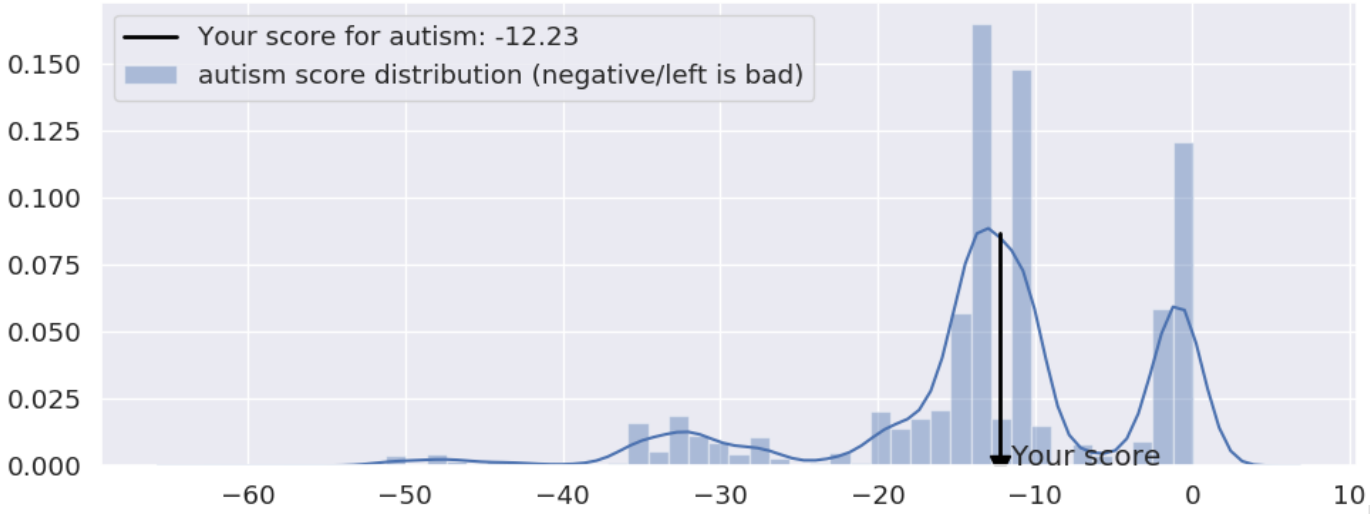
TOPIC: AUTISM



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★ 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)

★ rs4307059(C;T) info:1.34 33.69% **1.19x risk of Autism**

Slightly increased risk of Autism due to worse brain cell adhesion.
23andMe blog rs4307059 — compared to two copies of a C, each copy of the more common T version increased the odds of autism by 1.19 times.

Established associations:

autism **you have 1 copies of the risk allele T**

(risk allele=T, odds ratio/beta 1.19 with pval 2E-10, pubmedid=19404256)

Gene related topics:

autism(5.00) · brain(1.00)

Relevant papers and external links:

PMID 19404256
Common genetic variants on 5p14.1 associate with autism spectrum disorders.

PHARMGKB PA164741095
Evidence: PubMed ID:19404256This SNP on 5p14.1 was associated with autism spectrum disorder in a combined analysis of four GWAS data sets with a total of more than 10,000 subjects of European ancestry. This SNP is in an intergenic region between CDH10 and CDH9 that contains several highly conserved genomic elements.

OMIM 209850

dbSNP: rs4307059 GWAS Catalog: rs4307059 Genecard: nan chromosome 5

SNPedia: rs4307059(C;T)

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★ rs1143674(A;G) info:1.50 44.78% **1.3x increased autism risk**

rs1143674 is a SNP in the integrin alpha-4 ITGA4 gene on chromosome 2.

Across two sets of family samples (Irish and American), rs1143674 showed a slight association with increased risk for autism, with an odds ratio of 1.3 (p = 0.008).

Gene related topics:

PSYCH(0.03) · autism(0.03)

Relevant papers and external links:

PMID 18846500

dbSNP: rs1143674 Genecard: ITGA4 chromosome 2 SNPedia: rs1143674(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)

★ rs6314(C;T) bad:3.02 13.82% **higher risk for RA; better response to paroxetine as treatment for depression**

rheumatoid arthritis risk

rs6314, also known as C1354T or His452Tyr/H452Y, is a SNP in the serotonin 2A receptor HTR2A gene.

Based on a study of 166 Caucasian patients being treated for depression with paroxetine, rs6314 heterozygotes were associated with better response. There was a significantly higher frequency of heterozygotes in the remitter and response groups in comparison to the non-remitter (odds ratio 7.50, p=0.002) and non-response groups (odds ratio 5.25, p=0.01).

rs6314 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 rs6314(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.

"Haplotype association analysis suggests that the haplotype CCGCA (at SNPs rs3125, rs6314, rs1923886, rs2224721 and rs2770296) is protective against bipolar disorder (P = 0.021, odds ratio 0.63) and the rarer haplotype CCACG confers risk to the disorder (P = 0.0065, odds ratio 3.08)."

Genetic variation in the serotonin 2A receptor and suicidal ideation in a sample of 270 Irish high-density schizophrenia families.

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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) · 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 PA162181130
Evidence: PubMed ID:11311507; PubMed ID:15052272; PubMed ID:18253134In a study in a unipolar depressive population (n=166) this variant showed an association with remission and response following paroxetine therapy. HTR2A C1354T heterozygotes were significantly associated with improved response to paroxetine therapy. The SNP was also studied as a modifier of antidepressant response in a study in depressed patients (n=173) given a variety of antidepressant treatments with no association found, however another report found the HTR2A T1354 allele to be associated with response to fluoxetine.

PMID 18712714

PHARMGKB PA162181131
Evidence: PubMed ID:14699448; PubMed ID:15891581; PubMed ID:18602445This variant in the gene encoding the serotonin 2A receptor has been associated with human episodic memory performance and with differences in brain volume in memory-related brain regions.

dbSNP: rs6314 Genecard: HTR2A chromosome 13 SNPedia: rs6314(C;T)

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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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★ rs2329340(T;T) info:0.75 12.98% **MA four-marker haplotype in DDC was specific to migraine with Aura (rs2329340A, rs11974297C, rs2044859T, rs11761683G)**

A four-marker haplotype in DDC was specific to migraine with Aura (rs2329340A, rs11974297C, rs2044859T, rs11761683G)
a haplotype consisting of rs2329340(A), rs11974297(C), rs2044859(T) and rs11761683(G) associated with increased incidence of migraine with aura among a sample of 528 migraine patients (308 without aura, 220 with aura) and 528 sex-matched migraine-free controls

Neurotransmitter systems and neurotrophic factors in autism: association study of 37 genes suggests involvement of DDC.

Gene related topics:

migraine with aura(5.00) · migraine(5.00) · autism(1.00) · INFECTION(0.08) · malaria(0.08) · PSYCH(0.06) · adhd(0.06)

Relevant papers and external links:

PMID 19455600

PMID 22397633

dbSNP: rs2329340 Genecard: DDC chromosome 7 SNPedia: rs2329340(T;T)

★ rs7341475(G;G) info:1.32 72.46% **1.58x increased schizophrenia risk for women**

A total of 2,274 cases of schizophrenia were studied, resulting in an association between rs7341475, exclusively in women. Based on all populations the estimated relative risk for women carrying the common (G;G) genotype is 1.58 (p = 8.8 x 10(-7).

This finding was replicated in a large independent Ashkenazi Jewish collection (721 cases, 259 female; 1455 controls, 834 female), with confirmation that it applies to both schizophrenia and schizoaffective disorder.

Established associations:

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

(risk allele=G, odds ratio/beta 1.58 [1.31-1.89] with pval 9E-7, pubmedid=18282107)

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Gene related topics:

schizophrenia(5.00) · UNKNOWN(2.00) · otosclerosis(2.00) · PSYCH(0.22) · autism(0.22)

Relevant papers and external links:

PMID 18282107

PMID 20431428

PHARMGKB PA164740860

Evidence: PubMed ID:18282107; Web Resource:External Link results: Genome-Wide Association Identifies a Common Variant in the Reelin Gene That Increases the Risk of Schizophrenia Only in Women. (Initial Sample Size: 660 cases, 2,271 controls; Replication Sample Size: 2,274 cases, 4,401 controls); (Region: 7q22.1; Reported Gene(s): RELN; Risk Allele: rs7341475-G); (p-value= 8.99999999999999E-07).This variant is associated with Schizophrenia.

dbSNP: rs7341475 GWAS Catalog: rs7341475 Genecard: RELN chromosome 7

SNPedia: rs7341475(G;G)

★ 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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★ 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.

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)

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★ rs1734792(C;-) warning:2.64 33.56% **1.4x increased risk for lupus**

rs1734792 is one of several SNPs in the methyl CpG binding protein 2 (MECP2) that have been associated with risk for systemic lupus erythromatosis (SLE). The MECP2 gene is located on the X chromosome, which may be instructive since lupus is a predominantly female disease.

An initial survey of 600 Korean patients, followed by 1,000 Caucasian patients, ultimately led to a meta-analysis indicating an odds ratio for the rs1734792(A) risk allele of 1.42 (CI: 1.24-1.58, p = 3.3E-08).

Variants within MECP2, a key transcription regulator, are associated with increased susceptibility to lupus and differential gene expression in patients with systemic lupus erythematosus.

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · lupus(5.00) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · systemic lupus erythematosus(1.00) · lupus erythematosus(1.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

PMID 18320046

PMID 19333917

dbSNP: rs1734792 Genecard: MECP2 chromosome X SNPedia: rs1734792(C;C)

★ 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.

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Characterization of a functional promoter polymorphism of the human tryptophan hydroxylase 2 gene in serotonergic raphe neurons.

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)

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★ rs1734791(A;-) warning:2.50 31.78% **1.4x increased risk for lupus**

rs1734791 is one of several SNPs in the methyl CpG binding protein 2 (MECP2) that have been associated with risk for systemic lupus erythromatosis (SLE). The MECP2 gene is located on the X chromosome, which may be instructive since lupus is a predominantly female disease.

An initial survey of 600 Korean patients, followed by 1,000 Caucasian patients, ultimately led to a meta-analysis indicating an odds ratio for the rs1734791(A) risk allele of 1.39 (CI: 1.24â€“1.57, p = 7.2×10−8).

Variants within MECP2, a key transcription regulator, are associated with increased susceptibility to lupus and differential gene expression in patients with systemic lupus erythematosus.

A common MECP2 haplotype associates with reduced cortical surface area in humans in two independent populations.

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · lupus(5.00) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · systemic lupus erythematosus(1.00) · lupus erythematosus(1.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

PMID 18320046
PMID 19333917

PMID 19717458

dbSNP: rs1734791 Genecard: MECP2 chromosome X SNPedia: rs1734791(A;A)

★ 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)

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★ rs10513025(T;T) info:0.08 89.92% **normal risk of Autism**

Like almost everyone else, you have the normal version of this SNP. Unfortunately you didn't get the rare alternative mutations that would halve your risk of autism.
Your normal version (T;T) seems to reduce the expression of SEMA5A in the brains of people with autism. rs10513025 is a SNP located in chromosomal region 5p15, in between the SEMA5A and TAS2R1 genes.

Based on a linkage and association mapping study of 1,031 multiplex autism families, rs10513025 was found to be associated with autism, and the expression of SEMA5A was seen to be reduced in brains from autistic patients. Although this finding was replicated, the authors caution that this SNP explains a very small fraction of the heritability of a complex disorder like autism, presumably due to their finding that the most common allele (present at a frequency of ~96%) is the allele associated with higher risk. The much rarer allele, rs10513025(C), is associated with a 0.55x reduced risk for autism (p=9.6x10e-6).

Gene related topics:

autism(5.00)

Relevant papers and external links:

OMIM 209850

dbSNP: rs10513025 Genecard: nan chromosome 5 SNPedia: rs10513025(T;T)

★ rs7997012(G;G) info:0.86 52.89% **~18% less likely to respond to citalopram**

rs7997012, located in an intron of the HTR2A gene, was found to be associated with a somewhat increased rate of successful response to treatment of depression with the drug citalopram.

From this article :

"HTR2A encodes the serotonin 2A receptor, which is downregulated by citalopram. Participants who were homozygous for the A allele had an 18% reduction in absolute risk of having no response to treatment, compared with those homozygous for the other allele."

See also Anxiety Blog posting

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Gene related topics:

citalopram(5.00) · 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) · anxiety(1.00) · OTHER(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 16642436

PMID 16642436

OMIM 182135

PHARMGKB PA165109601

Evidence: PubMed ID:19636338Risk or phenotype-associated allele: A. Phenotype: Patients with the 5-HTR2A intron 2 (rs7997012) AA genotype suffered from more pronounced side effects compared to carriers of the GA or GG genotype. Study size: 124. Study population/ethnicity: Caucasian; Patients with psychiatric disorders. Significance metric(s): p = 0.018 and p = 0.002. Type of association: PD.

PHARMGKB PA161614203
Evidence: PubMed ID:16642436In one study, subjects homozygous for allele A had a reduction in absolute risk of having no response to treatment with citalopram for major depressive disorder. The allele was found to be more frequent in white than in black subjects, and this is consistant with racial differences in response to antidepressant treatment.

OMIM 182135

dbSNP: rs7997012 Genecard: HTR2A chromosome 13 SNPedia: rs7997012(G;G)

★ rs3813929(C;-) info:1.89 74.85% **possible weight gain if taking olanzapine**

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rs3813929, also known as -759C/T, is a SNP in the 5-hydroxytryptamine (serotonin) receptor 2C HTR2C gene.

A study of 107 patients with schizophrenia being treated with olanzapine reported a protective effect against weight-gain from the (T) allele of this SNP; zero patients (of 28) with a rs3813929(T) allele had a body mass index increase of >=10% (p=0.002), whereas (C;C) homozygotes did. This effect may also involve nearby SNP rs518147.

T allele showed borderline significant association with higher BMI and incidence of lifetime major depressive disorder among 4978 persons from the European Prospective Investigation into Cancer (EPIC)-Norfolk study, however, only the association with BMI remained borderline significant within the full EPIC-Norfolk cohort (20,981 persons)

The association between HTR2C polymorphisms and obesity in psychiatric patients using antipsychotics: a cross-sectional study.

Association of the HTR2C gene and antipsychotic induced weight gain: a meta-analysis.

The association between HTR2C gene polymorphisms and the metabolic syndrome in patients with schizophrenia.

Multivariate permutation analysis associates multiple polymorphisms with subphenotypes of major depression.

Focus on HTR2C: A possible suggestion for genetic studies of complex disorders.

HTR2C gene polymorphisms and the metabolic syndrome in patients with schizophrenia: a replication study.

Cohort profile: risk patterns and processes for psychopathology emerging during adolescence: the ROOTS project.

Association of polymorphisms of the serotonergic system with smoking initiation in Caucasians.

Pharmacogenetics and antipsychotics: therapeutic efficacy and side effects prediction.

Functional consequences of two HTR2C polymorphisms associated with antipsychotic-induced weight gain.

Gene related topics:

olanzapine(5.00) · weight(5.00) · METABOLIC(3.60) · weight gain(3.60) · PHARMACOGENOMIC(2.00) · weight gain antipsychotic drug induced(2.00) · weight gain antipsychotic induced(2.00) · OTHER(1.00) · weight loss(1.00) · CANCER(1.00) · body mass(1.00) · cancer(1.00) · bmi(1.00) · metabolic syndrome(1.00) · schizophrenia(1.00) · major depression(1.00) · body mass index(1.00) · obesity(1.00) · depression(1.00) · major depressive disorder(1.00) · smoking(1.00) · PSYCH(0.67) · bipolar affective disorder(0.67) · NEUROLOGICAL(0.33) · migraine with aura(0.33) · mood disorders(0.23) · suicide(0.22) · tardive dyskinesia(0.11) · personality(0.06) · attention deficit hyperactivity disorder(0.05) · personality traits(0.05) · depressive disorder major(0.05) · CHEMDEPENDENCY(0.04) · alcohol abuse(0.04) · autism(0.03)

Relevant papers and external links:

PMID 19434072

PMID 20065966

PHARMGKB PA165109600
Evidence: PubMed ID:19636338Risk or phenotype-associated allele: T. Phenotype: Short-term weight gain under olanzapine therapy was significantly lower for 5-HTR2C -759 T-allele carriers. Study size: 124. Study population/ethnicity: Caucasian; Patients with psychiatric disorders. Significance metric(s): p = 0.011. Type of association: PK.

PHARMGKB PA165108056
Evidence: PubMed ID:15666332Risk or phenotype-associated allele: T. Phenotype: The T allele was more frequently found in subjects not gaining 10% or more of their body weight, compared with those who did gain 10% or more of their body weight. None of the subjects who gained >10% of their initial body weight over 6 weeks carried a T allele (0/11). Study size: 42. Study population/ethnicity: Caucasian acutely ill patients with schizophrenia taking olanzapine. Significance metric(s): p = 0.0035. Type of association: CO.

PHARMGKB PA164924604
Evidence: PubMed ID:19622037This variant is associated with Olanzapine-induced weight gain. The -759T allele confers protective effect towards Olanzapine-induced weight gain in a study consisting of 107 patients with schizophrenia.

PMID 17016522

PMID 17291373

PMID 17632216

PMID 18081710

PMID 18802918

PMID 19142101

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PMID 19359258

PMID 20060656

PMID 21162693

PMID 21391883

dbSNP: rs3813929 Genecard: HTR2C chromosome X SNPedia: rs3813929(C;C)

★ 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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★ rs10227893(T;T) good:0.00 74.83% **common in complete genomics**

variations in rs17137124 and rs10227893 may impair speech

Gene related topics:

PSYCH(0.05) · autism(0.05)

Relevant papers and external links:

PMID 15877281

dbSNP: rs10227893 Genecard: FOXP2 chromosome 7 SNPedia: rs10227893(T;T)

★ rs28934907(C;-) good:0.00 **common in clinvar**

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

OMIM 300005

dbSNP: rs28934907 Genecard: MECP2 chromosome X SNPedia: rs28934907(C;C)

★ rs5917(G;G) good:0.00 99.48% **common**

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Gene related topics:

PHARMACOGENOMIC(3.00) · aspirin resistance(3.00) · CARDIOVASCULAR(2.78) · myocardial infarct atherosclerosis coronary(2.78) · atherosclerosis generalized(1.00) · myocardial infarction stroke(1.00) · myocardial infarct lymphoproliferative disorders restenosis(0.67) · myocardial infarct(0.31) · heart disease ischemic(0.30) · CANCER(0.21) · cancer(0.21) · subarachnoid hemorrhage(0.21) · coronary artery disease(0.14) · atherosclerosis coronary(0.13) · HEMATOLOGICAL(0.13) · platelet aggregation(0.13) · restenosis(0.12) · stroke ischemic(0.11) · thrombosis(0.11) · cardiovascular diseases(0.10) · REPRODUCTION(0.09) · pregnancy loss(0.09) · NORMALVARIATION(0.09) · normal variation(0.09) · stroke(0.09) · myocardial infarction(0.08) · cardiovascular(0.08) · brain ischemia stroke(0.08) · acute coronary syndrome(0.06) · PSYCH(0.05) · autism(0.05) · pregnancy loss recurrent(0.04) · METABOLIC(0.02) · waist hip ratio(0.02)

Relevant papers and external links:

OMIM 173470

dbSNP: rs5917 Genecard: ITGB3 chromosome 17 SNPedia: rs5917(G;G)

★ rs28934904(C;-) good:0.00 **common in clinvar**

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

OMIM 300005

dbSNP: rs28934904 Genecard: MECP2 chromosome X SNPedia: rs28934904(C;C)

★ rs61748421(C;-) good:0.00 **common in clinvar**

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

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Relevant papers and external links:

OMIM 300005

dbSNP: rs61748421 Genecard: MECP2 chromosome X SNPedia: rs61748421(C;C)

★ rs659734(T;T) good:0.00 84.41% **common in complete genomics**

Genetic variation in the serotonin 2A receptor and suicidal ideation in a sample of 270 Irish high-density schizophrenia families.

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) · 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 18712714

dbSNP: rs659734 Genecard: HTR2A chromosome 13 SNPedia: rs659734(T;T)

★ rs61751370(G;-) good:0.00 **common in clinvar**

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

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Relevant papers and external links:

dbSNP: rs61751370 Genecard: MECP2 chromosome X SNPedia: rs61751370(G;G)

★ rs28935168(C;-) good:0.00 **common in clinvar**

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

OMIM 300005

dbSNP: rs28935168 Genecard: MECP2 chromosome X SNPedia: rs28935168(C;C)

★ rs1734787(A;-) info:0.00 37.06% **normal**

rs1734787 is one of several SNPs in the methyl CpG binding protein 2 (MECP2) that have been associated with risk for systemic lupus erythromatosis (SLE). The MECP2 gene is located on the X chromosome, which may be instructive since lupus is a predominantly female disease.

An initial survey of 600 Korean patients, followed by 1,000 Caucasian patients, ultimately led to a meta-analysis indicating an odds ratio for the rs1734787(C) risk allele of 1.42 (CI: 1.26â€“1.60, p = 1.6Ã—10e-08).

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · lupus(1.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

PMID 18320046

dbSNP: rs1734787 Genecard: MECP2 chromosome X SNPedia: rs1734787(A;A)

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★ rs61748396(C;-) good:0.00 **common in clinvar**

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

OMIM 300005

dbSNP: rs61748396 Genecard: MECP2 chromosome X SNPedia: rs61748396(C;C)

★ rs61751362(C;-) good:0.00 **common in clinvar**

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

OMIM 300005

dbSNP: rs61751362 Genecard: MECP2 chromosome X SNPedia: rs61751362(C;C)

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★ rs1859767(A;A) good:0.00 38.12% **common in complete genomics**

Gene related topics:

PSYCH(0.03) · autism(0.03)

Relevant papers and external links:

dbSNP: rs1859767 Genecard: NRCAM chromosome 7 SNPedia: rs1859767(A;A)

★ rs740602(G;G) good:0.00 88.38% **common in complete genomics**

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) · 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:

dbSNP: rs740602 Genecard: COMT chromosome 22 SNPedia: rs740602(G;G)

★ **rs1804197(C;C)** good:0.00 77.90% **common in complete genomics**

rs1804197, a SNP in the adenomatous polyposis of the colon APC gene, is associated with autism .

Although many SNPs have been found in this gene in association with (successful) hunts for mutations predisposing individuals to colon cancers, this previously unidentified SNP is located in the 3' untranslated portion of the gene. Also in conjunction with a common four-SNP haplotype (TGAG for rs2229992, rs42427, rs459552, and rs465899), this SNP is reported to be associated with Asperger's sydrome in the Swedish population studied.

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Gene related topics:

CANCER(4.76) · adenomatous polyposis coli(4.76) · colon polyps(3.00) · adenomatous polyposis coli colorectal neoplasms(2.00) · colorectal adenomas(2.00) · familial adenomatous polyposis(2.00) · autism(1.00) · colorectal cancer(0.83) · adenoma colorectal neoplasms(0.28) · colorectal neoplasms(0.15) · pancreatic cancer(0.08) · IMMUNE(0.03) · inflammatory bowel disease(0.03)

Relevant papers and external links:

PMID 17221838

PMID 17221838

OMIM 114500

dbSNP: rs1804197 Genecard: APC chromosome 5 SNPedia: rs1804197(C;C)

★ **rs9907627(G;G)** good:0.00 **common in complete genomics**

Gene related topics:

CANCER(4.00) · neurofibromatosis1(4.00) · PSYCH(0.03) · autism(0.03)

Relevant papers and external links:

dbSNP: rs9907627 Genecard: NF1 chromosome 17 SNPedia: rs9907627(G;G)

★ rs61748408(C;-) good:0.00 **common in clinvar**

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Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

dbSNP: rs61748408 Genecard: MECP2 chromosome X SNPedia: rs61748408(C;C)

★ rs28934908(C;-) good:0.00 **common in clinvar**

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

OMIM 300005

dbSNP: rs28934908 Genecard: MECP2 chromosome X SNPedia: rs28934908(C;C)

★ rs28934906(C;-) good:0.00 **common in clinvar**

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

OMIM 300005

dbSNP: rs28934906 Genecard: MECP2 chromosome X SNPedia: rs28934906(C;C)

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★ rs61749715(C;-) good:0.00 **common in clinvar**

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

OMIM 300005

dbSNP: rs61749715 Genecard: MECP2 chromosome X SNPedia: rs61749715(C;C)

★ rs61751449(C;-) good:0.00 **common in clinvar**

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

OMIM 300005

dbSNP: rs61751449 Genecard: MECP2 chromosome X SNPedia: rs61751449(C;C)

★ rs28935468(C;-) good:0.00 **common in clinvar**

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Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

OMIM 300005

dbSNP: rs28935468 Genecard: MECP2 chromosome X SNPedia: rs28935468(C;C)

★ rs4531(G;G) good:0.00 87.37% **common on affy axiom data**

Cholinergic nicotinic receptor genes implicated in a nicotine dependence association study targeting 348 candidate genes with 3713 SNPs.

Gene related topics:

PSYCH(2.00) · UNKNOWN(2.00) · dopamine beta hydroxylase activity(2.00) · interpersonal sensitivity paranoid ideation psychoticism(2.00) · perceptual disorders(1.00) · nicotine(1.00) · nicotine dependence(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 17135278

dbSNP: rs4531 Genecard: DBH chromosome 9 SNPedia: rs4531(G;G)

★ rs4648317(C;C) good:0.00 60.83% **normal**

rs4648317, a SNP in the dopamine D2 receptor DRD2 gene, has been linked to higher rates of smoking and higher nicotine dependence scores, based on a study of ~220 adolescents. The risk allele rs4648317 (T).

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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) · nicotine(1.00) · nicotine dependence(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:

PMID 18434921

dbSNP: rs4648317 Genecard: DRD2 chromosome 11 SNPedia: rs4648317(C;C)

★ rs61751444(C;-) good:0.00 **common in clinvar**

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

dbSNP: rs61751444 Genecard: MECP2 chromosome X SNPedia: rs61751444(C;C)

★ rs12273539(C;C) good:0.00 71.38% **common in complete genomics**

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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) · 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:

PHARMGKB PA164857049
Evidence: PubMed ID:19414708This intronic variant (T/C) in the BDNF gene was associated with major depressive disorder in a study of 264 controls and 272 Mexican Americans with major depressive disorder. (OR= 1.75; p value=<0.001; risk allele=T)

dbSNP: rs12273539 Genecard: BDNF chromosome 11 SNPedia: rs12273539(C;C)

★ rs61748392(A;-) good:0.00 **common in clinvar**

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

OMIM 300005

dbSNP: rs61748392 Genecard: MECP2 chromosome X SNPedia: rs61748392(A;A)

★ rs61753965(C;-) good:0.00 **common in clinvar**

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Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

OMIM 300005

dbSNP: rs61753965 Genecard: MECP2 chromosome X SNPedia: rs61753965(C;C)

★ rs61753971(G;-) good:0.00 **common in clinvar**

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

OMIM 300005

dbSNP: rs61753971 Genecard: MECP2 chromosome X SNPedia: rs61753971(G;G)

★ rs2518224(A;A) info:0.00 88.68% **normal**

This SNP, located in an intron of the GRIK2 gene, has been linked in one study to increased thoughts of suicide in patients using the anti-depressant drug citalopram. The risk is only seen for individuals carrying two rs2518224(C) alleles, i.e. the rs2518224(C;C) homozygotes. The odds ratio for these individuals is 8.2. If the individual also has at least one rs4825476(G) allele, the odds ratio for having suicidal thoughts is increased to ~15x (CI: 3.7 - 60.6).

Gene related topics:

citalopram(1.00) · suicide(1.00) · NEUROLOGICAL(0.12) · huntington s disease(0.12) · PSYCH(0.05) · autism(0.05)

Relevant papers and external links:

PMID 17898344

PHARMGKB PA162263539
Evidence: PubMed ID:17898344This SNP in the GRIK2 gene was associated with treatment-emergent suicidal ideation during citalopram therapy in a clinically representative cohort of outpatients with major depressive disorder. DNA samples from 1,915 participants were genotyped.

dbSNP: rs2518224 Genecard: GRIK2 chromosome 6 SNPedia: rs2518224(A;A)

★ rs28934905(T;-) good:0.00 **common in clinvar**

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

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Relevant papers and external links:

OMIM 300005

dbSNP: rs28934905 Genecard: MECP2 chromosome X SNPedia: rs28934905(T;T)

★ rs155100(A;T) good:0.00 49.49% **common in complete genomics**

Gene related topics:

PSYCH(0.03) · autism(0.03)

Relevant papers and external links:

dbSNP: rs155100 Genecard: ITGA4 chromosome 2 SNPedia: rs155100(A;T)

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★ rs17110747(G;G) good:0.00 73.04% **common in complete genomics**

Gene related topics:

OTHER(1.00) · chronic fatigue syndrome(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:

dbSNP: rs17110747 Genecard: TPH2 chromosome 12 SNPedia: rs17110747(G;G)

★ rs11178997(T;T) info:0.00 72.05% **common for Caucasians on the 23andme platform**

common for Caucasians on the 23andme platform

rs4570625 and rs11178997 in TPH2's regulatory region display preferential transmission

paper Association with and bipolar disorder in a Northern Swedish, Isolated Population. 182 patients and 364 unrelated control individuals. P = .001

rs11178997 significantly reduced TPH2 transcriptional activity by 22% and 7% in cell lines might have implications for the development and function of the serotonergic system in the brain.

paper Carriers of the A-Allele of rs11178997 showed a decreased cortisol and ACTH stimulation. might be crucial factors for major depression

pdf paper associated with bipolar disorder

Minor allele yielded odds ratio of 1.5 (CI:1.2-1.9) for increased risk for bipolar disorder in a study of 883 Russian patients.

Transmission disequilibrium of polymorphic variants in the tryptophan hydroxylase-2 gene in attention-deficit/hyperactivity disorder.

Association of brain-specific tryptophan hydroxylase, TPH2, with unipolar and bipolar disorder in a Northern Swedish, isolated population.

Tryptophan hydroxylase 2 gene and alcohol use among college students.

Investigation of the tryptophan hydroxylase 2 gene in bipolar I disorder in the Romanian population.

Alternative splicing and extensive RNA editing of human TPH2 transcripts.

Pharmacogenetics of antidepressant response.

Common variants in the TPH2 promoter confer susceptibility to paranoid schizophrenia.

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Gene related topics:

OTHER(1.00) · chronic fatigue syndrome(1.00) · bipolar disorder(1.00) · brain(1.00) · alcohol(1.00) · schizophrenia(1.00) · major depression(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 17568567

PMID 17905754

OMIM 143465

OMIM 607478

OMIM 125480

OMIM 613003

PMID 16116490

PMID 17015812

PMID 18782386

PMID 18797398

PMID 20126463

PMID 21172166

PMID 22392150

dbSNP: rs11178997 Genecard: TPH2 chromosome 12 SNPedia: rs11178997(T;T)

★ rs8192466(C;C) good:0.00 99.84% **common**

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) · 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:

OMIM 113505

dbSNP: rs8192466 Genecard: BDNF chromosome 11 SNPedia: rs8192466(C;C)

★ rs61751439(C;-) good:0.00 **common in clinvar**

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

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Relevant papers and external links:

dbSNP: rs61751439 Genecard: MECP2 chromosome X SNPedia: rs61751439(C;C)

★ rs4846051(A;A) good:0.00 81.53% **common in complete genomics**

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) · 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) ·

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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:

PHARMGKB PA164918207
Evidence: PubMed ID:16439441The C allele of this variant was associated with increased risk of toxicity in African American Rheumatoid Arthritis patients receiving methotrexate.

dbSNP: rs4846051 Genecard: MTHFR chromosome 1 SNPedia: rs4846051(A;A)

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★ rs61750240(C;-) good:0.00 **common in clinvar**

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

OMIM 300005

dbSNP: rs61750240 Genecard: MECP2 chromosome X SNPedia: rs61750240(C;C)

★ rs736707(T;T) good:0.00 40.19% **common in complete genomics**

Gene related topics:

UNKNOWN(2.00) · otosclerosis(2.00) · PSYCH(0.22) · autism(0.22)

Relevant papers and external links:

dbSNP: rs736707 Genecard: RELN chromosome 7 SNPedia: rs736707(T;T)

★ rs25533(T;T) good:0.00 85.95% **common in complete genomics**

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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:

dbSNP: rs25533 Genecard: SLC6A4 chromosome 17 SNPedia: rs25533(T;T)

★ rs45590836(G;G) good:0.04 **common in complete genomics**

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) · 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) ·

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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:

OMIM 607093

OMIM 607093

dbSNP: rs45590836 Genecard: MTHFR chromosome 1 SNPedia: rs45590836(G;G)

★ rs6869645(C;C) good:0.03 85.29% **common in complete genomics**

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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) · 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: rs6869645 Genecard: SLC6A3 chromosome 5 SNPedia: rs6869645(C;C)

★ rs12364283(A;A) good:0.15 90.76% **common in complete genomics**

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) · 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 PA162355756
Evidence: PubMed ID:18077373; PubMed ID:18829695This promoter variant affects total DRD2 mRNA expression. The C allele is associated with enhanced total DRD2 mRNA expression and is associated with working memory activity in prefrontal cortex and striatum of schizophrenia patients.

dbSNP: rs12364283 Genecard: DRD2 chromosome 11 SNPedia: rs12364283(A;A)

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★ **rs1042714(G;G)** info:0.04 4.17%(rare) **complex; see details for increased risks**

Several susceptibilities have been linked to rs1042714, a SNP in the ADRB2 gene that is also known as the Q27E SNP. The rs1042714(C) allele encodes the glutamine (Gln; "Q"), and the rs1042714(G) allele encodes the glutamic acid (Glu; "E").

A study of 304 patients found that the Glu27 allele led to increased risk for idiopathic venous thromboembolism; the reported odds ratio was 1.40 (CI: 1.09-1.79, p=0.006) for carriers of at least one risk allele.

A study of 334 families with at least one child with autism found that increased risk associated with the rs1042714(G;G) homozygous genotype; the odds ratio reported was between 1.33-1.60 (CI: 1.07-2.58). The risk was approximately doubled among mothers who had clinical markers for pregnancy related stress.

In a study of 342 patients with type-2 diabetes, the rs1042714(G;G) genotype was associated with reduced risk compared to carriers of a rs1042714(C) allele, with an odds ratio of 0.56 (CI: 0.36-0.91).

A study of 294 Italian ischemic stroke patients found increased risk associated with the rs1042714(G;G) genotype, with an odds ratio of 1.68 (CI: 1.17-2.41, p=0.005).

A large study of almost 8,000 patients found no consistent evidence for association with obesity, type-2 diabetes or hypertension, however, there was some association between the rs1042714(G) allele and systolic blood pressure.

Among 215 adults treated with topical beta-blockers to reduce intraocular pressure (IOP), rs1042714(C;C) genotypes were significantly more likely to experience a (desirable) IOP decrease of 20% or more (odds ratio 2.00, CI: 1.00-4.02).

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) · thromboembolism(1.00) · stroke(1.00) · blood pressure(1.00) · systolic blood pressure(1.00) · venous thromboembolism(1.00) · autism(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)

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Relevant papers and external links:

PMID 16651467

PMID 17199132

PMID 17150099

PMID 17531924

PMID 17221209

PMID 18625943

OMIM 109690

PHARMGKB PA164857062
Evidence: PubMed ID:15153795; PubMed ID:15867853; PubMed ID:15987731Three recent meta-analyses have shown that the Gln27Glu and Gly16Arg polymorphisms are not associated with asthma.

PHARMGKB PA161145205
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 PA162167948
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 PA164857063
Evidence: PubMed ID:12835612; PubMed ID:15861037Heart failure patients homozygous for Gln27 were less likely to have improved left ventricular ejection fraction after carvedilol treatment compared to Glu27 carriers. However, in another study of heart failure patients, Gln27Glu polymorphism was not associated with

the improvement of left ventricular ejection fraction or decrease in heart rate due to a beta blocker in stable congestive heart failure.

dbSNP: rs1042714 Genecard: ADRB2 chromosome 5 SNPedia: rs1042714(G;G)

★ rs5945175(T;-) good:0.10 96.90%

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

dbSNP: rs5945175 Genecard: MECP2 chromosome X SNPedia: rs5945175(T;T)

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★ rs1076560(C;C) info:0.21 59.35% **normal**

rs1076560 is located in intron 6 of the dopamine receptor D2 gene.

In one study of Japanese males, rs1076560(A) alleles were 1.3 fold more associated with Alcoholism than the rs1076560(C) alleles.

The DRD2 risk allele A was more prevalent in the alcoholic patients than in the healthy controls. These data identify rs1076560 as a potentially important variable in the development of alcoholism.

rs1076560 and the DAT 3'-VNTR variant influences memory

Genetic variants altering dopamine D2 receptor expression or function modulate the risk of opiate addiction and the dosage requirements of methadone substitution

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) · 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:

PMID 17196743

PMID 19176830

PMID 19373123

PHARMGKB PA162360621
Evidence: PubMed ID:17196743This variant is a potential marker for alcoholism. A allele was more prevalent in the alcoholic patients than in the healthy controls in a study consists of 248 alcoholic patients and 322 healthy controls (all Japanese males).

PHARMGKB PA162355758
Evidence: PubMed ID:18077375; PubMed ID:18829695This variant is in intron 6, and T allele shifts mRNA splicing from the short form (D2S) to the long form (D2L). The T allele carriers are also associated with reduced working memory and attension perfermance.

dbSNP: rs1076560 Genecard: DRD2 chromosome 11 SNPedia: rs1076560(C;C)

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★ rs1800496(C;C) good:0.22 99.84% **common in complete genomics**

origins associated with an independent aspect of decision making in a learning paradigm

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) · decision making(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:

dbSNP: rs1800496 Genecard: DRD2 chromosome 11 SNPedia: rs1800496(C;C)

★ **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.

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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)

★ rs28934579(T;T) good:0.07 **common in complete genomics**

Gene related topics:

PSYCH(1.12) · affective disorder(1.12) · personality(0.06) · personality traits(0.05) · AGING(0.04) · longevity(0.04) · autism(0.03)

Relevant papers and external links:

OMIM 191290

dbSNP: rs28934579 Genecard: TH chromosome 11 SNPedia: rs28934579(T;T)

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★ rs1801028(C;C) good:0.35 93.75% **normal risk**

rs1801028 is a SNP in the dopamine D2 receptor DRD2 gene. A meta-analysis comprising 27 samples and over 3,707 schizophrenia patients concluded that Cys/Ser heterozygotes, i.e. rs1801028(C;G) genotypes, were at elevated risk for schizophrenia when compared to either homozygote genotype (rs1801028(C;C) or rs1801028(G;G)). The odds ratio was 1.4 ($p<0.005$).

An earlier meta-analysis comprising over 9,000 schizophrenia patients concluded pretty much the same thing: the Cys311 (rs1801028(G)) allele frequency led to an odds ratio of 1.43 (CI: 1.16-1.78, $p<0.001$) for this risk allele.

A study of ~120 Chinese patients with schizophrenia concluded that Cys/Ser heterozygotes may not respond to risperidone treatment as well as Ser/Ser homozygotes.

A paper has been published describing mistakes made in assigning allele status for this SNP:

(free full text) Misassigned alleles can annihilate efforts to control quality in otherwise well-designed genetic association analyses. To date, the issue remains underreported, as is exemplified by studies of a diallelic DRD2 missense variant in schizophrenia. For this variant, allele frequency data have been either

misassigned, or incorrectly cited on four consecutive occasions. Contrary to conjecture, low heterozygosity has not guarded against the error with regard to rs1801028, a SNP that features a canonical base pair transversion, G:C.

In the case of rs1801028, rs1801028(C) is the more common allele, encoding the amino acid Serine at position 311, whereas rs1801028(G) encodes Cysteine.

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) · 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 16402354

PMID 12707934

PMID 15140279

PMID 18154681

PHARMGKB PA161145173
Evidence: Web Resource:External Link variant has decreased affinity for dopamine; no clear concensus on association between Ser311Cys polymorphism and schizophrenia.

dbSNP: rs1801028 Genecard: DRD2 chromosome 11 SNPedia: rs1801028(C;C)

★ rs61754424(C;-) good:0.22 **common in clinvar**

rs61754424, also known as c.82C>T, p.Gln28Ter and Q16X, is a mutation in the MECP2 gene on the X chromosome.

The rare rs61754424(T) variant is listed in ClinVar as a pathogenic mutation leading to Rett syndrome, apparently based on it's listing in the RettBase as a nonsense mutation that is disease associated.

However, we have been unable to find verification for this. If you know of someone who is rs61754424(C;T), please contact us if you can say if the person is healthy, or, has symptoms of Rett syndrome.

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

dbSNP: rs61754424 Genecard: MECP2 chromosome X SNPedia: rs61754424(C;C)

★ rs6413429(G;G) info:0.31 91.18%

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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) · 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: rs6413429 Genecard: SLC6A3 chromosome 5 SNPedia: rs6413429(G;G)

★ rs769224(G;G) good:0.54 90.19%

The complex global pattern of genetic variation and linkage disequilibrium at catechol-O-methyltransferase.

Influence and interaction of genetic polymorphisms in catecholamine neurotransmitter systems and early life stress on antidepressant drug response.

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) · stress(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 18574484

PMID 21680027

dbSNP: rs769224 Genecard: COMT chromosome 22 SNPedia: rs769224(G;G)

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★ rs5031002(G;-) good:0.54 98.26% **common on affy axiom data**

Per the 23andMe blog, the minor allele of this SNP (A) was associated with increased LDL cholesterol (wrong allele listed on the 23andMe blog)

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 A +

Established associations:

low density lipoprotein cholesterol measurement **you have 0 copies of the risk allele A**

(risk allele=A, odds ratio/beta 0.3 [0.18-0.41] mmol/l increase with pval 2E-7, pubmedid=19060910)

Gene related topics:

METABOLIC(4.00) · REPRODUCTION(4.00) · androgen insensitivity syndrome(4.00) · infertility male oligospermia(4.00) · CANCER(2.78) · prostate cancer(2.78) · infertility male(2.37) · DEVELOPMENTAL(2.00)

· IMMUNE(2.00) · OTHER(2.00) · acne vulgaris(2.00) · androgen levels(2.00) · bone mass osteoporotic fractures(2.00) · cryptorchidism hypospadias(2.00) · neoplasms prostatic prostatic neoplasms(2.00) · oligospermia(1.80) · UNKNOWN(1.45) · alopecia(1.45) · male pattern baldness(1.00) · cholesterol(1.00) · ldl cholesterol(1.00) · azoospermia oligospermia(0.64) · polycystic ovarian syndrome polycystic ovary syndrome(0.64) · prostatic hyperplasia(0.57) · cryptorchidism(0.45) · AGING(0.44) · aging(0.44) · endometrial neoplasms(0.42) · hypospadias(0.36) · mammographic density(0.36) · ovarian failure premature(0.31) · azoospermia(0.27) · ovarian cancer(0.11) · polycystic ovary syndrome(0.08) · bone mineral density(0.07) · breast neoplasms mammary neoplasms(0.06) · metabolic syndrome(0.04) · prostatic neoplasms(0.04) · PSYCH(0.03) · autism(0.03) · endometriosis(0.02)

Relevant papers and external links:

PMID 19060910
Genome-wide association analysis of metabolic traits in a birth cohort from a founder population.

PHARMGKB PA164740276
Evidence: PubMed ID:19060910; Web Resource:External Link results: Genome-wide association analysis of metabolic traits in a birth cohort from a founder population. (Initial Sample Size: 4,763 individuals; Replication Sample Size: NR); (Region: Xq12; Reported Gene(s): AR; Risk Allele: rs5031002-A); (p-value= 0.0000002).This variant is associated with LDL cholesterol.

dbSNP: rs5031002 GWAS Catalog: rs5031002 Genecard: AR chromosome X

SNPedia: rs5031002(G;G)

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★ rs11214606(C;C) good:0.56 95.77% **common on affy axiom data**

ARVCF single marker and haplotypic association with schizophrenia.

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) · 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:

PMID 19508883

dbSNP: rs11214606 Genecard: DRD2 chromosome 11 SNPedia: rs11214606(C;C)

★ rs4633(C;C) info:0.65 39.49% **normal**

rs4633 is a variant at codon 62 of the COMT gene, however, it does not change the amino acid sequence of the COMT protein.

In a study of 150 (Caucasian) cases of endometrial cancer, a significant increase in rs4633(T;T) genotype was observed in patients compared to controls (OR = 2.39, CI: 1.31-4.37, p = 0.004). Furthemore, the frequency of the C-G haplotype of rs4633-rs4680 was significantly higher in controls (p < 0.0001) than in patients. This correlated with lower expression levels of the COMT protein in carriers of these alleles.

pubmed 1180576 Schizophrenia Susceptibility

Genetic basis for individual variations in pain perception and the development of a chronic pain condition

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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) · cancer(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 18324659

PMID 15537663

OMIM 116790

PHARMGKB PA164944060
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: rs4633 Genecard: COMT chromosome 22 SNPedia: rs4633(C;C)

★ rs3027935(C;-) good:0.35 80.74%

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

dbSNP: rs3027935 Genecard: MECP2 chromosome X SNPedia: rs3027935(C;C)

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★ rs5987201(C;-) good:0.35 82.17%

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

dbSNP: rs5987201 Genecard: MECP2 chromosome X SNPedia: rs5987201(C;C)

★ rs1611115(C;C) info:0.39 61.99% **normal**

rs1611115 (C-1021T) is a SNP near the dopamine beta-hydroxylase (DBH) gene.

per OMIM, the T allele is associated with lower DBH expression in plasma (16 of TT genotype had DBH activity of 4.1, 46 of CT genotype had DBH activity of 25.2, and 112 of CC genotype had DBH activity of 48.1 nmol/min/ml)

Association tests related to affective disorders such as ADHD were performed using four independent samples, healthy volunteers (N = 387), patients with affective disorders (N = 182), adult attention deficit hyperactivity disorder (ADHD) patients (N = 407), and patients with personality disorders (N = 637). Personality disorder patients carrying the DBH TT genotype exhibited higher neuroticism and novelty seeking scores as compared to individuals with the (C;C) or (C;T) genotype. Analyses on the level of the neuroticism and novelty seeking subscales revealed that the DBH (T;T) genotype was primarily associated with personality features related to impulsiveness and aggressive hostility. Also adult ADHD patients carrying the homozygous (T;T) genotypes displayed by significantly increased neuroticism scores; when both personality disorder and adult ADHD patient were analyzed together, (T;T) carriers also displayed by significantly lower conscientiousness levels.

This study concludes that rs1611115(T;T) homozygotes appear to be at increased risk for personality traits related to impulsiveness, aggression, and adult ADHD.

Dopamine-beta-hydroxylase in postural tachycardia syndrome. T allele associated with lower plasma DBH levels, and accounts for up to 52% of variation in plasma DBH, but not linked to increased susceptibility to postural tachycardia syndrome (POTS).

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Gene related topics:

PSYCH(2.00) · UNKNOWN(2.00) · dopamine beta hydroxylase activity(2.00) · interpersonal sensitivity paranoid ideation psychoticism(2.00) · perceptual disorders(1.00) · personality traits(1.00) · neuroticism(1.00) · affective disorder(1.00) · personality disorders(1.00) · novelty seeking(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 18982239

PMID 18982239

PMID 17625104

dbSNP: rs1611115 Genecard: DBH chromosome 9 SNPedia: rs1611115(C;C)

★ rs27048(C;T) info:0.51 43.64% **normal**

[rs27048]], 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, rs27072.

Variations in the DAT1 gene may affect the inattentive subtype of ADHD.

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) · seizures(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

PMID 20800641

dbSNP: rs27048 Genecard: SLC6A3 chromosome 5 SNPedia: rs27048(C;T)

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★ rs6267(G;G) good:0.92 97.34% **common**

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) · 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:

OMIM 116790

PHARMGKB PA164944058

Evidence: PubMed ID:18163386This study showed a highly significant association between a COMT haplotype of two functional SNPs (Ala72Ser and Val158Met) and aggressive behavior in schizophrenia.

dbSNP: rs6267 Genecard: COMT chromosome 22 SNPedia: rs6267(G;G)

★ rs61749721(C;-) good:0.50 **common in clinvar**

Mutation analysis of the MECP2 gene in patients of Slavic origin with Rett syndrome: novel mutations and polymorphisms.

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

OMIM 300005

PMID 17387578

dbSNP: rs61749721 Genecard: MECP2 chromosome X SNPedia: rs61749721(C;C)

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★ rs1718101(G;G) good:0.10 97.38%

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:

dbSNP: rs1718101 Genecard: CNTNAP2 chromosome 7 SNPedia: rs1718101(G;G)

★ rs518147(C;-) info:0.80 47.25% **less weight gain if taking olanzapine**

rs518147, also known as -697G/C, is a SNP in the 5-hydroxytryptamine (serotonin) receptor 2C HTR2C gene.

A study of 107 patients with schizophrenia being treated with olanzapine reported a protective effect against weight-gain from the (C) allele of this SNP; fewer patients (of 28) with a rs518147(C) allele had a body mass index increase of >=10% (p=0.0006), whereas (G;G) homozygotes did. This effect may also involve nearby SNP rs3813929.

The association between HTR2C polymorphisms and obesity in psychiatric patients using antipsychotics: a cross-sectional study.

The association between HTR2C gene polymorphisms and the metabolic syndrome in patients with schizophrenia.

Multivariate permutation analysis associates multiple polymorphisms with subphenotypes of major depression.

HTR2C gene polymorphisms and the metabolic syndrome in patients with schizophrenia: a replication study.

Association of polymorphisms of the serotonergic system with smoking initiation in Caucasians.

HTR2C promoter polymorphisms are associated with risperidone efficacy in Chinese female patients.

Functional consequences of two HTR2C polymorphisms associated with antipsychotic-induced weight gain.

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Gene related topics:

olanzapine(5.00) · weight(5.00) · METABOLIC(3.60) · weight gain(3.60) · PHARMACOGENOMIC(2.00) · weight gain antipsychotic drug induced(2.00) · weight gain antipsychotic induced(2.00) · OTHER(1.00) · weight loss(1.00) · body mass(1.00) · metabolic syndrome(1.00) · schizophrenia(1.00) · major depression(1.00) · body mass index(1.00) · obesity(1.00) · depression(1.00) · smoking(1.00) · PSYCH(0.67) · bipolar affective disorder(0.67) · NEUROLOGICAL(0.33) · migraine with aura(0.33) · mood disorders(0.23) · suicide(0.22) · tardive dyskinesia(0.11) · personality(0.06) · attention deficit hyperactivity disorder(0.05) · personality traits(0.05) · depressive disorder major(0.05) · CHEMDEPENDENCY(0.04) · alcohol abuse(0.04) · autism(0.03)

Relevant papers and external links:

PMID 19434072

PHARMGKB PA164924605
Evidence: PubMed ID:19622037This variant is associated with Olanzapine-induced weight gain. The -697C allele confers protective effect towards Olanzapine-induced weight gain in a study consisting of 107 patients with schizophrenia.

PMID 17016522

PMID 17632216

PMID 18081710

PMID 19142101

PMID 20060656

PMID 20415561

PMID 21391883

dbSNP: rs518147 Genecard: HTR2C chromosome X SNPedia: rs518147(C;C)

★ rs45471299(C;C) good:0.22 **common in clinvar**

Gene related topics:

PSYCH(1.12) · affective disorder(1.12) · personality(0.06) · personality traits(0.05) · AGING(0.04) · longevity(0.04) · autism(0.03)

Relevant papers and external links:

OMIM 191290

dbSNP: rs45471299 Genecard: TH chromosome 11 SNPedia: rs45471299(C;C)

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★ rs28934580(G;G) good:0.22 **common in clinvar**

Gene related topics:

PSYCH(1.12) · affective disorder(1.12) · personality(0.06) · personality traits(0.05) · AGING(0.04) · longevity(0.04) · autism(0.03)

Relevant papers and external links:

OMIM 191290

dbSNP: rs28934580 Genecard: TH chromosome 11 SNPedia: rs28934580(G;G)

★ rs28934581(A;A) good:0.22 **common in clinvar**

Gene related topics:

PSYCH(1.12) · affective disorder(1.12) · personality(0.06) · personality traits(0.05) · AGING(0.04) · longevity(0.04) · autism(0.03)

Relevant papers and external links:

OMIM 191290

dbSNP: rs28934581 Genecard: TH chromosome 11 SNPedia: rs28934581(A;A)

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★ rs61749738(C;-) good:0.59 **common/normal**

rs61749738, also known as c.719C>G, p.Thr240Ser and T240S, as well as c.683C>G, p.Thr228Ser and T228S, represents a rare mutation in the MECP2 gene, located on the X chromosome.

Different sources differ on whether the rare rs61749738(G) allele leads to Rett syndrome, one of the most common forms of mental retardation in females. Some sources report that this allele is benign; others conclude it is pathogenic, including the authors of .

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

PMID 25741868

dbSNP: rs61749738 Genecard: MECP2 chromosome X SNPedia: rs61749738(C;C)

★ 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

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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.

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.

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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

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 disease(0.50) · colonic neoplasms rectal neoplasms(0.50) · thrombosis venous

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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.

PHARMGKB PA162316602

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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.

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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 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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★ rs56164415(G;G) good:0.35 80.85%

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) · 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:

dbSNP: rs56164415 Genecard: BDNF chromosome 11 SNPedia: rs56164415(G;G)

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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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★ rs37022(T;T) good:1.02 41.52% **common in complete genomics**

D2 dopamine receptor gene haplotypes and their influence on alcohol and tobacco consumption magnitude in alcohol-dependent individuals.

Neurotransmission and bipolar disorder: a systematic family-based association study.

Association of tagging single nucleotide polymorphisms on 8 candidate genes in dopaminergic pathway with schizophrenia in Croatian population.

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

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) · 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 17526637
PMID 18444252
PMID 19673036
PMID 19693267

dbSNP: rs37022 Genecard: SLC6A3 chromosome 5 SNPedia: rs37022(T;T)

★ rs41282918(A;A) good:0.53 **common in complete genomics**

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) · 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)

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Relevant papers and external links:

PHARMGKB PA164857053
Evidence: PubMed ID:19414708This variant (A/C) in the 3'UTR of the BDNF gene was associated with major depressive disorder in a study of 264 controls and 272 Mexican Americans with major depressive disorder. (OR= 2.13; p value=0.01; risk allele=A)

dbSNP: rs41282918 Genecard: BDNF chromosome 11 SNPedia: rs41282918(A;A)

★ rs1042173(G;T) info:0.38 49.96% **normal**

rs1042173 is a SNP in the solute carrier family 6 (neurotransmitter transporter, serotonin), member 4 SLC6A4 gene.

A study of 275 patients seeking treatment for alcoholism concluded that Caucasians of with the rs1042173 (T;T) genotype consumed an average of 11.17 drinks per drinking day, compared with an average of 8.58 for carriers of a rs1042173(G) allele (p = 0.0034). While this held true for both men and women, this

association was not seen in Hispanics.

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)

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Relevant papers and external links:

PMID 19032574

dbSNP: rs1042173 Genecard: SLC6A4 chromosome 17 SNPedia: rs1042173(G;T)

★ 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
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)

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★ **rs4298437(C;C)** **good:0.19** **56.40%**

The risk allele is T (in dbSNP orientation), according to a table published by the National Human Genome Research Institute.

Established associations:

alzheimers disease **you have 0 copies of the risk allele A**
(risk allele=A complex/no impact summary)

Gene related topics:

UNKNOWN(2.00) · otosclerosis(2.00) · PSYCH(0.22) · autism(0.22)

Relevant papers and external links:

PMID 20452100
Alzheimer disease pathology in cognitively healthy elderly: a genome-wide study.

dbSNP: rs4298437 GWAS Catalog: rs4298437 Genecard: RELN chromosome 7

SNPedia: rs4298437(C;C)

★ 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.

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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.

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."

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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

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)

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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
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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★ rs9534505(G;G) good:0.48 81.24% **common in complete genomics**

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) · 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:

dbSNP: rs9534505 Genecard: HTR2A chromosome 13 SNPedia: rs9534505(G;G)

★ **rs2283123(C;C)** **info:1.08** **78.60%** **normal 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 a decreased risk of schizophrenia.

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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: rs2283123 Genecard: DBH chromosome 9 SNPedia: rs2283123(C;C)

★ **rs6350(C;C)** **info:1.39** **91.10%**

Genetic polymorphisms in monoamine neurotransmitter systems show only weak association with acute post-surgical pain in humans.

SLC6A3 and body mass index in the Prostate, Lung, Colorectal and Ovarian Cancer Screening Trial.

Familiality and molecular genetics of attention networks in ADHD.

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

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) · ovarian cancer(1.00) · pain(1.00) · CANCER(1.00) · body mass(1.00) · cancer(1.00) · nicotine(1.00) · body mass index(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 16848906

PMID 19183461

PMID 19418498

PMID 19693267

dbSNP: rs6350 Genecard: SLC6A3 chromosome 5 SNPedia: rs6350(C;C)

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★ rs3914132(T;T) info:0.25 65.11% **Normal (not lower) otosclerosis risk**

Each C allele at rs3914132 decreases the likelihood of developing otosclerosis, which can cause hearing loss.

Gene related topics:

UNKNOWN(2.00) · otosclerosis(2.00) · hearing loss(1.00) · PSYCH(0.22) · autism(0.22)

Relevant papers and external links:

OMIM 166800

PHARMGKB PA164857082

Evidence: PubMed ID:19230858In a GWAS done in Belgian-Dutch cases and controls (694 of each, split into a discovery and a replication set) and then in a second replication set of 455 French cases and 480 controls, rs3914132 was found to be associated with otosclerosis.

PHARMGKB PA163522158
Evidence: PubMed ID:19230858This variant in the RELN gene was highly associated with otosclerosis in a GWAS in 302 case and 302 controls.

PHARMGKB PA164739953
Evidence: PubMed ID:19230858; Web Resource:External Link results: A Genome-wide Analysis Identifies Genetic Variants in the RELN Gene Associated with Otosclerosis. (Initial Sample Size: 302 cases, 302 controls; Replication Sample Size: 847 cases, 872 controls); (Region: 7q22.1; Reported Gene(s): RELN; Risk Allele: rs3914132-?); (p-value= 0.00000002).This variant is associated with Otosclerosis.

dbSNP: rs3914132 Genecard: RELN chromosome 7 SNPedia: rs3914132(T;T)

★ rs17435(A;-) info:1.18 19.90% **normal**

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rs17435 is one of several SNPs in the methyl CpG binding protein 2 (MECP2) that have been associated with risk for systemic lupus erythromatosis (SLE). The MECP2 gene is located on the X chromosome, which may be instructive since lupus is a predominantly female disease.

An initial survey of 600 Korean patients, followed by 1,000 Caucasian patients, ultimately led to a meta-analysis indicating an odds ratio for the rs17435(T) risk allele of 1.39 (CI: 1.24â€“1.56, p = 1.2Ã—10e-08).

Variants within MECP2, a key transcription regulator, are associated with increased susceptibility to lupus and differential gene expression in patients with systemic lupus erythematosus.

Replication of recently identified systemic lupus erythematosus genetic associations: a case-control study.

A common MECP2 haplotype associates with reduced cortical surface area in humans in two independent populations.

Gene related topics:

NEUROLOGICAL(15.75) · rett syndrome(15.75) · DEVELOPMENTAL(3.00) · epilepsy rett syndrome(3.00) · mental disorder(3.00) · mental retardation rett syndrome(2.00) · systemic lupus erythematosus(1.00) · lupus erythematosus(1.00) · lupus(1.00) · mental retardation(0.60) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

PMID 18320046
OMIM 300809

PMID 19333917
PMID 19442287
PMID 19717458

dbSNP: rs17435 Genecard: MECP2 chromosome X SNPedia: rs17435(A;A)

★ rs2745557(A;G) info:0.09 24.93% **autism risk?**

A study of 151 Korean families reported that the rs2745557(A) allele of the PTGS2 gene, also known as cyclooxygenase-2 (Cox-2), was overtransmitted ($p<0.01$) in autism spectrum disorders.

Gene related topics:

autism(5.00) · CANCER(2.00) · CARDIOVASCULAR(2.00) · heart disease ischemic stroke(2.00) · helicobacter infections inflammation precancerous conditions stomach neoplasms(2.00) · adenoma colorectal neoplasms(0.28) · METABOLIC(0.24) · osteoarthritis knee(0.24) · skin cancer non melanoma(0.14) · carcinoma squamous cell esophageal neoplasms(0.12) · esophageal cancer(0.11) · HEMATOLOGICAL(0.07) · hemoglobins(0.07) · stomach cancer(0.06) · hematocrit(0.05) · IMMUNE(0.04) · sarcoidosis(0.04) · pancreatic neoplasms(0.03)

Relevant papers and external links:

PMID 18579107

dbSNP: rs2745557 Genecard: PTGS2 chromosome 1 SNPedia: rs2745557(A;G)

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★ rs2229860(C;C) good:0.29 99.72% **common in complete genomics**

Gene related topics:

UNKNOWN(2.00) · otosclerosis(2.00) · PSYCH(0.22) · autism(0.22)

Relevant papers and external links:

dbSNP: rs2229860 Genecard: RELN chromosome 7 SNPedia: rs2229860(C;C)

★ rs1328674(G;G) good:0.70 89.85% **average**

rs1328675 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 rs1328674(A) in dbSNP orientation. The overall risk for the haplotype CTCC of SNPs rs6311-rs1328674-rs6313-rs6314 is 1.68 (CI: 1.20 - 2.34, p = 0.02). This is actually quite close to the odds ratio calculated for rs1328674 by itself; OR of 1.62 (p = 0.007).

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 should be CACC, rather than CTCC as published.

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) · 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

dbSNP: rs1328674 Genecard: HTR2A chromosome 13 SNPedia: rs1328674(G;G)

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★ rs5918(T;T) good:1.34 83.02% **average**

The 'A2' allele of the platelet specific alloantigen system is encoded by rs5918(C), and it has been implicated as increasing the risk of myocardial infarctions, heart disease, and resistance to blood-thinning benefits of aspirin.

On its own, the A2 allele is implicated especially in early onset heart disease ; in combination with the 4G allele of the PAI1 gene, rs1799889, the increased risk of myocardial infarction in a Finnish study population was 4 fold higher (odds ratio = 4.5, p=0.001), particularly in males (odds ratio = 6.4, p=0.0005) . Olympic skater Sergei Grinkov, who had this risk factor, died of a heart attack at age 28.

A2 allele carriers also appear to be relatively resistant to the anti-thrombotic (i.e. anti-clotting) actions normally associated with aspirin use.

A protective effect of rs5918 has also been observed for the development of Non-Hodgkin Lymphoma, both for the SNP (which is also known as L59P) and for its gene, ITGB3. The odds ratio is 0.66 (CI: 0.52-0.85).

rs5918 is not associated with breast cancer risk for either BRCA1 or BRCA2 mutation carriers, based on a multi-center study including ~10,000 patients from 34 studies.

Gene related topics:

PHARMACOGENOMIC(3.00) · aspirin resistance(3.00) · CARDIOVASCULAR(2.78) · myocardial infarct atherosclerosis coronary(2.78) · atherosclerosis generalized(1.00) · myocardial infarction stroke(1.00) · lymphoma(1.00) · breast cancer(1.00) · heart disease(1.00) · myocardial infarct lymphoproliferative disorders restenosis(0.67) · myocardial infarct(0.31) · heart disease ischemic(0.30) · CANCER(0.21) · cancer(0.21) · subarachnoid hemorrhage(0.21) · coronary artery disease(0.14) · atherosclerosis coronary(0.13) · HEMATOLOGICAL(0.13) · platelet aggregation(0.13) · restenosis(0.12) · stroke ischemic(0.11) · thrombosis(0.11) · cardiovascular diseases(0.10) · REPRODUCTION(0.09) · pregnancy loss(0.09) · NORMALVARIATION(0.09) · normal variation(0.09) · stroke(0.09) · myocardial infarction(0.08) · cardiovascular(0.08) · brain ischemia stroke(0.08) · acute coronary syndrome(0.06) · PSYCH(0.05) · autism(0.05) · pregnancy loss recurrent(0.04) · METABOLIC(0.02) · waist hip ratio(0.02)

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Relevant papers and external links:

PMID 8598867

PMID 9700201

PMID 11723016

PMID 17827388

PMID 19876733

OMIM 173470

PHARMGKB PA165291873
Evidence: PubMed ID:20138334Risk or phenotype-associated genotype: T/T Phenotype: This study investigated the association of ITGB3 SNP with a greater prevalence of platelet hyperactivity (HPR) in stable coronary artery disease. HPR patients with inadequate aspirin inhibition were significantly more often homozygous PlA1/A1 (T/T) (65.4% vs. 47.7%, p=0.015). Study size: 188 Study population/ethnicity: patients with stable coronary artery disease, most of them were males (89.9%), current smokers (71.3%), type 2 diabetics (38.3%) or having hypertension (26.6%)

PHARMGKB PA165108281
Evidence: PubMed ID:11723016Risk or phenotype-associated allele: C. Phenotype: In a study of clotting times of microvascular injury, the P1A2 allele (C variant) was associated with shorter bleeding time and less response to aspirin. Study size: 24. Study population/ethnicity: Healthy males 21-24 years. Significance metric(s): p = 0.003. Type of association: PD.

PHARMGKB PA165108282

Evidence: PubMed ID:11545752Risk or phenotype-associated allele: C. Phenotype: Patients with the PI(A1A2) genotype (C allele carriers) had a higher risk of secondary coronary events which was reduced by pravastatin. There was also an interaction between PI(A1A2) and the ACE:I/D variant and pravastatin response. Study size: 767. Study population/ethnicity: Subset of Cholesterol And Recurrent Events (CARE) trial, men and women with myocardial infarction receiving placebo or pravastatin. Significance metric(s): . Type of association: CO.

dbSNP: rs5918 Genecard: ITGB3 chromosome 17 SNPedia: rs5918(T;T)

★ 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.

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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

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)

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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)

★ 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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★ 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)

★ rs11615016(A;A) good:0.44 95.03%

Gene related topics:

OTHER(1.00) · chronic fatigue syndrome(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:

dbSNP: rs11615016 Genecard: TPH2 chromosome 12 SNPedia: rs11615016(A;A)

★ rs7794745(A;A) good:0.71 24.46% **normal risk (for autism)**

A common SNP in the CNTNAP2 gene, rs7794745, is associated with increased risk for autism based on a study of 148 affected children from families with two more autistic children.

Related article

A genome-wide association study of autism reveals a common novel risk locus at 5p14.1.

Do candidate genes discriminate patients with an autism spectrum disorder from those with attention deficit/hyperactivity disorder and is there an effect of lifetime substance use disorders?

A common genetic variant in the neurexin superfamily member CNTNAP2 is associated with increased risk for selective mutism and social anxiety-related traits.

Genetic variation in CNTNAP2 alters brain function during linguistic processing in healthy individuals.

Gene related topics:

anxiety(1.00) · brain(1.00) · 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 18179894

OMIM 604569

PHARMGKB PA162168112

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Evidence: PubMed ID:18179894This common variant in CNTNAP2 is significantly associated with autism susceptibility in two independent family-based samples.

OMIM 604569

PMID 19456320

PMID 20446882

PMID 21193173

PMID 21987501

dbSNP: rs7794745 Genecard: CNTNAP2 chromosome 7 SNPedia: rs7794745(A;A)

★ 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.

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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
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.

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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)

★ 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.

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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.

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

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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
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)

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★ rs968671(A;A) good:0.75 93.48% **common in complete genomics**

Repeatedly associated with increased body mass index and hypertension according to

Gene related topics:

body mass(1.00) · hypertension(1.00) · body mass index(1.00) · CARDIOVASCULAR(0.19) · blood pressure determination(0.19) · PSYCH(0.09) · autism(0.09)

Relevant papers and external links:

PMID 19161620

dbSNP: rs968671 Genecard: GABRG3 chromosome 15 SNPedia: rs968671(A;A)

★ rs4251417(G;G) info:1.15 94.53%

Association between a functional polymorphism in the serotonin transporter gene and diarrhoea predominant irritable bowel syndrome in women.

Sexually dimorphic effects of four genes (COMT, SLC6A2, MAOA, SLC6A4) in genetic associations of ADHD: a preliminary study.

Variants of the serotonin transporter gene and NEO-PI-R Neuroticism: No association in the BLSA and SardiNIA samples.

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)

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Relevant papers and external links:

PHARMGKB PA164920374
Evidence: PubMed ID:19541292This study identified a two-SNP haplotype proxy for 5HTTLPR (the length polymorphism repeat in the promoter region of the SLC6A4 gene) ; the CA haplotype of SNPs rs4251417 and rs2020934 is coupled with the short allele of 5HTTLPR (r(2) = .72).

PMID 15361494

PMID 18937309

PMID 19199283

dbSNP: rs4251417 Genecard: SLC6A4 chromosome 17 SNPedia: rs4251417(G;G)

★ rs4565946(C;T) info:0.75 40.59%

rs4570625 and rs4565946 linked to the pathogenesis of early-onset obsessive compulsive disorder

Tourette's Syndrome

Gene related topics:

OTHER(1.00) · chronic fatigue syndrome(1.00) · obsessive compulsive disorder(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 16146581

dbSNP: rs4565946 Genecard: TPH2 chromosome 12 SNPedia: rs4565946(C;T)

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★ 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

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)

★ rs28914832(A;A) good:1.40 99.96% **common in complete genomics**

Allelic heterogeneity at the serotonin transporter locus (SLC6A4) confers susceptibility to autism and rigid-compulsive behaviors.

Enhanced activity of human serotonin transporter variants associated with autism.

Characterization of a functional polymorphism in the 3' UTR of SLC6A4 and its association with drinking intensity.

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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:

OMIM 182138

PMID 15995945

PMID 18957375
PMID 19032574

dbSNP: rs28914832 Genecard: SLC6A4 chromosome 17 SNPedia: rs28914832(A;A)

★ rs1861973(C;C) good:0.23 57.61%

nature The inheritance of the AC haplotype of rs1861972 – rs1861973, the C allele of rs1811399, and the C allele of rs1234747 may contribute to autism by affecting microRNA

rs1861972(A) + rs1861973(C) haplotype assoc. with protective effect against autism in a study of 818 Han Chinese (184 diag. autistics, 225 unrelated volunteers, 409 randomly selected)

Gene related topics:

PSYCH(0.17) · autism(0.17)

Relevant papers and external links:

PMID 18424904
OMIM 611016
OMIM 131310
OMIM 131310

dbSNP: rs1861973 Genecard: EN2 chromosome 7 SNPedia: rs1861973(C;C)

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★ 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)

★ 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)

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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 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)

★ rs6311(C;T) good:2.42 49.36% **Normal risk of sexual dysfunction when taking SSRI Antidepressants.**

rs6311 (-1438A>G / A-1438G or -1438G>A / G-1438A) is a SNP located upstream (and within the promoter region of) HTR2A.

Multiple Regulatory Variants Modulate Expression of 5-Hydroxytryptamine 2A Receptors in Human Cortex. The minor A allele of rs6311 reduces expression of a previously unannotated extension of the 5' untranslated region of HTR2A mRNA. rs6311 together with rs6314 have a modest effect on depression severity, when examined in the Sequenced Treatment Alternatives to Relieve Depression (STAR*D) study.

Three (rs643627-rs594242-rs6311: A-C-T), two (rs594242-rs6311: C-T) and a single functional (rs6311: T) marker were protective against suicidal behavior.
The complementary markers (rs594242-rs6311: G-C and rs6311: C) were associated with increased risk for non-violent and impulsive suicidal behavior.
Furthermore, CC-homozygotes for the functional SNP rs6311 reported more anger- and aggression-related behavior.

This SNP has also been reported to be part of a haplotype associated with risk for rheumatoid arthritis. In this report, the risk allele is rs6311(C).

may effect promoter activity (in the presence of a SV40 enhancer, promoter activity was significantly higher with the A allele than a G allele, but only in cell lines expressing endogenous HTR2A)

-1438 A allele creates a consensus binding site for Th1/E47, a transcription factor implicated in development of the nervous system

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

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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 23158458

PMID 16814396

PMID 18006541

PMID 15364038

PMID 18079067

PHARMGKB PA165111369
Evidence: PubMed ID:19997080Risk or phenotype-associated allele: A Phenotype: Carriers of the A variant of HTR2A:(-1438)G>A had lower Autism Treatment Evaluation Checklist (ATEC) scores than GG homozygotes, indicating improved symptoms and response to risperidone. Study size: 45 Study population/ethnicity: Children with Autism receiving risperidone Significance metric(s): p = 0.019 Type of association: PD

PMID 17440930

PHARMGKB PA161145082
Evidence: PubMed ID:17688403; PubMed ID:17804117Associated with anorexia and bulimia. The AA genotype was found to be a risk factor for tardive dyskinesia (TD).

PHARMGKB PA162190493
Evidence: PubMed ID:15364038; PubMed ID:18079067The A allele of -1438G/A is suggested to have increased promoter activity in functional in vitro studies. In silico analysis revealed that the -1438 A allele creates a consensus binding site for Th1/E47, a transcription factor implicated in the development of the nervous system. The SNP was also associated with Chronic fatigue syndrome together with rs 6313 and rs1923884.

dbSNP: rs6311 Genecard: HTR2A chromosome 13 SNPedia: rs6311(C;T)

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★ rs2351299(G;G) info:0.25 60.61% **normal risk of autism**

Investigation of autism and GABA receptor subunit genes in multiple ethnic groups.

Gene related topics:

autism(5.00)

Relevant papers and external links:

PMID 16770606

dbSNP: rs2351299 Genecard: GABRB1 chromosome 4 SNPedia: rs2351299(G;G)

★ rs6807362(C;G) info:0.25 49.37% **normal autism risk**

rs6807362 is a SNP in the 5-hydroxytryptamine (serotonin) receptor 3, family member C HTR3C gene.

In a study of 97 familes with at least one individual with autism, rs6807362 was significantly associated with the disorder (p = 0.0012).

Gene related topics:

autism(5.00)

Relevant papers and external links:

PMID 19035560

dbSNP: rs6807362 Genecard: HTR3C chromosome 3 SNPedia: rs6807362(C;G)

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★ rs2268494(T;T) good:0.37 86.81%

Gene related topics:

PSYCH(0.28) · autism(0.28)

Relevant papers and external links:

dbSNP: rs2268494 Genecard: OXTR chromosome 3 SNPedia: rs2268494(T;T)

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

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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★ 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)

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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.

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.

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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.

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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.

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)

★ 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.

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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)

★ 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)

★ 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

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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)

★ rs10513026(C;C) good:0.45 92.67% **common in complete genomics**

A genome-wide linkage and association scan reveals novel loci for autism.

Gene related topics:

autism(1.00)

Relevant papers and external links:

OMIM 609297
PMID 19812673

dbSNP: rs10513026 Genecard: nan chromosome 5 SNPedia: rs10513026(C;C)

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★ rs6460013(G;G) good:0.90 77.79%

Support for the homeobox transcription factor gene ENGRAILED 2 as an autism spectrum disorder susceptibility locus.

Gene related topics:

PSYCH(0.17) · autism(0.17)

Relevant papers and external links:

PMID 16252243

dbSNP: rs6460013 Genecard: EN2 chromosome 7 SNPedia: rs6460013(G;G)

★ rs25409(C;C) good:3.02 99.60%

Gene related topics:

PSYCH(0.17) · autism(0.17) · METABOLIC(0.14) · glucose(0.14) · NEUROLOGICAL(0.04) · epilepsy(0.04)

Relevant papers and external links:

OMIM 137192

OMIM 137192

dbSNP: rs25409 Genecard: GABRB3 chromosome 15 SNPedia: rs25409(C;C)

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