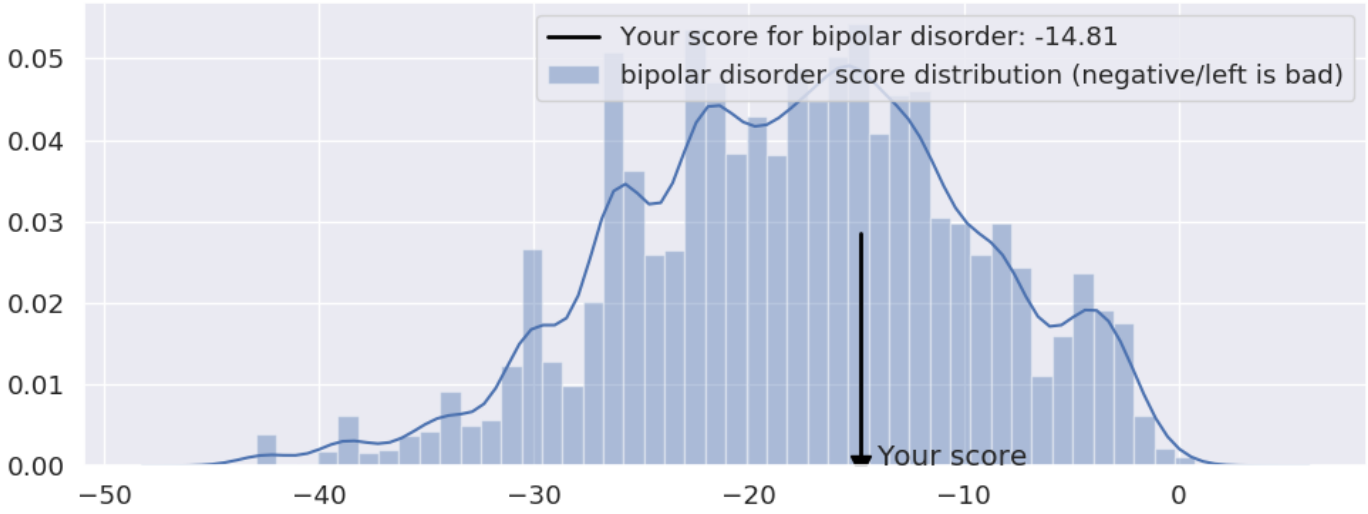
TOPIC: BIPOLAR DISORDER



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★ rs420259(T;T) info:1.81 41.29% **Normal risk of Bipolar Disorder.**

This is the normal version that most people have. It seems to have a higher risk of bipolar disorder than the rare version, based on preliminary research.

Linked to bipolar disorder in one of the most comprehensive studies in 2007. Risk allele with reference to dbSNP orientation is reported to be (T), with either one or two copies leading to an odds ratio of 2 (CI 1.6-2.7).

Some replications, but in 2011 23andMe published as failed to replicate in

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

Findings from bipolar disorder genome-wide association studies replicate in a Finnish bipolar family-cohort.

Simultaneous genotype calling and haplotype phasing improves genotype accuracy and reduces false-positive associations for genome-wide association studies.

Established associations:

bipolar disorder **you have 2 copies of the risk allele T**

(risk allele=T, odds ratio/beta 2.08 [1.60-2.71] with pval 6E-8, pubmedid=17554300)

Gene related topics:

bipolar disorder(5.00)

Relevant papers and external links:

PMID 17554300
OMIM 611247
PHARMGKB PA162356654
Evidence: PubMed ID:17554300; Web Resource:External Link Results: Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls (Initial Sample Size: 1,868 cases, 2,938 controls; Replication Sample Size: NR; Risk Allele: rs420259-A). This variant is associated with bipolar disorder.

PMID 18224312
PMID 19308021
PMID 19931040

dbSNP: rs420259 GWAS Catalog: rs420259 Genecard: PALB2 chromosome 16

SNPedia: rs420259(T;T)

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★ rs4027132(A;G) info:1.09 49.67% **1.39x increased risk of developing bipolar disorder**

rs4027132 has been reported in a large study to be associated with bipolar disorder.

The risk allele (oriented to the dbSNP entry) is (A), and the odds ratio associated with heterozygotes is 1.39 (CI 1.19-1.64), and for homozygotes, 1.51 (CI 1.27-1.79).

Gene related topics:

bipolar disorder(5.00)

Relevant papers and external links:

PMID 17554300

dbSNP: rs4027132 Genecard: nan chromosome 2 SNPedia: rs4027132(A;G)

★ rs683395(C;T) info:1.00 22.56% **1.5x risk**

rs683395 has been reported in a large study to be associated with bipolar disorder.

The risk allele (oriented to the dbSNP entry) is (G); the odds ratio associated with heterozygotes is 1.47 (CI 1.26-1.71), and for homozygotes, 1.3 (CI 0.69-2.46).

Findings from bipolar disorder genome-wide association studies replicate in a Finnish bipolar family-cohort.

Established associations:

bipolar disorder **you have 1 copies of the risk allele C**

(risk allele=C, odds ratio/beta 1.47 [1.26-1.71] with pval 5E-6, pubmedid=17554300)

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Gene related topics:

bipolar disorder(1.00)

Relevant papers and external links:

PMID 17554300

PHARMGKB PA162356655

Evidence: PubMed ID:17554300; Web Resource:External Link Results: Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls (Initial Sample Size: 1,868 cases, 2,938 controls; Replication Sample Size: NR; Risk Allele: rs683395-G). This variant is associated with bipolar disorder.

PMID 19308021

dbSNP: rs683395 GWAS Catalog: rs683395 Genecard: LAMP3 chromosome 3

SNPedia: rs683395(C;T)

★ rs140504(A;G) info:1.00 32.74% **1.4x increased risk for bipolar disorder**

rs140504, a SNP in the BCR gene on chromosome 22, has been associated with increased risk for bipolar disorder. The odds ratio for carriers of the minor allele (G) are reported as 1.45 (CI:1.11 - 1.84, p=0.0054) based on a study of 171 Japanese patients.

Gene related topics:

bipolar disorder(5.00) · CANCER(0.02) · leukemia(0.02)

Relevant papers and external links:

PMID 15866548
OMIM 125480

dbSNP: rs140504 Genecard: BCR chromosome 22 SNPedia: rs140504(A;G)

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★ rs10994336(C;T) warning:2.38 21.60% **1.45x increased odds of developing bipolar disorder**

23andMe blog rs10994336 or (rs4948418) Each T at this SNP increased the odds of developing bipolar disorder by 1.45 times compared to the CC genotype.

This is one of the SNPs reported by NutraHacker.

Established associations:

bipolar disorder **you have 1 copies of the risk allele T**
(risk allele=T, odds ratio/beta 1.45 with pval 9E-9, pubmedid=18711365)

Gene related topics:

METABOLIC(0.14) · cholesterol ldl(0.14) · PSYCH(0.10) · bipolar disorder(0.10) · CARDIOVASCULAR(0.06) · arteries(0.06)

Relevant papers and external links:

PMID 18711365
Collaborative genome-wide association analysis supports a role for ank3 and cacna1c in bipolar disorder.

OMIM 125480

OMIM 612357

PHARMGKB PA162356412
Evidence: PubMed ID:18711365; Web Resource:External Link Results: Collaborative genome-wide association analysis supports a role for ANK3 and CACNA1C in bipolar disorder (Initial Sample Size: 1,098 cases, 1,267 controls; Replication Sample Size: 4,387 cases, 6,209 controls; Risk Allele: rs10994336-T). This variant is associated with Bipolar disorder.

PHARMGKB PA162355588
Evidence: PubMed ID:18711365A genome-wide association analysis study in 4,387 cases and 6,209 controls identified a strong association of this variant in the ANK3 gene with bipolar disorder.

dbSNP: rs10994336 GWAS Catalog: rs10994336 Genecard: ANK3 chromosome 10

SNPedia: rs10994336(C;T)

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

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

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

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

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

Apoptotic engulfment pathway and schizophrenia.

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

Expanding the range of ZNF804A variants conferring risk of psychosis.

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

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

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

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

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

ZNF804A may be associated with executive control of attention.

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

Evidence of sex-modulated association of ZNF804A with schizophrenia.

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

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

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

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

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

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

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

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

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

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Established associations:

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

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

Gene related topics:

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

Relevant papers and external links:

PMID 20688871

PMID 18677311

OMIM 181500

OMIM 612361

PHARMGKB PA164856914

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

PHARMGKB PA162356547

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

PMID 19693005

PMID 19721717

PMID 19911060

PMID 20048749

PMID 20231838

PMID 20368704

PMID 20485477

PMID 20934520

PMID 20957649

PMID 21040459

PMID 21302348

PMID 21349497

PMID 21457757

PMID 21767209

PMID 21810628

PMID 21892778

PMID 21911029

PMID 22328493

PMID 22384243

PMID 22416237

PMID 22425527

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

SNPedia: rs1344706(T;T)

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★ rs1012053(A;A) warning:2.34 59.41% **Normal (higher) risk of Bipolar Disorder.**

This is the most common version of this SNP, so most people have this. But it is suspected of having a higher risk of Bipolar Disorder compared to the other rare versions of this SNP.
Bipolar disorder failed to replicate in

Established associations:

bipolar disorder **you have 2 copies of the risk allele A**

(risk allele=A, odds ratio/beta 1.59 [1.35-1.87] with pval 2E-8, pubmedid=17486107)

Gene related topics:

OTHER(1.00) · PSYCH(0.10) · bipolar disorder(0.10)

Relevant papers and external links:

PMID 17486107
A genome-wide association study implicates diacylglycerol kinase eta (dgkh) and several other genes in the etiology of bipolar disorder.

PHARMGKB PA162356620
Evidence: PubMed ID:17486107; Web Resource:External Link Results: A genome-wide association study implicates diacylglycerol kinase eta (DGKH) and several other genes in the etiology of bipolar disorder (Initial Sample Size: 461 cases, 563 controls; Replication Sample Size: 772 cases, 876 controls; Risk Allele: rs1012053-A).

dbSNP: rs1012053 GWAS Catalog: rs1012053 Genecard: DGKH chromosome 13

SNPedia: rs1012053(A;A)

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★ rs11622475(C;T) info:0.60 37.32% **1.1x risk of bipolar disorder**

rs11622475 has been reported in a large study to be associated with bipolar disorder.

The risk allele (oriented to the dbSNP entry) is (C); the odds ratio associated with heterozygotes is 1.13 (CI 0.89-1.44), and for homozygotes, 1.47 (CI 1.17-1.86).

Established associations:

bipolar disorder **you have 1 copies of the risk allele C**

(risk allele=C, odds ratio/beta 1.13 [0.89-1.44] with pval 8E-6, pubmedid=17554300)

Gene related topics:

bipolar disorder(1.00)

Relevant papers and external links:

PMID 17554300

dbSNP: rs11622475 GWAS Catalog: rs11622475 Genecard: TDRD9 chromosome 14

SNPedia: rs11622475(C;T)

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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)

★ rs1344484(T;T) info:0.48 29.30% **1.5x risk**

rs1344484 has been reported in a large study to be associated with bipolar disorder.

The risk allele (oriented to the dbSNP entry) is (T); the odds ratio associated with heterozygotes is 1.24 (CI 1.03-1.48), and for homozygotes, 1.52 (CI 1.27-1.82).

Findings from bipolar disorder genome-wide association studies replicate in a Finnish bipolar family-cohort.

Gene related topics:

bipolar disorder(1.00)

Relevant papers and external links:

PMID 17554300
PMID 19308021

dbSNP: rs1344484 Genecard: nan chromosome 16 SNPedia: rs1344484(T;T)

★ rs6932590(C;T) info:0.99 33.07% **1.1x increased risk for schizophrenia**

rs6932590 is a SNP that is postulated to have a role in conferring increased susceptibility for schizophrenia. rs6932590 lies in the major histocompatibility complex (MHC) region on chromosome 6, a

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region that codes for several genes with significant roles in autoimmunity, such as human leukocyte antigens. Specifically, rs6932590 lies in an intergenic region between PRSS16 (a serine protease expressed in the thymus that plays a role in positive T cell selection) and POM121L2 (a membrane glycoprotein) .

The T allele at rs6932590 purportedly confers an increased risk for schizophrenia, with reported odds ratios of 1.16 and 1.31 .

Evidence

One large-scale case-controlled genome-wide association study (GWAS) that examined differential genotypes in individuals with schizophrenia, found five significant schizophrenia-associated SNP markers in the MHC region: rs6913660, rs13219354, rs6932590, rs13211507, and rs3131296, with significant linkage disequilibrium between them. This GWAS was performed by the International Schizophrenia Consortium (ISC), Molecular Genetics of Schizophrenia (MGS), and SGENE and included, in all, 12,945 cases and 34,591 controls. Samples were taken from Caucasian populations across Europe (eight separate geographical locations) 23andMe blog. rs6932590 was found to be the most significant SNP between cases and controls, with a p-value of 1.4x10e-12. The T allele was found to confer increased risk for schizophrenia with an odds ratio of 1.16.

In a separate GWAS that was designed to examine only the top eight significant SNPs (i.e., candidates) reported by the previous ISC/SGENE/MGS study , 2,496 cases and 5,184 control subjects from the Shanghai, China, population were genotyped. rs6932590 was again found to be significant (corrected p-value of 0.00096), as well as other SNPs in the MHC region (rs3131296, rs3130375) . The 0.00096 p-value is much higher than the one reported by the aforementioned ISC/SGENE/MGS study, however, the investigators deemed it significant. This study also found the T allele at rs6932590 to confer an increased risk for schizophrenia, but with an odds ratio of 1.31. Overall, this study found only four of the eight candidate SNPs from the ISC/SGENE/MGS study to be significant (i.e., validated) in the genotyped Shanghai, China sample.

Importantly, in both of the above studies, rs2958182, a SNP that lies in the TCF4 gene (outside of the MHC region), was also found to be significant. TCF4 codes for a transcription factor that is primarily expressed in pre-B cells and potentially plays a role in cognitive development .

A recent expression quantitative trait locus (eQTL) study investigated whether the SNPs reported by ISC/MGS/SGENE affect gene expression in normal human cortical brain tissue. The study did not find that rs6932590 was associated with any significant differential expression . However, the investigators noted that MHC region SNPs such as rs6932590 may affect gene expression earlier in development, which is difficult to observe temporally, particularly in the elderly, post-mortem brain samples that were used in the eQTL study.

Biological Explanation

It is still unclear what role immune system genes may have on schizophrenia etiology, or whether the significant genes in the MHC region are even immune genes (there are other, non-immunity genes in the MHC region). One interesting finding is that a single allele variant in a schizophrenia SNP marker in the MHC

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region, rs3131296 (which has an r^2 of .86 with the HLA-DRB1*03 gene), while conferring risk for schizophrenia, confers increased protection for type-1 diabetes, celiac disease, and systemic lupus erythematosus .

There have been hypotheses that a winter or spring season of birth (i.e., an implicit increased risk of maternal influenza infection during pregnancy; which could lead to inflammation and aberrant neuronal development/growth) or early childhood infections (e.g., measles) may catalyze the development of schizophrenia. However, these hypotheses remain unproven and there was no evidence for them in the ISC/SGENE/MGS patient history data.

Competing/Complementary Theories

The polygenic inheritance model of schizophrenia is predicated on the idea that dozens of common variants combine to increase an individual's risk of schizophrenia. However, there has been limited success in finding significant common SNPs that are associated with schizophrenia. For example, in a study consisting of 1,870 schizophrenia cases and 2,002 controls that were genotyped for 789 SNPs that were hand-picked for their purported significance in schizophrenia susceptibility, no statistically significant associations were found . At present, the most optimistic estimates of the magnitude of the effect of the polygenic model on disease susceptibility variance is approximately 20% .

The most promising explanation of schizophrenia heritability may come from the significantly increased presence of rare structural variants such as deletions and duplications in individuals with schizophrenia . Certainly, it is possible that certain SNPs, say in brain development or the immune system, may operate in concert with the effects of rare structural variants.

Other theories to explain the missing heritability include effects from certain copy number variations (CNV) and effects from a few of many highly impactful, independent, rare alleles .

As of May 2011, the Psychiatric GWAS Consortium is working on the largest GWAS for schizophrenia ever performed, with 59,000 cases and 7,700 family trios .

References

Common variants conferring risk of schizophrenia.

Common variants in major histocompatibility complex region and TCF4 gene are significantly associated with schizophrenia in Han Chinese

Common polygenic variation contributes to risk of schizophrenia and bipolar disorder

Schizophrenia susceptibility alleles are enriched for alleles that affect gene expression in adult human brain

No significant association of 14 candidate genes with schizophrenia in a large European ancestry sample: implications for psychiatric genetics

Rare structural variants disrupt multiple genes in neurodevelopmental pathways in schizophrenia

Rare structural variants in schizophrenia: one disorder, multiple mutations; one mutation, multiple disorders

Large recurrent microdeletions associated with schizophrenia

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Schizophrenia: a common disease caused by multiple rare alleles

A framework for interpreting genome-wide association studies of psychiatric disorders

The human genome browser at UCSC

Cognitive and sensorimotor gating impairments in transgenic mice overexpressing the schizophrenia susceptibility gene Tcf4 in the brain

Established associations:

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

(risk allele=T, odds ratio/beta 1.16 with pval 1E-12, pubmedid=19571808)

Gene related topics:

schizophrenia(5.00) · celiac disease(1.00) · bipolar disorder(1.00) · brain(1.00) · measles(1.00) · systemic lupus erythematosus(1.00) · OTHER(1.00) · IMMUNE(1.00) · lupus erythematosus(1.00) · psychiatric disorders(1.00) · INFECTION(1.00) · lupus(1.00) · inflammation(1.00)

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Relevant papers and external links:

PMID 20673877
PMID 12045153
PMID 19571808
PMID 20673877
PMID 19571808
PMID 19571811
PMID 19571808
PMID 20673877
PMID 20434134
PMID 21339752
PMID 19571808
PMID 18198266
PMID 19571811
PMID 18369103
PMID 19883952
PMID 18668039
PMID 17329737
PMID 19002139
PMID 19571808

PMID 20673877
PMID 19571811
PMID 21339752
PMID 18198266
PMID 18369103
PMID 19883952
PMID 18668039
PMID 17329737
PMID 19002139
PMID 12045153
PMID 20434134
OMIM 181500

dbSNP: rs6932590 GWAS Catalog: rs6932590 Genecard: nan chromosome 6

SNPedia: rs6932590(C;T)

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★ 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.

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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★ rs16944(G;G) info:1.92 24.07% **Increased risk of mental disorders**

rs16944 is a SNP in the interleukin 1, beta gene, a member of the cytokine family involved in the inflammatory response. The rs16944 G;G genotype is associated with risk for multiple mental illnesses. An increased risk for schizophrenia was found in a genetic association study of 2111 Caucasian individuals with an odds ratio of 1.24 (95% CI 1.09, 1.41) . Studies examining the brains of schizophrenic and bipolar individuals found reduced gray matter and hypoactivity associated with the G;G genotype. Additionally, this genotype is associated with impaired ability to achieve remission in a study of 256 Caucasian individuals with depression (odds ratio = 1.74; 95% confidence interval 1.2-4.3).

The rs16944 A allele increases susceptibility to Osteoarthritis, with 1.80 times for heterozygotes (AG) and 2.90 times for homozygotes (AA)

Gray matter deficits in bipolar disorder rs16944(T;T)

Gene related topics:

mental disorders(5.00) · IMMUNE(4.49) · periodontitis(4.49) · CANCER(4.00) · helicobacter infections stomach neoplasms(4.00) · INFECTION(2.70) · helicobacter infections(2.70) · NEUROLOGICAL(2.40) · seizures febrile(2.40) · UNKNOWN(2.00) · atrophy helicobacter infections(2.00) · dental plaque gingivitis(2.00) · gastritis(2.00) · gastritis helicobacter infections stomach ulcer stomach ulcer(2.00) · stomach neoplasms(1.92) · ige(1.80) · stomatitis aphthous(1.80) · duodenal ulcer helicobacter infections(1.60) · adenocarcinoma helicobacter infections stomach neoplasms(1.50) · helicobacter infections peptic ulcer(1.29) · otitis media recurrence(1.29) · helicobacter pylori infection(1.12) · meningococcal infections(1.12) · alveolar bone loss(1.00) · gastritis atrophic stomach neoplasms(1.00) · gastroesophageal reflux helicobacter infections(1.00) · helicobacter infections purpura thrombocytopenic idiopathic(1.00) · helicobacter infections stomach ulcer stomach ulcer(1.00) · hepatocellular carcinoma(1.00) · bipolar disorder(1.00) · schizophrenia(1.00) · depression(1.00) · stomach cancer(0.79) · periodontal disease(0.74) · REPRODUCTION(0.69) · premature birth(0.69) · CARDIOVASCULAR(0.67) · heart disease(0.67) · METABOLIC(0.64) · degenerative arthropathy osteoarthritis(0.64) · gastric cancer(0.62) · h pylori infection(0.46) · VISION(0.40) · arthritis rheumatoid rheumatoid arthritis(0.40) · graves ophthalmopathy(0.40) · purpura thrombocytopenic idiopathic werlhof s disease(0.33) · chronic periodontitis(0.31) · meningococcal disease(0.29) · alzheimer s disease(0.26) · osteoarthritis knee(0.24) · mucocutaneous lymph node syndrome(0.22) · carcinoma squamous cell mouth neoplasms squamous cell carcinoma(0.21) · subarachnoid hemorrhage(0.21) · glomerulonephritis iga(0.20) · epilepsy temporal lobe(0.19) · multiple myeloma(0.19) · abortion habitual(0.19) · sepsis(0.18) · adenocarcinoma stomach neoplasms(0.18) · polycystic ovarian syndrome polycystic ovary syndrome(0.16) · osteoarthritis(0.14) · sepsis systemic infection(0.14) · skin cancer non melanoma(0.14) · inflammation(0.13) · arthritis rheumatoid(0.13) · RENAL(0.12) · chronic renal failure kidney failure chronic(0.12) · tuberculosis(0.12) · graves disease graves disease(0.11) · alopecia areata(0.11) · pulmonary disease chronic obstructive(0.10) · PSYCH(0.10) · mood disorders(0.10) · hepatitis b chronic(0.10) · juvenile arthritis(0.09) · rheumatoid spondylitis spondylitis ankylosing(0.09) · ankylosing spondylitis(0.08) · spondylitis ankylosing(0.07) · acute coronary syndrome(0.06) · inflammatory bowel disease(0.06) · c reactive protein(0.06) · arthritis(0.06) · CHEMDEPENDENCY(0.05) · alcohol(0.05) · graft versus host disease(0.05) · endometriosis(0.05) · celiac disease(0.04) · pregnancy loss recurrent(0.04) · NORMALVARIATION(0.04) · normal variation(0.04) · bone mineral density(0.04) · ulcerative colitis(0.04) · sarcoidosis(0.04) · esophageal cancer(0.03) · melanoma(0.03) · cervical cancer(0.02) · OTHER(0.02) · cystic fibrosis(0.02)

Relevant papers and external links:

PMID 16905295

PMID 22763186

PMID 19125864

PMID 20044070

PMID 15077300

PMID 19125864

PHARMGKB PA164926306
Evidence: PubMed ID:16257277This variant was significantly associated with acquired resistance to bisphosphonates treatment in Caucasian patients with Paget's disease of the bone

dbSNP: rs16944 Genecard: IL1B chromosome 2 SNPedia: rs16944(G;G)

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

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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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★ 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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★ **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

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)

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★ **rs1446109(A;A)** **info:0.00** **74.20%**

having minor alleles at all 3 of rs1446109-rs1007371-rs723524 may affect left-right asymmetrical brain function, such as handedness, and much of human cognition, behavior and emotion. pdf full paper

Genome-wide association and meta-analysis of bipolar disorder in individuals of European ancestry.

Gene related topics:

bipolar disorder(1.00) · brain(1.00) · CARDIOVASCULAR(0.06) · arteries(0.06)

Relevant papers and external links:

PMID 17667961
PMID 19416921

dbSNP: rs1446109 Genecard: CTNNA2 chromosome 2 SNPedia: rs1446109(A;A)

★ rs3761218(T;T) info:0.00 43.29% **normal**

rs3761218 has been reported in a large study to be associated with bipolar disorder.

The risk allele (oriented to the dbSNP entry) is (C); the odds ratio associated with heterozygotes is 0.97 (CI 0.81-1.15), and for homozygotes, 1.31 (CI 1.09-1.57).

Findings from bipolar disorder genome-wide association studies replicate in a Finnish bipolar family-cohort.

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Established associations:

bipolar disorder **you have 0 copies of the risk allele C**
(risk allele=C, odds ratio/beta 1.03 [1.15-1.23] with pval 7E-6, pubmedid=17554300)

Gene related topics:

bipolar disorder(1.00)

Relevant papers and external links:

PMID 17554300
PHARMGKB PA162356652
Evidence: PubMed ID:17554300; Web Resource:External Link Results: Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls (Initial Sample Size: 1,868 cases, 2,938 controls; Replication Sample Size: NR; Risk Allele: rs3761218-C). This variant is associated with bipolar disorder.

PMID 19308021

dbSNP: rs3761218 GWAS Catalog: rs3761218 Genecard: CDC25B chromosome 20

SNPedia: rs3761218(T;T)

★ rs131702(G;T) info:0.00 44.47%

rs131702, a SNP in the BCR gene on chromosome 22, has been associated with increased risk for bipolar disorder. The odds ratio for carriers of the minor allele (G) are reported as 1.49 (CI:1.15 - 1.93, p=0.0026) based on a study of 171 Japanese patients.

Gene related topics:

bipolar disorder(1.00) · CANCER(0.02) · leukemia(0.02)

Relevant papers and external links:

PMID 15866548

dbSNP: rs131702 Genecard: BCR chromosome 22 SNPedia: rs131702(G;T)

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★ rs10937823(C;C) good:0.00 73.14% **normal**

rs10937823 is a SNP associated with the gene SORCS2, variants of which may play a role in bipolar disorder. The allele most frequently occurring is C, and the variant is T. In the HapMap-CEU population, CC, CT, and TT genotype frequencies were 0.894, 0.097, and 0.009, respectively. However, in the HapMap-CHB population, CT was the most frequently expressed genotype; CC, CT, and TT genotype frequencies were 0.366, 0.439, and 0.195, respectively. In some sample populations, the TT genotype did not exist (TSI, MKK, LWK, ASW). [dbSNP]

The SORCS2 (sortillin-related VPS10 domain containing receptor 2) gene on chromosome 4p16.1 is found to be highly expressed in the mature human brain and kidney. High levels of expression are also seen in the brain during mouse development. [GeneCards] This gene family is responsible for trafficking lysosomal enzymes that are modified within the Golgi to their packaging vacuole. Sortillins have been found to facilitate neurotrophin binding to its receptor, which is necessary for mediating neuronal apoptosis, growth and survival. Given these roles, it has been proposed that alterations to the SORCS2 gene can lead to neuronal dysfunction.

In 2008, a genome-wide association study indicated the SORCS2 gene in bipolar disorder. The study conducted by Baum et al. analyzed 461 affected individuals and 563 matched controls (from the National Institutes of Mental Health Genetics Initiative, all of European ancestry), and a replication sample of 772 affected individuals and 876 matched controls (affected individuals recruited from German hospital admissions, and controls recruited from a German census). The C allele of rs10937823 was found at a frequency of 0.949 in cases and 0.912 in controls in NIMH cohort (p=0.001); 0.954 in cases and 0.931 in controls in the German cohort (p=0.004). The resulting combined odds ratio was 1.67 between the German and NIMH cohorts. Table 3 Furthermore, with multi-locus analysis, the authors found that the highest risk of bipolar disorder was associated with presence of both rs10937823 and rs1170191, a SNP in diacylglycerol kinase eta (DGKH Rs1170191), suggesting that these SNPs have a combinatorial effect in conferring risk (p=1.2x10-8).

A Finnish bipolar family study in 2009 replicated the association between rs10937823 and bipolar disorder. The researchers genotyped the 26 SNPs most strongly associated SNPs as found by the Wellcome Trust Case Control Consortium, and the aforementioned study by Baum et al. in 723 individuals from 180 families. The study classified individuals as falling into diagnostic classes of bipolar type I disorder (cases = 214; controls = 509) or a broad mood spectrum disorder encompassing all types of bipolar disorder (cases = 298; controls = 495). In an opposite finding to that of Baum et al., the minor allele (T) frequency in the unaffected individuals was 0.999; in cases, the frequency was 0.112. The lower frequency of the T allele of rs10937823 in cases versus controls was statistically significant (p=0.004187 using broad mood spectrum disorder; p=0.004703 using broad mood spectrum disorder).

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In 2011, In a Japanese cohort of 736 individuals, rs10937823 was again found to be significantly associated with bipolar disorder by Takata et al. (363 cases, 370 controls; p=0.0175). However, the results showed that the risk allele (more commonly associated allele in cases) was T, not C. The minor allele frequency in the bipolar disorder sample was 0.275, versus 0.253 in the control sample. An odds ratio for this SNP was not reported.

In 2011, Cristoforou et al. identified an association between rs10937823 and bipolar disorder in a Scottish population (506 cases, 607 controls; p=0.031; OR not reported). Like the Japanese study by Takata et al., the minor allele (T) was found to be associated with bipolar disease. Similar observations of this “flip-flop” phenomenon where opposite alleles appear to confer risk have been reported in other genes, such as COMT gene and schizophrenia, and GSTO1 gene and age of onset in Alzheimer’s Disease. The authors suggest that such different results could be due to allelic heterogeneity, the effects of multiple loci, or varying linkage disequilibrium amongst the subjects, if the findings across the studies and populations are indeed correct.

Gene related topics:

bipolar disorder(1.00) · survival(1.00) · brain(1.00) · OTHER(1.00) · schizophrenia(1.00)

Relevant papers and external links:

PMID 11165493
PMID 8187177
PMID 14985763
PMID 19002190
PMID 20351716
PMID 17486107
PMID 17486107
PMID 19308021
PMID 21507135
PMID 20351716
PMID 17273975

dbSNP: rs10937823 Genecard: SORCS2 chromosome 4 SNPedia: rs10937823(C;C)

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

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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)

★ rs2630351(G;G) good:0.00 82.80% **common in complete genomics**

Gene related topics:

NEUROLOGICAL(3.79) · tardive dyskinesia(3.79) · essential tremor(2.88) · PSYCH(2.37) · schizophrenia(2.37) · CHEMDEPENDENCY(2.00) · UNKNOWN(2.00) · akathisia drug induced(2.00) · cocaine abuse(2.00) · 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:

dbSNP: rs2630351 Genecard: DRD3 chromosome 3 SNPedia: rs2630351(G;G)

★ rs2609653(T;T) good:0.00 91.03% **normal**

rs2609653 has been reported in a large study to be associated with bipolar disorder.

The risk allele (oriented to the dbSNP entry) is (C); the odds ratio associated with heterozygotes is 1.43 (CI 1.19-1.71), and for homozygotes, 3.62 (CI 1.26-10.44).

Established associations:

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

(risk allele=T, Modelling analysis odds ratio/beta 1.13 [1.07-1.19] with pval 3E-6, pubmedid=23453885)

bipolar disorder **you have 2 copies of the risk allele T**

(risk allele=T, Modelling analysis odds ratio/beta 1.13 [1.07-1.19] with pval 3E-6, pubmedid=23453885)

unipolar depression **you have 2 copies of the risk allele T**

(risk allele=T, Modelling analysis odds ratio/beta 1.13 [1.07-1.19] with pval 3E-6, pubmedid=23453885)

attention deficit hyperactivity disorder **you have 2 copies of the risk allele T**

(risk allele=T, Modelling analysis odds ratio/beta 1.13 [1.07-1.19] with pval 3E-6, pubmedid=23453885)

autism spectrum disorder **you have 2 copies of the risk allele T**

(risk allele=T, Modelling analysis odds ratio/beta 1.13 [1.07-1.19] with pval 3E-6, pubmedid=23453885)

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Gene related topics:

bipolar disorder(1.00)

Relevant papers and external links:

PMID 23453885
Identification of risk loci with shared effects on five major psychiatric disorders: a genome-wide analysis.

PMID 17554300

dbSNP: rs2609653 GWAS Catalog: rs2609653 Genecard: nan chromosome 8

SNPedia: rs2609653(T;T)

★ rs7570682(G;G) info:0.00 44.88% **normal**

rs7570682 has been reported in a large study to be associated with bipolar disorder.

The risk allele (oriented to the dbSNP entry) is (A); the odds ratio associated with heterozygotes is 1.23 (CI 1.09-1.40), and for homozygotes, 1.64 (CI 1.28-2.12).

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Gene related topics:

bipolar disorder(1.00)

Relevant papers and external links:

PMID 17554300

dbSNP: rs7570682 Genecard: nan chromosome 2 SNPedia: rs7570682(G;G)

★ rs908867(G;G) good:0.00 77.16% **common in complete genomics**

rs908867 is a SNP near the brain-derived neurotrophic factor BDNF gene.

In a study of , the rs908867 was associated with antidepressant response, with heterozygote carriers showing a better response than homozygotes.

However, a critique of this study was published , indicating that this finding should be viewed as preliminary until more research is conducted for several reasons, to wit, "First, the study included not only major depression but also bipolar disorder patients; second, various antidepressants were used in this study, which could affect patients' responses; third, the frequency of the haplotype associated with treatment response is rare; and fourth, previous studies of the effects of single BDNF polymorphisms on antidepressant action have reported conflicting findings."

A brain-derived neurotrophic factor haplotype is associated with therapeutic response in obsessive-compulsive disorder.

Pharmacogenetics of antidepressant response.

Gene related topics:

bipolar disorder(1.00) · brain(1.00) · major depression(1.00) · depression(1.00)

Relevant papers and external links:

PMID 17505499
PMID 18781861
PMID 19589503
PMID 21172166

dbSNP: rs908867 Genecard: nan chromosome 11 SNPedia: rs908867(G;G)

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★ 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)

★ rs2289658(A;A) good:0.00 83.35% **common in complete genomics**

Gene related topics:

PSYCH(0.10) · bipolar disorder(0.10)

Relevant papers and external links:

dbSNP: rs2289658 Genecard: NTRK2 chromosome 9 SNPedia: rs2289658(A;A)

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★ rs1156044(T;T) good:0.00 64.79% **common in complete genomics**

rs1156044(A) was significantly associated with bipolar disorder (p = 0.013) but differed in allele (rs1156044 G allele) from that previously reported as associated with BD.

Gene related topics:

bipolar disorder(1.00)

Relevant papers and external links:

PMID 18199248

dbSNP: rs1156044 Genecard: NDUFV2 chromosome 18 SNPedia: rs1156044(T;T)

★ rs6280(T;T) info:0.00 26.38% **normal**

rs6280, also known as Ser9Gly, is a SNP in the dopamine receptor D3 DRD3 gene. The rs6280(C) allele encodes a glycine, and the (T) allele encodes a serine (in dbSNP orientation).

In a study of 88 patients being treated for schizophrenia with olanzapine, those who were rs6280(C;C) homozygotes had greater positive symptom remission (endpoint rating of minimal or none on all PANSS clinical response positive items, 39.1%), as compared with (C;T) or (T;T) genotypes (13.8%; p = 0.033).

Ser9Gly has been implicated in executive function in some studies, but the results are conflicting.
Gly/Gly carriers showed significantly (p = 0.002) poorer performance than Ser/Ser carriers on executive functioning tasks in a somewhat small Caucasian sample (84 patients with first-episode psychosis and 85 controls).
Gly/Ser heterozygotes had 23% more preservative errors on the WCST compared to Ser/Ser homozygotes in a small (216) healthy Han Chinese sample (p = 0.009). Differences between homozygotes were not statistically significant.
No association between WCST scores and Ser9Gly was found in 138 schizophrenic patients.

Associated in a family association study and pooled sample of 2,037 with nicotine dependence in Americans of European descent.

Preliminary evidence for an association between a dopamine D3 receptor gene variant and obsessive-compulsive personality disorder in patients with major depression. DRD3 polymorphism for an individual with Gly/Gly (C;C) genotype is 2.4 (P = 0.017) times more likely to be diagnosed with OCPD. Male gender was also found to be a significant predictor of OCPD diagnosis (OR = 2.82, P = 0.001). DRD3 may contribute to the development of OCPD. This association was tested using two independent groups of individuals with a history of depression, from a clinical sample (n = 149) and a family study (n = 213).

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Gene related topics:

NEUROLOGICAL(3.79) · tardive dyskinesia(3.79) · essential tremor(2.88) · PSYCH(2.37) · schizophrenia(2.37) · CHEMDEPENDENCY(2.00) · UNKNOWN(2.00) · akathisia drug induced(2.00) · cocaine abuse(2.00) · personality(1.00) · olanzapine(1.00) · nicotine(1.00) · major depression(1.00) · psychosis(1.00) · depression(1.00) · nicotine dependence(1.00) · cocaine related disorders(0.67) · schizophrenia tardive dyskinesia(0.57) · tourette syndrome(0.47) · alcohol dependence(0.22) · parkinson s disease(0.14) · personality disorders(0.12) · bipolar disorder(0.10) · obsessive compulsive disorder(0.06) · adhd attention deficit hyperactivity disorder(0.05) · personality traits(0.05) · alcohol abuse(0.04) · smoking behavior(0.04)

Relevant papers and external links:

PMID 18320559
PMID 18351593
PMID 2186374
PMID 15785860

PMID 18348205

PMID 16583407

PHARMGKB PA165111549
Evidence: PubMed ID:20029384Risk or phenotype-associated allele: Phenotype: This meta-analysis showed no difference in clozipine response between the Ser and Gly allele. Study size: 758 Study population/ethnicity: Patients with Schizophrenia Significance metric(s): OR = 0.82, 95% confidence interval (CI): 0.65-1.04; p = 0.1 Type of association: PD

PHARMGKB PA162316713
Evidence: PubMed ID:18320559A study on 88 olanzapine-treated patients with schizophrenia found that the Gly/gly genotype was significantly associated with greater positive symptom improvement.

PHARMGKB PA164807597
Evidence: PubMed ID:19396436This variant in the DRD3 gene was significantly associated with the therapeutic efficacy of pramipexole in Chinese patients with Parkinson's disease in a study of 30 patients.

PHARMGKB PA165111370
Evidence: PubMed ID:19997080Risk or phenotype-associated allele: C Phenotype: Carriers of the C variant (Gly) of DRD3:Ser9Gly had greater reductions in Autism Treatment Evaluation Checklist (ATEC) scores, indicating improved symptoms and response to risperidone, than TT homozygotes. Study size: 45 Study population/ethnicity: Children with Autism receiving risperidone Significance metric(s): p = 0.012 Type of association: PD

PHARMGKB PA165110867
Evidence: PubMed ID:16809426Risk or phenotype-associated allele: C. Phenotype: A study comparing 276 patients with essential tremor and 184 normal controls confirmed the association of the Gly-9 variant in the DRD-3 gene with risk and age-at-onset of essential tremor. Study size: 276. Study population/ethnicity: French.

OMIM 126451

dbSNP: rs6280 Genecard: DRD3 chromosome 3 SNPedia: rs6280(T;T)

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★ **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).

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)

★ **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)

★ **rs2230912(A;A)** good:0.00 86.62% **common on affy axiom data**

rs2230912, also known as Q460R, is a SNP in the purinergic receptor P2X, ligand-gated ion channel, 7 P2RX7 gene.

This SNP has been reported to be associated with increased risk for bipolar disorder in one study , but the association was not replicated in a large UK case-control sample (bipolar I disorder N = 687, unipolar recurrent major depression N = 1,036, controls N = 1,204).

The G allele is the putative risk allele.

Analysis of single nucleotide polymorphisms in genes in the chromosome 12Q24.31 region points to P2RX7 as a susceptibility gene to bipolar affective disorder.

P2RX7, a gene coding for a purinergic ligand-gated ion channel, is associated with major depressive disorder.

Association between depression and the Gln460Arg polymorphism of P2RX7 gene: a dimensional approach.

P2RX7 Gln460Arg polymorphism is associated with depression among diabetic patients.

Genetics of the P2X7 receptor and human disease.

Two haplotypes of the P2X(7) receptor containing the Ala-348 to Thr polymorphism exhibit a gain-of-function effect and enhanced interleukin-1beta secretion.

P2RX7 polymorphisms Gln460Arg and His155Tyr are not associated with major depressive disorder or remission after SSRI or ECT.

P2RX7 gene is associated consistently with mood disorders and predicts clinical outcome in three clinical cohorts.

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Gene related topics:

bipolar disorder(1.00) · bipolar affective disorder(1.00) · affective disorder(1.00) · mood disorder(1.00) · major depression(1.00) · mood disorders(1.00) · depression(1.00) · major depressive disorder(1.00) · CANCER(0.31) · leukemia lymphoid(0.31) · INFECTION(0.17) · tuberculosis(0.17) · leukemia lymphocytic chronic b cell(0.11) · leukemia(0.08)

Relevant papers and external links:

PMID 18268501
PMID 19160446
PMID 16673375
PMID 16822851
PMID 18543274
PMID 18801407
PMID 19319666
PMID 20360457

PMID 21335057
PMID 21438144

dbSNP: rs2230912 Genecard: P2RX7 chromosome 12 SNPedia: rs2230912(A;A)

★ 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.

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)

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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)

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★ **rs10134944(C;C)** info:0.00 72.46% **normal**

rs10134944 has been reported in a large study to be associated with bipolar disorder.

The risk allele (oriented to the dbSNP entry) is (T); the odds ratio associated with heterozygotes is 1.45 (CI 1.24-1.68), and for homozygotes, 1.32 (CI 0.74-2.33).

Established associations:

bipolar disorder **you have 0 copies of the risk allele T**
(risk allele=T, odds ratio/beta 1.45 [1.24-1.68] with pval 7E-6, pubmedid=17554300)

Gene related topics:

bipolar disorder(1.00) · PSYCH(0.08) · attention deficit disorder with hyperactivity(0.08)

Relevant papers and external links:

PMID 17554300

dbSNP: rs10134944 GWAS Catalog: rs10134944 Genecard: SLC35F4 chromosome 14

SNPedia: rs10134944(C;C)

★ 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)

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★ rs56142442(C;C) good:0.00 99.16% **common in complete genomics**

Gene related topics:

PSYCH(0.10) · bipolar disorder(0.10)

Relevant papers and external links:

dbSNP: rs56142442 Genecard: NTRK2 chromosome 9 SNPedia: rs56142442(C;C)

★ rs334558(G;G) info:0.00 35.84% **common**

Although not associated with increased risk for developing bipolar disorder, amongst those with it, the (G) allele of this SNP has been reported to have a somewhat protective effect in that individuals who have a (G) allele develop bipolar disorder at a later age than those with the more common (A;A) genotypes. [PMID 15351432, PMID 14729229]

Also known as the -50T/C SNP from this GSK3-beta (GSK3B) gene.

may effect lithium treatment response - C allele carriers showed considerably greater response with lithium treatment, with remissions of 56.25% after 4 weeks compared to 31% for T/T carriers

Gene related topics:

PSYCH(0.07) · bipolar disorder(0.07)

Relevant papers and external links:

PMID 17628506
OMIM 605004
PHARMGKB PA162369928
Evidence: PubMed ID:16315267; PubMed ID:18195729This variant is in the promoter region of GSK3B. T allele is associated with greater transcription activity compared with the A allele. This variant is associated with antidepressant treatment response in Chinese major depressive disorder. In a four-locus haplotype analysis (rs334558; rs13321783; rs2319398; rs6808874), the GSK3B TAGT carriers showed a poorer response to antidepressants than carriers with other haplotype groups.

dbSNP: rs334558 Genecard: GSK3B chromosome 3 SNPedia: rs334558(G;G)

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★ rs2289657(G;G) good:0.00 88.72% **common in complete genomics**

Gene related topics:

PSYCH(0.10) · bipolar disorder(0.10)

Relevant papers and external links:

dbSNP: rs2289657 Genecard: NTRK2 chromosome 9 SNPedia: rs2289657(G;G)

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

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)

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★ rs324032(A;A) good:0.02 72.73% **common in complete genomics**

Gene related topics:

NEUROLOGICAL(3.79) · tardive dyskinesia(3.79) · essential tremor(2.88) · PSYCH(2.37) · schizophrenia(2.37) · CHEMDEPENDENCY(2.00) · UNKNOWN(2.00) · akathisia drug induced(2.00) · cocaine abuse(2.00) · 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:

dbSNP: rs324032 Genecard: DRD3 chromosome 3 SNPedia: rs324032(A;A)

★ 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)

★ rs1018381(C;C) good:0.06 68.70% **normal cognitive ability**

g2b2mh blog In a population of healthy individuals, those that carry common variants (such as rs760761, rs1018381, rs2619522) located in the dysbindin (DTNBP1) gene, a risk factor for schizophrenia, show

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minor cognitive impairments such as decreased attentional capacity, worse performance on memory tasks, and alterations in schizotypal beliefs and experiences.

Gene related topics:

PSYCH(1.29) · schizophrenia(1.29) · memory(1.00) · CARDIOVASCULAR(0.29) · blood pressure determination(0.29) · NEUROLOGICAL(0.11) · cognitive ability(0.11) · bipolar disorder(0.07)

Relevant papers and external links:

OMIM 607145

dbSNP: rs1018381 Genecard: DTNBP1 chromosome 6 SNPedia: rs1018381(C;C)

★ **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)

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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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★ rs2630349(G;G) good:0.13 69.07% **common in complete genomics**

Gene related topics:

NEUROLOGICAL(3.79) · tardive dyskinesia(3.79) · essential tremor(2.88) · PSYCH(2.37) · schizophrenia(2.37) · CHEMDEPENDENCY(2.00) · UNKNOWN(2.00) · akathisia drug induced(2.00) · cocaine abuse(2.00) · 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:

dbSNP: rs2630349 Genecard: DRD3 chromosome 3 SNPedia: rs2630349(G;G)

★ 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)

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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)

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)

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★ **rs324650(T;T)** **info:0.08** **34.96%**

In 2009, a large, multi-sample replication study failed to find any significant association with Intelligence for this or other SNPs in CHRM2 .

Earlier work had observed that this snp (and perhaps its neighbors) influenced intelligence and alcohol dependence

CHRM2 gene predisposes to alcohol dependence, drug dependence and affective disorders: results from an extended case-control structured association study. And the earlier

and or this pdf poster show association between the CHRM2 gene and intelligence . In this study of ~300 Caucasians, the (T;T) genotype appeared to typically have a 4 point higher IQ than the (A;T) genotype, which had a 4 point higher IQ (on average) than the (A;A) genotype. A nearby SNP, rs324640, showed similar results to rs324650, and was in linkage disequilibrium (r2 = 0.90) with it. .

No evidence for association between 19 cholinergic genes and bipolar disorder.

Exploring the functional role of the CHRM2 gene in human cognition: results from a dense genotyping and brain expression study.

Uncovering genes for cognitive (dys)function and predisposition for alcoholism spectrum disorders: a review of human brain oscillations as effective endophenotypes.

Depression Case Control (DeCC) Study fails to support involvement of the muscarinic acetylcholine receptor M2 (CHRM2) gene in recurrent major depressive disorder.

Gene related topics:

PSYCH(2.00) · intelligence(2.00) · bipolar disorder(1.00) · affective disorder(1.00) · brain(1.00) · alcoholism(1.00) · OTHER(1.00) · alcohol(1.00) · alcohol dependence(1.00) · depression(1.00) · major depressive disorder(1.00)

Relevant papers and external links:

PMID 19418213
PMID 16000316
PMID 15229186
PMID 17160701
PMID 17081262
PMID 17081262
OMIM 103780
OMIM 608516
OMIM 118493
PMID 17373692
PMID 17996044
PMID 18634760
PMID 19181679

dbSNP: rs324650 Genecard: CHRM2 chromosome 7 SNPedia: rs324650(T;T)

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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)

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★ **rs165599(A;G)** **info:0.25** **49.98%**

anxiety-related personality traits, ADHD, schizophrenia

part of a three marker haplotype rs737865-rs4680-rs165599

epistasis between SNPs in COMT (rs2097603, Val158Met (rs4680), rs165599) and polymorphisms in other schizophrenia susceptibility genes

popular in pubmed

is associated with bipolar disorder and influences prefrontal aspects of verbal memory in bipolar patients and healthy controls.

Gene related topics:

UNKNOWN(6.00) · digeorge syndrome pharyngeal pouch syndrome(6.00) · PSYCH(5.72) · schizophrenia(5.72) · CANCER(3.00) · neoplasms pain(3.00) · fibromyalgia(2.67) · NEUROLOGICAL(2.00) · mamographic density(2.00) · pain temporomandibular joint disorders(2.00) · psychosis(1.67) · obsessive compulsive disorder(1.31) · CHEMDEPENDENCY(1.07) · substance related disorders(1.07) · panic disorder(1.03) · adhd attention deficit hyperactivity disorder(1.00) · eating disorders(1.00) · anxiety(1.00) · memory(1.00) · pain(0.94) · breast cancer(0.83) · marijuana abuse(0.67) · mood disorders(0.62) · parkinson s disease(0.62) · DEVELOPMENTAL(0.60) · leiomyoma uterine neoplasms(0.60) · mental retardation(0.60) · schizophrenia bipolar disorder(0.60) · OTHER(0.57) · sleep deprivation(0.57) · bipolar disorder(0.50) · depression(0.50) · fractures bone(0.50) · personality(0.38) · mammographic density(0.36) · personality traits(0.33) · heroin abuse(0.30) · alcoholism(0.29) · adhd(0.26) · cognitive ability(0.25) · heroin dependence(0.25) · bulimia(0.24) · endometrial cancer(0.23) · smoking(0.21) · anorexia nervosa(0.15) · suicide(0.14) · liver cancer(0.13) · tobacco use disorder(0.11) · narcolepsy(0.11) · METABOLIC(0.10) · weight gain(0.10) · smoking behavior(0.09) · endometrial neoplasms(0.07) · attention deficit hyperactivity disorder(0.05) · IMMUNE(0.05) · vitiligo(0.05) · autism(0.03) · REPRODUCTION(0.02) · endometriosis(0.02)

Relevant papers and external links:

PMID 17547583

OMIM 104300

OMIM 116790

PHARMGKB PA164888988
Evidence: PubMed ID:19451915In a study of 78 African American and 65 white patients diagnosed with schizophrenia or schizoaffective disorder, this SNP in the COMT gene was found to have significant associations with response to risperidone over 2-12 weeks in both African-American and white patients.

PHARMGKB PA162168065
Evidence: PubMed ID:16876132This variant may predict a favorable outcome for bupropion treatment for smoking cessation

dbSNP: rs165599 Genecard: COMT chromosome 22 SNPedia: rs165599(A;G)

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

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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)

Relevant papers and external links:

PMID 19032574

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

★ 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)

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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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★ **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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★ **rs10982256(T;T)** info:0.05 15.02% **normal**

rs10982256 has been reported in a large study to be associated with bipolar disorder.

The risk allele (oriented to the dbSNP entry) is (T); the odds ratio associated with heterozygotes is 1.26 (CI 1.08-1.47), and for homozygotes, 1.47 (CI 1.24-1.74).

Gene related topics:

bipolar disorder(1.00)

Relevant papers and external links:

PMID 17554300

dbSNP: rs10982256 Genecard: DFNB31 chromosome 9 SNPedia: rs10982256(T;T)

★ **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)

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Relevant papers and external links:

PMID 18006541

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

★ 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

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)

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★ 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)

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

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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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★ rs167771(A;A) info:1.19 34.65% **Possibly more "Insistence on Sameness" on ADI-R test. Possibly increased FTND nicotine dependence.**

Possibly more "Insistence on Sameness" on ADI-R test.
Possibly increased FTND nicotine dependence.
rs167771 was significantly associated with autism spectrum disorder in a study of 144 patients.

G allele associated with increased extra-pyramidal symptom risk as a result of risperidone treatment in a study of 321 psychiatric inpatients (81 presenting with EPS and 189 without)

Gene related topics:

nicotine(5.00) · nicotine dependence(5.00) · NEUROLOGICAL(3.79) · tardive dyskinesia(3.79) · essential tremor(2.88) · PSYCH(2.37) · schizophrenia(2.37) · CHEMDEPENDENCY(2.00) · UNKNOWN(2.00) · akathisia drug induced(2.00) · cocaine abuse(2.00) · autism(1.00) · cocaine related disorders(0.67) · schizophrenia tardive dyskinesia(0.57) · tourette syndrome(0.47) · alcohol dependence(0.22) · parkinson s disease(0.14) · personality disorders(0.12) · bipolar disorder(0.10) · obsessive compulsive disorder(0.06) · adhd attention deficit hyperactivity disorder(0.05) · personality traits(0.05) · alcohol abuse(0.04) · smoking behavior(0.04)

Relevant papers and external links:

PMID 19058789
PMID 19506579

PHARMGKB PA165109604
Evidence: PubMed ID:19506579Risk or phenotype-associated allele: G. Phenotype: The G variant of rs167771 was associated with increased risk for extrapyramidal symptoms in psychiatric patients receiving risperidone. Study size: 132. Study population/ethnicity: Patients with Schizophrenia or 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)

★ 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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★ **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)

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)

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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)

★ 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)

★ rs4276227(C;C) info:0.10 44.75% **1.5x risk**

rs4276227 has been reported in a large study to be associated with bipolar disorder.

The risk allele (oriented to the dbSNP entry) is (C); the odds ratio associated with heterozygotes is 1.2 (CI 0.99-1.46), and for homozygotes, 1.49 (CI 1.23-1.81).

Gene related topics:

bipolar disorder(1.00) · METABOLIC(0.06) · body composition(0.06)

Relevant papers and external links:

PMID 17554300

dbSNP: rs4276227 Genecard: CMTM8 chromosome 3 SNPedia: rs4276227(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)

★ rs821616(A;T) info:0.18 37.52% ?

Women with two rs821616(T) SNPs seem to experience more rapid cognitive decline as they age compared to men.

13 single-nucleotide polymorphisms (SNPs) in 723 members of 179 Finnish Bipolar disorder families.
rs821616 verbal fluency

Gene related topics:

bipolar disorder(1.00) · PSYCH(0.60) · schizophrenia bipolar disorder(0.60) · schizophrenia(0.35) · cognitive function(0.33) · NEUROLOGICAL(0.02) · amyotrophic lateral sclerosis(0.02)

Relevant papers and external links:

PMID 16054297
PMID 17673452

dbSNP: rs821616 Genecard: DISC1 chromosome 1 SNPedia: rs821616(A;T)

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★ 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)

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)

★ 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.

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Gene related topics:

PSYCH(9.95) · attention deficit hyperactivity disorder(9.95) · adhd attention deficit hyperactivity disorder(3.41) · schizophrenia methamphetamine abuse(2.00) · CHEMDEPENDENCY(1.25) · smoking behavior(1.25) · perceptual disorders(1.00) · bipolar disorder(1.00) · 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)

★ rs4680(G;G) good:1.99 39.79% **(warrior) multiple associations, see details**

You have the VAL/VAL version of the snp discussed in this news article. It is able to perform better in a test where the optimal strategy changes.

Placebo is less effective for you

External Link
rs4680 (Val158Met) is a well studied SNP in the COMT gene. 23andMe blog summarizes them as
rs4680(A) = Worrier. Met, more exploratory, lower COMT enzymatic activity, therefore higher dopamine levels; lower pain threshold, enhanced vulnerability to stress, yet also more efficient at processing information under most conditions
rs4680(G) = Warrior. Val, less exploratory, higher COMT enzymatic activity, therefore lower dopamine levels; higher pain threshold, better stress resiliency, albeit with a modest reduction in executive cognition performance under most conditions

Roughly speaking, the predominant wisdom (known colloquially as the warrior/worrier hypothesis; summary at) posits that people with Val alleles have increased COMT activity and lower prefrontal extracellular dopamine compared with those with the Met substitution. Val158 alleles may be associated with an advantage in the processing of aversive stimuli (warrior strategy), while Met158 alleles may be associated with an advantage in memory and attention tasks (worrier strategy). Under conditions of increased dopamine release (eg, stress), individuals with Val158 alleles may have improved dopaminergic transmission and better performance, while individuals with Met158 alleles may have less efficient neurotransmission and worse performance. Some evidence suggests that Val158 alleles are associated with schizophrenia, while Met158 alleles are associated with anxiety.

Specific studies include:

rs4680, a functional Val/Met polymorphism, showed modest association with Irish familial schizophrenia. Haplotype A-G-A for SNPs rs737865-rs4680-rs165599 was preferentially transmitted to the affected subjects.

A study of 400 individuals reported that an increase in plasma total homocysteine (tHcy) of 10.4% (CI: 0.01-0.21, p=0.03) for associated with rs4680(A;A) homozygotes compared with rs4680(G;G) subjects. The (A;A) genotype was also more common, but statistically not that significantly, in venous thrombosis patients (OR 1.61, CI: 0.97-2.65], p=0.06) compared to control subjects.

And from :
"Adolescent cannabis use was associated with increased risk of schizophreniform disorder in adulthood among Val/Val (i.e. rs4680(G;G)) individuals (OR = 10.9, 95% CI: 2.2â€“54.1) and, to a lesser extent, among Met/Val individuals (rs4680(A;G)); OR = 2.5, 95% CI: .78 â€“ 8.2), but not among Met/Met (rs4680 (A;A)) individuals (OR = 1.1, 95% CI: .21â€“5.4)."

Also potentially associated with schizophrenia

As part of a haplotype with rs4633, the (G) allele is also associated with endometrial cancer

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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."

In response to pain Met/Met allele carriers showed greater "sensory and affective ratings of pain and a more negative internal affective state. Opposite effects were observed in val158 homozygotes. The COMT val158met polymorphism thus influences the human experience of pain and may underlie interindividual differences in the adaptation and responses to pain and other stressful stimuli."

Reports that ease of entering hypnosis correlates with number of rs4680(G) alleles.

see gs226 for a report related to impulsiveness

External Link and blog discussion of data about rs4680 thinking about nothing in an fmri.

Breast Cancer Risk Modifiers

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Established associations:

blood metabolite measurement **you have 0 copies of the risk allele A**

(risk allele=A, X-01911 odds ratio/beta 0.044 [0.032-0.056] unit decrease with pval 1E-13, pubmedid=24816252; risk allele=A, X-11593--O-methylascorbate odds ratio/beta 0.049 [0.045-0.053] unit decrease with pval 5E-178, pubmedid=24816252)

Gene related topics:

UNKNOWN(6.00) · digeorge syndrome pharyngeal pouch syndrome(6.00) · PSYCH(5.72) · schizophrenia(5.72) · CANCER(3.00) · neoplasms pain(3.00) · fibromyalgia(2.67) · NEUROLOGICAL(2.00) · mamographic density(2.00) · pain temporomandibular joint disorders(2.00) · psychosis(1.67) · obsessive compulsive disorder(1.31) · CHEMDEPENDENCY(1.07) · substance related disorders(1.07) · panic disorder(1.03) · adhd attention deficit hyperactivity disorder(1.00) · eating disorders(1.00) · sleep(1.00) · cognitive function(1.00) · anxiety(1.00) · homocysteine(1.00) · memory(1.00) · stress(1.00) · cancer(1.00) · thrombosis(1.00) · venous thrombosis(1.00) · cocaine dependence(1.00) · pain(0.94) · breast cancer(0.83) · marijuana abuse(0.67) · mood disorders(0.62) · parkinson s disease(0.62) · DEVELOPMENTAL(0.60) · leiomyoma uterine neoplasms(0.60) · mental retardation(0.60) · schizophrenia bipolar disorder(0.60) · OTHER(0.57) · sleep deprivation(0.57) · bipolar disorder(0.50) · depression(0.50) · fractures bone(0.50) · personality(0.38) · mammographic density(0.36) · personality traits(0.33) · heroin abuse(0.30) · alcoholism(0.29) · adhd(0.26) · cognitive ability(0.25) · heroin dependence(0.25) · bulimia(0.24) · endometrial cancer(0.23) · smoking(0.21) · anorexia nervosa(0.15) · suicide(0.14) · liver cancer(0.13) · tobacco use disorder(0.11) · narcolepsy(0.11) · METABOLIC(0.10) · weight gain(0.10) · smoking behavior(0.09) · endometrial neoplasms(0.07) · attention deficit hyperactivity disorder(0.05) · IMMUNE(0.05) · vitiligo(0.05) · autism(0.03) · REPRODUCTION(0.02) · endometriosis(0.02)

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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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★ 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)

★ 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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★ **rs945032(G;G)** **info:0.32** **67.25%**

located in promoter region of BDKRB2 - associated with panic disorder, bipolar disorder, and substance abuse

Identification of functional SNPs in the 5-prime flanking sequences of human genes.

Gene related topics:

bipolar disorder(1.00) · substance abuse(1.00) · panic disorder(1.00)

Relevant papers and external links:

PMID 19086053
PMID 15717931

dbSNP: rs945032 Genecard: BDKRB2 chromosome 14 SNPedia: rs945032(G;G)

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★ 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)

★ rs6323(G;-) info:2.09 14.07% **Increased monoamine oxidase A activity**

rs6323 (R297R / Arg297Arg) is a SNP in the MAOA (monoamine oxidase A) gene. Monoamine oxidase A degrades serotonin, dopamine, epineprine, and norepinephrine. The G allele encodes for the higher activity form of the enzyme.

Subjects with major depressive disorder with the highest activity form of the enzyme (G or G/G) had a significantly lower magnitude of placebo response.

Gene related topics:

PSYCH(3.00) · antisocial personality disorder(3.00) · bp major depressive(2.00) · personality disorders(1.12) · aggressive behavior(1.00) · mood disorder(0.60) · UNKNOWN(0.56) · sudden infant death(0.56) · CHEMDEPENDENCY(0.51) · smoking behavior(0.51) · personality traits(0.47) · suicide(0.32) · panic disorder(0.31) · depression(0.30) · personality(0.25) · major depressive disorder(0.24) · alcoholism(0.24) · bipolar disorder(0.24) · adhd attention deficit hyperactivity disorder(0.21) · attention deficit hyperactivity disorder(0.21) · smoking(0.21) · alcohol abuse(0.17) · adhd(0.15) · NEUROLOGICAL(0.13) · restless legs syndrome(0.13) · autism(0.12) · parkinson s disease(0.12) · migraine(0.11) · depressive disorder major(0.10) · mood disorders(0.10) · obsessive compulsive disorder(0.06) · dementia(0.04)

Relevant papers and external links:

PMID 19593178

dbSNP: rs6323 Genecard: MAOA chromosome X SNPedia: rs6323(G;G)

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★ rs27072(C;T) good:2.07 32.60% **Lower risk of alcohol withdrawal seizures, and perhaps lower odds of ADHD.**

Alcoholics with this are less likely to have seizures during alcohol withdrawal.

rs27072, a SNP in the dopamine transporter SLC6A3 gene, has been associated with more severe symptoms upon alcohol withdrawal, such as seizures, in a study of 250 Caucasian alcohol-dependent patients. Two haplotypes appear to be tagged by this SNP and a neighbor, rs27048. The rs27072 polymorphism has been shown to be significantly associated with ADHD in clinical samples of Canadian children aged 6 to 16 [Feng et al., 2005] and the significant association was also reported by Ouellet-Morin et al. in a Canadian twin study, 2008 (Am. J. of Medical Genetics Part B, 147B:1442-1449 (2008).

The interaction between the dopamine transporter gene and age at onset in relation to tobacco and alcohol use among 19-year-olds.

Association of promoter variants of human dopamine transporter gene with schizophrenia in Han Chinese.

Dopamine Transporter Gene Variant Affecting Expression in Human Brain is Associated with Bipolar Disorder.

Sequence variation in the 3'-untranslated region of the dopamine transporter gene and attention-deficit hyperactivity disorder (ADHD).

Association of the dopamine transporter gene and ADHD symptoms in a Canadian population-based sample of same-age twins.

Candidate gene studies of ADHD: a meta-analytic review.

Association between polymorphism of the dopamine transporter gene and early smoking onset: an interaction risk on nicotine dependence.

Gene and gene by sex associations with initial sensitivity to nicotine in nonsmokers.

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

Haplotypes of dopamine and serotonin transporter genes are associated with antisocial personality disorder in alcoholics.

Pharacogenetic effects of dopamine transporter gene polymorphisms on response to chlorpromazine and clozapine and on extrapyramidal syndrome in schizophrenia.

The role of dopamine transporter (SLC6A3) and dopamine D2 receptor/ankyrin repeat and kinase domain containing 1 (DRD2/ANKK1) gene polymorphisms in personality traits.

Alcohol Dependence

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Gene related topics:

PSYCH(9.95) · attention deficit hyperactivity disorder(9.95) · seizures(5.00) · adhd attention deficit hyperactivity disorder(3.41) · schizophrenia methamphetamine abuse(2.00) · CHEMDEPENDENCY(1.25) ·

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

Relevant papers and external links:

PMID 18070248

OMIM 143465

OMIM 126455

PMID 19740369

PMID 19879111

PMID 21525861

PMID 16082693

PMID 18165969

PMID 19506906

PMID 14685824

PMID 18690117

PMID 19693267

PMID 20505557

PMID 20580759

PMID 21354244

dbSNP: rs27072 Genecard: SLC6A3 chromosome 5 SNPedia: rs27072(C;T)

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★ **rs1049353(G;G)** info:1.93 75.80% **more resistant to treatment for depression**

Based on a study of 256 Caucasian patients being treated for depression, carriers of a rs1049353(G) allele were less likely to respond favorably, particularly if they were females with comorbid anxiety.

rs806368(C) together with rs1049353(A) or rs1049353(G) make up a haplotype associated with PTSD, with the C-A haplotype somewhat associated (p=0.04) with higher risk of PTSD and the C-G haplotype associated (p=0.01) with a lower risk of PTSD.

Cannabis receptor haplotype associated with fewer cannabis dependence symptoms in adolescents.

No evidence for an involvement of variants in the cannabinoid receptor gene (CNR1) in obesity in German children and adolescents.

The G1422A variant of the cannabinoid receptor gene (CNR1) is associated with abdominal adiposity in obese men.

No association of CNR1 gene variations with susceptibility to schizophrenia.

Polymorphisms of the cannabinoid 1 receptor gene and cognitive impairment in multiple sclerosis.

Association between single nucleotide polymorphisms in the cannabinoid receptor gene (CNR1) and impulsivity in southwest California Indians.

Cannabinoid receptor 1 gene association with nicotine dependence.

Cannabinoid type-1 receptor gene polymorphisms are associated with central obesity in a Southern Brazilian population.

Lack of association of genetic variants in genes of the endocannabinoid system with anorexia nervosa.

Evidence for association between polymorphisms in the cannabinoid receptor 1 (CNR1) gene and cannabis dependence.

Genetic variation in cannabinoid receptor 1 (CNR1) is associated with derangements in lipid homeostasis, independent of body mass index.

Interaction between two independent CNR1 variants increases risk for cocaine dependence in European Americans: a replication study in family-based sample and population-based sample.

CB1 expression is attenuated in Fallopian tube and decidua of women with ectopic pregnancy.

Candidate genes for cannabis use disorders: findings, challenges and directions.

Association of CNR1 and FAAH endocannabinoid gene polymorphisms with anorexia nervosa and bulimia nervosa: evidence for synergistic effects.

The implication of CNR1 gene's polymorphisms in the modulation of endocannabinoid system effects.

Eating disorders: the current status of molecular genetic research.

Investigation of CNR1 and FAAH endocannabinoid gene polymorphisms in bipolar disorder and major depression.

CNR1 gene polymorphisms in addictive disorders: a systematic review and a meta-analysis.

G1359A polymorphism of the cannabinoid receptor gene (CNR1) and insulin resistance in patients with diabetes mellitus type 2.

Role of genetic variation in the cannabinoid type 1 receptor gene (CNR1) in the pathophysiology of human obesity.

Endocannabinoid Pro129Thr FAAH functional polymorphism but not 1359G/A CNR1 polymorphism is associated with antipsychotic-induced weight gain.

Are endocannabinoid type 1 receptor gene (CNR1) polymorphisms associated with obesity and metabolic syndrome in postmenopausal Polish women?

A common variation in the cannabinoid 1 receptor (CNR1) gene is associated with pre-eclampsia in the Central European population.

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Cannabinoid type 1 receptor gene polymorphisms are not associated with olanzapine-induced weight gain.

Cannabinoid Receptor Genotype Moderation of the Effects of Childhood Physical Abuse on Anhedonia and Depression.

Gene related topics:

depression(5.00) · bipolar disorder(1.00) · anxiety(1.00) · eating disorder(1.00) · insulin resistance(1.00) · cognitive impairment(1.00) · diabetes mellitus type 2(1.00) · body mass(1.00) · olanzapine(1.00) · multiple sclerosis(1.00) · anorexia nervosa(1.00) · diabetes mellitus(1.00) · metabolic syndrome(1.00) · nicotine(1.00) · weight(1.00) · eating disorders(1.00) · schizophrenia(1.00) · METABOLIC(1.00) · adiposity(1.00) · major depression(1.00) · body mass index(1.00) · obesity(1.00) · bulimia(1.00) · insulin(1.00) · impulsivity(1.00) · nicotine dependence(1.00) · cocaine dependence(1.00) · weight gain(1.00) · CHEMDEPENDENCY(0.67) · marijuana abuse(0.67)

Relevant papers and external links:

PMID 18579347
PMID 18213623
PMID 16917946
PMID 17292652
PMID 17873324
PMID 17881126
PMID 17942526
PMID 18179391
PMID 18606954
PMID 18776593
PMID 19014633
PMID 19016476
PMID 19018721
PMID 19052543
PMID 19093002
PMID 19335651
PMID 19659925
PMID 19886064
PMID 20033240
PMID 20080186
PMID 20192949
PMID 20204253
PMID 20415562
PMID 20631561
PMID 20838400
PMID 21129839
PMID 21695734

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PMID 22393204

dbSNP: rs1049353 Genecard: CNR1 chromosome 6 SNPedia: rs1049353(G;G)

★ rs1006737(G;G) info:0.56 48.79%

23andMe blog rs1006737 or (rs2159100) Each T at this SNP increased the odds of bipolar disorder by 1.18 times compared to having two CC copies

Established associations:

unipolar depression **you have 0 copies of the risk allele A**

(risk allele=A, odds ratio/beta 1.18 bipolar with pval 3E-8, pubmedid=20351715)

bipolar disorder **you have 0 copies of the risk allele A**

(risk allele=A, odds ratio/beta 1.18 with pval 7E-8, pubmedid=18711365; risk allele=A, odds ratio/beta 1.18 bipolar with pval 3E-8, pubmedid=20351715)

schizophrenia **you have 0 copies of the risk allele A**

(risk allele=A, odds ratio/beta 1.103 [1.08-1.13] with pval 5E-12, pubmedid=23974872)

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Gene related topics:

PSYCH(0.13) · bipolar disorder(0.13) · CARDIOVASCULAR(0.03) · tunica media(0.03)

Relevant papers and external links:

PMID 23974872
Genome-wide association analysis identifies 13 new risk loci for schizophrenia.

PMID 18711365
Collaborative genome-wide association analysis supports a role for ank3 and cacna1c in bipolar disorder.

PMID 20351715
Meta-analysis of genome-wide association data of bipolar disorder and major depressive disorder.

OMIM 125480

OMIM 612372

PHARMGKB PA162355589
Evidence: PubMed ID:18711365A genome-wide association analysis study in 4,387 cases and 6,209 controls identified a strong association of this variant in the CACNA1C gene with bipolar disorder.

dbSNP: rs1006737 GWAS Catalog: rs1006737 Genecard: CACNA1C chromosome 12

SNPedia: rs1006737(G;G)

★ 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.

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

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dbSNP: rs6265 GWAS Catalog: rs6265 Genecard: BDNF chromosome 11

SNPedia: rs6265(G;G)

★ rs9378249(T;T) good:0.90 **common in complete genomics**

Evidence for somatic gene conversion and deletion in bipolar disorder, Crohn's disease, coronary artery disease, hypertension, rheumatoid arthritis, type-1 diabetes, and type-2 diabetes.

Gene related topics:

bipolar disorder(1.00) · arthritis(1.00) · rheumatoid arthritis(1.00) · hypertension(1.00) · coronary artery disease(1.00)

Relevant papers and external links:

PMID 21291537

dbSNP: rs9378249 SNPedia: rs9378249(T;T)

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★ rs6458307(C;C) info:0.52 40.30% **1.4x risk**

rs6458307 has been reported in a large study to be associated with bipolar disorder.

The risk allele (oriented to the dbSNP entry) is (T); the odds ratio associated with heterozygotes is 0.84 (CI 0.75-0.96), and for homozygotes, 1.39 (CI 1.13-1.69).

Gene related topics:

bipolar disorder(1.00)

Relevant papers and external links:

PMID 17554300
PHARMGKB PA162356506

Evidence: PubMed ID:17554300; Web Resource:External Link results: Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls (Initial Sample Size: 1,868 cases, 2,938 controls; Replication Sample Size: NR). This variant is associated with Bipolar disorder.

dbSNP: rs6458307 Genecard: nan chromosome 6 SNPedia: rs6458307(C;C)

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

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

From this article's abstract:

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

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

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

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

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

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

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

Genetic differences in human circadian clock genes among worldwide populations.

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

Circadian polymorphisms associated with affective disorders.

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

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

Genotyping sleep disorders patients.

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

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

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

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

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

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

Attention-Deficit Hyperactivity Disorder

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Gene related topics:

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

Relevant papers and external links:

PMID 17221848
PMID 20364331
PMID 17948273
PMID 20065968
PMID 17428266
PMID 18071340
PMID 18228528
PMID 18663240
PMID 18957941
PMID 19166596
PMID 20180986
PMID 20368993

PMID 20396431
PMID 20600471
PMID 22310473
PMID 22709985
PMID 20961464
PMID 23399917
PMID 24467926

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

★ rs131690(A;G) info:0.94 49.42% **1.5x increased risk for bipolar disorder**

rs131690, a SNP in the BCR gene on chromosome 22, has been associated with increased risk for bipolar disorder. The odds ratio for carriers of the minor allele (G) are reported as 1.50 (CI:1.14 - 2.03, p=0.0063) based on a study of 171 Japanese patients.

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Gene related topics:

bipolar disorder(5.00) · CANCER(0.02) · leukemia(0.02)

Relevant papers and external links:

PMID 15866548

dbSNP: rs131690 Genecard: BCR chromosome 22 SNPedia: rs131690(A;G)

★ rs2953145(C;G) info:1.04 29.28% **1.8x risk**

rs2953145 has been reported in a large study to be associated with bipolar disorder.

The risk allele (oriented to the dbSNP entry) is (G); the odds ratio associated with heterozygotes is 1.84 (CI 1.31-2.58), and for homozygotes, 2.14 (CI 1.53-2.98).

Established associations:

bipolar disorder **you have 1 copies of the risk allele G**

(risk allele=G, odds ratio/beta 1.84 [1.31-2.58] with pval 7E-6, pubmedid=17554300)

Gene related topics:

bipolar disorder(1.00)

Relevant papers and external links:

PMID 17554300

PHARMGKB PA162356651

Evidence: PubMed ID:17554300; Web Resource:External Link Results: Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls (Initial Sample Size: 1,868 cases, 2,938 controls; Replication Sample Size: NR; Risk Allele: rs2953145-C). This variant is associated with bipolar disorder.

dbSNP: rs2953145 GWAS Catalog: rs2953145 Genecard: RNPEPL1 chromosome 2

SNPedia: rs2953145(C;G)

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★ rs5750285(C;C) good:1.08 21.59% **Better response to lithium treatment**

[PMID 18408563] Stargazin involvement with bipolar disorder and response to lithium treatment.

Gene related topics:

bipolar disorder(1.00)

Relevant papers and external links:

PHARMGKB PA161659506

Evidence: PubMed ID:18408563The C allele of this variant is associated with increased response of patients with bipolar disorder to lithium treatment relative to that observed in patients carrying two copies of the G allele. This association was seen in one cohort of 195 patients, but not in a separate cohort of 134 patients.

dbSNP: rs5750285 Genecard: nan chromosome 22 SNPedia: rs5750285(C;C)

★ rs4713902(C;C) info:1.41 3.88%(rare) **normal**

rs4713902, a SNP in the FKBP5 gene, is associated with increased risk for bipolar disorder in a study of 500+ Caucasian patients.

The most common allele, rs4713902(T), was associated with highest risk. Giving that allele a relative odds ratio of 1.0, the odds for the (C;T) and (C;C) genotypes were 0.69x (CI:0.55-0.88, p=0.001) and 0.47 (CI: 0.30-0.74, p=0.001).

Gene related topics:

bipolar disorder(1.00) · PSYCH(0.07) · depression(0.07)

Relevant papers and external links:

PMID 18180755

dbSNP: rs4713902 Genecard: FKBP5 chromosome 6 SNPedia: rs4713902(C;C)

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★ rs2251219(A;G) good:1.61 43.74% **Lower bipolar disorder risk?**

The C-allele of rs2251219 was significantly under-represented in bipolar disorder cases compared to controls (p=0.002, OR = 0.57; Table 2), replicating our main meta-analysis result.
Replication Study and Meta-Analysis in European Samples Supports Association of the 3p21.1 Locus with Bipolar Disorder.

Gene related topics:

bipolar disorder(5.00) · METABOLIC(0.12) · adiponectin(0.12)

Relevant papers and external links:

PMID 20081856
PMID 22560537

dbSNP: rs2251219 Genecard: PBRM1 chromosome 3 SNPedia: rs2251219(A;G)

★ rs1064395(G;G) good:2.61 56.67% **Normal risk of bipolar disorder or schizophrenia**

rs1064395 is a single nucleotide variant (SNV) found in the neurocan gene (NCAN) that has been implicated as a predictor of both bipolar disorder (BD) and schizophrenia. BD is characterized by a fluctuation between manic episodes and severe depression. Schizophrenia is characterized by hallucinations, both visual and auditory, paranoia, disorganized thinking and lack of normal social skills.

NCAN is a chondroitin sulfate proteoglycan that is involved in cell adhesion and migration. It contains 14 exons and spans 41 kb of the genome at the location 19p12. [OMIM] It has also been found that, in mice, this gene is expressed in a localized way in the cortical and hippocampal regions of the brain which are responsible for cognition and regulation of emotions.

In 2011, a study by Cichon et al identified rs1064395 in a genome-wide association study (GWAS) for Bipolar Disorder (BD). This study was done by analyzing around 500,000 autosomal SNPs and 12,000 X-chromosomal SNPS in 682 patients with BD and 1300 controls. The patients studied were from Europe, the USA, and Australia. The study then went forward to test the top 48 hits from their initial GWAS with an independent cohort of 1729 cases and 2313 controls, which narrowed down their significant hits to a list of eight. These eight were then used in a meta analysis. The rs1064395 was highly significant with a p-value of 3.02X10-8 and an odds ratio of 1.31, with A being the risk allele. The authors then further validated with a much larger sample size (6030 cases and 31,749 controls) and found a p-value of 2.74X10-4 and an odds ratio of 1.12 which, combined with their previous study lead to an overall p-value of 2.14X10-9 and an odds ratio of 1.17.

In 2012, a study by Mühleisen et al investigated this same SNP in case control studies involving schizophrenic patients. They designed a patient-control study using samples from patients with European ancestry. The initial study included a cohort of 5061 patients and 9655 controls. The risk A-allele was found more frequently in schizophrenic patients then in controls with a p-value of 2.28X10 -8 and an odds ratio of 1.11. They followed up this initial experiment using a independent cohort from the Schizophrenia Psychiatric GWAS Consortium that included 5537 patients and 8043 controls. This follow up inquiry yielded a p-value of 0.0239 and an odds ratio of 1.07. Thus, these authors concluded that, not only is rs1064395 predictive of BD, it is also predictive of schizophrenia.

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Established associations:

bipolar disorder **you have 0 copies of the risk allele A**

(risk allele=A, odds ratio/beta 1.17 with pval 2E-9, pubmedid=21353194)

Gene related topics:

bipolar disorder(5.00) · schizophrenia(5.00) · brain(1.00) · hallucinations(1.00) · depression(1.00) · METABOLIC(0.13) · ldl cholesterol(0.13)

Relevant papers and external links:

PMID 21353194

PMID 21353194

PMID 22497794

dbSNP: rs1064395 GWAS Catalog: rs1064395 Genecard: NCAN chromosome 19

SNPedia: rs1064395(G;G)

★ rs1375144(T;T) good:1.94 21.93% **Normal risk of developing bipolar disorder**

rs1375144, a SNP in the DPP10 gene, has been reported in a large study to be associated with bipolar disorder.

The risk allele (oriented to the dbSNP entry) is (C) ; the odds ratio associated with heterozygotes is 1.32 (CI 1.07-1.63), and for homozygotes, 1.59 (CI 1.29-1.96). [Note: the 2009 citation below did not see a statistically significant difference between cases and controls for this allele in their study.]

Findings from bipolar disorder genome-wide association studies replicate in a Finnish bipolar family-cohort.

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Gene related topics:

bipolar disorder(5.00) · CARDIOVASCULAR(0.03) · c reactive protein(0.03) · HEMATOLOGICAL(0.02) · hemoglobin a glycosylated(0.02)

Relevant papers and external links:

PMID 17554300

PMID 19308021

dbSNP: rs1375144 Genecard: DPP10 chromosome 2 SNPedia: rs1375144(T;T)

★ rs7757037(A;A) good:2.01 22.34% **0.63x decreased risk for bipolar disorder**

rs7757037, a SNP in the FKBP5 gene, is associated with increased risk for bipolar disorder in a study of 500+ Caucasian patients.

The most common allele, rs7757037(G), was associated with highest risk. Giving that allele a relative odds ratio of 1.0, the odds for the (A;G) and (A;A) genotypes were 0.68x (CI:0.53-0.87, p=0.007) and 0.63x (CI: 0.45-0.89, p=0.007).

Gene related topics:

bipolar disorder(5.00) · PSYCH(0.07) · depression(0.07)

Relevant papers and external links:

PMID 18180755

dbSNP: rs7757037 Genecard: FKBP5 chromosome 6 SNPedia: rs7757037(A;A)

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