IMMUNE(5.47) · asthma(5.47) · nocturnal asthma(4.00) · OTHER(3.00) · beta agonist dependence(3.00) · desensitization(3.00) · INFECTION(2.00) · METABOLIC(2.00) · PSYCH(2.00) · asthma bhr total ige airway obstruction fev1 fvc increased(2.00) · asthma smokers(2.00) · bronchial asthma(2.00) · decline in lung function(2.00) · decreased airway responsiveness(2.00) · high total ige(2.00) · high total ige asthma atopy(2.00) · parasitic infection(2.00) · pef(2.00) · regional fat distribution(2.00) · response to beta2 agonist therapy(2.00) · steroid dependent asthma(2.00) · steroid requiring asthma in sedentary women(2.00) · stress(2.00) · obesity(1.26) · CARDIOVASCULAR(1.23) · hypertension(1.23) · asthma severity(1.07) · asthma total ige(1.00) · asthma total ige spt(1.00) · atopy bhr(1.00) · hypertension obesity weight gain(1.00) · tachycardia ventricular(1.00) · blood pressure arterial(0.96) · REPRODUCTION(0.80) · fev1(0.80) · obstetric labor premature(0.80) · obesity overweight(0.69) · bhr(0.67) · bmi(0.56) · atopic asthma(0.43) · heart failure(0.41) · pulmonary disease chronic obstructive(0.28) · copd chronic obstructive pulmonary disease(0.25) · hypertension hypertrophy left ventricular left ventricular hypertrophy(0.24) · asthma atopy(0.18) · metabolic syndrome(0.17) · cardiomyopathy dilated dcm dilated cardiomyopathy(0.14) · acute coronary syndrome(0.14) · cerebral palsy(0.09) · VISION(0.08) · glaucoma(0.08) · premature birth(0.08) · NEUROLOGICAL(0.08) · migraine disorders(0.08) · chronic obstructive pulmonary disease copd(0.07) · chronic obstructive pulmonary disease(0.07) · glaucoma open angle(0.07) · cystic fibrosis(0.05) · heart rate(0.04) · adhd attention deficit hyperactivity disorder(0.02)

Relevant papers and external links:

PMID 19800676

PHARMGKB PA164857060
Evidence: PubMed ID:7915137Gly16 receptor had an enhanced agonist-promoted downregulation relative to Arg16 in in vitro studies using chinese hamster fibroblasts with expressed ADRB2.

PHARMGKB PA161145204
Evidence: Web Resource:External Link variant has been studied in terms of asthma and its drug response and in terms of heart disease and vascular phenotypes and their drug responses.

PHARMGKB PA162167949

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Evidence: PubMed ID:18615004None of the ADRB2 haplotypes tested that contained this SNP were associated with differential primary outcome (i.e., the first incidence of death, nonfatal myocardial infarction, or nonfatal stroke) in hypertensive patients with coronary artery disease.

PHARMGKB PA162675224
Evidence: PubMed ID:18156033The ADRB2:Arg16Gly variant was not associated with asthma exacerbation in patients treated with inhaled corticosteroids plus longacting beta agonists.

PHARMGKB PA162675225
Evidence: PubMed ID:16322642Relative to ADRB2:16Gly/Gly patients with asthma, ADRB2:16Arg/Arg patients with asthma may have an impaired therapeutic response to salmeterol in either the absence or presence of concurrent inhaled corticosteroid use.

PHARMGKB PA164857059
Evidence: PubMed ID:15500895Subjects who are homozygous for Arg16 and who were on regular albuterol treatment were reported to have lower morning peak flows compared with those who were not on regular albuterol treatment, suggesting that regular albuterol therapy may not be appropriate for Arg16 homozygous subjects.

PHARMGKB PA164857061
Evidence: PubMed ID:11586955In healthy subjects, the Arg16 variant was associated with enhanced isoproterenol-mediated desensitization of the vasculature and enhanced isoproterenol induced-venodilation, suggesting that it may be an important determinant of the vascular response to stress.

OMIM 109690

dbSNP: rs1042713 Genecard: ADRB2 chromosome 5 SNPedia: rs1042713(G;G)

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★ **rs1800437(G;G)** **info:0.00** **70.37%** **average**

SNP rs1800437, located in the coding region of the GIPR gene and known as Glu354Gln, shows a marginal association with cardiovascular disease (CVD) in a study of ~200 patients from a Dutch population. Heterozygotes are reported to be at lower risk for CVD, with an odds ratio of 0.72 (CI: 0.52 - 0.97, p=0.03). However, no significant association was seen for homozygous carriers of the minor allele.

Established associations:

anthropometry **you have 2 copies of the risk allele G**

(risk allele=G, Obesity class I odds ratio/beta 1.1 with pval 3E-14, pubmedid=23563607)

Gene related topics:

cardiovascular disease(1.00) · CARDIOVASCULAR(1.00) · cardiovascular(1.00)

Relevant papers and external links:

PMID 23563607
Genome-wide meta-analysis identifies 11 new loci for anthropometric traits and provides insights into genetic architecture.

PMID 17624916

dbSNP: rs1800437 GWAS Catalog: rs1800437 Genecard: GIPR chromosome 19

SNPedia: rs1800437(G;G)

★ rs2905072(T;T) info:0.00 62.24% **common in complete genomics; bipolar disorder (you have the risk allele T)**

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Established associations:

bipolar disorder **you have 2 copies of the risk allele T**
(risk allele=T, odds ratio/beta 1.21 [1.11-1.32] with pval 6E-6, pubmedid=19416921)

Relevant papers and external links:

PMID 19416921
Genome-wide association and meta-analysis of bipolar disorder in individuals of european ancestry.

dbSNP: rs2905072 GWAS Catalog: rs2905072 Genecard: GFI1B chromosome 9

SNPedia: rs2905072(T;T)

★ rs12708716(A;A) info:0.00 42.14% **1.6x risk of type-1 diabetes**

rs12708716 has been reported in a large study to be associated with type-1 diabetes.

The risk allele (oriented to the dbSNP entry) is (A); the odds ratio associated with heterozygotes is 1.19 (CI 0.97-1.45), and for homozygotes, 1.55 (CI 1.27-1.89).

In an expanded follow-up study of >6,000 controls and 6,000 patients, the heterozygote odds ratio for this SNP was recalculated to be 0.81 (CI 0.77-0.86).

Tested in a large multiple sclerosis data set consisting of 2369 trio families, 5737 cases and 10 296 unrelated controls, SNP rs12708716 was associated with disease risk (p = 1.6 x 10e-16)

Established associations:

autoantibody measurement **you have 0 copies of the risk allele G**

(risk allele=G, T1D odds ratio/beta 1.2 with pval 5E-14, pubmedid=21829393)

type i diabetes mellitus **you have 2 copies of the risk allele A**

(risk allele=A, odds ratio/beta 1.23 [1.16-1.30] with pval 3E-18, pubmedid=17554260; risk allele=G complex/no impact summary; risk allele=A, odds ratio/beta 1.19 [0.97-1.45] with pval 5E-7, pubmedid=17554300; risk allele=G, T1D odds ratio/beta 1.2 with pval 5E-14, pubmedid=21829393)

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Gene related topics:

multiple sclerosis(1.00) · IMMUNE(0.15) · diabetes mellitus type 1(0.15) · type 1 diabetes(0.06)

Relevant papers and external links:

PMID 18978792
Meta-analysis of genome-wide association study data identifies additional type 1 diabetes risk loci.

PMID 21829393
Genome-wide association analysis of autoantibody positivity in type 1 diabetes cases.

PMID 17554300

PMID 17554260

PMID 18987646

OMIM 222100

OMIM 611303

PHARMGKB PA164740754

Evidence: PubMed ID:18978792; Web Resource:External Link results: Meta-analysis of genome-wide association study data identifies additional type 1 diabetes risk loci. (Initial Sample Size: 3,561 cases, 4,646 controls; Replication Sample Size: 6,225 cases, 6,946 controls, 3,064 trios); (Region: 16p13.13; Reported Gene(s): CLEC16A; Risk Allele: rs12708716-G); (p-value= 0.0000000000007).This variant is associated with Type 1 diabetes.

PHARMGKB PA162356641
Evidence: PubMed ID:17554300; Web Resource:External Link Results: Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls (Initial Sample Size: 1,963 cases, 2,938 controls; Replication Sample Size: (see Todd 2007); Risk Allele: rs12708716-A). This variant is associated with type 1 diabetes.

PHARMGKB PA162356630
Evidence: PubMed ID:17554260; Web Resource:External Link Results: Robust associations of four new chromosome regions from genome-wide analyses of type 1 diabetes (Initial Sample Size: 1,963 cases, 2,938 controls; Replication Sample Size: 4,000 cases, 5,000 controls, 2,997 trios; Risk Allele: rs12708716-A).

PHARMGKB PA162355909
Evidence: PubMed ID:17554300A genome-wide association study in 2,000 individuals for each of 7 major diseases and a shared set of 3,000 controls found an association of this SNP with type 1 diabetes.

dbSNP: rs12708716 GWAS Catalog: rs12708716 Genecard: CLEC16A chromosome 16

SNPedia: rs12708716(A;A)

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POPULAR FAVORITES

POPULAR SNPS

Others marked them as favorite. What about you, do you have the good or bad variant ?

- rs53576 - linked to socially related personality traits and behaviors
- rs1800497 - avoidance of errors
- rs1801133 - MTHFR/efficiency in processing folic acid
- rs4680 - the warrior/worrier hypothesis

rs17070145 - memory performance
rs140701 - anxiety disorders
rs1801131 - MTHFR/folate metabolism
rs2802292 - longevity
rs3184504 - risk for celiac disease
rs909525 - repeats of the MAOA warrior gene
rs1333049 - risk for coronary artery disease

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