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# Promethease Rep

## Version: 0.1.164

## Generated: 2015-11-13 05:52

### Infile: AncestryDNA raw data AP.txt
### infiles /tmp/tmpc6nCfK

### Reference Population TSI

### 12952 genotypes annotated

## Good news

...more... (hide)

| | |
|---|---|
| rs3732379(C;T)<br>Magnitude: 2.8<br>Frequency: 38.1%<br>Repute:Good<br>References:10 | reduced risk of acute coronary events heterozygosity was associated with a markedly reduced risk of acute coronary events<br>...more... (hide)<br>see also rs3732378 [OMIM:HUMAN IMMUNODEFICIENCY VIRUS TYPE 1, RAPID PROGRESSION TO AIDS] |
| rs4680(G;G)<br>Magnitude: 2.5<br>Frequency: 29.2%<br>Repute:Good<br>References:279 | (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 http://www.nytimes.com/2013/02/10/magazine/why-can-some-kids-handle-pressure-while-others-fall-apart.html?ref=magazine&pagewanted=all<br>...more... (hide)<br>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 |

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| | 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 compar... |
| rs1815739(C;C)<br>Magnitude: 2.2<br>Frequency: 19.5%<br>Repute:Good<br>References:5 | Better performing muscles. Likely sprinter.<br>This genotype indicates better performing muscles, particularly for sprinting and power sports. Fast-twitch muscle fibers are able to produce alpha-actinin-3. Professional sprinters usually have this, although it is less common for endurance athletes.<br>...more... (hide)<br>This SNP, in the ACTN3 gene, encodes a premature stop codon in a muscle protein called alpha-actinin-3. The polymorphism alters position 577 of the alpha-actinin-3 protein. In the (C;C) genotype is often called RR. The truncated (T;T) is often called XX. (T;T) is under-represented in elite strength athletes, consistent with previous reports indicating that alpha-actinin-3 deficiency appears to impair muscle performance. The most common nucleotide at this position, (C), encodes an arginine (amino acid code R), the alternative T allele encodes a stop codon (X). Hence, the SNP is referred to as R577X, with homozygotes being either RR or XX and heterozygotes being RX. XX individuals completely lack the expression of alpha-actinin-3. An earlier report studying a relatively small number of Austr... |
| rs1837253(C;T)<br>Magnitude: 2.2<br>Frequency: 46.0%<br>Repute:Good<br>References:7 | reduced risk (0.84x) for late-onset (adult) asthma Found in 38% of all Caucasions, this genotype is associated with a reduced risk (relative risk = 0.84) for late-onset (adult) asthma.<br>...more... (hide)<br>rs1837253 is a SNP found a bit upstream of the thymic stromal lymphopoietin TSLP gene on chromosome 5. The TSLP protein plays a role in inflammation of cells in the trachea and alveoli therefore may have a causative role in certain types of asthma. The rs1837253(T) allele has been found to reduce the risk of developing late-onset (adult) asthma in several studies, and it is recognized by the CPMC Study as one they report to their participants. [GWAS:None] |
| rs6323(G;G)<br>Magnitude: 2.1<br>Frequency: 16.8%<br>Repute:Good<br>References:13 | Increased monoamine oxidase A activity...more... (hide)<br>rs6323 (R297R / Arg297Arg) is a SNP in the MAOA (monoamine oxidase A) gene. Monoamine oxidase A degrades |

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| | 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. |
| rs9264942(C;T)<br>Magnitude: 2.1<br>Frequency: 48.7%<br>Repute:Good<br>References:21 | 60% reduction in HIV viral load The rs9264942(C;T) genotype is reported to be associated with a 60% reduction in viral load in HIV-infected individuals. See also rs9264942 and HIV.<br>...more... (hide)<br>This SNP (C/T) is in 5' region of the HLA-C gene, 35 kb away from transcription initiation in or around the HLA-C gene. 'People with this tiny sequence variation, dubbed rs9264942, appear to have up to 90% less virus in their systems than those who carry other polymorphisms. About 10% of Europeans appear to carry two copies of rs9264942, which leads to an average 90% viral load reduction. About 50% of Europeans carry one copy, which gives a 60% reduction. By comparison, less than 40% of people of African descent appear to carry a single copy of the polymorphism.' Genetic variation may lower HIV load by 90% They are referring to the (C;C) genotype giving a 90% reduction and the (C;T) giving a 60% reduction. dbSNP page This SNP is also reported to account for 6.5% of the 15% variation in vir... |
| rs9273363(C;C)<br>Magnitude: 2.1<br>Frequency: 49.2%<br>Repute:Good<br>References:3 | Much lower 0.15x risk of Type 1 Diabetes. According to 23andMe, this SNP greatly reduces your risk of Type 1 Diabetes. Type 1 Diabetes is an autoimmune disease where your immune system attacks your own body's insulin producing cells in the pancreas, preventing your blood sugar from being able to enter cells that need energy. This has been verified for white people. 23andMe originally wanted to use rs9272346 instead, but it was deemed unreliable.<br>...more... (hide)<br>This is one of the SNPs that 23andMe uses for Type 1 diabetes. According to 23andMe, this SNP changes your risk of Type 1 Diabetes. That is an autoimmune disease where your immune system attacks your own body's insulin producing cells in the pancreas, preventing your blood sugar from being able to enter cells that need energy. This has been verified for white people. 23andMe originally wanted to use rs9272346 instead, but it was deemed unreliable. [GWAS:Chronic lymphocytic leukemia] |
| rs3129934(C;C) | Normal lower risk of Multiple Sclerosis....more... (hide) |

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| Magnitude: 2.1<br>Frequency: 66.1%<br>Repute:Good<br>References:7 | rs3129934 is a SNP near the HLA-class II region that may be associated with several autoimmune diseases. In a study of two cohorts (Spanish and American multiple sclerosis patients, each numbering several hundred), rs3129934 was the SNP most associated with increased risk for multiple sclerosis. The odds ratio was 3.3 (CI: 2.3 - 4.9, p = 9 x 10e-11). Seven other SNPs were also identified in this study as associated with increased risk for MS. associated with type-1 diabetes. 1.6x risk of type-1 diabetes? 23andMe uses rs9273363 instead of rs3129934 in its reports on type 1 diabetes risk. This SNP was formerly used as a proxy for rs9272346, with this explanation: 'Note: the 2007 Wellcome Trust paper reported a strong association between type 1 diabetes and the SNP rs9272346. Our quality cont... |
| gs101<br>Magnitude: 2<br>Repute: Good | probably able to digest milk<br>77% of Europeans with this are able to digest lactose and dairy products. People without this are more likely to experience lactose intolerance. |
| gs156<br>Magnitude: 2<br>Repute: Good | NAT2 Rapid metabolizer.<br>NAT2 Rapid metabolizer source This is ostensibly the 'normal' type of metabolizer, who carries two rapid (active) acetylator alleles such as NAT2*4. NAT2 rapid acetylators can be typically be administered drugs which are substrates of the NAT2 enzyme following standard dosing practices. This prediction and that of gs154 of NAT2 metabolizer phenotype is based on using 2 SNPs, rs1041983 and rs1801280. An alternative prediction of NAT2 metabolizer status based on using more SNPs (6) is used by genosets gs138, gs139, and gs140. |
| rs6855911(G;G)<br>Magnitude: 2<br>Frequency: 6.2%<br>Repute:Good<br>References:10 | rare, but 0.62x decreased risk for gout...more... (hide)<br>risk for cardiovascular morbidity and mortality rs6855911 (p = 1.84E-16) although no risk for coronary artery disease or myocardial infarction was observed, rs6855911 was associated with gout. The minor allele had a protective effect, with an allelic odds ratio of 0.62 (CI: 0.52-0.75, p = 3.2x10(-7)). |

[PharmGKB:Non-Curated GWAS Results: The GLUT9 Gene Is Associated with Serum Uric Acid Levels in Sardinia and Chianti Cohorts (Initial Sample Size: 4,305 Sardinian individuals; Replication Sample Size: 1,301 Tuscan individuals; Risk Allele: rs6855911-A). This variant is associated with serum urate levels.] [GWAS:Serum urate]

rs800292(T;T)
Magnitude: 2
Frequency: 6.2%
Repute:Good
References:28

5% decreased risk of macular degeneration ...more... (hide)

rs800292 is a SNP in the complement factor H CFH gene; it has been linked to blindness in age related macular degeneration. This SNP is also known as 184G>A, I62V, or Val62Ile. A haplotype of rs1061170 rs3753394 rs800292 rs1329428 (TGTC) was found to confer a significantly increased likelihood of exudative AMD CFH variations appear to contribute to ARMD in Caucasians, but not in Japanese A study of Chinese AMD patients reports that carriers of both rs11200638 and rs800292 risk alleles pushes the odds ratio for AMD up to 23x. Overall, an 'extremely high' population attributable risk (PAR) of 78% reported for these SNPs. rs800292 was associated with polypoidal choroidal vasculopathy (PCV) in a study of 130 Japanese patients. age related macular degeneration Susceptibility genes for age-re...

rs1012053(A;C)
Magnitude: 2
Frequency: 20.4%
Repute:Good
References:7

0.625x reduced risk of Bipolar Disorder...more... (hide)

Bipolar disorder failed to replicate in [PharmGKB:Non-Curated GWAS 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).] [GWAS:Bipolar disorder]

rs11614913(T;T)
Magnitude: 2
Frequency: 22.1%
Repute:Good
References:66

decreased risk for colorectal cancer and lung cancer source ...more... (hide)

miRNA sequences and non-small cell lung cancer rs11614913 SNP in hsa-mir-196a2 was associated with survival in individuals with NSCLC. Specifically, survival was significantly decreased in individuals who were homozygous CC at SNP rs11614913. microRNA 1,009 breast cancer cases and 1,093 controls in a population of Chinese women *rs11614913 in hsa-mir-196a2 T>C 1.23; 95% confidence interval [CI], 1.02-

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| | 1.48 *rs3746444 in hsa-mir-499 A>G OR 1.25; 95% CI, 1.02-1.51 Single nucleotide polymorphisms of microRNA machinery genes modify the risk of renal cell carcinoma. Genetic variations in microRNA-related genes are novel susceptibility loci for esophageal cancer risk. Signatures of purifying and local positive selection in human miRNAs. MicroRNA polymorphisms: the future of pharmacogenomics... |
| rs6983267(T;T)<br>Magnitude: 2<br>Frequency: 23.9%<br>Repute:Good<br>References:149 | normal risk for cancers; aspirin reduces risk for colorectal cancer This genotype is not on its own associated with higher cancer risk. Additionally, aspirin has been reported to somewhat lower the risk for colorectal cancer in individuals of this genotype.<br>...more... (hide)<br>rs6983267 is a SNP on chromosome 8q24, associated with increased risk for several cancers, particularly prostate cancer. In studies dividing the 8q24 region, this SNP falls in region 3. This SNP has also been reported to influence the cancer-risk-decreasing effect of aspirin. In a study of over 3,600 Caucasians with prostate cancer, rs6983267 is one of five SNPs used (with family history as a sixth factor) to cumulatively predict overall risk. On their own, the rs6983267(G;G) and (G;T) risk genotypes yield an odds ratio for developing prostate cancer of 1.37 (CI: 1.18-1.59, p=3.4-10e-5) and may account for 22.2% of population attributable risk. The increased risk of developing prostate cancer associated with rs6983267 now appears to be independent of the risk associated with its close neig... |
| rs7216389(C;C)<br>Magnitude: 2<br>Frequency: 23.9%<br>Repute:Good<br>References:26 | 0.69x lower risk of Childhood Asthma.<br>...more... (hide)<br>rs7216389, a SNP in the ORMDL3 gene on chromosome 17q21, was associated with susceptibility to childhood asthma in a study of ~1,000 British patients. The variation appears to be linked to altered levels of the ORMDL3 mRNA, which was shown in a cohort study of ~5,000 British and German patients to be correlated to childhood asthma. A large study of Scottish asthma patients replicated these results, finding that a single copy of the T allele conferred an odds ratio of 1.50 (CI: 1.24-1.81) and 2 copies conferred an odds ratio of 2.11 (CI: 1.71-2.61), respectively. [PharmGKB:Non-Curated GWAS Results: Genetic variants regulating ORMDL3 expression contribute to the risk of childhood asthma (Initial Sample Size: 994 cases, 1,243 controls; Replication Sample Size: 2,320 cases, 3,301 controls; Ri... |

rs10784502(C;C)
Magnitude: 2
Frequency: 26.5%
Repute:Good
References:0

better intracranial volume?...more... (hide)

associated with intracranial volume (12q14.3; N = 15,782; P = 1.12 × 10−12) http://www.genomesunzipped.org/2012/04/another-iq-gene-new-methods-old-flaws.php (C;C) has higher cranial capacity (about 2 teaspoons) and over 2% higher IQ. (C;T) is half this positive effect. Intelligence

rs13192841(A;G)
Magnitude: 2
Frequency: 31.0%
Repute:Good
References:11

somewhat reduced risk (~0.7x) for lupus
see discussion at rs13192841
...more... (hide)

rs13192841 is a SNP in an intergenic region of chromosome 6. Three independent SNPs in the TNFAIP3 region (rs13192841, rs2230926 and rs6922466) were associated with systemic lupus erythematosus (SLE) among individuals of European ancestry in a study of 1,200+ patients. For rs13192841, the minor allele was associated with reduced risk (OR 0.72 CI: 0.62–0.83, p = 7.9 × 10e−6. The authors point out that rs13192841 is a 'perfect proxy' for rs10499194, which has a similiar reduced risk for rheumatoid arthritis associated with it's minor allele. An association was found between Systemic lupus erythematosus (SLE) and rs13192841. [OMIM:SYSTEMIC LUPUS ERYTHEMATOSUS, SUSCEPTIBILITY TO, 13; SLEB13] [GWAS:Multiple sclerosis]

rs7538876(G;G)
Magnitude: 2
Frequency: 31.2%
Repute:Good
References:7

0.78x decreased Basal Cell Carcinoma
risk. Reduced risk of this kind of skin
cancer.
...more... (hide)

2 SNPs located in different regions of chromosome 1 are likely to be associated with increased risk for basal cell carcinoma (BCC), the most common form of skin cancer. *each A at rs7538876 yields a 1.28x increased risk of developing BCC (p = 4.4 x 10e-12) *each G at rs801114 also yields a 1.28x increased odds of developing BCC (p = 5.9 x 10e-12) *2.68x increased risk of BCC for homozygous carriers of both SNPs This study was based on an analysis of ~2,000 Icelandic and Eastern European skin cancer patients (as well as a large number of controls). The authors estimate that approximately 1.6% of all individuals of European ancestry are homozygous for both risk alleles, and thus that percentage of the population is at 2.68x higher risk for BCC compared to individuals who are rs7538876(G;G) a...

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| rs189798(C;T)<br>Magnitude: 2<br>Frequency: 33.6%<br>Repute:Good<br>References:1 | decreased high myopia risk Less risk of high myopia (in French people). 34.2% of French people without high myopia have T here, but only 21.1% of controls do.(table)<br>...more... (hide) |
| rs9642880(G;G)<br>Magnitude: 2<br>Frequency: 35.4%<br>Repute:Good<br>References:14 | Slightly lower risk of Bladder Cancer....more... (hide)<br>rs9642880 is a SNP located in chromosome region 8q24, a region that has been linked to prostate, breast, and colon cancer. A study ultimately totaling over 4,000 bladder cancer patients of European ancestry (and 40,000 controls) concluded that individuals carrying two copies of rs9642880(T) were at 1.49x increased risk (allele-specific odds ratio = 1.22, p = 9 x 10e-12) for the disease. This SNP was not associated, however, with risk for prostate, breast, or colon cancer. Another study in Chinese populations found similar results in regard to bladder cancer. rs9642880(T;T) estimated risk of developing 1.49 times that of noncarriers. 1,803 urinary bladder cancer cases and 34,336 controls. No association with prostate, colorectal and breast cancers. In contrast to GSTM1 SNPs, the rs9642880(T... |
| rs1061147(C;C)<br>Magnitude: 2<br>Frequency: 38.1%<br>Repute:Good<br>References:6 | Reduced 0.34x risk of Age Related Macular Degeneration. When the retina cells in the eye get damaged with age, it is known as Age Related Macular Degeneration. This SNP reduces the risk to about a third of normal.<br>...more... (hide)<br>may influence age related macular degeneration (damage to the eye with ageing) when part of the haplotype of rs1061170 rs3753394 rs800292 rs1329428 (TGTC), but does not appear to do so alone. age related macular degeneration [GWAS:Age-related macular degeneration] |
| rs1051730(C;C)<br>Magnitude: 2<br>Frequency: 38.9%<br>Repute:Good<br>References:95 | Smokes normal (lower) number of cigarettes if a smoker. Less nicotine dependence. Also smokes each cigarette less deeply. Therefore there is a reduced risk of lung cancer and a reduced (0.8x) risk of Peripheral Artery Disease. This doesn't affect whether people start smoking though.<br>...more... (hide)<br>rs1051730, also known as D398N, is a SNP in the nicotinic acetylcholine receptor alpha 3 subunit CHRNA3 gene. In two recent (2008) studies, together comprising over 6,000 lung |

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| | cancer patients of European ancestry, the rs1051730(T) allele was very significantly associated with increased risk. Having one copy (i.e. being a rs1051730(C;T) genotype) increased risk for lung cancer about 1.3x, and having two copies (rs1051730(T;T) individuals) represented 1.8x increased risk. Up to 14% of lung cancer incidence may be attributable to this allele. An independent study published at the same time concluded that (T) allele carriers for SNP rs1051730 are not at higher risk of becoming smokers compared to (C) carriers. However, if they do smoke, (T) carriers are quite likely to smoke more cigarettes th... |
| rs5882(A;G)<br>Magnitude: 2<br>Frequency: 39.8%<br>Repute:Good<br>References:31 | Lower risk of dementia and Alzheimer's disease. Higher good cholesterol. One copy of a longevity gene. This seems to raise HDL ('good') cholesterol, and reduce the risk of dementia and Alzheimer's disease. It may slightly increase longevity. It has been particularly studied in Ashkenazi Jews.<br>...more... (hide)<br>This SNP rs5882 in the CETP gene referred to as ILE405VAL and RSAI405V has been related to ageing and exceptional long life. A study in Ashkenazi Jews showed that individuals homozygotic for the G allele have significantly longer lifespans. New research also indicates that G homozygotes have an average of 70% less risk of dementia and Alzheimer's disease. The study also found that heterozygotes have a lower risk of dementia and Alzheimer's disease but more study is needed to determine the risk reduction for heterozygotes. See Association of a Functional Polymorphism in the Cholesteryl Ester Transfer Protein (CETP) Gene With Memory Decline and Incidence of Dementia and CETP Variant Linked to Slower Cognitive Decline and Reduced Dementia Risk . [PharmGKB:Curated Risk or phenotype-associated... |
| rs1864163(A;G)<br>Magnitude: 2<br>Frequency: 41.4%<br>Repute:Good<br>References:7 | associated with higher HDL cholesterol...more... (hide)<br>G allele is associated with 4.12mg/dl increase in HDL cholesterol (good cholesterol). [GWAS:HDL cholesterol] |
| rs801114(T;T)<br>Magnitude: 2<br>Frequency: 42.0%<br>Repute:Good<br>References:7 | 0.78x decreased Basal Cell Carcinoma risk. ...more... (hide)<br>2 SNPs located in different regions of chromosome 1 are likely to be associated with increased risk for basal cell carcinoma |

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| | (BCC), the most common form of skin cancer. *each A at rs7538876 yields a 1.28x increased risk of developing BCC (p = 4.4 x 10e-12) *each G at rs801114 also yields a 1.28x increased odds of developing BCC (p = 5.9 x 10e-12) *2.68x increased risk of BCC for homozygous carriers of both SNPs This study was based on an analysis of ~2,000 Icelandic and Eastern European skin cancer patients (as well as a large number of controls). The authors estimate that approximately 1.6% of all individuals of European ancestry are homozygous for both risk alleles, and thus that percentage of the population is at 2.68x higher risk for BCC compared to individuals who are rs7538876(G;G) a... |
| rs763110(C;T)<br>Magnitude: 2<br>Frequency: 48.6%<br>Repute:Good<br>References:19 | ~0.80x reduced cancer risk...more... (hide)<br><br>rs763110, also known as -844C>T, is a SNP in the FAS ligand FASLG gene. A meta-analysis of 19 published studies, including 11,105 cancer cases and 11,372 controls, concluded that the rs763110(C;T) and (T;T) genotypes were associated with a significantly reduced (about 0.80x) cancer risk of all cancer types. |
| rs3738579(C;T)<br>Magnitude: 2<br>Frequency: 49.6%<br>Repute:Good<br>References:0 | 0.5x decreased risk for cervical cancer, HNSCC, and breast cancer<br>...more... (hide)<br><br>rs3738579 represents a SNP in the 5' UTR region upstream of the RNASEL gene. A study of patients diagnosed with carcinoma of the uterine cervix, head and neck squamous cell carcinomas (HNSCC), and breast cancer found 1.5x-2x increased risk for all three cancer types for the rs3738579(T;T) genotype, while finding decreased risk (0.5x) for rs3738579(C;T) heterozygotes. rs3738579(C;C) homozygotes had 0.6x less risk for cervical cancer but increased risk for HNSCC (1.4x) and breast cancer (1.8x). Although statistics were not reported per genotype, a combination of data from all three cancer forms over all genotypes provided strong statistical evidence for rs3738579 as a cancer marker, with a p-value of 4.43Ã—10eâˆ’5. |
| rs4149268(G;G)<br>Magnitude: 2<br>Frequency: 52.2%<br>Repute:Good<br>References:10 | associated with higher HDL cholesterol...more... (hide)<br><br>G allele is associated with 0.82mg/dl increase in HDL cholesterol (good cholesterol). [GWAS:HDL cholesterol] |

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| rs10936599(C;C)<br>Magnitude: 2<br>Frequency: 53.1%<br>Repute:Good<br>References:14 | longer telomeres, longer life?...more... (hide)<br><br>[GWAS:Celiac disease] | |
| rs7903146(C;C)<br>Magnitude: 2<br>Frequency: 54.9%<br>Repute:Good<br>References:293 | Normal (lower) risk of Type 2 Diabetes and Gestational Diabetes. Normal insulin secretion in the pancreas. This has been confirmed in most races.<br>...more... (hide)<br>This SNP in TCF7L2 influences the risk of Type-2 diabetes (T2D). This SNP is also known as IVS3C>T. rs7903146(C;T) rs7903146(T;T) strongly predicted future type-2 diabetes. Considered in context with rs7903146 rs12255372 rs10885406. Note: this is one of two SNPs within the TCF7L2 gene that have been reported to be associated with type-2 diabetes, the other being rs4506565. They have approximately equal power to estimate risk for type-2 diabetes, and the results from one correlate 92% of the time with the other. Full text of the paper is available from from Plos Medicine. Or from NCBI as . TCF7L2 polymorphisms and progression to diabetes in the Diabetes Prevention Program. reconfirmed in a diverse population Associated in a study of ~1000 Hispanic-Americans with reduced insulin secretion... | |
| rs6505162(A;C)<br>Magnitude: 2<br>Frequency: 58.0%<br>Repute:Good<br>References:14 | 0.58x decreased risk for esophageal cancer<br><br>rs6505162 is a SNP located in the pre-miRNA region of mir423. The major allele is rs6505162(C). A study of 346 Caucasians with esophageal cancer found that compared with the homozygous wild-type genotype of rs6505162, individuals with the heterozygous and homozygous variant genotype had a significantly reduced esophageal cancer risk with odds ratios of 0.58 (CI: 0.41-0.82) and 0.43 (CI: 0.27-0.68), respectively. After stratifying by gender, smoking, and age, the authors discovered that the protective effect was significant only for individuals under 64 years of age, regardless of sex or whether they smoked. Other unfavorable genotypes (ie SNPs) were also identified that were not as significant as rs6505162, and the overall risk of esophageal cancer increased with increasing numbers of unfa... | ...more... (hide) |
| rs4143094(G;G)<br>Magnitude: 2<br>Frequency: 58.5%<br>Repute:Good | no increased risk of colorectal cancer correlated with consumption of processed meats<br>...more... (hide) | |

References:1

'"rs4143094"' is a SNP identified by a genome-wide diet-gene interaction analysis (GxE), and was found to be associated with increased risk of colon cancer correlated to the dietary variable of processed meat consumption. It is located on chromosome 10p14, 7.2kb upstream of GATA binding protein 3 ("GATA3"), in the promoter region of the gene rs4143094 was identified in a screen of 2.7 million SNPs for the risk of colorectal cancer, by combining 10 studies comparing 9287 cases and 9117 controls. Data was obtained from the Genetics and Epidemiology of Colorectal Cancer Consortium (GECCO) and analyzed by conventional case-control logistic regression for multiplicative interactions between SNPs and dietary factors. rs4143094 was the most significantly implicated SNP in 10p14 for processed ...

rs3819331(T;T)
Magnitude: 2
Frequency: 67.3%
Repute:Good
References:0

lower risk of autism...more... (hide)

Examination of tetrahydrobiopterin pathway genes in autism. Examination of association to autism of common genetic variation in genes related to dopamine.

rs11045585(A;A)
Magnitude: 2
Frequency: 72.3%
Repute:Good
References:8

24% chance (lower than average) of docetaxel-induced leukopenia/neutropenia
...more... (hide)

rs12762549 and rs11045585 can be used to predict whether docetaxel will induce leukopenia/neutropenia, according to a study of ~100 Japanese patients. When patients were classified into three groups by the scoring system based on the genotypes of these two SNPs, patients with a score of 1 or 2 were shown to have a significantly higher risk of docetaxel-induced leukopenia/neutropenia as compared to those with a score of 0 (P = 0.0000057; odds ratio [OR], 7.00; 95% CI [confidence interval], 2.95-16.59). This prediction system correctly classified 69.2% of severe leukopenia/neutropenia and 75.7% of non-leukopenia/neutropenia into the respective categories. [Effect percentages provided in table are raw population fractions taken from the referenced study based on rs11045585 only. Total populat...

rs16973225(A;A)
Magnitude: 2
Frequency: 89.1%
Repute:Good
References:1

common/normal; aspirin use reduces colorectal cancer risk a bit see discussion at rs16973225
...more... (hide)

rs16973225 is a SNP on chromosome 15q25.2 near the IL16

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| | gene. In a long-term study of ~17,000 individuals and aspirin or NSAID use, the few (9%) with either an (A;C) or (C;C) genotype showed no risk reduction for colorectal cancer compared with (A;A) individuals among those taking anti-inflammatory drugs, who did benefit (prevalence, 28% vs 38%; (A;A) odds ratio 0.66 when taking aspirin/NSAID, CI: 0.62-0.71, p = 1.9 × 10e−30). If this data is robust and all other factors are equal, this indicates that most people will lower their colorectal cancer risk by taking aspirin or NSAIDs, since most people have rs16973225(A;A) genotypes. However, the few people who are rs16973225(A;C) or are rs16973225(C;C) will not change their risk one way or the other by taking aspirin. This same study reached ... |
| rs671(G;G)<br>Magnitude: 2<br>Frequency: 100.0%<br>Repute:Good<br>References:52 | Alcohol Flush: Normal, doesn't flush. Normal hangovers. Normal risk of Alcoholism. Normal risk of Esophageal Cancer. Disulfiram is effective for alcoholism. Two working copies of Aldehyde Dehydrogenase (ALDH2) mean you won't turn bright red when you drink alcohol, unlike most Asians. After your body turns alcohol into the dangerous acetaldehyde, this gene quickly turns the acetaldehyde into vinegar. It also stops the acetaldehyde in cigarettes from causing Esophageal Cancer. It reduces hangovers compared to Asians, but less hangovers and flushing obviously increases the risk of alcoholism. Alcoholism can be treated effectively with the drug Disulfiram (Antabuse) which would stop this gene from working and cause the Asian flush in non-Asians. PMC2659709 [http://www.ncbi.nlm.nih.gov/pmc/art... ...more... (hide)<br>rs671 is a classic SNP, well known in a sense through the phenomena known as the 'alcohol flush', also known as the 'Asian Flush' or 'Asian blush', in which certain individuals, often of Asian descent, have their face, neck and sometimes shoulders turn red after drinking alcohol. The rs671(A) allele of the ALDH2 gene is the culprit, in that it encodes a form of the aldehyde dehydrogenase 2 protein that is defective at metabolizing alcohol. This allele is known as the ALDH*2 form, and individuals possessing either one or two copies of it show alcohol-related sensitivity responses including facial flushing, and severe hangovers (and hence they are usually not regular drinkers). Perhaps not surprisingly they appear to suffer less from alcoholism and alcohol-related liver disease. Disulfiram (... |

rs12979860(C;C)
Magnitude: 2
Repute:Good
References:182

~80% of such hepatitis C patients respond to treatment (hide)
...more...

rs12979860 is a SNP near the IL28B gene, encoding interferon-lambda-3 (IFN-lambda-3). Several studies now indicate that this SNP predicts hepatitis C treatment-induced viral clearance. It is associated with an approximately twofold change in response to pegylated interferon-alpha (PEG-IFN-alpha) plus ribavirin (RBV) treatment, both among patients of European ancestry (p = 1.06 x 10e-25) and African-Americans (p = 2.06 x 10e-3). These studies indicate that the virus was eradicated in ~80% of (C;C) patients, compared to only about 25% of those with (T;T), while (C;T) response was intermediate. FDA PGx mentions about rs12979860 with respect to Boceprevir, Simeprevir, Sofosbuvir, and Telaprevir 23andMe blog rs8099917(C) less likely to respond to Hepatitis C treatment. A functional SNP rs368234...

rs10033464(G;G)
Magnitude: 1.9
Frequency: 82.3%
Repute:Good
References:13

0.92x decreased risk of Atrial Fibrillation and cardioembolic stroke. This is the normal version which makes the heart form properly and makes people slightly less likely to get Atrial Fibrillation (quivering of the top chambers of the heart caused by chaotic electrical signals) and less likely to have a cardioembolic stroke (blocked blood flow to the brain). But also check the other SNP nearby (rs2200733) which affects Atrial Fibrillation even more. 23andMe gave this a 4 star confidence rating as it has been heavily studied. It has been confirmed in Europeans.
...more... (hide)

Two SNPs from chromosome 4q25, rs2200733 and rs10033464, were found to be associated with atrial fibrillation in a study of both European and Asian populations. The odds ratio associated with one or more copies of either risk allele was ~1.4x. In 1,661 Icelandic ischemic stroke samples and two large European replication sets combined, rs10033464 was associated with cardioembolic stroke (CES) (odds ratio 1.27, p = 6.1 x 10e-4). rs2200733 and rs10033464 are strongly associated with AF in four cohorts of European descent. Variation in the 4q25 chromosomal locus predicts atrial fibrillation after coronary artery bypass graft surgery. Chromosome 4q25 variants and atrial fibrillation recurrence after catheter ablation. Common variants in KCNN3 are associated with lone atrial fibrillation. Associ...

rs2303138(G;G)
Magnitude: 1.9

Normal risk (0.1%) for Ankylosing Spondylitis. Normal, very low risk of the

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|---|---|
| Frequency: 85.0%<br>Repute:Good<br>References:0 | rare autoimmune disease that causes inflammation of the spine and joints. Based on preliminary research.<br>...more... (hide)<br>Each A allele at rs2303138 in the LNPEP gene is associated with 1.5x higher odds of ankylosing spondylitis, according to a 2007 report. |
| rs1052576(A;A)<br>Magnitude: 1.7<br>Frequency: 29.2%<br>Repute:Good<br>References:6 | 0.5x reduced risk of multiple myeloma, general reduced risk of cancer A number of small studies with fairly low p-values, but with low heterogeneity across studies, show a protective effect for the A allele which is greater in homozygotes.<br>...more... (hide)<br>also known as Q221R rs1052576 is located on chromosome 1p36.21 in the coding region of CASP9. The CAG to CGG transition results in the allele Q221R. This SNP is associated with ADHD, general cancer risk, multiple myeloma, non-small cell lung cancer and non-Hodgkin lymphoma .Generally the A allele confers protection from a variety of cancers. An intact apoptotic pathway is essential for many cellular processes including development, immune response and prevention of cellular transformation. Caspases are cysteinyl aspartate proteases involved in signal transduction cascades that result in programmed cell death. CASP9 is an initiator caspase and part of the intrinsic pathway, which is an evolutionarily conserved apoptotic pathway. It is responsible for cleaving and activating the executioner ... |
| rs420259(C;T)<br>Magnitude: 1.5<br>Frequency: 31.0%<br>Repute:Good<br>References:5 | Possibly reduced risk of Bipolar Disorder. This appears to lower the risk of Bipolar Disorder compared to the more common version of the gene, based on preliminary research.<br>...more... (hide)<br>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. [PharmGKB:Non-Curated GWAS Results: Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls (Initial Sample... |

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| rs4489954(G;T)<br>Magnitude: 1.5<br>Frequency: 45.5%<br>Repute:Good<br>References:3 | 0.69x risk risk of developing restless legs syndrome<br>...more... | (hide) |
| | rs4489954, a SNP located in a region of chromosome 15q, has been linked to a lower frequency of restless legs syndrome, a common sleep disorder, with an overall odds ratio of 0.69 (CI: 0.69-0.82) for the (T) minor allele. [PharmGKB:Curated In replicated GWAS case-control studies of Caucasian(European and French-Canadian) familial and sporadic RLS, the G allele of rs4489954 was significantly associated with Restless Legs Syndrome.] | |
| rs3851179(A;G)<br>Magnitude: 1.5<br>Frequency: 46.9%<br>Repute:Good<br>References:31 | 0.85x decreased risk for Alzheimer's disease | ...more...<br>(hide) |
| | rs3851179 is a SNP upstream of the PICALM gene. A study of over 5,000 Alzheimer's disease patients (and over 10,000 controls) found a slight protective effect of the (A) allele of this SNP. rs3851179(A) carriers had a slightly decreased risk for Alzheimer's disease, with an odds ratio of 0.85 (CI: 0.8-0.9, p=1.9x10e-8). The association of this SNP and late-onset Alzheimer's disease was replicated over a total of another 1829 cases (and 2576 controls). [GWAS:Alzheimer's disease] | |
| rs3784709(C;T)<br>Magnitude: 1.5<br>Frequency: 47.8%<br>Repute:Good<br>References:3 | 0.71x risk of developing restless legs syndrome | ...more...<br>(hide) |
| | rs3784709, a SNP located in a region of chromosome 15q, has been linked to a lower frequency of restless legs syndrome, a common sleep disorder, with an overall odds ratio of 0.71 (CI: 0.60-0.83) for the (T) minor allele. [PharmGKB:Curated In replicated GWAS case-control studies of Caucasian(European and French-Canadian) familial and sporadic RLS, the C allele of rs3784709 was significantly associated with Restless Legs Syndrome.] | |
| rs1063192(C;T)<br>Magnitude: 1.5<br>Frequency: 47.8%<br>Repute:Good<br>References:17 | 0.71x reduced risk of myocardial infarction...more... | (hide) |
| | rs1063192 is a SNP in the cyclin-dependent kinase inhibitor 2B CDKN2B gene. A study of 432 Han Chinese patients with myocardial infarctions concluded that male subjects carrying a rs1063192(C) allele were at 0.71x decreased risk (for MI). | |

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| | Myocardial Infarction [GWAS:Vertical cup-disc ratio] |
| rs1026732(A;G)<br>Magnitude: 1.5<br>Frequency: 48.2%<br>Repute:Good<br>References:4 | 0.70x risk for restless legs...more... (hide)<br><br>rs1026732, a SNP located in a region of chromosome 15q, has been linked to a lower frequency of restless legs syndrome, a common sleep disorder, with an overall odds ratio of 0.70 (CI: 0.59-0.82) for the (A) minor allele. [PharmGKB:Curated In replicated GWAS case-control studies of Caucasian(European and French-Canadian) familial and sporadic RLS, the G allele of rs1026732 was significantly associated with Restless Legs Syndrome.] |
| rs11136000(C;T)<br>Magnitude: 1.5<br>Frequency: 48.7%<br>Repute:Good<br>References:34 | 0.84x decreased risk for Alzheimer's disease ...more... (hide)<br><br>rs11136000 is a SNP associated with the clusterin CLU gene, which has also been known as the APOJ gene. A study of over 5,000 Alzheimer's disease patients (and over 10,000 controls) found a slight protective effect of the (T) allele of this SNP. rs11136000(T) carriers had a slightly decreased risk for Alzheimer's disease, with an odds ratio of 0.84 (CI: 0.8-0.9, p=1.4x10e-9). The association of this SNP and late-onset Alzheimer's disease was replicated over a total of another 1829 cases (and 2576 controls). A MRI study of ~400 young (average age: 23) healthy carriers of rs11136000(C) alleles found a distinct profile of lower white matter integrity in these individuals that may increase their vulnerability to developing Alzheimer's later in life. To put it another way, 50 years before the a... |
| rs11635424(A;G)<br>Magnitude: 1.5<br>Frequency: 48.7%<br>Repute:Good<br>References:3 | 0.70x risk for restless legs...more... (hide)<br><br>rs11635424, a SNP located in a region of chromosome 15q, has been linked to a lower frequency of restless legs syndrome, a common sleep disorder, with an overall odds ratio of 0.70 (CI: 0.59-0.82) for the (A) minor allele. [PharmGKB:Curated In replicated GWAS case-control studies of Caucasian(European and French-Canadian) familial and sporadic RLS, the G allele of rs11635424 was significantly associated with Restless Legs Syndrome.] |
| rs12593813(A;G) | 0.71x risk for restless legs...more... (hide) |

Magnitude: 1.5
Frequency: 50.0%
Repute:Good
References:6

rs12593813, a SNP located in a region of chromosome 15q, has been linked to a lower frequency of restless legs syndrome, a common sleep disorder, with an overall odds ratio of 0.71 (CI: 0.60-0.83) for the (A) minor allele. Restless Legs Syndrome: Preliminary Research [PharmGKB:Curated GWAS Results: Genome-wide association study of restless legs syndrome identifies common variants in three genomic regions (Initial Sample Size: 401 cases, 1,644 controls; Replication Sample Size: 1,158 cases, 1,178 controls; Risk Allele: rs12593813-G).] [GWAS:None] [GWAS:Restless legs syndrome]

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rs4149274(C;C)
Magnitude: 1.5
Frequency: 62.1%
Repute:Good
References:2

Associated with higher HDL (good) cholesterol. ...more... (hide)

C allele is associated with 1.51mg/dl increase in HDL cholesterol (good cholesterol).

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rs10050860(C;C)
Magnitude: 1.3
Frequency: 55.8%
Repute:Good
References:9

Normal risk (about 0.1%) for Ankylosing Spondylitis. Normal, but still very low, risk (1 in a thousand) of the rare autoimmune disease that causes inflammation of the spine and joints. Based on preliminary research.
...more... (hide)

rs10050860 is one of several SNPs in the ERAP1 gene that has been shown in a large (over 1,000 Caucasian patients) study to be associated with ankylosing spondylitis. The odds ratio is 0.71 (p=7.7x10e-9). Note that the text description (but not the result) at 23andMe is wrong. It is the uncommon T allele that gives protection. Association of an ERAP1 ERAP2 haplotype with familial ankylosing spondylitis. Investigating the genetic association between ERAP1 and ankylosing spondylitis. Serum cytokine receptors in ankylosing spondylitis: relationship to inflammatory markers and endoplasmic reticulum aminopeptidase polymorphisms. ERAP1 polymorphisms and haplotypes are associated with ankylosing spondylitis susceptibility and functional severity in a Spanish population. Associations between ERAP1...

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gs184
Magnitude: 1.2
Repute: Good

Able to taste bitterness.
Normal tasting ability for bitterness. This makes turnip, cabbage, brussels sprouts, broccoli, and other cruciferous vegetables taste more interesting and less horribly

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| | bland. Coffee and dark beers also tastes more bitter. You can taste propylthiouracil (PROP), PTC, and related chemicals. On the other hand, you can't taste the unpleasant bitterness of the tropical fruit from the Bignay tree, it will taste sweet to you. You probably eat healthier. |
| rs6048(A;G)<br>Magnitude: 1.2<br>Frequency: 18.5%<br>Repute:Good<br>References:3 | slightly lower risk (10-20%) of deep vein thrombosis (hide)<br>...more...<br>Gene variants associated with deep vein thrombosis. Updated analysis of gene variants associated with deep vein thrombosis. [OMIM:?] |
| rs4988235(C;T)<br>Magnitude: 1.1<br>Frequency: 36.3%<br>Repute:Good<br>References:16 | likely to be able to digest milk as an adult<br>Approximately 80% of individuals with this genotype are able to digest milk (lactose) as adults.<br>...more... (hide)<br>Also known as 'C/T(-13910)', and located in the MCM6 gene but with influence on the lactase LCT gene, rs4988235 is one of two SNPs that is associated with the primary haplotype associated with hypolactasia, more commonly known as lactose intolerance in European Caucasian populations. , In these populations, the rs4988235(T) allele is both the more common allele and the one associated with lactase persistence; individuals who are rs4988235(C;C) are likely to be lactose intolerant. In populations of sub-Saharan Africans, though, the rs4988235(T) allele is so rare that it's unlikely to be predictive of lactase persistence, and other SNPs are predictive instead. * See also OMIM 601806.0001 Measuring European population stratification with microarray genotype data. Adult-type hypolactasia is no... |
| rs2293347(G;G)<br>Magnitude: 1.1<br>Frequency: 81.4%<br>Repute:Good<br>References:9 | among NSCLC patients, better Gefitinib response; common<br>...more... (hide)<br>rs2293347, also known as D994D, is a SNP in the epidermal growth factor receptor EGFR gene. In a study of therapeutic outcome and survival in 84 advanced non-small-cell lung cancer (NSCLC) patients treated with Gefitinib, the response rate for rs2293347(G;G) patients was almost double that of other genotypes for this SNP (71.2% versus 37.5%, p=0.0043). The rs2293347(G;G) genotype was also associated with longer progression-free survival compared with the rs2293347(A;G) or (A;A) genotypes (11 months versus 3 months, p=0.0018). |

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| rs2351299(G;T)<br>Magnitude: 1<br>Frequency: 27.0%<br>Repute:Good<br>References:1 | possible reduced risk of Autism...more...<br><br>Investigation of autism and GABA receptor subunit genes in multiple ethnic groups. | (hide) |
| rs2736990(T;T)<br>Magnitude: 1<br>Frequency: 27.4%<br>Repute:Good<br>References:6 | Normal risk of developing Parkinson's Disease<br><br>rs2736990 is a SNP associated with Parkinson's disease (PD), a neurodegenerative disorder characterized by the degradation of dopamine-producing neurons in the substantia nigra. Rs273990 is located on chromosome 4q22 in the synuclein-alpha SNCA locus. The function of SNCA is not fully understood, but it predominately found in the presynaptic terminals of neurons, and it is thought that it plays a role in maintaining adequate supplies of synaptic vesicles. SNCA is also a major component of Lewy Bodies (LB), protein accumulations often found in neurons in patients with PD. The best evidence for the association of rs2736990 with PD was provided in a genome-wide association study (GWAS) that found an increased frequency of the minor C allele (T is the major allele) in position 90897564 a... | ...more...<br>(hide) |
| rs3087243(G;G)<br>Magnitude: 1<br>Frequency: 28.3%<br>Repute:Good<br>References:82 | Normal risk for autoimmune diseases...more...<br><br>The rs3087243 SNP is also known in the literature as the CT60 G>A or the +6230G>A polymorphism, and it is located in the CTLA4 gene. In Asian (Japanese) populations, the presence of an rs3087243(G) allele represents a 1.3 fold increased risk of autoimmune thyroid disease, and for those with autoimmune thyroid disease, a 1.5 fold increased risk of type-1 diabetes. However, in individuals without autoimmune thyroid disease, no association was seen between this SNP and type-1 diabetes. The authors speculate that earlier studies may have reported associations between this SNP and type-1 diabetes that were actually primarily based on the association with autoimmune thyroid disease. This same SNP, rs3087243, has also been implicated as a (minor) risk factor for developing rheumatoid arthri... | (hide) |
| rs828907(G;G)<br>Magnitude: 1<br>Frequency: 28.3% | Normal risk of developing bladder cancer...more... | (hide) |

Repute:Good
References:4

rs828907, also known as G-1401T, is a SNP in the Ku80 gene. A study of 288 Chinese patients with bladder cancer and 288 age- and gender-matched healthy controls found that the rs828907(T) allele conferred a significant (p=0.0055) increased risk of bladder cancer. In association with smoking, the (G;T) and (T;T) genotypes conferred a 2x risk (CI:1.232-3.419) for bladder cancer. correlated with oral cancer susceptibility

rs10492519(A;A)
Magnitude: 1
Frequency: 29.2%
Repute:Good
References:1

Normal risk of developing prostate cancer...more... (hide)

rs10492519 is one of seven SNPs found in a combined study of over 1,000 patients to be associated with increased risk for prostate cancer. The risk allele for this SNP is (G); and while the odds ratio was not specifically reported, the probability of false significance (not permuted though) was given as p=5.6 x 10e-6, using an additive model of risk.

rs10260404(T;T)
Magnitude: 1
Frequency: 31.9%
Repute:Good
References:10

Normal risk of developing ALS...more... (hide)

rs10260404, a SNP in the region of the DPP6 gene on chromosome 7, has been associated with the sporadic form of ALS (Lou Gehrig's disease) in a study of 1000+ European patients. The overall odds ratio for the risk allele rs10260404(C) is 1.30 (CI: 1.18-1.43, p=0.017). When broken down by genotype, the odds ratios for heterozygotes are 1.20 (CI: 1.06-1.41), and for rs10260404(C;C) homozygotes, 1.60 (CI: 1.32-1.92). A 'C-C' haplotype for this SNP and that of it's neighbor rs10239794 is also highly (statistically; p=10e-9) associated with ALS. However, in an expanded study pooling 4 populations (Irish, Dutch, US, Polish) rs10260404 failed to reach Bonferroni significance. although it did remain significant in the (expanded) Irish-only population. [PharmGKB:Non-Curated GWAS results: Screening...

rs12752888(C;T)
Magnitude: 1
Frequency: 33.6%
Repute:Good
References:1

Normal progression to Alzheimer's disease from mild cognitive impairment. ...more... (hide)

rs12752888 is a SNP on chromosome 1 upstream of the gene ACOT11. ACOT11 has been associated with obesity and is a member of a family of enzymes that help catalyze the de-esterification of fatty acids. One study used genotyping

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| | information from 489 patients with mild cognitive impairment (MCI) from the Vitamin E trial. The GWAS employed used Alzheimer's disease progression measured quantitatively from Clinical Dementia Rating-sum of boxes (CRD-SB) scores over time. Additionally, the study utilized information about 200 Alzheimer's disease patients, 300 MCI patients, and 200 controls from the Alzheimer's Disease Neuroimaging Initiative (ADNI) consortium. Significance was assessed based on the interaction term between genotype and time in a Cox proportional hazards model. rs127528... |
| rs12037606(G;G)<br>Magnitude: 1<br>Frequency: 39.8%<br>Repute:Good<br>References:0 | Normal risk of developing Crohn's disease...more... (hide)<br><br>rs12037606 has been reported in a large study to be associated with Crohn's disease. The risk allele (oriented to the dbSNP entry) is (A); the odds ratio associated with heterozygotes is 1.22 (CI 1.07-1.40), and for homozygotes, 1.52 (CI 1.28-1.82). |
| rs13181(T;T)<br>Magnitude: 1<br>Frequency: 40.7%<br>Repute:Good<br>References:43 | Normal risk for cutaneous melanoma...more... (hide)<br><br>rs13181, also known as c.2251A>C, p.Lys751Gln and K751Q, is a variant in the ERCC2 gene on chromosome 19. A meta-analysis of 20 studies, comprising 2,308 cases, ultimately concluded that the rs13181(G) allele is associated with increased cutaneous melanoma risk (odds ratio 1.12, CI: 1.03-1.21, p = 0.01; population attributable risk = 9.6%). A preliminary (as yet unreplicated) study of 430 patients with ovarian cancer concluded that rs13181(C;C) is associated with an increased risk of ovarian cancer (OR 5.01 relative to rs13181(A;A), CI: 3.37-7.43; p < 0.0001). Interestingly, heterozygotes were reported to have a lower odds ratio for developing ovarian cancer (of 0.43; CI: 0.27–0.62, p < 0.0001) compared to rs13181(A;A) individuals. Polymorphisms in XPD and TP53 and mutation in hum... |
| rs4792311(G;G)<br>Magnitude: 1<br>Frequency: 40.7%<br>Repute:Good<br>References:1 | Normal risk of prostate cancer...more... (hide)<br><br>Sequence variants of elaC homolog 2 (Escherichia coli) (ELAC2) gene and susceptibility to prostate cancer in the Health Professionals Follow-Up Study. Development of a fingerprinting panel using medically relevant polymorphisms. |

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| | Genetic variants and prostate cancer risk: candidate replication and exploration of viral restriction genes. Population based allele frequencies of disease associated polymorphisms in the Personalized Medicine Research Project. [OMIM:PROSTATE CANCER, SUSCEPTIBILITY TO] |
| rs699473(T;T)<br>Magnitude: 1<br>Frequency: 41.1%<br>Repute:Good<br>References:7 | Normal brain tumor risk...more... (hide)<br><br>A study of non-Hispanic Caucasians with various types of brain tumors concluded that there was an increased risk of glioma (odds ratio 1.3, CI: 1.0-1.7) and meningioma (odds ratio 1.7, CI: 1.1-2.7) associated with the rs699473(C) allele. |
| rs807701(T;T)<br>Magnitude: 1<br>Frequency: 41.5%<br>Repute:Good<br>References:2 | Normal dyslexia risk...more... (hide)<br><br>Rs807701, a SNP in the DCDC2 gene, is in a region that crops up in several independent studies as likely to associated with dyslexia. The risk allele in the Caucasian populations studied is (C). One study reports that the odds ratio for rs807701 genotypes increases if calculated from subsets of more severely dyslexic individuals as compared to more heterogenous, larger groups of dyslexic individuals. The genotype relative risk (GRR) for rs807701(C;C) increased from 1.88 (95% CI 0.89-3.97; P=.058) for the larger group up to 5.04 (95% CI 1.35-18.88; P=.002) for the most severely affected group. Combined with another SNP marker in the DCDC2 gene, rs793862, the (haplotype) GRR also increased for the homozygous haplotype rs793862(A)-rs807701(C), from 4.11 (95% CI 2.77-6.08; P<.0001) for the ... |
| rs729302(A;A)<br>Magnitude: 1<br>Frequency: 46.0%<br>Repute:Good<br>References:17 | Normal risk of developing rheumatoid arthritis ...more... (hide)<br><br>A variant located 64 bp upstream of the first untranslated exon (exon 1A), consisting of a 5 bp insertion/deletion CGGGG, may be the causative SNP in this region that is most responsible for increasing SLE risk, however it lacks an rs# (i.e. it isn't registered in dbSNP). A meta-analysis comprising 5 case-control studies, totaling 6,582 rheumatoid arthritis cases and 5,375 controls, concluded that several IRF5 gene SNPs were indeed (still) significantly associated with the disease. The rs729302(C) allele was associated with a slight protective effect (odds ratio 0.89, CI: 0.8-0.977, p = 0.015). |

[GWAS:Systemic lupus erythematosus]

rs492602(C;T)
Magnitude: 1
Frequency: 46.0%
Repute:Good
References:10

Normal B12 levels...more... (hide)

From : We identified a strong association (p = 5.36 x 10(-17)) between rs492602 in the FUT2 gene and plasma vitamin B(12) levels in a genome-wide scan (n = 1,658) and an independent replication sample (n = 1,059) from the Nurses' Health Study. Women homozygous for the rs492602(C) allele (in dbSNP orientation) had higher B(12) levels. This allele is in strong linkage disequilibrium with the FUT2 nonsecretor variant encoding W143X, suggesting a plausible mechanism for altered B(12) absorption and plasma levels. [PharmGKB:Curated GWAS Results: Common variants of FUT2 are associated with plasma vitamin B12 levels (Initial Sample Size: 1,658 women; Replication Sample Size: 1,059 women; Risk Allele: rs492602-G). This variant is associated with Plasma level of vitamin B12.] [OMIM:FUCOSYLTRANSFER...

rs4939827(C;T)
Magnitude: 1
Frequency: 46.9%
Repute:Good
References:42

0.86x decreased risk for colorectal cancer...more... (hide)

rs4939827 is one of 3 SNPs in the SMAD7 gene associated with risk for colorectal cancer, based on a large study (7,400+ cases) conducted in the UK. The odds ratios show a decreased risk for the minor rs4939827(C) allele; the OR for (C;C) homozygotes is 0.73 (CI: 0.66-0.8), and for (C;T) heterozygotes 0.86 (CI: 0.79-0.92), overall p=1x10<sup>-12</sup>. [PharmGKB:Non-Curated GWAS Results: Genome-wide association scan identifies a colorectal cancer susceptibility locus on 11q23 and replicates risk loci at 8q24 and 18q21 (Initial Sample Size: 981 cases, 1,002 controls; Replication Sample Size: 16,476 cases, 15,351 controls; Risk Allele: rs4939827-T).] [OMIM:MOTHERS AGAINST DECAPENTAPLEGIC, DROSOPHILA, HOMOLOG OF, 7; SMAD7] [GWAS:Colorectal cancer] [GWAS:Colorectal cancer]

rs2952768(C;T)
Magnitude: 1
Frequency: 48.6%
Repute:Good
References:1

slightly less drug dependence, decreased effectiveness of analgesics
...more... (hide)

Genome-wide association study identifies a potent locus associated with human opioid sensitivity rs2952768(C) was

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| | associated with more analgesic requirements ... exhibited less vulnerability to severe methamphetamine or alcohol dependence, fewer eating disorders, and a lower 'Reward Dependence' score on a personality questionnaire. The (C;C) genotype was significantly associated with the elevated expression of a neighboring gene, CREB1. SNPs in this locus are the most potent genetic factors associated with human opioid sensitivity known to date, affecting both the efficacy of opioid analgesics such as morphine and fentanyl and liability to severe substance dependence. These findings may provide valuable information for the personalized treatment of pain and drug dependence. Oth... |
| rs700651(A;A)<br>Magnitude: 1<br>Frequency: 48.6%<br>Repute:Good<br>References:6 | Normal risk of aneurysm...more... (hide)<br><br>23andMe blog aneurysm *rs700651 2 G 1.18x risk 1.56x risk *rs10958409 8 A 1.37x risk 1.79x risk *rs1333040 9 T 1.29x risk 1.67x risk Increased risk of intracranial (brain aneurysm) with certain SNPs. rs700651(G;G) associated with 1.56x risk of developing an intracranial aneurysm (IA). Heterozygote risk is 1.18x. Other SNPs associated with increased risk of developing IA include rs1333040 and rs10958409. rs10958409 is located on chromosome 9; rs1333040 is located on chromosome 8; rs700651 is located on chromosome 2. Unfortunately, the first sign of an intracranial aneurysm is rupture. Catastrophic bleeding and death may result. Specific knowledge of risk may influence screening decision. More information: Intracranial aneurysm screening: indications and advice for practice. . [PharmGKB:Non... |
| rs10096097(A;A)<br>Magnitude: 1<br>Frequency: 49.6%<br>Repute:Good<br>References:1 | normal Anorexia Nervosa risk...more... (hide)<br><br>rs10096097 is a SNP near the membrane bound O-acyltransferase domain containing 4 MBOAT4 gene. A study of 543 German anorexia nervosa patients observed some association between the disorder and rs10096097(G;G) (nominal two-sided p = 0.031). |
| rs6812193(C;T)<br>Magnitude: 1<br>Frequency: 51.3%<br>Repute:Good<br>References:7 | normal risk of developing Parkinson's Disease ...more... (hide)<br><br>People with two copies of the T allele at rs6812193 appear to have a slightly lower risk of Parkinson's disease. The same |

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| | researchers identified another risk allele for Parkinson's disease at rs11868035. plos [GWAS:Parkinson's disease] |
| rs3814570(C;C)<br>Magnitude: 1<br>Frequency: 53.1%<br>Repute:Good<br>References:0 | Normal risk of developing Crohn's disease...more... (hide)<br><br>rs3814570 is a SNP in the transcription factor 7-like 2 (T-cell specific, HMG-box) TCF7L2 gene. A study of 784 Crohn's disease patients with ileal involvement concluded that rs3814570 was associated with increased risk, but not with risk for colonic Crohn's disease or ulcerative colitis. The odds ratio was 1.27 (CI: 1.07 - 1.52, p = 0.00737). Evidence for association between polycystic ovary syndrome (PCOS) and TCF7L2 and glucose intolerance in women with PCOS and TCF7L2. |
| rs2300478(T;T)<br>Magnitude: 1<br>Frequency: 56.2%<br>Repute:Good<br>References:8 | Normal risk of developing restless legs syndrome ...more... (hide)<br><br>rs2300478, a SNP located in the MEIS1 gene, has been linked to restless legs syndrome, a common sleep disorder, with an overall odds ratio of 1.78 (CI: 1.52-2.09) for the (G) risk allele . The association between this SNP and RLS has been replicated in three European populations, within family lines but not in sporadic cases . The highest association to restless legs syndrome is a haplotype consisting of the rs6710341(A) and rs12469063(G) alleles. This haplotype gives rise to an odds ratio of 2.75 (CI: 2.23-3.41) . Variants in MEIS1 rs2300478, BTBD9 rs9357271, and MAP2K5/SKOR1 rs1026732 confer a significant risk of RLS in a US population. In another study, rs2300478 was not associated with attention deficit hyperactivity disorder (ADHD). Association studies of variants in MEIS1, BTBD9, and... |
| rs3790565(T;T)<br>Magnitude: 1<br>Frequency: 57.1%<br>Repute:Good<br>References:2 | Normal risk of developing primary biliary cirrhosis ...more... (hide)<br><br>Primary biliary cirrhosis associated with HLA, IL12A, and IL12RB2 variants. C allele associated with increased risk of Primary biliary cirrhosis |
| rs12469063(A;A)<br>Magnitude: 1<br>Frequency: 57.5%<br>Repute:Good | Normal risk of developing restless legs syndrome ...more... (hide) |

References:4

rs12469063, a SNP located in the MEIS1 gene, has been linked to restless legs syndrome, a common sleep disorder, with an overall odds ratio of 1.78 (CI: 1.52-2.10) for the (G) risk allele. The association from this region that gives the highest association to restless legs syndrome, however, is a haplotype consisting of the rs6710341(A) and rs12469063(G) alleles. This haplotype gives rise to an odds ratio of 2.75 (CI: 2.23-3.41). MEIS1 intronic risk haplotype associated with restless legs syndrome affects its mRNA and protein expression levels. Replication of restless legs syndrome loci in three European populations. Genotyping sleep disorders patients. [OMIM:?]

rs9357271(T;T)
Magnitude: 1
Frequency: 57.5%
Repute:Good
References:6

Normal risk of developing restless legs syndrome ...more... (hide)

rs9357271, a SNP located in the BTBD9 gene region, has been linked to a lower frequency of restless legs syndrome, a common sleep disorder, with an overall odds ratio of 0.63 (CI: 0.52-0.75) for the (C) minor allele. This SNP may also be associated with Tourette syndrome (TS), particularly TS without obsessive-compulsive disorder, based on a study of ~600 patients. [PharmGKB:Curated In replicated GWAS case-control studies of Caucasian(European and French-Canadian) familial and sporadic RLS, the T allele of rs9357271 was significantly associated with Restless Legs Syndrome.] [OMIM:RESTLESS LEGS SYNDROME, SUSCEPTIBILITY TO, 6] [GWAS:None]

rs3745516(G;G)
Magnitude: 1
Frequency: 58.4%
Repute:Good
References:4

Normal risk of developing primary biliary cirrhosis ...more... (hide)

Each A allele of this SNP in the SPIB gene is associated with 1.46 times higher odds of primary biliary cirrhosis. [GWAS:Primary biliary cirrhosis]

rs7574865(G;G)
Magnitude: 1
Frequency: 60.2%
Repute:Good
References:108

normal risk of rheumatoid arthritis, SLE, Sjögren's syndrome, type-1 diabetes, and primary biliary cirrhosis. ...more... (hide)

rs7574865, a SNP in the third intron of the STAT4 gene, has been reported in a large study of Swedes to be associated with both rheumatoid arthritis (RA) and lupus (SLE). Among other studies, it has been confirmed in a meta-analysis of 8 studies totaling 7,381 patients and over 10,000 controls from both European and Asian populations. The risk allele (oriented to

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| | the dbSNP entry) is (T); the odds ratio associated the presence of a risk allele was 1.3 for rheumatoid arthritis and 1.55 for lupus (SLE). The paper states that, 'Homozygosity of the risk allele, as compared with absence of the allele, was associated with a more than doubled risk for lupus and a 60% increased risk for rheumatoid arthritis.' A study of 124 Caucasian patients with primary Sjögren's syndrome, an autoimmune dise... | |
| rs199533(C;C)<br>Magnitude: 1<br>Frequency: 61.9%<br>Repute:Good<br>References:2 | Normal risk of developing Parkinson's Disease<br><br>[GWAS:Parkinson's disease] | ...more...<br>(hide) |
| rs10501570(T;T)<br>Magnitude: 1<br>Frequency: 62.5%<br>Repute:Good<br>References:2 | Normal risk of developing Parkinson's Disease<br><br>Detecting disease-associated genotype patterns. Phactr2 and Parkinson's disease. [PharmGKB:Non-Curated GWAS results: Genome-wide genotyping in Parkinson's disease and neurologically normal controls: first stage analysis and public release of data (Initial Sample Size: 267 cases, 270 controls; Replication Sample Size: NR). This variant is associated with Parkinson's disease.] [GWAS:Parkinson's disease] | ...more...<br>(hide) |
| rs1975197(C;C)<br>Magnitude: 1<br>Frequency: 66.4%<br>Repute:Good<br>References:7 | Normal risk of developing restless legs syndrome<br><br>rs1975197, a SNP in the motor neuron PTPRD gene, has been linked to restless legs syndrome based on a study of several European populations totaling ~2,500 patients. The odds ratio associated with the rs1975197(T) risk allele is 1.31 (CI: 1.20-1.44, adjusted p = 5 x10e-9). Note that a nearby SNP, rs4626664, was also shown to independently confer risk for restless legs syndrome in this same study. Restless Legs Syndrome: Preliminary Research [PharmGKB:Curated GWAS Results: PTPRD (protein tyrosine phosphatase receptor type delta) is associated with restless legs syndrome (Initial Sample Size: 628 cases, 1,644 controls; Replication Sample Size: 1,835 cases, 3,111 controls; Risk Allele: rs1975197-T). This variant is associated with Restless legs syndrome.] [OMIM:RESTLESS LEGS SYNDROME, SUSCEP... | ...more...<br>(hide) |
| rs9561778(G;G)<br>Magnitude: 1<br>Frequency: 66.4% | Normal risk of adverse drug reations from cyclophosphamide treatments<br>...more... (hide) | |

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| Repute:Good<br>References:3 | rs9561778 is a SNP in the ATP-binding cassette, sub-family C (CFTR/MRP), member 4 ABCC4 gene. A stepwise case-control study of ~400 breast cancer Japanese patients receiving cyclophosphamide-based combination chemotherapy, roughly divided equally between those having or not having serious adverse drug reactions (ADRs), concluded that carriers of rs9561778(T) allele had a ~2x higher risk of ADRs. The ADRs for these patients tended to be gastrointestinal toxicity and leukopenia/neutropenia. [PharmGKB:Curated Risk or phenotype-associated allele: T. Phenotype: In a stepwise case-control association study of breast cancer patients receiving cyclophosphamide combination therapy, rs9561778 in ABCC4 was associated with cyclophosphamide-induced ADRs. Study size: 403. Study population/ethnicity: Ja... | |
| rs6710341(A;A)<br>Magnitude: 1<br>Frequency: 70.5%<br>Repute:Good<br>References:3 | Normal risk of developing restless legs syndrome<br><br>rs6710341, a SNP located in the MEIS1 gene, has been linked to restless legs syndrome, a common sleep disorder, with an overall odds ratio of 0.96 (CI: 0.77-1.17) for the (G) risk allele. The association from this region that gives the highest association to restless legs syndrome, however, is a haplotype consisting of the rs6710341(A) and rs12469063(G) alleles. This haplotype gives rise to an odds ratio of 2.75 (CI: 2.23-3.41). | ...more...<br>(hide) |
| rs10464059(G;G)<br>Magnitude: 1<br>Frequency: 72.3%<br>Repute:Good<br>References:1 | Normal risk of developing Parkinson's Disease<br><br>[GWAS:Parkinson's disease] | ...more...<br>(hide) |
| rs16953002(G;G)<br>Magnitude: 1<br>Frequency: 73.5%<br>Repute:Good<br>References:1 | Slightly lower risk of melanoma in individuals with low BMI Highly significant, replicated studies, p=3.6e-12. Risk allele A appears to represent a greater risk in those with low BMI, which is plausible due to the known role of FTO in obesity. This SNP is in intron 8, whereas BMI SNP's are usually in intron 1. The association was much stronger for those in the lowest quartile of BMI (1.66 per A allele risk) vs those in the other 3 quartiles (1.03 per A allele risk - no longer statistically significant). Risk for those in higher 3 | |

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| | quartiles of BMI is likely not substantially affected. ...more... (hide) [GWAS:Melanoma] | |
| rs2697962(G;G) Magnitude: 1 Frequency: 74.3% Repute:Good References:0 | Normal risk of developing Parkinson's Disease<br>rs2697962 is in linkage disequilibrium with a polymorphism that increases susceptibility to Parkinson's disease 1.67 times for carriers of the A allele | ...more... (hide) |
| rs4626664(G;G) Magnitude: 1 Frequency: 75.5% Repute:Good References:5 | Normal risk of developing restless legs syndrome<br>rs4626664, a SNP in the motor neuron PTPRD gene, has been linked to restless legs syndrome based on a study of several European populations totaling ~2,500 patients. The odds ratio associated with the rs4626664(A) risk allele is 1.44 (CI: 1.31-1.59, adjusted p = 5 x 10e-10). Note that a nearby SNP, rs1975197, was also shown to independently confer risk for restless legs syndrome in this same study. Restless Legs Syndrome: Preliminary Research [PharmGKB:Curated GWAS Results: PTPRD (protein tyrosine phosphatase receptor type delta) is associated with restless legs syndrome (Initial Sample Size: 628 cases, 1,644 controls; Replication Sample Size: 1,835 cases, 3,111 controls; Risk Allele: rs4626664-A). This variant is associated with Restless legs syndrome.] [OMIM:RESTLESS LEGS SYNDROME, SUSC... | ...more... (hide) |
| rs2295490(A;A) Magnitude: 1 Frequency: 76.8% Repute:Good References:8 | normal Normal risk of early-onset type II diabetes<br>rs2295490 is a SNP in the tribbles homolog 3 TRIB3 gene, also known as Q84R. The (A) allele encodes the Gln (Q), while the (G) allele encodes the Arg (R). A study involving four different case-control samples comprising a total of 5469 Caucasisans determined that rs2295490(G) was nominally associated with type-2 diabetes (odds ratio 1.17, CI: 1.00-1.36, p = 0.04). When stratified by age of diabetes onset, these carriers had an increased risk of early-onset T2D (OR 1.32, CI: 1.10-1.58, p = 0.002). this gain of function mutation may affect insulin signaling and increase the risk of insulin resistance [PharmGKB:Curated This variant in the TRIB3 gene is associated with early-onset T2D in Whites. This Q84R | ...more... (hide) |

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| | %22gain of function%22 polymorphism, impairs insulin signaling.] |
| rs10488631(T;T)<br>Magnitude: 1<br>Frequency: 77.0%<br>Repute:Good<br>References:29 | Normal risk of developing SLE, primary biliary cirrhosis, and Sjögren's syndrome<br>...more... (hide)<br><br>rs10488631, a SNP located 3' of the IRF5 gene, has been reported as a possibly causative SNP for systemic lupus erythematosus (SLE), based on a study of ~700 Swedish patients. The risk allele is rs10488631(C), with a reported odds ratio of 2.07 (CI: 1.63-2.62, p = 9x10e-10). The C allele is also associated with primary biliary cirrhosis, with each C increasing the odds about 1.6 times. Each C allele increases risk of Sjögren's syndrome by 1.7 times. SLE rs10488631 and rs7582694 [OMIM:INTERFERON REGULATORY FACTOR 5; IRF5] [GWAS:Systemic lupus erythematosus] |
| rs4273077(A;A)<br>Magnitude: 1<br>Frequency: 78.6%<br>Repute:Good<br>References:3 | Normal risk for development of MGUS<br>This was defined as an OR of 1.0 for the development of MGUS (Monoclonal Gammopathy of Uncertain Significance).<br>A is the major allele and not the risk allele.<br>...more... (hide)<br>[GWAS:Hematological and biochemical traits] |
| rs1223271(G;G)<br>Magnitude: 1<br>Frequency: 80.4%<br>Repute:Good<br>References:1 | Normal risk of developing Parkinson's Disease<br><br>[GWAS:Parkinson's disease] |
| rs6532197(A;A)<br>Magnitude: 1<br>Frequency: 84.1%<br>Repute:Good<br>References:1 | Normal risk of developing Parkinson's Disease<br><br>[GWAS:Parkinson's disease] |
| rs4979462(C;C)<br>Magnitude: 1<br>Frequency: 98.5%<br>Repute:Good<br>References:3 | Normal risk of developing primary biliary cirrhosis<br><br>[GWAS:Primary biliary cirrhosis] |
| rs182549(C;T)<br>Magnitude: 1<br>Repute:Good | Can digest milk.<br>...more... (hide) |

References:9

Also known as 'G/A(-22018)' and located in the MCM6 but with influence on the lactase LCT gene, rs182549 is one of two SNPs that is associated with the primary haplotype associated with hypolactasia, more commonly known as lactose intolerance in European Caucasian populations. In these populations, the rs182549(C) allele (as named in accordance with dbSNP) is both the more common allele and the one associated with lactose intolerance. In populations of sub-Saharan Africans, though, the rs182549(C) allele is unlikely to be predictive of lactose intolerance, and other SNPs are predictive instead. * See also OMIM 601806.0002 Gender differences in genetic risk profiles for cardiovascular disease. Natriuretic peptide system gene variants are associated with ventricular dysfunction after coronar...

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| gs158<br>Magnitude: 0<br>Repute: Good | CYP1A2 normal metabolizer<br>CYP1A2 normal metabolizer |

## Bad news

...more... (hide)

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| rs1421085(C;C)<br>Magnitude: 3.1<br>Frequency: 19.5%<br>Repute:Bad<br>References:51 | ~1.7x increased obesity risk...more... (hide)<br><br>rs1421085 is a SNP located in the first intron of the FTO (fat mass and obesity associated) gene on chromosome 16. This SNP showed the most association with obesity in the original work by Dina et al . The rs1421085 T-to- C single-nucleotide alteration underlies the association between FTO and obesity by disrupting ARID5B-mediated repression of IRX3 and IRX5. This disruption leads to a developmental shift from browning to whitening programs and loss of mitochondrial thermogenesis Within the FTO gene, many SNPs appear to be co-inherited. The SNP showing the strongest association with body weight (i.e. body mass index, BMI) is not rs1421085, although this SNP is one of co-inherited SNPs in the FTO gene region. For more information, refer to the FTO gene or the most studied of FTO SNPs, rs993... |
| gs151<br>Magnitude: 3<br>Repute: Bad | CYP2C19 Intermediate Metabolizer<br>CYP2C19 Intermediate Metabolizer. Your body breaks down some medicines at a slightly slower than normal rate (which is represented by gs150). Individuals with gs152 genotypes have even slower metabolism. *anti-epileptics (such as |

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| | diazepam, phenytoin, and phenobarbitone) *anti-depressants (such as amitriptyline and clomipramine) *anti-platelet drug clopidogrel (Plavix) *anti-ulcer proton pump inhibitors like omeprazole (trade names Losec and Prilosec), esomeprazole (trade name Nexium), and lansoprazole (Prevacid) *hormones (estrogen, progesterone). |
| rs1801133(T;T)<br>Magnitude: 3<br>Frequency: 8.8%<br>Repute:Bad<br>References:224 | homozygous for C677T of MTHFR = 10-20% efficiency in processing folic acid = high homocysteine, low B12 and folate levels This is the homozygous form of the C677T allele for the MTHFR gene. It is found in approximately * 15% of Hispanic Americans * 12% of Caucasian Americans * 12% of Japanese * 6% of Germans * 3% of Asians * 2% of African Americans * 1% of Sub-Saharan Africans Numerous health effects are associated with this variant. * Hyperhomocysteinemia and neurologic disorders: a review. (2014). A review. Certain genetic factors also cause elevated homocysteine levels, such as C667T substitution of the gene encoding methylenetetrahydrofolate reductase. Elevated homocysteine has been observed in several medical conditions, such as cardiovascular disorders, atherosclerosis, myocardial i...<br>...more... (hide)<br>rs1801133 is a SNP that is relatively common and has been studied for (relatively) a long time. Also known as C677T, Ala222Val, and A222V, it encodes a variant in the MTHFR gene, which encodes an enzyme involved in folate metabolism. Homozygous rs1801133(T;T) individuals have ~30% of the expected MTHFR enzyme activity, and rs1801133(C;T) heterozygotes have ~65% activity, compared to the most common genotype, rs1801133(C;C). This reduced activity (i.e. this SNP) has been linked at least once to each of the following disorders (though not necessarily reproducibly): * autism * cancer, including ** gastric cancer ** lung cancer ** head and neck cancer ** renal cancer * cleft lip and cleft palate * coronary artery disease * dementia * depression * hyperhomocysteinemia * migraine * neural tube d... |
| rs4244285(A;G)<br>Magnitude: 3<br>Frequency: 19.0%<br>Repute:Bad | poorer metabolizer of several popular medicines; patients prescribed Plavix get less benefit, and have higher risk for adverse cardiovascular events |

References:30

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rs4244285 is a SNP in the CYP2C19 gene, potentially encoding the CYP2C19*2 variant. This variant is the most common reason for poor metabolism of compounds like mephenytoin (an anti-convulsant), some antidepressants, the anti-platelet drug Plavix, and some drugs used for ulcer conditions of various types. The risk allele is rs4244285(A). As a nonfunctioning CYP2C19, this variant would be expected to be a poor metabolizer of several commonly prescribed drugs, including anti-ulcer drugs like omeprazole (trade names Losec and Prilosec), esomeprazole (trade name Nexium), and lansoprazole (trade name Prevacid). In Caucasians, SNPs in CYP2C19 are relatively rare (in contrast to SNPs in CYP2D6), but SNPs in this gene are common in Asians. Ulcer treatment with omeprazole to reduce Helicobacter pyl...

rs1121980(T;T)
Magnitude: 3
Frequency: 21.2%
Repute:Bad
References:28

2.76x risk for obesity...more... (hide)

rs1121980, a SNP in the FTO gene, showed the strongest association of several SNPs in the region with early onset obesity in a study of ~1,000 Caucasians. The odds ratio for the rs1121980(T) risk allele was 1.66 (CI: 1.37â€“2.01); and the odds ratios for heterozygotes and homozygotes (rs1121980(T;T)) were estimated (with a correct p<0.0001) at 1.67 (CI: 1.22â€“2.27) and 2.76 (CI: 1.88â€“4.03), respectively. Note that although rs1121980 showed the strongest association, it is in a block in the FTO gene of at least 5 other SNPs that are in linkage disequilibrium (r<sup>2</sup>>=0.88 for all). The other 5 SNPs are: * rs9939973 * rs7193144 * rs9940128 * rs8050136 * rs9939609 This SNP is also associated with (severe) obesity i...

rs6920220(A;G)
Magnitude: 3
Frequency: 27.7%
Repute:Bad
References:19

1.2x risk Rheumatoid Arthritis...more... (hide)

rs6920220 has been reported in a large study to be associated with rheumatoid arthritis. The risk allele (oriented to the dbSNP entry) is (A); the odds ratio associated with heterozygotes is 1.20 (CI 1.06-1.36), and for homozygotes, 1.72 (CI 1.33-2.22). rheumatoid arthritis *rs6920220 [P= 2.6 x 10(-6), OR 1.22 (1.13-1.33)]. *rs5029937 *rs13207033 protective [P= 0.0001, OR 0.86 (0.8-0.93)] perfectly correlated with rs10499194 The combination of the carriage of both risk alleles of rs6920220 and rs5029937 together with the absence of the protective allele of rs13207033 was strongly associated with RA when compared to carriage of none [OR of 1.86 (95% CI) (1.51-

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| | 2.29)]. This equates to an effect size of 1.50 (95% CI 1.21-1.85) Lack of association or interactions between the IL-4, IL-4Ralpha an... |
| rs1045642(T;T)<br>Magnitude: 3<br>Frequency: 29.2%<br>Repute:Bad<br>References:106 | altered drug metabolism and bioavailability; moderately increased risk for certain cancers This is the homozygous variant of C3435T, on the gene encoding P-glycoprotein (called MDR1 or ABCB1). P-glycoprotein is a protein involved in pumping foreign substances out of cells. It mainly affects areas of the body that have a barrier or excretory function: blood-tissue barrier, the gastrointestinal tract, liver and kidney. Poor functioning of this cellular defense mechanism can affect elimination of carcinogens and movement of drugs in the system. == Drug Treatment Response & Toxicity == '"ABCB1 polymorphism as prognostic factor in breast cancer patients treated with docetaxel and doxorubicin neoadjuvant chemotherapy."' (2014). Among 216 patients, those with the 3435TT genotype had a longer o...<br>...more... (hide)<br>rs1045642, also known as C3435T, is a SNP located in the ABCB1 gene. It is often studied in conjunction with rs2032582. C3435T has been mentioned by: * A 'Silent' Polymorphism in the MDR1 Gene Changes Substrate Specificity (for example, to verapamil) * (R)-lansoprazole (Prevacid) concentrations are significantly increased in CYP2C19 extensive metabolizers with ABCB1 C3435T C allele. * In a Korean population, plasma concentrations of fexofenadine (Allegra) were 17% lower in 2677AA/3435CC subjects and 47% higher in the 2677TT/3435TT subjects compared to 2677GG/3435CC subjects. * In contrast, in this study no association was observed between the C3435T polymorphism and fexofenadine plasma or urine concentrations in a German Caucasian population. * A meta-analysis including 9 case-control stud... |
| rs2981582(C;T)<br>Magnitude: 3<br>Frequency: 41.6%<br>Repute:Bad<br>References:34 | 1.3x higher risk of ER+ breast cancer...more... (hide)<br>rs2981582 in the FGFR2 gene was one of the four strongest associations found in a genome-wide association study (GWAS) of over 4,000 breast cancer samples. The T allele was more strongly related to ER-positive (per-allele odds ratio 1.31 |

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| | (CI: 1.27-1.36)) than ER-negative (odds ratio 1.08 (CI:1.03-1.14)) disease (p for heterogeneity = 10(-13)). While on its own still of fairly small effect, this was the most significant of 7 SNPs to help estimate risk of breast cancer. Family history and/or BRCA1 or BRCA2 testing status are more significant factors, which were not part of this panel. Based on a study of 1,049 Chinese breast cancer patients, carriers of risk alleles at three SNPs (rs2981582, rs1219648 and rs2420946) were at 1.36x increased risk for breast cancer (CI: 1.13-1.62, p = 0.001). A... |
| rs13266634(C;C)<br>Magnitude: 3<br>Frequency: 56.6%<br>Repute:Bad<br>References:118 | increased risk for type-2 diabetes...more... (hide)<br><br>rs13266634 is a SNP in the zinc transporter protein member 8 SLC30A8 gene that has primarily been associated with type-2 diabetes in several studies. This SNP is also known as the Arg325Trp or R325W variant; the (C) allele encodes the arginine (R), and the (T) allele encodes the tryptophan (W). significantly associated p = 0.0073; in 1,630 Japanese subjects with type-2 diabetes and in 1,064 controls The major alleles of the SLC30A8 SNP rs13266634 and the HHEX SNP rs7923837 associate with reduced insulin secretion, but not with insulin resistance. 46% of European non-diabetic offspring of type-2 diabetes patients are rs13266634(C;C) homozygotes; they are diabetes-prone and characterised by a 19% decrease in first-phase insulin release following an intravenous glucose load. * Note: this SNP,... |
| rs2011077(G;G)<br>Magnitude: 3<br>Frequency: 64.6%<br>Repute:Bad<br>References:1 | 6.2x increased risk of developing prostate cancer, a 3x increased risk of developing BPH, and a 5.5x increased risk of developing metastatic prostate cancer In asian populations up to 25% of people have this genotype. In european populations approximate 5% of people have this genotype. In african populations less than 1% of people have this genotype.<br>...more... (hide)<br>A study of ~500 Japanese prostate cancer patients found that individuals with a rs2011077(G;G) genotype had a 6.2- and 3-fold increased risk of prostate cancer and benign prostate hyperplasia (BPH), and a 5.5-fold increased risk of metastatic prostate cancer, compared to the (A;A) genotype. |
| gs155<br>Magnitude: 2.5<br>Repute: Bad | CYP3A5 non-expressor<br>This is common for Caucasians, but rare for African populations. As a CYP3A5 non-expressor you are unable to |

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| | metabolize some common medications. This influences the synthesis of cholesterol, steroids and other lipids. The enzyme metabolizes drugs such as olanzapine, tacrolimus, nifedipine and cyclosporine as well as the steroid hormones testosterone, progesterone and androstenedione. |
| gs281<br>Magnitude: 2.5<br>Repute: Bad | part of the 88% of the population claimed not to maintain weight loss unless you perform high energy exercise An interesting hypothesis, but not well validated, this patent is summarized in a simplified blog post and suggests a genotype to suggest diet and lifestyle changes which may be beneficial. Based on 2 snps *rs4994 *rs1042713 |
| rs7089424(G;G)<br>Magnitude: 2.5<br>Frequency: 8.9%<br>Repute:Bad<br>References:6 | moderately (~4x) increased risk for acute lymphoblastic leukemia see references at rs7089424, including meta-analyses<br>...more... (hide)<br><br>Replication analysis confirms the association of ARID5B with childhood B-cell acute lymphoblastic leukemia. Variation at 7p12.2 and 10q21.2 influences childhood acute lymphoblastic leukemia risk in the Thai population and may contribute to racial differences in leukemia incidence. [OMIM:?] [GWAS:Acute lymphoblastic leukemia (childhood)] |
| rs6441286(G;G)<br>Magnitude: 2.5<br>Frequency: 13.4%<br>Repute:Bad<br>References:4 | 3.08x chance of developing primary biliary cirrhosis (hide)<br>...more...<br><br>23andMe blog *Each G at rs2856683 increased the odds of primary biliary cirrhosis by 1.75x. *Each G at rs6441286 increased the odds of primary biliary cirrhosis by 1.54x. *Each A at rs3790567 increased the odds of primary biliary cirrhosis by 1.51x. [PharmGKB:Curated A GWAS of 2072 Canadian and U.S. subjects (536 patients with primary biliary cirrhosis and 1536 controls) showed that primary biliary cirrhosis was significantly and reproducibly associated with this SNP (rs6441286) at the IL12A locus (P=2.42x10(-14); odds ratio, 1.54).] [OMIM:?] [GWAS:Primary biliary cirrhosis] |
| rs891512(A;G)<br>Magnitude: 2.5<br>Frequency: 33.0%<br>Repute:Bad<br>References:6 | Higher blood pressure than G;G...more... (hide) |

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| | rs891512 (also known as G24943A, IVS25+15 [G→A]) is an intronic variant in the NOS3 gene significantly associated with blood pressure. The intronic variant, IVS25+15 [G-->A], was significantly associated with blood pressure; GG homozygotes had significantly lower levels of diastolic blood pressure (DBP) (-2.8 mm Hg; P = 0.016) and systolic blood pressure (SBP) (-1.9 mm Hg; P = 0.018) than A-allele carriers. eNOS polymorphism rs891512 (G24943A) is associated with hypertension in Chilean individuals (p<0.05). Two specific SNPs in significant LD with each other (rs891512 and rs1808593) were significantly associated with ABI (ankle-brachial index) in both subsets. protective haplotype C-T-A (rs2070744-rs1799983-rs891512) was observed (P = 0.01) with a pronounced effect against suic... | |
| rs10490924(G;T)<br>Magnitude: 2.5<br>Frequency: 38.1%<br>Repute:Bad<br>References:58 | 2.7x risk for age related macular degeneration | ...more...<br>(hide) |
| | rs10490924 was identified as a risk factor from chromosome 10 related to age related macular degeneration. The risk allele is (T). Odds ratios for heterozygotes and homozygotes are, respectively, 2.69 (CI: 2.22-3.27) and 8.21 (CI: 5.79-11.65). Disease risk in combination with the rs1061170 SNP in the CFH gene is dramatically increased. Homozygotes for both the rare/risk alleles at both loci are estimated to be at 57 fold higher risk for age related macular degeneration than individuals homozygous for the common alleles at both loci. A subsequent study indicated that the risk based on solely the ARMS2 SNP rs10490924 is significantly higher in smokers than in non-smokers. Based on a study of ~750 patients, rs10490924 is significantly higher in participants with choroidal neovascularization t... | |
| rs2180439(C;T)<br>Magnitude: 2.5<br>Frequency: 39.8%<br>Repute:Bad<br>References:5 | Increased risk of Male Pattern Baldness. One copy of the risk allele, and one copy of the normal allele. Verified in Europeans and Asians. Family Tree DNA, which tends to skew heterozygous results like this in the most positive light, reports this as less likely to go bald, but 23andMe doesn't mention it.<br>...more... (hide) | |
| | spitoon rs2180439(T;T) have 2x increased odds of male pattern baldness. [PharmGKB:Non-Curated Susceptibility variants for male-pattern baldness on chromosome 20p11. (Initial Sample Size: 296 cases, 347 controls; Replication Sample Size: 319 cases, 234 controls); (Region: 20p11.22; Reported Gene: PAX1, BQ013595, BE789145; Risk Allele: rs2180439-C) This variant is associated with Male-pattern | |

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| | baldness.] [OMIM:ALOPECIA, ANDROGENETIC, 3; AGA3] [GWAS:Male-pattern baldness] |
| rs339331(T;T)<br>Magnitude: 2.5<br>Frequency: 41.6%<br>Repute:Bad<br>References:8 | Prostate cancer risk...more... (hide)<br><br>[GWAS:None] |
| rs2943634(C;C)<br>Magnitude: 2.5<br>Frequency: 44.2%<br>Repute:Bad<br>References:12 | higher risk of ischemic stroke...more... (hide)<br><br>rs2943634 is a SNP found to be reproducibly associated with heart disease rs2943634 associated with high density lipoprotein (HDL) cholesterol Association studies of 22 candidate SNPs with late-onset Alzheimer's disease. Evaluation of the association of genetic variants on the chromosomal loci 9p21.3, 6q25.1, and 2q36.3 with angiographically characterized coronary artery disease. Genome-wide association of early-onset myocardial infarction with single nucleotide polymorphisms and copy number variants. Genome-wide association studies for atherosclerotic vascular disease and its risk factors. Genetics of coronary artery disease: focus on genome-wide association studies. Evaluation of population impact of candidate polymorphisms for coronary heart disease in the Framingham Heart Study Offspri... |
| rs3738919(C;C)<br>Magnitude: 2.5<br>Frequency: 46.0%<br>Repute:Bad<br>References:1 | 1.94x risk of developing rheumatoid arthritis ...more... (hide)<br><br>rs3738919, a SNP located in the ITGAV gene, was identified in a European study to be associated with rheumatoid arthritis (RA). The risk allele for rs3738919 is the more common allele, (C). For the three European Caucasian populations studied (372 RA patients + 330 controls), and combining the (C;C) and (A;C) genotypes in comparison to the (A;A) genotype, the odds ratio for RA = 1.94 (CI: 1.3â€“2.9, p = 0.002). There was no significant difference in RA risk between those carrying one or two (C) alleles. An editorial about this finding has been published. |
| rs776746(G;G)<br>Magnitude: 2.5<br>Frequency: 92.8%<br>Repute:Bad | CYP3A5*3 homozygote; CYP3A5 non-expressor Impacts metabolism of tacrolimus, an immunosuppressive drug used in organ transplantation. This is the |

References:35

most common phenotype in caucasian and many other populations. Only in africans is expressing the gene common.
...more... (hide)

rs776746, also known as 6986A>G, is a SNP encoding the (nonfunctional) CYP3A5*3 allele of the CYP3A5 gene. CYP3A5*3 has been studied especially in connection with the metabolism of tacrolimus, an immunosuppressive drug used in organ transplantation. The optimal therapeutic dose is in a relatively narrow window, so blood concentration monitoring is recommended. CYP3A5*3 carriers tend to build up higher levels of tacrolimus than CYP3A5*1 carriers, unless their tacrolimus intake is reduced. Risk of testicular germ cell cancer in relation to variation in maternal and offspring cytochrome p450 genes involved in catechol estrogen metabolism. Polymorphisms in the cytochrome P450 genes CYP1A2, CYP1B1, CYP3A4, CYP3A5, CYP11A1, CYP17A1, CYP19A1 and colorectal cancer risk. Prevalence of common dis...

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rs3825942(C;C)
Magnitude: 2.3
Frequency: 68.3%
Repute:Bad
References:46

common but 10x higher glaucoma risk in most (but not all) populations This genotype is found in 2 out of 3 people, but has been repeatedly linked to a much higher risk of glaucoma. However a 2010 paper in a south african population reports reduced risk, casting some doubts.
...more... (hide)

rs3825942, also known as G153D, a SNP causing an amino acid change in the lysyl oxidase 1 LOXL1 gene, has been linked to exfoliation glaucoma (also known as exfoliation syndrome). This form of glaucoma causes up to 10% of the cases of blindness in many countries, including the US. From the abstract of this study: 'Approximately 25% of the general population is homozygous for the highest risk haplotype [C;C here combined with other risk SNPs] and their risk of suffering XFG (exfoliation glaucoma) is over 100 times that of those only carrying low-risk haplotypes.' The risk allele for this SNP is rs3825942(C), as oriented with respect to the dbSNP entry, and it confers a estimated relative risk (by itself) of 27 compared to the (T) allele. The odds ratio is 20.10 (CI 10.80-37.41). [Note that ...

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rs2165241(C;T)
Magnitude: 2.2
Frequency: 43.4%
Repute:Bad
References:37

4.4x increased risk of exfoliation glaucoma glaucoma risk likely to actually be based on nearby rs3825942
...more... (hide)

This SNP, located in an intron of the LOXL1 gene, was initially reported to be associated with exfoliation glaucoma. However, it was shown in the same study to no longer be significant once two other SNPs, which cause actual changes in the LOXL1

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| | protein, were identified. More specifically, the risk allele rs2165241(T) was found to be associated with glaucoma only because it effectively predicted (with 90% probability) the actual high-risk haplotype consisting of rs1048661(G) and rs3825942(C), as oriented with respect to their entries in dbSNP. However, a meta-analysis published in 2010 concluded that it's likely that SNPs rs1048661 and rs2165241 are not directly implicated in the pathogenesis of glaucoma; only the nearby rs3825942 seemed to be the disease-associated SNP. rs2165241 was sign... |
| rs2231137(G;G)<br>Magnitude: 2.2<br>Frequency: 96.9%<br>Repute:Bad<br>References:6 | ~1.5-3x increased risk for ischemic stroke...more... (hide)<br><br>rs2231137, also known as Val12Met, is a SNP in the ATP-binding cassette, sub-family G (WHITE), member 2 ABCG2 gene. The (G) allele encodes the Val. In a study of incident ischemic stroke during 14 years of follow-up in a population-based study of older adults known as the Cardiovascular Health Study (CHS), rs2231137 was associated with stroke in both white (hazard ratio, 1.46, CI: 1.05 - 2.03) and black (hazard ratio, 3.59, CI, 1.11 - 11.6) participants. The risk of ischemic stroke was higher in Val allele homozygotes than in Met allele carriers. The adjusted hazard ratio for Val allele homozygotes, compared with Met allele carriers, was 1.50 (90% CI, 1.06 to 2.12) in whites and 3.62 (90% CI, 1.11 to 11.9) in black participants (Table 4). [PharmGKB:Curated control size=7; M12 cases=6; PK=... |
| rs1050152(T;T)<br>Magnitude: 2.1<br>Frequency: 11.5%<br>Repute:Bad<br>References:24 | 2.1x increased risk of Crohn's disease...more... (hide)<br><br>rs1050152, a SNP in the SLC22A4 gene known as L503F, has been associated with an autoimmune disease, in this case, Crohn's disease, odds ratio = 2.1 (CI = 1.31â€“3.39, p = 0.002), based on a study of 203 cases and 200 controls. The risk allele is rs1050152(T). A nearby SNP (rs2631367) in the promoter region of the SLC22A5 gene defines a haplotype along with rs3792876, with odds ratio reported as similar for either SNP or the haplotype. Referring to the TC risk haplotype, the population risk attributable to heterozygotes was 19%, and for homozygous haplotype carriers, 27%. [PharmGKB:Non-Curated Higher gabapentin exposure and lower renal clearance/tubular secretion in individuals carrying this variant] [OMIM:?] |
| rs2294008(T;T) | increased risk of gastric and bladder ...more... |

Magnitude: 2.1
Frequency: 16.8%
Repute:Bad
References:42

cancer (hide)

rs2976392 and diffuse-type gastric cancer (allele-specific odds ratio (OR) = 1.62, 95% CI = 1.38-1.89, P = 1.11 x 10(-9)). Substitution of the C allele with the risk allele T at a SNP in the first exon (rs2294008, which has r(2) = 0.995, D' = 0.999 with rs2976392) reduces transcriptional activity of an upstream fragment of the gene [OMIM:PROSTATE STEM CELL ANTIGEN; PSCA] [GWAS:Bladder cancer]

rs2301436(A;A)
Magnitude: 2.1
Frequency: 25.7%
Repute:Bad
References:11

...more... (hide)

DeCode reports that rs2301436 is associated with susceptibility to Crohn's disease. [PharmGKB:Non-Curated GWAS Results: Genome-wide assocation defines more than 30 distinct susceptibility loci for Crohn's disease (Initial Sample Size: 3,230 cases 4,829 controls; Replication Sample Size: 2,325 cases 1,809 controls 1,339 affected trios; Risk Allele: rs2301436-T).] [GWAS:Crohn's disease] [GWAS:Crohn's disease]

rs6457617(T;T)
Magnitude: 2.1
Frequency: 26.5%
Repute:Bad
References:21

5.2x risk of rheumatoid arthritis...more... (hide)

rs6457617 has been reported in a large study to be associated with rheumatoid arthritis. This SNP is reported to be the most statistically significant of many SNPs similarly located in the MHC region. The risk allele (oriented to the dbSNP entry) is (T); the odds ratio associated with heterozygotes is 2.36 (CI 1.97-2.84), and for homozygotes, 5.21 (CI 4.31-6.30). [GWAS:Rheumatoid arthritis]

rs4149056(C;T)
Magnitude: 2.1
Frequency: 28.3%
Repute:Bad
References:62

reduced breakdown of some drugs; 5x increased myopathy risk for statin users
...more... (hide)

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

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| | and natural compounds. The rs4149056(C) SNP defines the SLCO1B1*5 allele. The rs4149056(C) allele gives rise to an amino acid change (from valine to alanine at residue 174) which has reduced uptake/transport activity. Therefore, drugs metabolized by OATP1B1 tend to build up to higher circulating concentrations than they would otherwise.. The drugs known (or in some cases, thought) to be transported less well ... |
| rs4363657(C;T)<br>Magnitude: 2.1<br>Frequency: 31.0%<br>Repute:Bad<br>References:10 | 4.5x increased myopathy risk for statin users ...more... (hide)<br><br>rs4363657 is a SNP in the SLCO1B1 gene, a gene which encodes a protein involved in the liver's uptake of certain drugs, including the statins used to lower cholesterol levels. rs4363657 in nearly complete linkage disequilibrium with rs4149056 SNP (r<sup>2</sup>=0.97), which has been linked to statin metabolism. rs4149056(C) odds ratio for myopathy among 20,000 individuals taking either 40 or 80mg of simvastatin daily was 4.5 (CI: 2.6-7.7) per copy of the C allele, and 16.9 (CI: 4.7-61.1) in (C;C) as compared with (T;T) homozygotes. See also rs4149056 for a more detailed description of the effect of SLCO1B1 gene SNPs on the metabolism of many drugs. SLCO1B1 genetic variant associated with statin-induced myopathy: a proof-of-concept study using the clinical practice research data... |
| rs944289(C;T)<br>Magnitude: 2.1<br>Frequency: 42.5%<br>Repute:Bad<br>References:16 | 1.3x increased thyroid cancer risk...more... (hide)<br><br>Each A at rs965513 increased the odds of thyroid cancer by 1.75 times. Each T at rs944289 increased the odds of thyroid cancer by 1.37 times. A genoset, Gs137, has also been created in SNPedia to represent the increased risk reported for carriers of both variants. See also: 23andMe blog [PharmGKB:Non-Curated GWAS results: Common variants on 9q22.33 and 14q13.3 predispose to thyroid cancer in European populations. (Initial Sample Size: 192 cases, 37,196 controls; Replication Sample Size: 432 cases, 1,727 controls); (Region: 14q13.3; Reported Gene(s): NKX2-1; Risk Allele: rs944289-T); (p-value= 0.000000002).This variant is associated with Thyroid cancer.] [OMIM:THYROID CARCINOMA, PAPILLARY] |
| rs560887(G;G)<br>Magnitude: 2.1<br>Frequency: 44.6%<br>Repute:Bad<br>References:39 | Avg. fasting Plasma Glucose 5.18 mmol/L (93 mg/dl). Slightly higher blood glucose. Fasting Plasma Glucose is the amount of sugar in your blood when you wake up. It's required to power your body, but too much |

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| | is very bad for you. Fasting plasma glucose between 5.6 mmol/L and 6.9 mmol/L (101 – 125 mg/dl) is considered pre-diabetes. Fasting plasma glucose higher than 7 mmol/L (126 mg/dl) is considered diabetes.<br>...more... (hide)<br>rs560887, p = 4 x 10(-7)) *654 normoglycemic subjects, replicated in 9,353 subjects *fasting plasma glucose (FPG) (beta = -0.06 mmol/l per A-allele, combined p = 4 x 10(-23)) and with pancreatic beta-cell function (Homa-B model, combined p = 3 x 10(-13)) in three populations; however it was not associated with type 2 diabetes risk. (ageing) Blood Glucose [PharmGKB:Non-Curated Phenotype 1: In a meta-analysis of 21 GWAS cohorts followed by analysis in additional individuals, this SNP was found to be associated with fasting glucose level. Study size: 119,169. Significance metric(s): p = 8.7 x 10(-218). Phenotype 2: In the same study, this SNP was found to be associated with HOMA-B (homeostasis model assessment of beta-cell function). Study size: 94,839. Significance metric(s): p = 1.5 x 10(-... |
| rs3761847(A;G)<br>Magnitude: 2.1<br>Frequency: 54.9%<br>Repute:Bad<br>References:40 | 1.32 x risk of rheumatoid arthritis. A common genetic variant at the TRAF1-C5 locus on chromosome 9 is associated with an increased risk of anti-CCP-positive rheumatoid arthritis. rs3761847, a SNP located between two genes associated with chronic inflammation (TRAF1 and C5), is associated with increased risk of anti CCP-positive rheumatoid arthritis. The risk allele is (G); the odds ratio reported is 1.32 (95% confidence interval, 1.23 to 1.42; P=4x10-14) .<br>...more... (hide)<br>rs3761847, a SNP located between two genes associated with chronic inflammation (TRAF1 and C5), is associated with increased risk of anti CCP-positive rheumatoid arthritis. The risk allele is (G); the odds ratio reported is 1.32 (95% confidence interval, 1.23 to 1.42; P=4x10-14) . rs3761847(G;G) associated with an increased risk of death (hazard ratio 3.96 [95% confidence interval 1.24-12.6], P = 0.020) as compared with (A;A) homozygote status. The excess mortality was attributed to deaths due to malignancies and sepsis but not cardiovascular disease (CVD). [PharmGKB:Non-Curated GWAS Results: TRAF1-C5 as a risk locus for rheumatoid arthritis--a genomewide study (Initial Sample Size: 1,493 cases, 1,831 controls; Replication Sample Size: 1,053 cases, 1,858 controls; Risk Allele: rs3761847-G... |
| rs646776(A;A) | 1.2x risk of coronary artery disease...more... (hide) |

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| Magnitude: 2.1<br>Frequency: 54.9%<br>Repute:Bad<br>References:38 | rs599839 and rs4970834 explain about 1% of the variation in circulating LDL-cholesterol levels. 'When we look at this particular genetic variance, of all the cholesterol variation among the population, 1% of it can be attributed to this particular locus,' said Sandhu. 'This is equivalent to more established genes for LDL regulation, particularly APOE.' rs646776 also linked. Pharmacogenetic meta-analysis of genome-wide association studies of LDL cholesterol response to statins found each minor allele to increase statin effectiveness by 1.3% (rs646776, β=−0.013, s.e.=0.002, P=1.05 × 10−9 and rs12740374, β=−0.013, s.e.=0.002, P=1.05 × 10−9). 23andMe blog coronary artery disease and heart attack SNP Risk Version Effect *rs646776 T 1.19 *rs1746... |
| rs5443(C;T)<br>Magnitude: 2.1<br>Frequency: 55.6%<br>Repute:Bad<br>References:40 | some risk linked to a number of metabolic conditions including obesity, coronary artery disease, insulin resistance and therefore diabetes, left ventricular hypertrophy, and hypertension. It has also been linked to how well a patient responds to Viagra (sildenafil).<br>...more... (hide)<br>rs5443, a SNP in the G-protein beta3 subunit (GNB3) gene that is more commonly known as the C825T variant, has been linked to a number of metabolic conditions including obesity, coronary artery disease, insulin resistance and therefore diabetes, left ventricular hypertrophy, and hypertension. It has also been linked to how well a patient responds to Viagra (sildenafil). Several studies have been unable to replicate one or more of the associations in at least some populations between this SNP and these conditions. The more notable studies include: *rs5443(T) allele carriers are 2-3 fold more likely to be obese in Caucasian, Chinese, and African American populations. *rs5443(T) carriers are clearly at higher risk for hypertension, but this review indicates that whether they are also at incre... |
| rs5186(A;C)<br>Magnitude: 2.1<br>Repute:Bad<br>References:65 | ~1.4x increased risk of hypertension...more... (hide)<br>rs5186, a SNP known as +1166A/C or A1166C, is located in the 3' untranslated region of the angiotensin II receptor type 1 gene AGTR1, which is also known as AT2R1 or AT1R. It is among the most studied of over 50 SNPs in AGTR1. The rs5186(C) allele is associated with increased risk for essential hypertension in Caucasian populations with an odds ratio of |

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| | 7.3 (homozygote (C;C) compared to (A;C) and (A;A), CI: 1.9-31.9,p=0.0015). There are likely to be ethnic differences in risk; while the rs5186(C) allele was associated with hypertension in a Chinese population , it was not been observed as a risk in a Japanese population. Age and gender may also influence risk, as discussed in a review of AGTR1 SNPs and their role in hypertension and related disorders. Pregnant women who are rs5186(C) allel... |
| gs221<br>Magnitude: 2<br>Repute: Bad | Autoimmune disorder risk in Europeans<br>This genoset tags the DQ2.2 haplotype in Europeans, and thus increased risk gluten intolerance and for autoimmune disorders such as celiac disease. |
| gs279<br>Magnitude: 2<br>Repute: Bad | Mild trimethylaminuria<br>Carrying two copies of a somewhat common allele of the FMO3 gene, defined as E308G and E258K, has been reported to lead to mild trimethylaminuria. |
| rs6449213(C;C)<br>Magnitude: 2<br>Frequency: 1.8%<br>Repute:Bad<br>References:17 | ~4x higher risk for hyperuracemia...more... (hide)<br><br>This is basically a surrogate for rs7442295, and thus associated with higher serum urate levels. [OMIM:SOLUTE CARRIER FAMILY 2 (FACILITATED GLUCOSE TRANSPORTER), MEMBER] [GWAS:Uric acid levels] |
| rs4673(T;T)<br>Magnitude: 2<br>Frequency: 9.7%<br>Repute:Bad<br>References:11 | 3.9x increased risk for cerebral ischemia...more... (hide)<br><br>rs4673, also known as C242T or H72Y, is a SNP in the NAD(P)H oxidase p22(phox) subunit CYBA gene. A study of German patients with ischemic stroke or transient ischemic attack under the age of 50 (n = 161) concluded that rs4673(T;T) individuals were almost 4 times more likely to experience cerebral ischemia (odds ratio 3.85, CI: 1.39-10.64) compared to rs4673(C;T) or (C;C) individuals after adjusting for classical risk factors. A meta-analysis concluded that the T allele effect may vary between ethnicities in both direction of effect and significance; Asian populations may have reduced coronary artery disease risk associated with the T allele. Genetic polymorphisms and susceptibility to lung disease. Genetic association of glutathione peroxidase-1 with coronary artery calcification in type ... |

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| rs7250872(T;T)<br>Magnitude: 2<br>Frequency: 9.8%<br>Repute:Bad<br>References:1 | Increased risk of developing bipolar disorder<br><br>[GWAS:Bipolar disorder] | ...more...<br>(hide) |
| rs2156921(G;G)<br>Magnitude: 2<br>Frequency: 13.6%<br>Repute:Bad<br>References:1 | 1.29x increased risk for depression...more...<br><br>rs2156921, a SNP in the BCR gene on chromosome 22, has been associated with increased risk for depression. The odds ratio for carriers of the minor allele (G) are reported as 1.29 (p=0.031) based on a study of 329 Japanese patients. | (hide) |
| rs909525(A;G)<br>Magnitude: 2<br>Frequency: 20.4%<br>Repute:Bad<br>References:5 | Probably one Warrior Gene and one non-Warrior Gene. Women with this combination usually have the 3 repeat MAOA Warrior Gene on one X chromosome and the 4 or 5 repeat MAOA non-Warrior Gene on the other X chromosome. The 3 repeat Warrior Gene makes people more aggressive and antisocial. If you are a man, there was a problem reading this SNP (or you are XXY).<br>...more... (hide)<br>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 | |
| rs1800896(A;A)<br>Magnitude: 2<br>Frequency: 21.2%<br>Repute:Bad<br>References:121 | 1.8x increased prostate cancer risk...more...<br><br>This SNP is upstream of the IL10 gene, and is also known as the -1082G>A IL10 SNP. Dust mite exposure modifies the effect of functional IL10 polymorphisms on allergy and asthma exacerbations. Dust mite exposure significantly modified the relation between 3 SNPs in IL10 (rs1800896, rs3024492, and rs3024496). Homozygosity for the minor allele of each of the 3 SNPs was associated with increased risk of occurrence ( | (hide) |

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| | approximately 3-fold to 39-fold increase). A study of 258 prostate cancer cases concluded that the rs1800896(A) allele, known to result in lower levels of this anti-inflammatory cytokine, was positively associated with risk (AG vs. GG, odds ratio of 1.69, CI: 1.10-2.60; AA vs. GG, OR of 1.81, CI: 1.11-2.96). |
| rs1361600(G;G)<br>Magnitude: 2<br>Frequency: 22.1%<br>Repute:Bad<br>References:4 | ~2x increased risk for adult-onset asthma in Japanese populations<br>...more... (hide)<br><br>rs1361600, also known as -603A>G, is a SNP in the promoter region of the coagulation factor III (aka thromboplastin or tissue factor) F3 gene. Two unrelated Japanese populations both showed an association between rs1361600(G;G) and adult-onset asthma, with an odds ratio of ~2 (p=.02 or .06). |
| rs9303277(T;T)<br>Magnitude: 2<br>Frequency: 23.0%<br>Repute:Bad<br>References:12 | Increased risk of developing primary biliary cirrhosis (hide)<br>...more...<br><br>[OMIM:?] [GWAS:Primary biliary cirrhosis] |
| rs4968451(A;C)<br>Magnitude: 2<br>Frequency: 26.5%<br>Repute:Bad<br>References:3 | 1.61x increased risk for meningioma...more... (hide)<br><br>rs4968451, which is in a breast cancer susceptibility gene, was associated with increased risk of meningioma, a common form of brain tumor, in a combination of five studies totaling 631 European patients. 'Meningioma risks associated with hetero- and homozygosity for the minor allele of were 1.61 (95% CI = 1.26 to 2.06) and 2.33 (95% CI = 1.25 to 4.34), respectively, and were thus compatible with a multiplicative model of action.' The risk allele and minor allele is rs4968451(C). Around 30% of Europeans carry one of risk genotypes, i.e. rs4968451(A;C) or (C;C); and perhaps 16% of meningiomas might be associated with this SNP. |
| rs663048(G;T)<br>Magnitude: 2<br>Frequency: 28.3%<br>Repute:Bad<br>References:1 | Significantly increased risk of developing lung cancer (hide)<br>...more...<br><br>rs663048 associated with lung cancer The homozygotes for the variant allele had more than a 3-fold risk compared with the wild-type homozygotes [combined odds ratio (OR), 3.32; 95% confidence interval (95% CI), 1.81-7.21]. Heterozygotes also |

had a significantly elevated risk of lung cancer from the combined replication studies with an OR of 1.15 (95% CI, 1.04-1.59). The effect remained significant after adjusting for age, gender, and pack-years of tobacco smoke.007;67(17):8406-11].

rs1801394(G;G)
Magnitude: 2
Frequency: 28.6%
Repute:Bad
References:16

1.4x higher risk for meningiomas This common variant (HapMap allele frequency of 31.3%) in a protein involved in folate (B9) and cobalamin (B12) metabolism and is often reported as 'MTRR I22M' (for the Ile to Met substitution at amino acid 22). Mothers homozygous for this variant were found in a few studies to have an increased risk for Down syndrome babies (risk of 0.4%, average risk in population is 0.25%), but other studies (including in Italian, Irish, French, and Indian-Gujarati women) found no increased risk. Notably, age plays a far larger role in the rate of Down syndrome (risk is 4.5% for a mother 45-years-of-age), and, it is unknown how this variant may combine with the effect of age. There are conflicting reports associating this variant with incidence of neural tube defects, p...
...more... (hide)

rs1801394, also known as A66G or Ile22Met, is a SNP in the methionine synthase MTRR gene. This gene encodes one of the two enzymes involved in the production of methionine (the other is MTR). The protein encoded by an rs1801394 allele has a lower affinity for MTR () and is inconsistently associated with homocysteine level, although it is a risk factor for neural tube defects () and Down syndrome () in conditions of higher homocysteine. Based on a study of British patients with primary brain tumors, (1,005 glioma cases and 631 meningioma cases), rs1801394(G;G) individuals were at higher risk for meningioma (odds ratio 1.41, CI: 1.02-1.94). In general, genotypes associated with increased 5,10-methylenetetrahydrofolate levels are associated with elevated risk for these types of brain cancer. ...

rs520354(A;A)
Magnitude: 2
Frequency: 30.1%
Repute:Bad
References:2

increased risk in men for biliary conditions...more... (hide)

rs520354 is a SNP in an intron of the apolipoprotein B APOB gene; it is also known as the IVS6+360C>T variant. In a study of ~1,000 Chinese patients with various biliary tract conditions, including cancer and gallstones, men carrying a rs520354(A)

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| | allele had a 2x risk of bile duct cancer (CI: 1.2-3.4). |
| rs4027132(A;A)<br>Magnitude: 2<br>Frequency: 31.0%<br>Repute:Bad<br>References:2 | 1.51x increased risk of developing bipolar disorder (hide)<br>...more...<br><br>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). [GWAS:None] |
| rs27388(A;A)<br>Magnitude: 2<br>Frequency: 32.1%<br>Repute:Bad<br>References:1 | Increased risk of developing schizophrenia ...more... (hide)<br><br>rs27388 is a SNP in the MEGF10 gene. In a case-control study involving 652 Irish patients, rs27388(A) allele carriers were at increased risk for schizophrenia. (Note that the (A) allele is quite common in many populations.) No association between schizophrenia and rs27388 of the MEGF10 gene in Chinese case-control sample. |
| rs2305795(A;A)<br>Magnitude: 2<br>Frequency: 32.7%<br>Repute:Bad<br>References:2 | 1.3x higher risk of narcolepsy compared to (A;G) genotype<br>...more... (hide)<br><br>Common variants in P2RY11 are associated with narcolepsy. Found that rs2305795 G allele appears to lower the risk of narcolepsy. This SNP is also in the intronal area of the EIF3G gene. |
| rs2707466(G;G)<br>Magnitude: 2<br>Frequency: 34.5%<br>Repute:Bad<br>References:2 | weaker bones...more... (hide)<br><br>Osteoporosis |
| rs6603272(G;T)<br>Magnitude: 2<br>Frequency: 35.9%<br>Repute:Bad<br>References:3 | 2.74x increased risk of developing schizophrenia ...more... (hide)<br><br>This snp is in the Pseudoautosomal region. rs6603272, part of a haplotype block spanning introns 4, 5 and 6 of the IL3RA gene, has been reported in a whole genome association study to be associated with schizophrenia. (The other SNPs in this |

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| | block are rs6422441 and rs17883192.) The risk allele (oriented to the dbSNP entry) is (G); the odds ratio associated with this allele is 2.74. |
| rs1136287(C;T)<br>Magnitude: 2<br>Frequency: 36.3%<br>Repute:Bad<br>References:10 | 1.5x increased risk of wet ARMD...more... (hide)<br><br>rs1136287, also known as Met72Thr, is a SNP in the PEDF gene. Analysis of 86 Taiwanese Chinese patients with wet age related macular degeneration (ARMD) found the (T) allele to be more frequent in patients than in controls (50% vs 31%; p =.0005). The rs1136287(T;T) genotype was also more prevalent in patients than in controls (odds ratio 3.9, p = .0015). |
| rs1495741(A;G)<br>Magnitude: 2<br>Frequency: 36.3%<br>Repute:Bad<br>References:14 | NAT2 intermediate metabolizer (predicted)<br>see discussion at rs1495741<br>...more... (hide)<br><br>With 99% sensitivity and 95% specificity, at least in Caucasian populations, rs1495741 is reported to classify the NAT2 phenotype of an individual; it therefore is a candidate to replace the 7-SNP panel used in genosets such as gs138, gs139, and gs140. The rs1495741(A;A), (A;G) and (G;G) genotypes predicted slow, intermediate, and rapid NAT2 acetylation phenotypes, respectively. [GWAS:None] |
| rs3212227(A;C)<br>Magnitude: 2<br>Frequency: 36.3%<br>Repute:Bad<br>References:71 | Significantly increased risk of developing cervical cancer (hide)<br>...more...<br><br>IL12B gene SNP, part of a haplotype with rs6887695 associated with psoriasis. A study of 500+ patients with psoriatic arthritis (PsA) concluded that the odds ratio for rs3212227(A;A) carriers was 1.43 (CI: 1.17-1.76). The effect sizes observed in patients with PsA appear to be smaller than those previously reported in patients with psoriasis, suggesting that both loci are primarily associated with psoriasis susceptibility rather than primarily with psoriatic arthritis. rs568408 GA/AA and rs3212227 AC/CC variant genotypes were associated with a significantly increased risk of cervical cancer [adjusted odds ratio, 1.43; 95% confidence interval (CI), 1.06-1.93; and adjusted odds ratio, 1.30; 95% CI, 0.97-1.75, respectively]. subjects carrying variant genotypes of both loci had a 1.82-fold (95... |
| rs6997709(G;T)<br>Magnitude: 2 | 1.2x higher risk for hypertension...more... (hide) |

Frequency: 37.2%
Repute:Bad
References:2

rs6997709 has been reported in a large study to be associated with high blood pressure. The risk allele (oriented to the dbSNP entry) is (G); the odds ratio associated with heterozygotes is 1.20 (CI 0.94-1.52), and for homozygotes, 1.49 (CI 1.18-1.89). In a study of 7,551 Koreans, this SNP was associated with systolic and diastolic blood pressures in a quantitative trait association test but no associations were found using a case-control association test.

rs31489(C;C)
Magnitude: 2
Frequency: 37.5%
Repute:Bad
References:8

Increased risk for lung cancer and emphysema The rs31489 C-allele correlated with reduced forced expiratory volume in 1 s (p=0.006). Homozygous carriers of the rs31489 C-allele exhibited increased susceptibility to bronchial obstruction (OR 1.82, 95% CI 1.24–2.69; p=0.002). A similar association was observed for diffusing capacity of the lung for carbon monoxide (p=0.004). Consistent with this, CC-carriers had an increased risk of emphysema (OR 2.04, 95% CI 1.41–2.94; p=1.73×10−4) and displayed greater alveolar destruction. Finally, CC-carriers also had an increased risk for lung cancer (OR 1.90, 95% CI 1.21–2.99; p=0.005), and were more susceptible to developing both lung cancer and bronchial obstruction than lung cancer alone (OR 2.11, 95% CI 1.04–4.26...
...more... (hide)
[GWAS:Lung adenocarcinoma]

rs2201841(C;T)
Magnitude: 2
Frequency: 38.1%
Repute:Bad
References:37

1.5x increased risk for Crohn's disease; 2x increased risk for Graves' disease
...more... (hide)

SNP rs2201841, in the IL23R gene, is associated with increased risk for Crohn's disease in both Jewish and non-Jewish populations. Another study found that the 'A allele' and 'AA genotype' were significantly overrepresented in Graves' disease patients with Graves ophthalmopathy, based on 216 North American patients. The odds ratios reported were 2.04 for the allele (p=1x10<sup>-4</sup>), and for the so-called 'AA' genotype (presumably rs2201841(T;T) when correctly oriented to the dbSNP orientation), 2.4 (p=1x10<sup>-4</sup>). 23andMe blog psoriasis Europeans *rs2201841(G) 1.13x risk

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| | [PharmGKB:Non-Curated GWAS results: Genome-wide scan reveals association of psoriasis with IL-23 and NF-kappaB pathways. (Initial Sample Size: 1,359 cases, 1,400 controls; Replication S... |
| rs10889677(A;C)<br>Magnitude: 2<br>Frequency: 38.9%<br>Repute:Bad<br>References:42 | 1.5x increased risk for certain autoimmune diseases; 2x increased risk for Graves disease<br>...more... (hide)<br><br>SNP rs10889677, in the IL23R gene, is associated with increased risk for Crohn's disease in both Jewish and non-Jewish populations. The same risk allele for this SNP has been associated with increased risk for ankylosing spondylitis in a large study of over 1,000 Caucasian patients. The odds ratio is 1.3 (p=1.3x10e-6). In a study of 216 North American patients with Graves' disease, the C allele of rs10889677 was 2.03x overrepresented (p=1.3x10<sup>â€“4</sup>), and the homozygous rs10889677(C;C) genotype was also overrepresented (2.36x; p=1.4x10<sup>-4</sup>) in Graves ophthalmopathy patients. A genome-wide association study using DNA samples from 1,052 individuals with ulcerative colitis and preexisting data from 2,571 controls, all of European ance... |
| rs3775291(G;G)<br>Magnitude: 2<br>Frequency: 38.9%<br>Repute:Bad<br>References:21 | common but higher risk of age related macular degeneration<br>...more... (hide)<br><br>rs3775291 is a SNP in the TLR3 gene associated with an amino acid change in the corresponding protein. In the orientation as shown in dbSNP, the more common rs3775291(G) allele encodes a leucine while the rarer rs3775291(A) allele encodes a phenylalanine. rs3775291 has been associated with one form of age related macular degeneration (ARMD), specifically, the form known as 'dry' ARMD, also known as geographic atrophy. A study of 3 case-control groups of Americans of European descent, comprising about 900 ARMD patients in total, indicated that having one rs3775291(A) allele reduces the odds of having dry ARMD about 30%, and being a rs3775291(A;A) homozygote cuts your odds by more than half. Specifically, the odds ratio of having dry ARMD with one minor allele is 0.712 (CI: 0.50-1.0), and fo... |
| rs1050631(C;T)<br>Magnitude: 2<br>Frequency: 38.9%<br>Repute:Bad<br>References:1 | Mean Survival Time of 25 months for esophageal squamous-cell carcinoma<br>...more... (hide) |

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| | rs1050631 is a SNP on chromosome 18q12 in exon 6 of SLC39A6 gene, associated with length of survival in esophageal squamous-cell carcinoma(ESCC). The SNP is located in the 5' UTR of SLC39A6. In a study, a genome-wide scan of SNPs to identify variants associated with length of survival in 1331 individuals with ESCC was performed, and was validated by two independent sets, including 1962 individuals with ESCC (1308 and 654 individuals in two different replications). The hazard ratio for death (odds ratio) (T; T) and (C; T) from ESCC in the 1331 test cases was 1.34 (95% confidence interval (CI) = 1.17−1.53; P = 7.78 E-6). For the combined data, hazard ratio of death similarly was found to be 1.34 (95% CI = 1.17−1.53; P = 7.78 E−6). The combined discovery and replicatio... |
| rs5848(A;G)<br>Magnitude: 2<br>Frequency: 38.9%<br>Repute:Bad<br>References:26 | increased dementia risk Increased risk of frontotemporal dementia, alzheimer's disease and parkinson's disease.<br>...more... (hide)<br><br>rs5848(T;T) found a 3.2-fold increased risk vs C-allele carriers (95% CI: 1.50-6.73, p=0.003) for ubiquitin- and TAR DNA-binding protein 43 (TDP-43)-positive frontotemporal dementia (FTLD-U also knows as FTLD-TDP43), a progressive neurodegenerative disease affecting approximately 10% of early-onset dementia patients. Detailed haplotype analysis was conducted, indicating rs5848 as most likely causal variant. FTLD-TDP43 is divided into four subtypes, some sources suggest association with type 3 only . Association between rs5848(T;T) and frontotemporal dementia is now generally assumed in literature, despite some studies failing to reproduce it. First meta-analysis of rs5848 association with Alzheimer's disease combining previous five studies for a total of 2502 Alzheimer's disease cases and ... |
| rs10984447(A;G)<br>Magnitude: 2<br>Frequency: 39.8%<br>Repute:Bad<br>References:4 | 1.17x increased risk for multiple sclerosis...more... (hide)<br><br>rs10984447 has been reported in a large study to be associated with multiple sclerosis. The risk allele (oriented to the dbSNP entry) is (A); the odds ratio associated with this allele is 1.17 (CI 1.09-1.25). [PharmGKB:Curated GWAS Results: Risk alleles for multiple sclerosis identified by a genomewide study (Initial Sample Size: 931 trios, 2,431 controls; Replication Sample Size: 609 trios, 2,322 cases, 2,987 controls; Risk Allele: rs10984447-A).] [GWAS:Multiple sclerosis] |
| rs2987983(C;T) | 1.2x increased risk of prostate cancer This |

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| Magnitude: 2<br>Frequency: 39.8%<br>Repute:Bad<br>References:17 | genotype is at increased risk of prostate cancer Recommendation: To reduce the risk eat a diet rich in phytoestrogens such as flax seed<br>...more... (hide)<br>In one large Swedish study, men with one or two rs2987983(C) alleles were reported to be at somewhat increased (odds ratio 1.22) risk for Prostate cancer compared to men homozygous for the wild type rs2987983(T) allele, however the risk can be reduced by adding phytoestrogens to the diet. [PharmGKB:Curated Haplotype tagging SNP.] [OMIM:ESTROGEN RECEPTOR 2; ESR2] |
| rs744373(C;T)<br>Magnitude: 2<br>Frequency: 39.8%<br>Repute:Bad<br>References:15 | 1.17x risk of Alzheimer's...more... (hide)<br>rs6733839 and rs744373 are less than 2,000 base pairs apart,and they have similar ORs and MAFs. Therefore, it seems reasonable to suspect that these 2 SNPs are actually reporting the same signal. [GWAS:None] |
| rs11672691(A;G)<br>Magnitude: 2<br>Frequency: 41.6%<br>Repute:Bad<br>References:1 | 1.18x increased risk for PCSM in patients with prostate cancer For a person already diagnosed with prostate cancer, this relatively common genotype is associated with a more aggressive form of the cancer and an increased risk (by 1.18x) of dying from causes related to the cancer.<br>...more... (hide)<br>rs11672691 is a SNP on chromosome 19 located between the ATP5SL and CEACAM21 genes and within LOC100505495, a possible noncoding RNA. A large study of over 10,000 patients with prostate cancer concluded that each rs11672691(G) allele was associated with an increased risk (by 1.18x) of prostate cancer specific mortality (PCSM), i.e. death from the cancer. A meta-analysis of 4 studies totaling ~6000 patients with prostate cancer and follow up in 49,000 samples concluded that rs11672691 was associated with increased susceptibility (odds ratio 1.12, CI:1.03–1.21, p = 1.4 x 10e-8) but also with aggressive prostate cancer and therefore a poorer prognosis. [GWAS:Prostate cancer] |
| rs1219648(A;G)<br>Magnitude: 2<br>Frequency: 41.6%<br>Repute:Bad<br>References:44 | 1.20x risk for breast cancer...more... (hide)<br>Based on a study of ~2500 female patients of European ancestry with sporadic postmenopausal breast cancer, this |

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| | SNP in the FGFR2 showed the greatest risk. The risk allele is rs1219648(G), with a pooled odds ratio of 1.64 (CI:1.42-1.90) for rs1219648(G;G) homozygotes, and an odds ratio of 1.20 (CI: 1.07-1.42) for rs1219648(A;G) heterozygotes, compared with rs1219648(A;A) homozygotes. rs1219648 represents the SNP in the FGFR2 gene with the strongest association with breast cancer. However, nearby SNPs are almost as predictive. In particular, the following SNP alleles all have linkage values of 0.96 or greater with the rs1219648(G) allele in European populations: *rs2981579(A) *rs2420946(T) *rs11200014(A) An experimental rationale is presented indicating that this SNP is part of a haplotype t... |
| rs16944(G;G)<br>Magnitude: 2<br>Frequency: 41.6%<br>Repute:Bad<br>References:102 | Increased risk of mental disorders...more... (hide)<br><br>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 (A... |
| rs11190870(C;T)<br>Magnitude: 2<br>Frequency: 42.5%<br>Repute:Bad<br>References:4 | Possibly increased risk of scoliosis...more... (hide)<br><br>The T allele of this SNP is associated with an increased risk of scoliosis in adolescent Japanese girls. [http://www.nature.com/ng/journal/v43/n12/full/ng.974.html] [GWAS:None] |
| rs2420946(C;T)<br>Magnitude: 2<br>Frequency: 43.4%<br>Repute:Bad<br>References:15 | 1.20x risk for breast cancer...more... (hide)<br><br>This SNP is basically a proxy for SNP rs1219648, which represents the SNP in the FGFR2 gene with the strongest association with breast cancer. Based on a study of 1,049 |

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| | Chinese breast cancer patients, carriers of risk alleles at three SNPs (rs2981582, rs1219648 and rs2420946) were at 1.36x increased risk for breast cancer (CI: 1.13-1.62, p = 0.001). |
| rs699(C;T)<br>Magnitude: 2<br>Frequency: 43.4%<br>Repute:Bad<br>References:49 | increased risk of hypertension...more... (hide)<br><br>rs699 is a SNP in the angiotensin AGT gene that encodes a functional change. In most published literature, the name for this SNP is M235T, or perhaps Met235Thr, however its amino acid 268 (not 235) that varies based on the numbering in todays databases. rs699 is also occasionally known as C4072T. In any case, the rs699(C) allele encodes the threonine variant, which is associated with higher plasma angiotensin levels, and ultimately higher blood pressure leading to increased risk for hypertension associated disorders. This association was first reported in 1992 , and many studies, though not all, have replicated these findings. rs699 is also (generally) reported to be in tight linkage with rs5051, a neighboring SNP in the promoter of the AGT gene. rs699(C) has also been implicated as a risk... |
| rs710521(A;A)<br>Magnitude: 2<br>Frequency: 45.1%<br>Repute:Bad<br>References:9 | 1.4x increased risk of developing bladder cancer. Slightly increased risk of bladder cancer, particularly in Europeans and in people who already have tumors with low risk of progression.<br>...more... (hide)<br>rs710521 is a SNP located in chromosome region 3q38, near the TP63 gene. A study ultimately totaling over 4,000 bladder cancer patients of European ancestry (and 40,000 controls) concluded that individuals carrying two copies of rs710521(A) were at 1.4x increased risk (allele-specific odds ratio = 1.19, p = 1 x 10e-7) for the disease. An additional 1,425 bladder cancer patients, supplemented by a meta-analysis of 5,695 additional cases (and 40,000+ controls), did replicate a (somewhat) increased risk associated with the rs710521(A) allele. The overall odds ratio was 1.18 (OR: 1.12 - 1.25, p < 0.0001). Another study in Chinese populations did not find an increased risk of bladder cancer for this SNP. [PharmGKB:Non-Curated GWAS Results: Sequence variant on 8q24 confers susceptibility to ... |
| rs16942(A;G)<br>Magnitude: 2<br>Frequency: 45.1%<br>Repute:Bad<br>References:4 | slightly increased risk for breast cancer...more... (hide)<br><br>This SNP, a variant in the BRCA1 gene, is 1 of 25 SNPs |

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| | reported to represent independently minor, but cumulatively significant, increased risk for breast cancer. For this particular SNP, the risk (minor) allele is (G). For details of all 25 SNPs in this group, along with the two methods used to calculate overall risk estimates for breast cancer, refer to the SNPedia breast cancer entry or BRCA1 and BRCA2. |
| rs10086908(C;T)<br>Magnitude: 2<br>Frequency: 47.8%<br>Repute:Bad<br>References:6 | 1.7x increased risk for prostate cancer...more... (hide)<br><br>rs10086908 is a SNP in the 8q24 chromosomal region, which has been linked in several studies to prostate cancer. In a study of 1,563 patients of European ancestry, rs10086908 was designated as the representative of a prostate cancer risk region termed 'locus 3', with an odds ratio of 1.70 (CI: 1.39-2.07) for carriers of a risk genotype. Two other regions of chromosome 8q24 were also studied. Confirmation study of prostate cancer risk variants at 8q24 in African Americans identifies a novel risk locus. Homozygous deletions and recurrent amplifications implicate new genes involved in prostate cancer. |
| rs1160312(A;G)<br>Magnitude: 2<br>Frequency: 47.8%<br>Repute:Bad<br>References:5 | 1.6x increased risk of Male Pattern Baldness. ...more... (hide)<br><br>nature Baldness OR = 1.60, P = 1.1 times 10-14). see also gs122 The risk allele is assumed to be rs1160312(A). Note that a haplotype consisting of rs201571(T) - rs6036025(G) has a perfect correlation (r2 = 1) with rs1160312(A), at least in the Caucasian population studied. [PharmGKB:Non-Curated Male-pattern baldness susceptibility locus at 20p11. (Initial Sample Size: 578 cases, 547 controls; Replication Sample Size: 1,351 cases, 2,485 controls); (Region: 20p11.22; Reported Gene: PAX1; Risk Allele: rs1160312-A) This variant is associated with Male-pattern baldness.] [OMIM:ALOPECIA, ANDROGENETIC, 3; AGA3] [GWAS:Male-pattern baldness] |
| rs2073963(G;T)<br>Magnitude: 2<br>Frequency: 47.8%<br>Repute:Bad<br>References:1 | increased risk of baldness...more... (hide)<br><br>Baldness paper from 23andMe June 2012 [GWAS:None] |
| rs5174(A;G)<br>Magnitude: 2<br>Frequency: 47.8% | 1.3x increased risk for heart disease...more... (hide) |

Repute:Bad
References:8

rs5174 encodes a variant of the LRP8 gene, encoding the low density lipoprotein receptor-related 8 protein (or the apolipoprotein e receptor). The variant affects the protein, changing an arginine to a glutamine; depending on the publication, it can be referred to by terms like R952Q or Arg952Gln. In three Caucasian populations combined, the odds ratio for coronary artery disease or myocardial infarction (and thus heart disease) is 1.31 (adjusted p=0.0003). Note that this association was only seen in populations with familial and premature disease, as a study of 1,231 patients with primarily late-onset, sporadic heart disease did not show any correlation with this SNP. rs5174 (or its equivalent, rs5177) was analyzed in 1,210 patients with familial MI and 1,015 controls from the German MI F...

rs3923809(A;A)
Magnitude: 2
Frequency: 50.4%
Repute:Bad
References:8

1.9x risk for developing restless legs syndrome rs3923809 is a A/G variation on human chromosome 6. Associated with: Restless legs syndrome. This SNP, located in an intron of the BTBD9 gene, has a variant that is seen somewhat more frequently in individuals with restless legs syndrome. The risk allele is rs3923809(A); carriers of two such alleles, i.e. those with rs3923809(A;A) genotypes, are estimated to be 1.9 fold more likely to have restless legs syndrome than rs3923809(G;G) individuals. The authors of this study suggest that perhaps half of the cases of restless legs syndrome may involve the rs3923809 risk genotypes. Note that 70-80% of all individuals in European populations carry one or two copies of the (A) major allele, yet restless legs syndrome is thought to affect less than 10...
...more... (hide)

This SNP, located in an intron of the BTBD9 gene, has a variant that is seen somewhat more frequently in individuals with restless legs syndrome. The risk allele is rs3923809(A); carriers of two such alleles, i.e. those with rs3923809(A;A) genotypes, are estimated to be 1.9 fold more likely to have restless legs syndrome than rs3923809(G;G) individuals. The authors of this study suggest that perhaps half of the cases of restless legs syndrome may involve the rs3923809 risk genotypes. Consistent with this finding, another report about rs3923809 links the (G) minor allele to a lower frequency of restless legs syndrome with an overall odds ratio of 0.57 (CI: 0.48-0.68). Note that 70-80% of all individuals in European

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| | populations carry one or two copies of the (A) major allele, yet restless le... | |
| rs1024611(C;T)<br>Magnitude: 2<br>Frequency: 50.4%<br>Repute:Bad<br>References:19 | increased risk of exercise induced ischemia | ...more...<br>(hide) |
| | rs1024611, also known as the -2578A>G SNP due to its position in the promoter of the monocyte chemoattractant protein-1 MCP-1 CCL2 gene, influences the production of its corresponding protein, a chemokine involved in inflammatory responses. In a study focusing on 679 apparently healthy siblings of people with premature heart disease, investigators found that carriers of an rs1024611(C) allele - oriented as in dbSNP, not as published - independently predicted the risk of exercise induced ischemia in general. The odds ratio was reported to be 1.86 (CI: 1.14-3.04, p=0.014), regardless of race, age, sex and other factors. However, it is not clear if this risk carries over to individuals who lack siblings with heart disease. discussed in the 23andMe blog as being relevant to HIV rs1024611 is... | |
| rs493258(A;G)<br>Magnitude: 2<br>Frequency: 50.4%<br>Repute:Bad<br>References:4 | 1.15x risk of Age Related Macular Degeneration | ...more...<br>(hide) |
| | Evaluation of new and established age-related macular degeneration susceptibility genes in the Women's Health Initiative Sight Exam (WHI-SE) Study. [GWAS:Age-related macular degeneration] | |
| rs2383207(A;G)<br>Magnitude: 2<br>Frequency: 52.2%<br>Repute:Bad<br>References:31 | increased risk for heart disease...more... | (hide) |
| | discussed in this blog post as possibly playing a role in coronary heart disease This SNP was also associated with increased risk for coronary artery disease in a Korean population. The association remained significant after adjusting for significant clinical covariates (P=0.001 to 0.024). We identified one risk haplotype (GGGG; P=0.017) and one protective haplotype (AAAA; P=0.007) for development of CAD. Further analysis suggested that the SNPs probably confer susceptibility to CAD in a dominance model (covariates-adjusted P=0.001 to 0.024; OR=2.37 to 1.54). Also found to be significant in a study of 416 Italian myocardial infarction patients. Found to be associated with coronary heart disease in a study of 1360 Chinese Han patients, with an odds ratio of | |

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| | 1.52 (CI: 1.13 - 2.04) for rs2383... |
| rs351855(C;T)<br>Magnitude: 2<br>Frequency: 53.4%<br>Repute:Bad<br>References:16 | 1.2x increased risk for prostate cancer...more... (hide)<br><br>rs351855, a SNP in the fibroblast growth factor receptor 4 (FGFR4) gene, is also known as the Gly388Arg variant. The rs351855(T) allele encodes the risk (Arg) allele. The Arg form of this SNP is likely to cause a harder to treat version of node-positive breast cancer, including reducing the efficacy of Herceptin, based on a study of 372 patients. A study of ~500 Japanese prostate cancer patients found that individuals with a rs351855(T;T) genotype had a 2.2- and 1.9-fold increased risk of prostate cancer and benign prostate hyperplasia (BPH), and a 1.8-fold increased risk of metastatic prostate cancer compared to the (C;C) genotype. A meta-analysis published in 2011, surveying a total of 2,618 cases of prostate cancer, concluded that the odds ratio per rs351855(T) allele was 1.17 (CI: 1.07... |
| rs3758391(C;C)<br>Magnitude: 2<br>Frequency: 54.9%<br>Repute:Bad<br>References:4 | Normal (worse) aging. This is the less beneficial version of the SIRT1 longevity gene, which doesn't extend lifespan. It is normal in non-Asians. It has more cognitive decline with age. Taking resveratrol is also believed to increase longevity via the SIRT1 gene, so it might (or might not) be possible to mitigate this with resveratrol supplements or some other SIRT1 activator.<br>...more... (hide)<br>rs3758391, a SNP in the SIRT1 gene, has been reported to potentially have some role in either longevity, or with higher statistical significance, better cognitive function in older individuals. SIRT1 is thought to regulate neuronal metabolism and survival in response to stress, and the equivalent gene in other species influences maximal life span. The study reporting improved cognition for carriers of rs3758391(T) alleles was based on over 1,000 Finnish adults over the age of 85, and a weak trend towards lower cardiovascular disease was also reported for this allele. |
| rs2383206(A;G)<br>Magnitude: 2<br>Frequency: 56.9%<br>Repute:Bad<br>References:31 | 1.4x increased risk for heart disease...more... (hide)<br><br>rs10757274 and rs2383206 can significantly increase the risk of heart diseaseAbout one in every four Caucasians are |

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| | thought to carry the variants, and their risk of coronary heart disease is increased by 30 to 40%. rs10757278] in the same region has been linked to diabetes The chromosomal region where these SNPs are located is 9p21, and has no known genes. a [http://suicyte.wordpress.com/2007/05/26/soul-searching-i/ blog post about investigating rs10757274 and rs2383206 This SNP was also associated with increased risk for coronary artery disease in a Korean population. Also found to be significant in a study of 416 Italian myocardial infarction patients. A study of 1,000+ patients with early-onset angiographic coronary artery disease (CAD) concluded that rs2383206(G) was associated with an... |
| rs13153971(C;T)<br>Magnitude: 2<br>Frequency: 57.5%<br>Repute:Bad<br>References:0 | Normal (higher) risk of Asthma. This is normal for white people. But this (C;T) has 1.58x the risk compared to T;T (normal for non-white people). Tested in Koreans. ...more... (hide)<br>Asthma |
| rs358806(C;C)<br>Magnitude: 2<br>Frequency: 59.3%<br>Repute:Bad<br>References:2 | 1.78x increased risk of developing Type-2 diabetes ...more... (hide)<br>rs358806 has been reported in a large study to be associated with type-2 diabetes. The risk allele is (C); the odds ratio associated with heterozygotes is 0.86 (CI 0.75-0.97), and for homozygotes, 1.78 (CI 1.34-2.36). [PharmGKB:Non-Curated GWAS results: Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls (Initial Sample Size: 1,924 cases, 2,938 controls; Replication Sample Size: (see Zeggini 2007)). This variant is associated with Type 2 diabetes.] [GWAS:Type 2 diabetes] |
| rs6897932(C;C)<br>Magnitude: 2<br>Frequency: 59.3%<br>Repute:Bad<br>References:53 | 1.5x increased risk for multiple sclerosis...more... (hide)<br>The (C) allele of rs6897932, located in the alternatively spliced exon 6 of IL7RA gene and encoding the amino acid threonine rather than isoleucine at amino acid position 244, is associated with a slight increase (18%) in risk of developing multiple sclerosis. [PMID 17660817; Nature Genetics 39, 1083 - 1091 (2007) SG Gregory et al.] Note that the (C) allele is the most common at this position in all known populations and influences the ratios of the alternative isoforms (membrane bound and soluble) of the gene. blog post giving perspective on the significance of this snp a significant risk factor for multiple sclerosis in four independent (overall P = 2.9 x 10(-7)) |

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| | influences the amount of soluble and membrane-bound isoforms of the protein by putatively disrupting an exonic splicing silenc... |
| rs10248420(A;A)<br>Magnitude: 2<br>Frequency: 67.3%<br>Repute:Bad<br>References:3 | 7x less likely to respond to certain antidepressants (hide)<br>...more...<br><br>rs10248420 is a SNP in the ABCB1 gene (also known as the MDR1 gene), which encodes a protein that transports certain molecules across the blood-brain barrier. SNPs in ABCB1 may thus influence the intracerebral concentrations of certain drugs and thus their efficacy or potential for adverse side effects. rs10248420 is one of 9 SNPs found within a tight linkage block (r<sup>2</sup> >= 0.8 ) such that the minor allele at any one of them predicts (with ~80%+ accuracy) that the other SNPs will also be the minor allele. The list of the 9 SNPs is shown below. When treated for depression with substrates of the protein encoded by ABCB1, carriers of one or two minor alleles at these ABCB1 SNPs have been reported to respond better than non-carriers. The antidepressant drugs that are kn... |
| rs9652490(A;A)<br>Magnitude: 2<br>Frequency: 67.3%<br>Repute:Bad<br>References:22 | ~2x increased risk for Parkinson's disease, and, essential tremor<br>...more... (hide)<br><br>rs9652490 is a SNP in the LINGO1 gene. Note: it is unclear why one set of authors report the risk allele for essential tremors to be (G), while the others just as clearly report the risk allele to be (A). A follow-up case-control study replicated the original association of rs9652490(A;A) to an ~2x increased risk for Parkinson's disease, but also found the same risk for essential tremor (ET), thus linking the two diseases. The ET study was based on ~300 patients. 23andMe blog 'In a combined analysis of 752 subjects with essential tremor from Iceland, Austria, Germany and the United States and 15,797 controls, Stefansson et al found that the G version of rs9652490 in the LINGO1 gene is associated with increased odds of the condition. Carrying one G increases the odds of essential tremor by ... |
| rs27072(C;C)<br>Magnitude: 2<br>Frequency: 67.9%<br>Repute:Bad<br>References:7 | 2x risk of severe alcohol withdrawal.<br>Possible increased odds of ADHD.<br>Alcoholics with this are twice as likely to have seizures during alcohol withdrawal.<br>...more... (hide)<br>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 |

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| | 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 schiz... |
| rs7250581(G;G)<br>Magnitude: 2<br>Frequency: 69.0%<br>Repute:Bad<br>References:2 | 1.4x risk 1.4x higher risk for coronary artery disease (hide)<br>...more...<br><br>rs7250581 has been reported in a large study to be associated with heart disease, in particular, coronary artery disease. The risk allele (oriented to the dbSNP entry) is (G); the odds ratio associated with heterozygotes is 1.06 (CI 0.79-1.43), and for homozygotes, 1.40 (CI 1.05-1.86). |
| rs1385699(T;T)<br>Magnitude: 2<br>Frequency: 70.8%<br>Repute:Bad<br>References:5 | increased risk of baldness slightly higher risk of baldness (hide)<br>...more...<br><br>rs1385699, also known as Arg57Lys, is a SNP in the ectodysplasin A2 receptor EDAR2 gene. This gene is located on the X chromosome. The rs1385699(T) allele, which is the most common in European populations, was found to be associated with androgenetic alopecia in a case-control study of 400 males. |
| rs11983225(T;T)<br>Magnitude: 2<br>Frequency: 71.7%<br>Repute:Bad<br>References:1 | 7x less likely to respond to certain antidepressants (hide)<br>...more...<br><br>rs11983225 is a SNP in the ABCB1 gene (also known as the MDR1 gene), which encodes a protein that transports certain molecules across the blood-brain barrier. SNPs in ABCB1 may thus influence the intracerebral concentrations of certain drugs and thus their efficacy or potential for adverse side effects. rs11983225 is one of 9 SNPs found within a tight linkage block (r<sup>2</sup> >= 0.8 ) such that the minor allele at any one of them predicts (with ~80%+ accuracy) that the other SNPs will also be the minor allele. The list of the 9 SNPs is shown below. When treated for depression with substrates of the protein encoded by ABCB1, carriers of one or two minor alleles |

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| | at these ABCB1 SNPs have been reported to respond better than non-carriers. The antidepressant drugs that are kn... |
| rs2032583(T;T)<br>Magnitude: 2<br>Frequency: 71.7%<br>Repute:Bad<br>References:11 | 7x less likely to respond to certain antidepressants. This version of a blood brain barrier protein blocks many common antidepressants from entering the brain, including: amitriptyline (Elavil), citalopram (Celexa), paroxetine (Paxil), and venlafaxine (Effexor). That makes those antidepressants 7 times less effective.<br>...more... (hide)<br>rs2032583 is a SNP in the ABCB1 gene (also known as the MDR1 gene), which encodes a protein that transports certain molecules across the blood-brain barrier. SNPs in ABCB1 may thus influence the intracerebral concentrations of certain drugs and thus their efficacy or potential for adverse side effects. rs2032583 is one of 9 SNPs found within a tight linkage block (r<sup>2</sup> >= 0.8 ) such that the minor allele at any one of them predicts (with ~80%+ accuracy) that the other SNPs will also be the minor allele. The list of the 9 SNPs is shown below. When treated for depression with substrates of the protein encoded by ABCB1, carriers of one or two minor alleles at these ABCB1 SNPs have been reported to respond better than non-carriers. The antidepressant drugs that are know... |
| rs2235040(G;G)<br>Magnitude: 2<br>Frequency: 71.7%<br>Repute:Bad<br>References:3 | 7x less likely to respond to certain antidepressants (hide)<br>...more...<br>rs2235040 is a SNP in the ABCB1 gene (also known as the MDR1 gene), which encodes a protein that transports certain molecules across the blood-brain barrier. SNPs in ABCB1 may thus influence the intracerebral concentrations of certain drugs and thus their efficacy or potential for adverse side effects. rs2235040 is one of 9 SNPs found within a tight linkage block (r<sup>2</sup> >= 0.8 ) such that the minor allele at any one of them predicts (with ~80%+ accuracy) that the other SNPs will also be the minor allele. The list of the 9 SNPs is shown below. When treated for depression with substrates of the protein encoded by ABCB1, carriers of one or two minor alleles at these ABCB1 SNPs have been reported to respond better than non-carriers. The antidepressant drugs that are know... |
| rs2235067(G;G)<br>Magnitude: 2<br>Frequency: 71.7%<br>Repute:Bad<br>References:0 | 7x less likely to respond to certain antidepressants (hide)<br>...more...<br>rs2235067 is a SNP in the ABCB1 gene (also known as the |

MDR1 gene), which encodes a protein that transports certain molecules across the blood-brain barrier. SNPs in ABCB1 may thus influence the intracerebral concentrations of certain drugs and thus their efficacy or potential for adverse side effects. rs2235067 is one of 9 SNPs found within a tight linkage block (r<sup>2</sup> >= 0.8 ) such that the minor allele at any one of them predicts (with ~80%+ accuracy) that the other SNPs will also be the minor allele. The list of the 9 SNPs is shown below. When treated for depression with substrates of the protein encoded by ABCB1, carriers of one or two minor alleles at these ABCB1 SNPs have been reported to respond better than non-carriers. The antidepressant drugs that are know...

rs4148739(A;A)
Magnitude: 2
Frequency: 71.7%
Repute:Bad
References:2

7x less likely to respond to certain antidepressants (hide)
...more...

rs4148739 is a SNP in the ABCB1 gene (also known as the MDR1 gene), which encodes a protein that transports certain molecules across the blood-brain barrier. SNPs in ABCB1 may thus influence the intracerebral concentrations of certain drugs and thus their efficacy or potential for adverse side effects. rs4148739 is one of 9 SNPs found within a tight linkage block (r<sup>2</sup> >= 0.8 ) such that the minor allele at any one of them predicts (with ~80%+ accuracy) that the other SNPs will also be the minor allele. The list of the 9 SNPs is shown below. When treated for depression with substrates of the protein encoded by ABCB1, carriers of one or two minor alleles at these ABCB1 SNPs have been reported to respond better than non-carriers. The antidepressant drugs that are know...

rs2811712(A;A)
Magnitude: 2
Frequency: 86.7%
Repute:Bad
References:1

Normal (higher) risk of physical impairment with age. This causes physical problems for older people, and Cerebral Small Vessel Disease in the brain.
...more... (hide)

rs2811712 is a SNP associated with physical function in older individuals, and thus indirectly associated with longevity. In a study ultimately totaling over 3,300 elderly Caucasian individuals, the less common rs2811712(G) allele was associated with reduced physical impairment. Overall, severely limited physical function was present in 15.0% of the more common rs2811712(A;A) homozygotes, versus 7.0% of the rs2811712(G;G) homozygotes, yielding a per risk allele odds ratio of 1.48 (CI: 1.17-1.88, p=0.001). Association of single-nucleotide polymorphisms in the cell cycle genes with breast cancer in the British population. Consortium analysis of 7 candidate SNPs for ovarian cancer. Candidate gene analysis using imputed genotypes: cell cycle single-nucleotide polymorphisms and ovarian cancer r...

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| rs2188380(A;A)<br>Magnitude: 2<br>Frequency: 100.0%<br>Repute:Bad<br>References:1 | slightly higher risk for gout Over half of the people in any population have this genotype, which is reported to be at slightly higher risk (>2x) for gout compared to people who have zero (A) alleles for this SNP.<br>...more... (hide)<br>gout association, based on ; note that this publication refers to risk allele (T) based on the minus strand, whereas for dbSNP and SNPedia, rs72552713 is defined as on the plus strand, so therefore the risk allele is (A). |
| rs2230201(G;G)<br>Magnitude: 2<br>Frequency: 100.0%<br>Repute:Bad<br>References:1 | >1.4x risk of lupus...more... (hide)<br>In ~500 Japanese SLE patients, the (G) allele of rs2230201, a SNP in the C3 gene, is associated with higher risk for systemic lupus erythematosus (SLE). The odds ratio is 1.19 (CI: 1.01-1.41, p=0.038). |
| rs4444903(A;G)<br>Magnitude: 2<br>Repute:Bad<br>References:19 | 3.5x risk of hep-cancer in cirrhosis patients; higher glioma risk<br>...more... (hide)<br>rs4444903 is a SNP, also known as +61, located in the promoter region of the epidermal growth factor EGF gene that influences the amount of EGF produced. The rs4444903(G) allele appears to produce higher amounts of EGF than the (A) allele. A study of 2 Caucasian populations of patients with alcoholic cirrhosis indicates that, for these patients, the presence of the risk allele rs4444903(G) increases the chances of developing hepatocellular carcinoma (liver cancer). After adjusting for age, sex, race, etiology, and severity of cirrhosis the number of (G) copies led to an odds ratio for either (G;G) or (A;G) genotypes versus the (A;A) genotype of 3.49 (CI: 1.29-9.44, p=.01). In another study, analysis of 197 glioma patients showed that the rs4444903(G) allele conferred higher risks for gliom... |
| rs3746444(C;T)<br>Magnitude: 2<br>Repute:Bad<br>References:64 | ~1.2x increased risk for cancer...more... (hide)<br>microRNA 1,009 breast cancer cases and 1,093 controls in a population of Chinese women *rs3746444 in hsa-mir-499 A>G OR 1.25; 95% CI, 1.02-1.51 Genetic variants of miRNA sequences and non-small cell lung cancer survival. MicroRNA polymorphisms: the future of pharmacogenomics, molecular epidemiology and individualized medicine. Comprehensive |

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| | analysis of the impact of SNPs and CNVs on human microRNAs and their regulatory genes. SNPs in human miRNA genes affect biogenesis and function. Common genetic variants in pre-microRNAs are associated with risk of coal workers' pneumoconiosis. MicroRNA polymorphisms: a giant leap towards personalized medicine. Common genetic variants in pre-microRNAs and risk of gallbladder cancer in North Indian population. Association of pre-microRNAs genetic varia... |
| rs17435(T;T)<br>Magnitude: 2<br>Frequency: 17.9%<br>Repute:Bad<br>References:4<br>ambig | 1.4x increased risk for lupus...more... (hide)<br><br>rs17435 is one of several SNPs in the methyl CpG binding protein 2 (MECP2) that have been associated with risk for systemic lupus erythromatosis (SLE). The MECP2 gene is located on the X chromosome, which may be instructive since lupus is a predominantly female disease. An initial survey of 600 Korean patients, followed by 1,000 Caucasian patients, ultimately led to a meta-analysis indicating an odds ratio for the rs17435(T) risk allele of 1.39 (CI: 1.24â€“1.56, p = 1.2Ã—10e-08). Variants within MECP2, a key transcription regulator, are associated with increased susceptibility to lupus and differential gene expression in patients with systemic lupus erythematosus. Replication of recently identified systemic lupus erythematosus genetic associations: a case-contro... |
| rs4073582(G;G)<br>Magnitude: 1.9<br>Frequency: 39.8%<br>Repute:Bad<br>References:1 | slightly higher risk for gout Although ~80% of almost any group of people are of this genotype, it is associated with about twice the risk for developing gout as are people with the (A;A) genotype for this SNP.<br>...more... (hide)<br>gout association, based on |
| rs402710(C;C)<br>Magnitude: 1.9<br>Frequency: 44.4%<br>Repute:Bad<br>References:26 | Common, but higher lung cancer risk ?...more... (hide)<br><br>[PharmGKB:Non-Curated GWAS results: Lung cancer susceptibility locus at 5p15.33. (Initial Sample Size: 2,971 cases, 3,746 controls; Replication Sample Size: 2,899 cases, 5,573 controls); (Region: 5p15.33; Reported Gene(s): TERT, CLPTM1; Risk Allele: rs402710-C); (p-value= 0.000004).This variant is associated with Lung cancer.] [OMIM:LUNG CANCER SUSCEPTIBILITY 3; LNCR3] [GWAS:Lung cancer] |
| rs1801260(T;T) | Normal (higher) risk of ADHD symptoms. |

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| Magnitude: 1.8<br>Frequency: 54.0%<br>Repute:Bad<br>References:9 | This is the most common form of the CLOCK gene. In addition to possibly affecting evening preference, it has been linked to increased ADHD symptoms.<br>...more... (hide)<br>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 < 0.00... |
| rs2003046(C;C)<br>Magnitude: 1.8<br>Frequency: 59.8%<br>Repute:Bad<br>References:0 | Normal (higher) risk of Male Pattern Baldness. Discovered by 23andWe based on customer surveys, and considered preliminary research.<br>...more... (hide)<br>This is the 23andWe discovery: 'Male Pattern Baldness: Preliminary Research'. It affects appearance. |
| rs11842874(A;A)<br>Magnitude: 1.8<br>Frequency: 86.7%<br>Repute:Bad<br>References:2 | common but +17% risk Osteoarthritis risk<br>1.17 [1.11–1.23], p = 2.1 × 10−8<br>...more... (hide)<br>rs11842874 is a SNP in the MCF.2 cell line derived transforming sequence-like MCF2L gene on ch 13. Based initially on a GWAS study of 3,000 patients of European ancestry with osteoarthritis and then subsequently replicated for a total of 19,000+ cases, the rs11842874(A) allele (major allele) was associated with increased risk, at an odds ratio of 1.17 (CI: 1.11–1.23, p = 2.1 × 10e?8). The authors note that MCF2L regulates a nerve growth factor (NGF), and treatment with a humanized monoclonal antibody against NGF is associated with reduction in pain and improvement in function for knee osteoarthritis patients. [GWAS:None] |
| rs7192(G;T)<br>Magnitude: 1.7<br>Frequency: 42.0%<br>Repute:Bad<br>References:12 | 1.7x increased risk for developing a peanut allergy In populations of European ancestry, the rs7192(T) allele is associated with somewhat increased risk for developing peanut allergies.<br>...more... (hide) |

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| | rs7192, also known as Leu242Val or L242V, is a SNP in the HLA-DRA gene on chromosome 6. A study of several thousand children and their parents, most with food allergies, concluded that two SNPs, rs7192 and rs9275596, are associated with increased susceptibility to developing a peanut allergy in populations of European ancestry. The odds ratio reported for rs7192(T) were 1.7 (p=10e-8). Detecting significant single-nucleotide polymorphisms in a rheumatoid arthritis study using random forests [OMIM:?] [GWAS:None] |
| rs4132601(G;T)<br>Magnitude: 1.7<br>Frequency: 46.0%<br>Repute:Bad<br>References:4 | somewhat (1.7x) increased risk for acute lymphoblastic leukemia see discussion at rs4132601<br>...more... (hide)<br>23andMe blog Acute lymphoblastic leukemia *each rs4132601(G) 1.69x odds of ALL *each rs7089424(G) 1.65x odds of ALL *each rs2239633(G) 1.34x odds of ALL A meta-analysis published in 2014 based on a total of 15 case-control studies with 8333 cases and 36036 controls concluded that rs4132601 was associated with increased ALL risk in Caucasians and Hispanics but not among Asians. [OMIM:?] [GWAS:Acute lymphoblastic leukemia (childhood)] |
| rs2227692(C;T)<br>Magnitude: 1.6<br>Frequency: 14.1%<br>Repute:Bad<br>References:1 | 1.6x higher risk for diffuse gastric cancer...more... (hide)<br>rs2227692, also known as c.1162+162C>T, is a SNP in the SERPINE1 gene. A study of 612 Korean patients with gastric cancer concluded that the rs2227692 C/T and T/T genotypes had 1.6x greater risk for diffuse gastric cancer than the C/C genotype (p = .00084). |
| rs2549782(G;G)<br>Magnitude: 1.6<br>Frequency: 24.1%<br>Repute:Bad<br>References:8 | 1.6x increased risk for preeclampsia in most populations Note: this is relevant to the fetal genotype, not the maternal genotype<br>...more... (hide)<br>rs2549782 is a SNP in the endoplasmic reticulum aminopeptidase 2 ERAP2 gene. A case-control study of 1103 Chilean maternal-fetal dyads and 1637 unpaired African American samples (836 maternal, 837 fetal) concluded that the fetal minor allele rs2549782(G) was associated with 1.3x increased risk (CI: 1.075-1.619) for pre-eclampsia in the African American population (p = 0.009), but not in the Chilean population. }} |
| rs2736100(G;G)<br>Magnitude: 1.6 | 1.6x higher risk for glioma development...more... (hide) |

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| Frequency: 26.5%<br>Repute:Bad<br>References:62 | 23andMe blog rs2736100 C 1.27 Glioma Lung cancer susceptibility locus at 5p15.33. Common genetic variants on 5p15.33 contribute to risk of lung adenocarcinoma in a Chinese population. Variants in the CDKN2B and RTEL1 regions are associated with high-grade glioma susceptibility. Deciphering the impact of common genetic variation on lung cancer risk: a genome-wide association study. Role of 5p15.33 (TERT-CLPTM1L), 6p21.33 and 15q25.1 (CHRNA5-CHRNA3) variation and lung cancer risk in never-smokers. Individuals susceptible to lung adenocarcinoma defined by combined HLA-DQA1 and TERT genotypes. New insights into susceptibility to glioma. Replication of lung cancer susceptibility loci at chromosomes 15q25, 5p15, and 6p21: a pooled analysis from the International Lung Cancer Consortium. Evaluatio... |
| rs3764880(A;A)<br>Magnitude: 1.6<br>Frequency: 69.2%<br>Repute:Bad<br>References:2 | 1.2 - 1.8x increased tuberculosis risk...more... (hide)<br><br>rs3764880 is a SNP in the TLR8 gene, located on the X chromosome. rs3764880 is also known as Met1Val. A study of both Indonesian and Russian tuberculosis patients (and controls) concluded that the minor (A) allele of rs3764880 was associated with increased risk for tuberculosis. In the Indonesian population of 375 patients, the odds ratio was 1.8 (CI: 1.2–2.7); in the Russian population of 1,837 patients, the odds ratio was 1.2 (CI: 1.02–1.48); the combined evidence for association was p = 1.2×10e?3 – 6×10e?4. Note that the association was found primarily in males (although also to a lesser extent in homozygous females), presumably due to the presence of only one X chromosome in males. A functional toll-like receptor 8 variant is associated with HIV disease rest... |
| gs247<br>Magnitude: 1.5<br>Repute: Bad | Parkinson's Disease Risk<br>23andMe Patent Granted : rs10513789(T) increased risk of developing Parkinson's Disease GenomicsLawReport points out that the originally proposed version considered all of these to indicate increased risk of PD *rs6599389(A) *rs873785(A) *rs11248060(T) *rs6812193(C) *rs4130047(C) *rs7451962(A) *rs4397141(C) gs248, gs249 and gs250 |
| rs1801020(T;T)<br>Magnitude: 1.5 | 1.31x increased risk of heart disease...more... (hide) |

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| Frequency: 8.0%<br>Repute:Bad<br>References:12 | Affects heart disease risk for both men and women, based on a study of two Finnish population cohorts (HR = 1.31 (1.08-1.60) for CVD, uncorrected p = 0.006 multiplicative model). A meta-analysis of publications through July 2009, totaling 4,386 cases (vs 40,089 controls) concluded that apart from a very weak association with myocardial infarction for the rs1801020 (T;T) + (C;T) compared with (C;C) genotype, (odds ratio 1.13, CI: 1.00 - 1.27), the evidence for an association between F12 -4C>T and venous thromboembolism and myocardial infarction was weak. [OMIM:COAGULATION FACTOR XII; F12] |
| rs6896702(C;T)<br>Magnitude: 1.5<br>Frequency: 25.7%<br>Repute:Bad<br>References:0 | Slightly increased risk of developing Parkinson's Disease (hide)<br>...more...<br><br>rs6896702 is in linkage disequilibrium with a polymorphism that increases susceptibility to Parkinson's disease 1.74 times for carriers of the T allele |
| rs4585(T;T)<br>Magnitude: 1.5<br>Frequency: 27.4%<br>Repute:Bad<br>References:3 | slightly poorer (0.75x) response to metformin in type 2 diabetics<br>...more... (hide)<br><br>rs4585 has been found to modify the metformin response in patients with type 2 diabetes. Metformin Response |
| rs464049(T;T)<br>Magnitude: 1.5<br>Frequency: 28.3%<br>Repute:Bad<br>References:1 | increased risk of schizophrenia in limited study ...more... (hide)<br><br>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. |
| rs966221(C;C) | 1.5x increased stroke risk certain ...more... |

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| Magnitude: 1.5<br>Frequency: 28.6%<br>Repute:Bad<br>References:10 | populations<br><br>A study of 639 Chinese stroke patients concluded that the rs966221(C) allele is associated with increased risk for atherothrombotic strokes (odds ratio 1.51, CI: 1.09-2.10) in this population. | (hide) |
| rs4538475(A;G)<br>Magnitude: 1.5<br>Frequency: 29.2%<br>Repute:Bad<br>References:2 | Slightly increased risk of developing Parkinson's Disease<br>...more...<br><br>[OMIM:?] [GWAS:Parkinson's disease] | (hide) |
| rs6908425(C;T)<br>Magnitude: 1.5<br>Frequency: 33.6%<br>Repute:Bad<br>References:12 | 1.63x increased risk of developing Crohn's disease<br>...more...<br><br>rs6908425 has been reported in a large study to be associated with Crohn's disease. The risk allele (oriented to the dbSNP entry) is (C); the odds ratio associated with heterozygotes is 1.63 (CI 1.38-2.25), and for homozygotes, 1.95 (CI 1.43-2.67). [OMIM:?] [GWAS:Crohn's disease] | (hide) |
| rs642961(A;G)<br>Magnitude: 1.5<br>Frequency: 33.9%<br>Repute:Bad<br>References:12 | 1.68x increased risk of cleft lip...more...<br><br>risk of cleft lip 23andMe considers rs861020 a proxy Associated with European Facial Appearance *rs7590268 *rs16903544 *rs987525 *rs9574565 *rs227731 *rs642961 *rs1258763 Genome scan, fine-mapping, and candidate gene analysis of non-syndromic cleft lip with or without cleft palate reveals phenotype-specific differences in linkage and association results. Lack of association between IRF6 polymorphisms (rs2235371 and rs642961) and non-syndromic cleft lip and/or palate in a Brazilian population. Current concepts in genetics of nonsyndromic clefts. Studies of palatine rugae and interferon regulatory factor 6 variations in a group of families with sporadic hypodontia. IRF6 gene variants in Central European patients with non-syndromic cleft lip with or without cleft palate. Family-based study sh... | (hide) |
| rs2280714(A;A)<br>Magnitude: 1.5<br>Frequency: 35.4%<br>Repute:Bad | 1.4x increased risk of SLE...more... | (hide) |

References:22

The rs2004640(A) allele had a higher frequency in SLE cases (0.385) than controls (0.321; odds ratio (OR) = 1.32, P = 0.0003). In combined analysis, including all seven independent cohorts from the three studies so far, robust and consistent associations of the rs2004640(A) allele with SLE were observed. The estimate of risk was OR = 1.44 (CI: 1.34-1.55), with an overall P = 1.85 x 10(-23) for the rs2004640(A) allele. The haplotype (rs2004640T-rs2280714A) involved in both the alternative splice donor site and the elevated expression of IRF5 also had a highly significant association with SLE (pooled, P = 2.11 x 10(-16)). Our results indicate that the genetic effect on the risk of SLE mediated by IRF5 variants can be generally accepted in both white and Asian populations. A meta-analysis com...

rs13149290(C;T)
Magnitude: 1.5
Frequency: 36.7%
Repute:Bad
References:1

Slightly increased risk of developing prostate cancer (hide)
...more...

rs13149290 is one of seven SNPs found in a combined study of over 1,000 patients to be associated with increased risk for prostate cancer. The risk allele for this SNP is (C); and while the odds ratio was not specifically reported, the probability of false significance (not permuted though) was given as p=2.5 x 10e-5, using a dominant model of risk.

rs4464148(C;T)
Magnitude: 1.5
Frequency: 37.5%
Repute:Bad
References:14

1.10x increased risk for colorectal cancer...more... (hide)

rs4464148 is one of 3 SNPs in the SMAD7 gene associated with risk for colorectal cancer, based on a large study (7,400+ cases) conducted in the UK. The odds ratios show an increased risk for the minor rs4464148(C) allele; the OR for (C;C) homozygotes is 1.35 (CI: 1.2-1.51), and for (C;T) heterozygotes 1.10 (CI: 1.09-1.21), overall p=7x10<sup>-8</sup>.

rs10980705(C;T)
Magnitude: 1.5
Frequency: 38.9%
Repute:Bad
References:3

2.3x increased risk for knee osteoarthritis...more... (hide)

rs10980705, also known as -2820G/A, is a SNP upstream of the lysophosphatidic acid receptor 1 LPAR1 gene. In a study of 2 Japanese populations of patients with knee osteoarthritis (OA), the rs10980705(T) allele (in dbSNP orientation) was associated with increased risk in two Japanese populations,

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| | with an odds ratio of 2.3 (CI: 1.6-3.3, p=2.6x10<sup>-5</sup>). The association appears to hold only for the homozygous (T;T) genotype, and the authors state they believe this variant may be the 'disease-causing SNP'. However, 2% of individuals without knee OA are (T;T) homozygotes, and, over 50% of all patients with knee OA are (C;C) homozygotes. In a study of five collections of samples, and a meta-analysis combining them all, no particularly significant association was seen for rs10... |
| rs7850258(G;G)<br>Magnitude: 1.5<br>Frequency: 40.7%<br>Repute:Bad<br>References:2 | Slightly higher odds of developing primary hypothyroidism.<br>...more... (hide)<br><br>rs7850258 is located on chromosome 9 and is 67 kb from the nearest gene, "FOXE1". "FOXE1", also known as thyroid transcription factor 2, is an intronless gene with a crucial role in thyroid morphogenesis. This SNP is associated with primary hypothyroidism. Primary hypothyroidism is the most common thyroid disorder, affecting 1-5% of the population, and is characterized by deficiencies of thyroid hormones T3 (triiodothyronine) and T4 (thyroxine). Using electronic medical record-linked DNA biobanks, a study with 1,317 cases and 5,053 controls of European American ancestry identified rs7850258 to have the strongest association with hypothyroidism. The odds ratio for the minor allele A was 0.74 (p-value = 3.96 x 10-9). This SNP was in strong pair-wise linkage disequilibrium (r2 > 0.98) ... |
| rs1375144(C;T)<br>Magnitude: 1.5<br>Frequency: 40.7%<br>Repute:Bad<br>References:1 | 1.32x increased risk of developing bipolar disorder (hide)<br>...more...<br><br>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 (T); the odds ratio associated with heterozygotes is 1.32 (CI 1.07-1.63), and for homozygotes, 1.59 (CI 1.29-1.96). Findings from bipolar disorder genome-wide association studies replicate in a Finnish bipolar family-cohort. [GWAS:None] |
| rs10993994(C;T)<br>Magnitude: 1.5<br>Frequency: 43.4%<br>Repute:Bad<br>References:56 | increased prostate cancer risk (odds ratio 1.2) Compared to the (C;C) genotype for this SNP, (C;T) genotypes are estimated to be at 1.2x increased risk for prostate cancer.<br>...more... (hide)<br>rs10993994 linked to prostate cancer cancer-genetics these snps influence genetic risk for prostate cancer *the haplotype |

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|---|---|
| | rs6983267 rs1016343 rs4242384 *rs7501939 *rs1859962 *rs2660753 *rs9364554 *rs6465657 *rs10993994 *rs7931342 *rs2735839 *rs5945619 [PharmGKB:Non-Curated GWAS Results: Multiple loci identified in a genome-wide association study of prostate cancer (Initial Sample Size: 1,172 cases, 1,157 controls; Replication Sample Size: 3,941 cases, 3,964 controls; Risk Allele: rs10993994-T).] [OMIM:MICROSEMINOPROTEIN, BETA; MSMB] [GWAS:Prostate cancer] [GWAS:Prostate cancer (aggressive)] |
| rs2007153(G;G)<br>Magnitude: 1.5<br>Frequency: 43.4%<br>Repute:Bad<br>References:0 | increased risk of schizophrenia in limited study ...more... (hide)<br><br>Associated with schizophrenia in a study of Croatians. Due to the limited number and diversity of the studied population, more study is needed to confirm any link. The G allele was associated with an increased risk of schizophrenia. |
| rs30187(C;T)<br>Magnitude: 1.5<br>Frequency: 44.2%<br>Repute:Bad<br>References:28 | 1.4x higher risk for Ankylosing Spondylitis. Higher, but still very low, risk (0.14%) of this rare autoimmune disease that causes inflammation of the spine and joints. ...more... (hide)<br>rs30187 is one of several SNPs in the ERAP1 gene that has been shown in a large (over 1,000 Caucasian patients) study to be associated with ankylosing spondylitis. Each T allele appears to increase the odds by about 1.4x (p=3.4x10e-10). A study of 872 Korean patients found that each T allele was associated with 1.5x higher odds of ankylosing spondylitis. [GWAS:None] |
| rs10883365(A;G)<br>Magnitude: 1.5<br>Frequency: 45.1%<br>Repute:Bad<br>References:8 | 1.2x increased risk for developing Crohn's disease ...more... (hide)<br><br>rs10883365 has been reported in a large study to be associated with Crohn's disease. The risk allele (oriented to the dbSNP entry) is (G); the odds ratio associated with heterozygotes is 1.2 (CI 1.03-1.39), and for homozygotes, 1.62 (CI 1.37-1.92). The association between rs10883365 and Crohn's disease was replicated in a Japanese population. Pharmacogenetics: data, concepts and tools to improve drug discovery and drug treatment. Genetic determinants of ulcerative colitis include the ECM1 locus and five loci implicated in Crohn's disease. Worldwide population differentiation at disease-associated SNPs. Gene variants influencing measures of inflammation or predisposing to autoimmune and inflammatory diseases are not associated |

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| | with the risk of type 2 diabetes. Unbiased estimation of odds ra... |
| rs1801274(C;T)<br>Magnitude: 1.5<br>Frequency: 45.1%<br>Repute:Bad<br>References:57 | complex; generally greater risk for cancer progression (hide)<br>...more...<br><br>rs1801274 is a SNP in the Fc fragment of IgG, low affinity IIa, receptor (CD32) FCGR2A gene. rs1801274(C) encodes the arginine (R) allele, with the (T) allele encoding the variant histidine (H). The (H) isoform is considered high-binding to IgG2 and IgG3, while the (R) isoform is considered low-binding. This SNP is known in the literature by many names, including A519C and H131R. What's the importance of this? FcgR isoforms expressed on immune system cells have been linked to the pathogenic consequences triggered by autoantibodies or immune complexes in autoimmune diseases such as rheumatoid arthritis (RA) and systemic lupus erythematosus (SLE), as well as to the efficacy of some immunotherapeutic treatments such as rituximab. Many studies have been published about this FCGR2A SNP, roughly... |
| rs12431733(C;T)<br>Magnitude: 1.5<br>Frequency: 46.9%<br>Repute:Bad<br>References:1 | Slightly increased risk of developing Parkinson's Disease (hide)<br>...more...<br><br>[GWAS:Parkinson's disease] |
| rs11171739(C;T)<br>Magnitude: 1.5<br>Frequency: 47.8%<br>Repute:Bad<br>References:9 | 1.34x risk of developing Type-1 diabetes...more... (hide)<br><br>rs11171739 has been reported in a large study to be associated with type-1 diabetes. The risk allele (oriented to the dbSNP entry) is (C); the odds ratio associated with heterozygotes is 1.34 (CI 1.17-1.54), and for homozygotes, 1.75 (CI 1.48-2.06). In an expanded follow-up study of >6,000 controls and 6,000 patients, in which rs2292239 was ultimately chosen to replace the nearby rs11171739 originally determined to be associated with type-1 diabetes, the heterozygote odds ratio for SNP rs11171739 was recalculated to be 1.22 (CI 1.17â€“1.29). [PharmGKB:Non-Curated A genome-wide association study in 2,000 individuals for each of 7 major diseases and a shared set of 3,000 controls found an association of this SNP with type 1 diabetes.] [GWAS:Type 1 diabetes] |
| rs872071(A;G) | ~1.5x increased risk for chronic (hide) |

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| Magnitude: 1.5<br>Frequency: 48.7%<br>Repute:Bad<br>References:17 | lymphocytic leukemia<br>...more... | |
| | rs872071 is a SNP associated with the interferon regulatory factor 4 IRF4 gene. In a GWAS study including over 1,500 Caucasian patients with chronic lymphocytic leukemia (CLL), the rs872071(G) allele showed the strongest association (p = 1.91 x 10e-20) of all SNPs studied. The reported odds ratio was 1.54. See also: 23andMe blog A subsequent study () has found a similar association extending to another 800+ Spanish and Swedish CLL patients. rs872071 was also seen to be associated with CLL in Hong Kong Chinese (71 patients), with a reported odds ratio of 1.78 (CI: 1.25-2.53). A related hematologic malignancy, Hodgkin's lymphoma, may also be influenced by rs872071. Two case-control series totalling 529 and 2192 patients found an odds ratio of 1.21 (CI: 1.05-1.39, p = 0.009). [PharmGKB:Non-C... | |
| rs10757272(C;T)<br>Magnitude: 1.5<br>Frequency: 49.6%<br>Repute:Bad<br>References:3 | 1.30x increased risk for Coronary artery disease | ...more...<br>(hide) |
| | rs10757272 increases susceptibility to Coronary artery disease 1.30 times for heterozygotes (CT) and 1.54 times for homozygotes (TT) | |
| rs486907(A;G)<br>Magnitude: 1.5<br>Frequency: 50.4%<br>Repute:Bad<br>References:10 | 1.5x increased prostate cancer risk...more... | (hide) |
| | rs486907 is a SNP in the RNase L RNASEL gene that has been associated with cancer risk. The basic rationale behind most of these studies is that RNase L is responsible for deactivating RNA-based viruses that are associated with certain cancers, and therefore SNPs that lead to lower RNase L activity may lead to increased cancer risk. This SNP is also known as R462Q or Arg462Gln. In a study of prostate cancer patients, rs486907(A;G) heterozygotes were calculated to be at 1.5x increased risk, and rs486907(A;A) homozygotes 2x risk (p=0.007). This SNP was also associated with hereditary-prostate-cancer (HPC) predisposition with an odds ratio of 1.97 (p=0.07) in a study of 116 affected Finnish families. See also: [OMIM:PROSTATE CANCER, SUSCEPTIBILITY TO] | |
| rs763361(C;T)<br>Magnitude: 1.5<br>Frequency: 50.9%<br>Repute:Bad | Slightly increased risk for multiple autoimmune diseases, such as type-1 diabetes Higher risk of type-1 diabetes.<br>...more... (hide) | |

References:35

rs763361, also known as Gly307Ser, is a SNP in the immune response CD226 gene. The rs763361(T) allele (in dbSNP orientation) encodes the Ser. Based on multiple studies (by one group) now totaling over 2,000 patients, the rs763361(T) allele is associated with increased risk for multiple autoimmune diseases, including type-1 diabetes (p = 3 x 10e-9), multiple sclerosis (p = 4 x 10e-4), and possibly rheumatoid arthritis (p = 0.017). Based on tag SNP analysis, the authors conclude that this SNP could be the causal variant. Tested in a large multiple sclerosis data set consisting of 2369 trio families, 5737 cases and 10 296 unrelated controls, SNP rs763361 was associated with disease risk (p = 5.4 x 10e-8) [PharmGKB:Non-Curated GWAS Results: Robust associations of four new chromosome regions f...

rs4845618(G;T)
Magnitude: 1.5
Frequency: 51.3%
Repute:Bad
References:5

1.7x increased melanoma risk...more... (hide)

A study of 219 female melanoma patients and an equal number of controls concluded that 4 linked SNPs within the IL6R gene were associated with increased melanoma risk, but only when heterozygous. The odds ratio associated with the minor allele of this SNP, rs4845618, was 1.72 (CI: 1.04-2.84). The 4 linked SNPs from identified in this study are: rs6684439, rs4845618, rs4845622, and rs8192284.

rs3825776(A;G)
Magnitude: 1.5
Frequency: 51.3%
Repute:Bad
References:2

1.3x increased risk for ALS...more... (hide)

rs3825776, a SNP in the region of the LIPC gene on chromosome 15, has been associated with the sporadic form of ALS (Lou Gehrig's disease) in a study of 1000+ European patients. The odds ratio for the risk allele rs3825776(G) is 1.34 (CI: 1.12 - 1.46).

rs7774434(C;T)
Magnitude: 1.5
Frequency: 51.9%
Repute:Bad
References:2

Slightly increased risk of developing primary biliary cirrhosis
...more... (hide)

On the association between rheumatoid arthritis and classical HLA class I and class II alleles predicted from single-nucleotide polymorphism data. A two-stage search strategy for detecting multiple loci associated with rheumatoid arthritis. [GWAS:Primary biliary cirrhosis]

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| rs140701(A;G)<br>Magnitude: 1.5<br>Frequency: 52.2%<br>Repute:Bad<br>References:12 | Increased risk for anxiety disorders...more... (hide)<br><br>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). |
| rs6601764(C;T)<br>Magnitude: 1.5<br>Frequency: 54.9%<br>Repute:Bad<br>References:0 | 1.16x increased risk of developing Crohn's disease (hide)<br>...more...<br><br>rs6601764 has been reported in a large study to be associated with Crohn's disease. The risk allele (oriented to the dbSNP entry) is (C); the odds ratio associated with heterozygotes is 1.16 (CI 1.01-1.33), and for homozygotes, 1.52 (CI 1.28-1.80). |
| rs2241880(C;T)<br>Magnitude: 1.5<br>Frequency: 56.2%<br>Repute:Bad<br>References:29 | 1.4x increased risk for Crohn's disease in Caucasians (hide)<br>...more...<br><br>rs2241880, a SNP in the ATG16L1 gene encoding a threonine to alanine substitution ('T300A') in a protein known to be involved in the function of the epithelial cells lining the intestine, has been associated with Crohn's disease in several recent studies. In another recent (2007) report, rs2241880 is confirmed to be associated with both Crohn's disease and ileal disease, but additionally, the authors calculate risk for individuals who are homozygotes for this SNP plus 2 others (in the IBD5 and NOD2 genes). Individuals homozygous for the risk alleles for all 3 of these SNPs are estimated to be at 20 fold higher risk (CI ~9-49) for Crohn's disease. From the largest most recent survey, the Crohn's disease-associated SNPs for IBD5 and NOD2 are, respectively, rs6596075 and rs17221417. associate... |
| rs2881766(T;T)<br>Magnitude: 1.5<br>Frequency: 64.6%<br>Repute:Bad<br>References:4 | Slightly increased risk for pregnancy-induced hypertension<br>...more... (hide)<br><br>rs2881766 is a SNP in the promoter region of the estrogen receptor alpha ESR1 gene. Carriers of rs2881766(T) alleles are at somewhat (slightly) higher risk for pregnancy-induced |

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| | hypertension. |
| rs4570625(G;G)<br>Magnitude: 1.5<br>Frequency: 64.6%<br>Repute:Bad<br>References:18 | maybe: higher scores on anxiety-related personality traits; greater placebo response One study of (only) 25 people shows this genotype was a significant predictor of clinical placebo response, being associated with greater improvement in anxiety symptoms. A criticism of this study has also been published, and it has not been replicated since the original publication years ago. This SNP has been linked to several psychiatric and/or behavioral phenomena, including: *higher scores on anxiety-related personality traits *obsessive compulsive disorder *panic disorder with a possible gender-dependent effect<br>...more... (hide)<br>rs4570625, also known as G-703T, is a SNP in the tryptophan hydroxylase 2 TPH2 gene. This SNP has been linked to several psychiatric and/or behavioural phenomena, including: * rs4570625 and rs4565946 linked to the pathogenesis of early-onset obsessive compulsive disorder * rs4570625 and rs11178997 in TPH2's regulatory region display preferential transmission * rs4570625(G;G) homozygosity was a significant predictor of clinical placebo response, being associated with greater improvement in anxiety symptoms. g2b2mh is rs4570625(G;G) is a significant predictor of clinical placebo response? maybe not panic disorder with a possible gender-dependent effect Transmission disequilibrium of polymorphic variants in the tryptophan hydroxylase-2 gene in attention-deficit/hyperactivity disorder. Associa... |
| rs501120(A;A)<br>Magnitude: 1.5<br>Frequency: 64.6%<br>Repute:Bad<br>References:10 | common, but >1.3x increased risk for heart disease (hide)<br>...more...<br>rs501120 is a SNP found to be associated with heart disease in two populations by the German MI (Myocardial infarction) Family Study group. The risk allele in dbSNP orientation is rs501120(A). The odds ratio per allele is 1.33 (CI: 1.20-1.48, adjusted p=0.0248). Association of genetic variants and incident coronary heart disease in multiethnic cohorts: the PAGE study. Comparative analysis of genome-wide association studies signals for lipids, diabetes, and coronary heart disease: Cardiovascular Biomarker Genetics Collaboration. Genetic risk score and risk of myocardial infarction in Hispanics. \|OA=1 }} Coronary artery disease-related genetic variant on chromosome 10q11 is associated |

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|---|---|
| | with carotid intima-media thickness and atherosclerosis. Genetics and cardiovascular disease: Design and dev... |
| rs7341475(G;G)<br>Magnitude: 1.5<br>Frequency: 74.3%<br>Repute:Bad<br>References:12 | 1.58x increased schizophrenia risk for women ...more... (hide)<br><br>A total of 2,274 cases of schizophrenia were studied, resulting in an association between rs7341475, exclusively in women. Based on all populations the estimated relative risk for women carrying the common (G;G) genotype is 1.58 (p = 8.8 x 10(<sup>-7</sup>). This finding was replicated in a large independent Ashkenazi Jewish collection (721 cases, 259 female; 1455 controls, 834 female), with confirmation that it applies to both schizophrenia and schizoaffective disorder. [PharmGKB:Non-Curated GWAS results: Genome-Wide Association Identifies a Common Variant in the Reelin Gene That Increases the Risk of Schizophrenia Only in Women. (Initial Sample Size: 660 cases, 2,271 controls; Replication Sample Size: 2,274 cases, 4,401 controls); (Region: 7q22.1; Reported Gene(s): RELN; Ris... |
| rs165599(G;G)<br>Magnitude: 1.5<br>Repute:Bad<br>References:64 | May indicate increased susceptibility to schizophrenia (hide)<br>...more...<br><br>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. [PharmGKB:Curated In 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.] [OMIM:CATECHOL-O-METHYLTRANSFERASE; COMT] |
| rs3843763(C;T)<br>Magnitude: 1.5<br>Repute:Bad<br>References:2 | Slightly lower HDL (%22Good%22) Cholesterol. This slightly reduces levels of good HDL cholesterol in your blood. And that's usually bad, because HDL cholesterol actually cleans your arteries and transports cholesterol out of your body. Things that can help raise HDL cholesterol if it is too low include: quitting smoking, losing weight, exercise, and niacin (vitamin B3). |

...more... (hide)

The (T) allele of rs3843763 was associated with risk for lower high-density lipoprotein (HDL) cholesterol plasma levels in 3 independent population samples, including both Caucasians and African-Americans. [PharmGKB:Curated This SNP has been associated with small changes in HDL cholesterol levels. The T allele has been correlated with lower HDL.] [OMIM:PHOSPHOLIPID TRANSFER PROTEIN; PLTP]

rs5219(C;T)
Magnitude: 1.5
Repute:Bad
References:79

1.3x increased risk for type-2 diabetes...more... (hide)

rs5219, also known as E23K, is a SNP in the KCNJ11 gene. The association between type-1 diabetes and rs5219 is mentioned as being replicated in ; there is also a 90% correlation between rs5219 and rs5215. This SNP is one of 4 relatively common SNPs reported to represent risk for type-2 diabetes in the DESIR prospective study of 3,877 Caucasian participants. Under an additive model, the odds ratio for the minor (risk) allele (T) is 1.15 (CI: 0.95-1.4, p=0.2, ie not significant). But for the 4 SNPs, each risk allele increased type-2 diabetes risk by 1.34x (p=2x10e-6), with an odds ratio of 2.48 (CI: 1.59-3.86) for carriers of 4 or more compared to those with one or none (risk alleles). Genotyping of ~4000 Chinese as well a meta-analysis concluded that rs5219 was associated with type-2 diabet...

rs8050136(A;A)
Magnitude: 1.4
Frequency: 17.7%
Repute:Bad
References:118

1.4x increased risk for T2D in some populations ...more... (hide)

This SNP is in a linkage block in the FTO gene with rs1121980; see rs1121980 for the association of this block with early onset obesity since it showed the strongest association. This SNP did not show any association with obesity, type-2 diabetes or any other related traits in a study of 3,210 Chinese subjects. Furthermore, the minor allele frequency was much lower in Chinese populations compared to populations of European descent. However, a more recent study of 4,189 Chinese Han subjects combined with a meta-analysis of published studies concluded that rs8050136(A) was associated with both increased risk for type-2 diabetes and obesity. rs8050136 associated with increased food intake, but not of impaired energy expenditure. [PharmGKB:Non-Curated GWAS results: Genome-wide association yie...

rs12770228(A;G)
Magnitude: 1.4
Frequency: 39.8%

1.4x increased risk for meningioma...more... (hide)

Repute:Bad
References:2

rs12770228 is a SNP in a region on ch 10p12.31 designated as the chromosome 10 open reading frame 114 C10orf114; it is just upstream of the MLLT10 gene. A study of ~1,600 patients with meningioma, all of European ancestry, concluded that increased risk for the disease was associated with rs12770228(A) alleles. The odds ratio was 1.37 (CI: 1.18-1.59, p = 4 x 10e-4). A nearby SNP, rs11012732, was also linked to meningioma risk.

rs1126497(C;T)
Magnitude: 1.4
Frequency: 54.9%
Repute:Bad
References:3

1.4x increased risk for breast cancer...more... (hide)

rs1126497, also known as Thr115Met, is a SNP in the epithelial cell adhesion molecule EPCAM gene. In a study of case control study of 1643 individuals with breast cancer and 1818 control subjects in Eastern and Southern Chinese populations, the rs1126497(T) allele was associated with a 1.4x increased risk (CI: 1.16-1.57), and it was also associated with early breast cancer onset (p=0.0023). Functional polymorphism in the EpCAM gene is associated with occurrence and advanced disease status of cervical cancer in Chinese population.

rs6010620(G;G)
Magnitude: 1.4
Frequency: 55.0%
Repute:Bad
References:31

1.4x higher risk for glioma development;
but this is the common allele
...more... (hide)

'"rs6010620"' is a SNP associated with atopic dermatitis (AD). Also known as eczema, AD is one of the most common chronic inflammatory skin disorders with a prevalence estimated up to 20% in children and 3% in adults . It is a complex and highly heterogeneous disease that likely involves various genetic factors. The SNP rs6010620 locates on chromosome 20q13.33, at which there are eight genes — "PRIC285", "STMN3", "RTEL1", "TNFRSF6B", "ARFRP1", "ZGPAT", and "LIME1" —, within a single 200-kb LD block around rs6010620. Among them, "TNFRSF6B" and "ZGPAT" might be the susceptibility candidates whose expressions correlate with the SNP as suggested by expression quantitative trait loci (eQTL) analysis . rs6010620's association with AD was first identified in ...

rs6495446(C;C)
Magnitude: 1.4
Frequency: 61.6%
Repute:Bad

2.5x increased risk for chronic kidney
disease in a small unreplicated study
...more... (hide)

References:1

rs6495446, an intronic SNP in the MTHFS gene, was associated with increased risk for chronic kidney disease based on a study associated with the Framingham Heart Study of ~1,000 Caucasian individuals. The odds ratio rs6495446(C) allele was 1.24 (CI: 1.09-1.41, p=0.001).news 23andMe reports this same information with a different emphasis: each T allele lowers the odds of chronic kidney disease by 0.8 times.

rs3131296(G;G)
Magnitude: 1.4
Frequency: 83.0%
Repute:Bad
References:7

1.4x increased risk for schizophrenia...more... (hide)

23andMe blog schizophrenia *rs3131296(C) increased the odds of schizophrenia by 1.19x *rs12807809(T) increased the odds of schizophrenia by 1.15x *rs9960767(C) increased the odds of schizophrenia by 1.23 times. A study of ~2,500 Han Chinese patients with schizophrenia did find rs3131296 to be significantly associated in this population. [GWAS:Schizophrenia]

rs17465637(C;C)
Magnitude: 1.34
Frequency: 50.0%
Repute:Bad
References:18

1.34x higher risk for myocardial infarction...more... (hide)

rs17465637 is a SNP found to be associated with heart disease by the German MI (Myocardial infarction) Family Study group This 2011 study concluded that the only MIA3 gene SNP to be associated with increased risk for myocardial infarction is rs17465637, with an odds ratio of 1.17 (CI: 1.04-1.32) and 1.37 (CI: 1.08-1.74) for carriers of one or two (C) alleles, respectively. 23andMe blog coronary artery disease and heart attack SNP Risk Version Effect *rs646776 T 1.19 *rs17465637 C 1.14 *rs1746048 C 1.17 *rs6725887 C 1.17 *rs11206510 T 1.15 *rs3184504 T 1.13 *rs2306374 C 1.15 *rs3782886 C 1.44 [PharmGKB:Curated GWAS results: Genome-wide association of early-onset myocardial infarction with single nucleotide polymorphisms and copy number variants. (Initial Sample Size: 2,967 cases, 3,075 con...

rs1260326(C;T)
Magnitude: 1.3
Frequency: 50.0%
Repute:Bad
References:48

slightly higher risk for gout...more... (hide)

The T allele of rs1260326 has been associated with type 2 diabetes and hypertriglyceridemia. A fine mapping approach indicates that this SNP, rs1260326, shows the strongest association signal with metabolic phenotypes in the region that

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| | also harbors the tightly linked (r(2)=0.93) SNP rs780094, which has been previously associated with triglyceride and glucose concentrations. This SNP, rs1260326, encodes a common missense glucokinase regulatory protein gene GCKR variant. gout association, based on Loci related to metabolic-syndrome pathways including LEPR,HNF1A, IL6R, and GCKR associate with plasma C-reactive protein: the Women's Genome Health Study. Polymorphisms of the HNF1A gene encoding hepatocyte nuclear factor-1 alpha are associated with C-reactive protein. The common P446L polymorphi... |
| rs1434536(A;G)<br>Magnitude: 1.3<br>Frequency: 56.6%<br>Repute:Bad<br>References:1 | 1.29x increased breast cancer risk...more... (hide)<br><br>rs1434536 is a SNP reported to potentially represent a miR-125b binding site. A study of 459 probands from a breast cancer affected sibling pair cohort were recruited from the ECOG study, and in conjunction with 1,145 patient samples from the CGEMS study, led to the conclusion that rs1434536 was associated with increased risk for breast cancer. The odds ratios for rs1434536(A;G) and (A;A) individuals were 1.29 (CI: 0.95-1.74) and 1.94 (CI: 1.40-2.71), respectively (p=1.6x10e-4). A miR-125b binding site polymorphism in bone morphogenetic protein membrane receptor type IB gene and prostate cancer risk in China. |
| rs13387042(A;A)<br>Magnitude: 1.25<br>Frequency: 31.9%<br>Repute:Bad<br>References:41 | 1.24x increased risk for breast cancer...more... (hide)<br><br>rs13387042(A;A) has an estimated 1.44-fold greater risk of breast cancer than the GG genotype. This initial association was later tested among patients in the Breast Cancer Association Consortium, including 31,510 women with invasive breast cancer, 1101 women with ductal carcinoma in situ (DCIS), and 35,969 controls. In Caucasian women of European origin, each A allele is associated with 1.12 higher odds (CI: 1.09-1.15). This association held regardless of ER or PR status. [PharmGKB:Non-Curated GWAS results: A multistage genome-wide association study in Breast Cancer identifies two new risk alleles at 1p11.2 and 14q24.1 (RAD51L1). (Initial Sample Size: 1,145 cases, 1,142 controls; Replication Sample Size: 8,625 cases, 9,657 controls); (Region: 2q35; Reported Gene(s): Intergenic; Risk Alle... |
| rs748404(T;T)<br>Magnitude: 1.25<br>Frequency: 57.7% | slightly increased risk (1.25) for lung cancer ...more... (hide) |

Repute:Bad
References:1

In a GWAS dataset of 1,447 lung cancer cases, and then replicated in another 1300 cases, the rs748404(T) allele was significantly correlated with increased risk (odds ratio 1.15, p = 1.1 × 10e-9), in agreement with a previous meta-analysis.

rs498872(C;T)
Magnitude: 1.2
Frequency: 39.8%
Repute:Bad
References:16

1.2x higher risk for glioma development...more... (hide)

23andMe blog rs498872 A 1.18 Glioma [GWAS:Glioma]

rs10865331(A;G)
Magnitude: 1.2
Frequency: 45.1%
Repute:Bad
References:8

1.2x higher risk for ankylosing spondylitis...more... (hide)

rs10865331 is a SNP in chromosomal region 2p15. After initial GWAS reports linked this SNP to ankylosing spondylitis (AS), a follow-up study in 456 Spanish patients replicated the association between the rs10865331(A) allele and higher risk for AS (p = 0.039). In a 2010 report, the AA genotype was associated with 1.3x odds of ankylosing spondylitis, and the GG genotype with 0.8x odds, compared to the AG genotype. [OMIM:?] [GWAS:Ankylosing spondylitis]

rs2056116(A;G)
Magnitude: 1.2
Frequency: 45.1%
Repute:Bad
References:3

1.18x risk for breast cancer...more... (hide)

rs2056116 is a somewhat unusual SNP, in that it is from an 'ultraconserved element' of the genome, meaning it is from a stretch of over 200 base pairs that is absolutely identical between equivalent regions of human, mouse, and rat genomes. The functions of these elements is unknown, but they are thought to have regulatory roles of various sorts. They usually have less SNPs on average than other regions of the genome. The (G) allele of rs2056116 was found to be associated with higher risk for breast cancer, with an odds ratio of 1.18 (CI: 1.06-1.30, p=0.002). The odds ratio for the (G;G) genotype compared to the (A;A) genotype was 1.41 (CI: 1.15-1.74, p=0.0011). When patients were divided into younger vs older women, the odds ratios were increased for women under 50 years of age; for the (...

rs4977756(A;G)
Magnitude: 1.2
Frequency: 47.8%

1.2x higher risk for glioma development...more... (hide)

Repute:Bad
References:15

rs4977756 is a SNP near the CDKN2BAS gene. A (GWA) study for open-angle glaucoma (OAG) blindness ultimately based on ~1500 cases concluded that the rather common rs4977756(A) allele was statistically associated with the condition, with a per allele odds ratio of 1.39 (CI: 1.28–1.51, p = 1.35 × 10e?14). [OMIM:?] [GWAS:Glioma]

rs2076295(G;T)
Magnitude: 1.2
Frequency: 54.9%
Repute:Bad
References:1

One copy of the risk allele (G), slightly increased risk for pulmonary fibrosis
...more... (hide)

The SNP "'rs2076295"' on chromosome 6p24 is located within the intronic region of the DSP gene, which encodes the protein desmoplakin. Desmoplakin is a component of the desmosome, and is an adhesive intercellular molecule that tightly links adjacent cells for cell-cell adhesion. It is part of a dynamic structure that involves other proteins, such as plakoglobin and plakophilins, which tether the cytoskeleton to the cell membrane . The minor allele rs2076295(G) is reported by dbSNP to have an allele frequency (MAF) of 0.454. A recent case-control genome-wide association study (GWAS) involving 1,616 cases and 4,683 controls (and a replication study with 876 cases and 1,890 controls) was performed on non- Hispanic white individuals with fibrotic idiopathic interstitial pneumonia (IIP) ; not...

rs11037909(T;T)
Magnitude: 1.2
Frequency: 55.8%
Repute:Bad
References:12

1.47x type II diabetes risk...more... (hide)

rs11037909 increases susceptibility to Type II Diabetes 1.27 times for heterozygotes (CT) and 1.47 times for homozygotes (TT)

rs2223841(A;A)
Magnitude: 1.2
Frequency: 69.2%
Repute:Bad
References:0

more likely to go bald before age 40 more likely to go bald before age 40
...more... (hide)

male-pattern baldness. Full text available. *G less likely to go bald before age 40 *A more likely to go bald before age 40

rs3802842(A;C)
Magnitude: 1.17
Frequency: 34.5%
Repute:Bad
References:31

1.17x increased risk of colorectal cancer...more... (hide)

rs3802842 is a SNP in a region of ch 11q23.1 that has been associated with some cancers. IN a study of over 10,000 cases of colorectal cancer, rs3802842(C) was associated with increased risk (odds ratio 1.17, CI: 1.12-1.22, p = 1.08 x 10e-12). Additional SNPs in chromosomal regions 8q21, 8q24, 10p14, 11q, 15q13.3 and 18q21 can be combined additively to produce an overall risk, with an average odds ratio per risk allele of 1.19. [PharmGKB:Non-Curated GWAS Results: Genome-wide association scan identifies a colorectal cancer susceptibility locus on 11q23 and replicates risk loci at 8q24 and 18q21 (Initial Sample Size: 981 cases, 1,002 controls; Replication Sample Size: 16,476 cases, 15,351 controls; Risk Allele: rs3802842-C).] [OMIM:COLORECTAL CANCER, SUSCEPTIBILITY TO, 7; CRCS7] [GWAS:None]...

rs610932(C;C) ...more... (hide)
Magnitude: 1.1
Frequency: 17.0%
Repute:Bad
References:6

Alzheimer's disease associated, based on large 2011 study Genome-wide association study identifies variants at CLU and PICALM associated with Alzheimer's disease. The prevalence of CD33 and MS4A6A variant in Chinese Han population with Alzheimer's disease. [GWAS:None]

rs3818361(C;T) 1.15x increased risk for late-onset Alzheimer's ...more... (hide)
Magnitude: 1.1
Frequency: 31.9%
Repute:Bad
References:13

rs3818361 is a SNP associated with the complement component (3b/4b) receptor 1 CR1 gene. The association of this SNP and late-onset Alzheimer's disease was replicated over a total of another 1829 cases (and 2576 controls). The odds ratio for rs3818361(T) allele was 1.15 (note the p value was 0.014). [GWAS:None]

rs2651899(A;G) 1.1x higher risk for migraines...more... (hide)
Magnitude: 1.1
Frequency: 44.2%
Repute:Bad
References:7

rs2651899 is a SNP on ch 1p36.32 in the PRDM16 gene. A large GWAS study of over 5,000 patients with migraines and a meta-analysis concluded that this SNP was one of three influencing the condition, albeit with only slight risk increases on its own. The odds ratio for the slightly rarer rs2651899(G) allele was 1.11 (CI: 1.07 - 1.15, p = 3.8 x 10e-9). A 2013 study published by the American Headache Society reports significant influence of rs1835740, LRP1 rs11172113 and

PRDM16 rs2651899 polymorphisms on migraine susceptibility in the Northern Indian population. Genome-wide-associated variants in migraine susceptibility: a replication study from North India. [GWAS:None]

rs889312(A;C)
Magnitude: 1.1
Frequency: 44.6%
Repute:Bad
References:49

Very slightly higher risk for breast cancer...more... (hide)

rs889312 is a SNP near the MAP3K1 gene. rs889312 was one of the four strongest associating SNPs found in a genome-wide association study of over 4,000 breast cancer samples. . From a DeCode user report, it appears that the risk allele is C; this A;A individual reportedly has 0.93x risk of breast cancer. In a study of 1,267 breast cancer patients, rs889312 heterozygotes and minor allele homozygotes were less likely to be lymph node positive at breast cancer diagnosis (P = 0.044) relative to major allele homozygote carriers. A meta-analysis including 26,015 cases and 33,962 controls concludes that the rs889312(C) allele is clearly a significant - but quite weak - contributor to increased risk for breast cancer. breast cancer *rs2981582, rs3803662, and rs889312) showed weak but significant as...

rs4977574(A;G)
Magnitude: 1.1
Frequency: 49.6%
Repute:Bad
References:28

Some studies - but not others - report a slightly increased risk for myocardial infarction
...more... (hide)

rs4977574 increases susceptibility to Coronary artery disease 1.30 times for heterozygotes (A;G) and 1.54 times for homozygotes (G;G) rs4977574 was found to be associated with myocardial infarction based on a study by the eMERGE network in which electronic medical records were searched to find clinical associations associated with selected SNPs. . In this study of 2,000 patients, rs4977574(G;G) homozygotes had an increased risk (odds ratio) for myocardial infarction of 1.84 (CI: 1.14−3.09, p = 0.011) [PharmGKB:Curated GWAS results: Genome-wide association of early-onset myocardial infarction with single nucleotide polymorphisms and copy number variants. (Initial Sample Size: 2,967 cases, 3,075 controls; Replication Sample Size: 9,746 cases, 9,746 controls); (Region: 9p21.3; Reported...

rs1344706(G;T)
Magnitude: 1.1
Frequency: 52.3%
Repute:Bad
References:41

1.1x increased risk for schizophrenia...more... (hide)

rs1344706 is a SNP in the ZNF804A gene, and it is probably

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| | 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 sc... |
| rs3758549(C;C)<br>Magnitude: 1.1<br>Frequency: 61.9%<br>Repute:Bad<br>References:11 | increased risk of parkinson's disease<br>While this does indicate increased risk of<br>in one Parkinson's disease study. It is so<br>common that it makes more sense to think<br>of the other 2 genotypes as rare risk<br>reducers.<br>...more... (hide)<br>CC linked to parkinson's disease We provide evidence for a novel, strong and reproducible association of the PITX3 promoter SNP rs3758549 : C>T (p=0.004) with PD. The C-allele appears to be a recessive risk allele with an estimated population frequency of 83%. |
| rs925391(C;C)<br>Magnitude: 1.1<br>Frequency: 83.1%<br>Repute:Bad<br>References:0 | more likely to go bald; common...more... (hide)<br>23andMe seems to consider each A at rs925391 to have lowered men's odds of baldness by 11x. baldness *lowest P value of 2.1×10-12 for rs10521339 *family based rs938059 shows the lowest P value (4.03×10-6) (table 3) *rs6152 (P=6.66×10-10) |
| rs11209026(G;G)<br>Magnitude: 1.1<br>Frequency: 91.8%<br>Repute:Bad<br>References:103 | Normal, but higher risk for certain autoimmune diseases. Almost everyone has this form, but it indicates a slightly higher risk of Ankylosing Spondylitis, Crohn's Disease (1.09x), Psoriasis (1.06x), and Ulcerative Colitis (1.06x). The risk is much greater than for the rare version. This is the interleukin-23 (IL-23) receptor. Media:http://www.the-rheumatologist.org/details/article/2528901/Neutralizing_IL-23_and_Its_Targets_May_Improve_Ankylosing_Spondylitis.html] Neutralizing IL-23 and Its Targets May Improve Ankylosing Spondylitis by Lara C. Pullen, PhD in The Rheumatologist newsmagazine ]<br>...more... (hide)<br>A relatively rare allele at SNP rs11209026, in the IL23R gene, |

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| | appears to provide a fairly strong protective effect against the development of Crohn's disease in both Jewish and non-Jewish populations. Note that in this study the (A;A) and (A;G) genotypes were combined for statistical reasons (there were too few (A;A) individuals to study by themselves), so the risk is shown as equal for both. associated With Inflammatory Bowel Diseases but Not With Celiac Disease rs11209026 had a protective effect for IBD in the case-control analysis (odds ratio [OR] 0.19, 95% confidence interval [CI] 0.10-0.37, P= 6.6E-09). Both CD (OR 0.14, CI 0.06-0.37, P= 3.9E-07) and UC (OR 0.33, CI 0.15-0.73, P= 1.4E-03) were associated with IL23R. rs2241880 was associated with CD susceptibility (OR 1.36, CI 1.12-1.... | |
| rs7412(C;C)<br>Magnitude: 1.1<br>Repute:Bad<br>References:34 | more likely to gain weight if taking olanzapine | ...more...<br>(hide) |
| | The ancestral allele is C. The rs7412(T) allele, also known as Arg176Cys, generally indicates the presence of an Apoε2 allele; see the ApoE page for a full discussion of the ApoE alleles and their association with Alzheimer's disease. Another SNP related to ApoE is rs429358. In a study of 67 mostly Caucasian patients prescribed the atypical antipsychotic olanzapine, carriers of a rs7412(C) allele were more likely to gain significant weight compared to rs7412(T;T) carriers, as assessed by physiogenomic analysis of corresponding weight profiles. Two other SNPs, rs5092 and rs4765623, were also significantly associated with weight profiles in these patients. Current limitations of SNP data from the public domain for studies of complex disorders: a test for ten candidate genes for obesity ... | |
| rs2546890(A;A)<br>Magnitude: 1<br>Frequency: 29.2%<br>Repute:Bad<br>References:7 | Higher risk of multiple sclerosis...more... | (hide) |
| | 23andMe blog multiple sclerosis rs2546890 IL12B G 0.87 [GWAS:None] | |
| rs6932590(C;T)<br>Magnitude: 1<br>Frequency: 29.2%<br>Repute:Bad<br>References:6 | 1.1x increased risk for schizophrenia...more... | (hide) |
| | 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 region that codes for several genes with significant roles in autoimmunity, such as human leukocyte | |

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| | 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 ... |
| rs987525(A;C)<br>Magnitude: 1<br>Frequency: 38.9%<br>Repute:Bad<br>References:12 | 2.5x increased risk for cleft lip...more... (hide)<br><br>A genome-wide association study involving 224 cases and 383 controls of Central European origin found that significant association between rs987525 and risk for nonsyndromic cleft lip with or without cleft palate (NSCL/P). The odds ratio for rs987525(A;G) individuals was 2.57 (CI: 2.02-3.26), and for rs987525(A;A), 6.05 (CI: 3.88-9.43). The calculated population attributable fraction for this marker is 0.41, suggesting that this study has identified a major genetic cause for NSCL/P. 23andMe blog article Associated with European Facial Appearance *rs7590268 *rs16903544 *rs987525 *rs9574565 *rs227731 *rs642961 *rs1258763 Family-based study shows heterogeneity of a susceptibility locus on chromosome 8q24 for nonsyndromic cleft lip and palate. Association between genetic variants of reported c... |
| rs2282679(A;C)<br>Magnitude: 1<br>Frequency: 43.4%<br>Repute:Bad<br>References:23 | somewhat lower vitamin D levels...more... (hide)<br><br>rs2282679, located in the group-specific component (vitamin D binding protein) GC gene on chromosome 4p12, has been linked by several studies to vitamin D serum concentrations. In both studies, the allele associated with lower vitamin D, and thus the potential for vitamin D insufficiency, is rs2282679(C). Carriers of two such alleles have lower vitamin D than carriers of one allele, who in turn on average have lower vitamin D levels than rs2282679(A;A) individuals. A near-perfect proxy (i.e. substitute) for rs2282679 is reported to be rs4588. [GWAS:Vitamin D levels] |

## These seem interesting, but have not been flagged as clearly Good or Bad

...more... (hide)

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| gs145<br>Magnitude: 4 |  <br>Female<br>Female. |
| rs1426654(A;A)<br>Magnitude: 2.7<br>Frequency: 100.0%<br>References:18 | probably light-skinned, European ancestry<br>This SNP influences skin pigmentation and indicates indicates light-skinned European ancestry. estimates that the rs1426654(A) allele (light skin pigmentation) spread through the European population around 6,000 - 12,000 years ago. Prior to that, 'European ancestors' were most likely relatively brown-skinned.<br>...more... (hide)<br>This SNP influences skin pigmentation. The allele, A111T, rs1426654(A), indicates light-skinned european ancestry. It appears as if this SNP is a relatively new one in human evolution; one estimate is that the rs1426654(A) allele, in other words, light skin pigmentation, spread through the European population around 6,000 - 12,000 years ago. Prior to that, 'European ancestors' were most likely relatively brown-skinned. Another study () has concluded that almost individuals carrying the A111T variant can trace ancestry back to a single person who most likely lived at least 10,000 years ago. This SNP is one of three from the SLC24A5 gene that can be analyzed to categorize the ancestry of a person as either European, African, or Asian, based on a 2009 study. The 3 SNPs are: * rs1426654 * rs25... |
| gs259<br>Magnitude: 2.5 | homozygous for eye color haplotype #3<br>According to one publication this is blue eye color haplotype #3, however, feedback from Promethease users suggest it is not a reliable predictor of eye color. |
| rs1667394(A;A)<br>Magnitude: 2.1<br>Frequency: 72.1%<br>References:8 | blond hair & blue eyes is 4x more likely...more... (hide)<br>rs1667394 increases susceptibility to Blond rather than brown hair 4.94 times for carriers of the A allele rs1667394 increases |

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| | susceptibility to Blue rather than brown eyes 29.43 times for carriers of the A allele rs1667394 increases susceptibility to Blue rather than green eyes 6.74 times for carriers of the A allele For green versus blue eye color rs12913832 in OCA2/HERC2 has a score of 51.5 and an estimated allelic OR of 8.43 . The SNP rs1667394 in this same region has an estimated OR of (4.85–10.06). [OMIM:HECT DOMAIN AND RCC1-LIKE DOMAIN 2; HERC2] [GWAS:Eye color] | |
| gs288<br>Magnitude: 2 | You have two long form 5-HTTLPR.<br>You likely have two long-form 5-HTTLPR (serotonin-transporter-linked polymorphic region). Variations in the region have been extensively investigated in connection with neuropsychiatric disorders. Identification of tag haplotypes for 5HTTLPR for different genome-wide SNP platforms | |
| rs2547231(G;G)<br>Magnitude: 2<br>Frequency: 1.8%<br>References:1 | Decreased levels of some blood metabolites<br><br>[GWAS:Blood metabolite levels] | ...more...<br>(hide) |
| rs5888(T;T)<br>Magnitude: 2<br>Frequency: 27.7%<br>References:15 | lower HDL cholesterol but higher risk for age-related macular degeneration<br>...more... (hide)<br><br>rs5888 is a SNP in the scavenger receptor class B, member 1 SCARB1 gene. In a case-control study of two Caucasian populations totaling 2,498 patients, rs5888(C;T) heterozygotes had an increased odds ratio of 2.9 (CI: 1.6-5.3, p<0.002) for age-related macular degeneration (ARMD) based on a pooled analysis. Subgroup analysis indicated that the odds ratio for exudative forms of ARMD was even higher (3.6; CI: 1.7-7.6, p>0.0015) for rs5888(C;T) heterozygotes. | |
| rs17576(A;A)<br>Magnitude: 2<br>Frequency: 38.1%<br>References:19 | normal risk for lung cancer; higher risk for MI<br><br>rs17576, also known as Gln279Arg or Q279R, is a SNP in exon 6 of the matrix metalloproteinase-9 MMP9 gene. The rs17576(G) allele encodes the Arg (R). In a study of 385 male veterans with greater than 20 pack-years of cigarette smoking, rs17576(G) allele carriers were at higher risk for chronic obstructive pulmonary disease (COPD). The rs17576(G;G) homozygous genotype was at 3-fold increased risk for COPD. A study of 744 Chinese patients with lung cancer found that the rs17576(G;G) genotype was associated with higher risk of lung cancer with metastasis (adjusted OR, 1.79, CI: 1.03-3.08) | ...more...<br>(hide) |

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| | compared to the (A;A) genotype. rs17576 is also one of two SNPs in the MMP9 gene associated with increased risk for myocardial infarction, but not coronary artery disease. The relatively weak odds ratios for... |
| rs10494366(T;T)<br>Magnitude: 2<br>Frequency: 40.7%<br>References:30 | Shorter QT interval...more... (hide)<br><br>rs10494366, a SNP in the NOS1AP gene encoding the nitric oxide synthase I adaptor protein, accounts for some of the variation seen in abnormal heart rhythms, in particular, the QT interval. Based on studies totaling ~4,000 individuals of Caucasian ancestry, homozygotes for one allele have shorter QT intervals, while homozygotes for the other allele have a longer QT interval. A follow-up study determined that one rs10494366(G) allele was associated with a 3.8-ms (CI: 3.0 - 4.6ms, p=7.8x10(-20)) increase in QT interval duration, and two (G) alleles had twice that increase. No increase in risk for sudden death due to a cardiac problem was associated with this SNP, though. rs10494366 minor homozygotes had a 9.3 msec longer QT interval compared to major homozygotes (p=5.7x10(-5)); rs10918594 mi... |
| rs9923231(C;T)<br>Magnitude: 2<br>Frequency: 40.7%<br>References:53 | reduced warfarin dose if treated for VTE...more... (hide)<br><br>Several SNPs in the VKORC1 gene have been linked to warfarin sensitivity, with perhaps the most common being this SNP rs9923231. Note that the orientation as published in scientific articles is often on the opposite strand compared to the orientation in dbSNP, so you will sometimes see it as a G>T snp. It is also known as *-1639G>A with the minus indicating that this is in an upstream promoter *3673 based on its position in GenBank accession number AY587020. *VKORC1*2. The main findings related to the treatment of venous thromboembolism (aka VTE; from hypercoagulability) with the blood thinner warfarin for this SNP are that carriers of the rs9923231(T) allele require significantly reduced doses of warfarin, and are (otherwise) at a higher risk of serious bleeding. Clinical studies de... |
| rs2298566(A;C)<br>Magnitude: 2<br>Frequency: 42.5%<br>References:3 | increased risk of coronary heart disease;<br>better response to statins<br>...more... (hide)<br><br>rs2298566 is a SNP in the SNX19 gene. The risk allele in terms of heart disease is rs2298566(C). This SNP is one of the 5 used by Celera's genetic risk score (GRS) for coronary heart disease (CHD). *rs20455, in the KIF6 gene *rs3900940, in the |

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| | MYH15 gene *rs7439293, in the PALLD gene *rs2298566, in the SNX19 gene *rs1010, in the VAMP8 gene For each of the five variants, the GRS was increased by 1 if the subject was homozygous for the risk variant, unchanged if heterozygous, and decreased by 1 if the individual did not carry the variant. Therefore, individuals carrying all 10 possible risk variants (two copies of each of the five SNPs) were assigned a GRS of 5 and those carrying no risk variants a GRS of -5. A high GRS was defined as 3 or higher. Approximately 4% of the white cohort in ARIC... |
| rs4481887(A;G)<br>Magnitude: 2<br>Frequency: 44.2%<br>References:2 | more likely to be able to smell asparagus<br>metabolites in urine 1.67x more likely than people with (G;G) to be able to smell the methanethiol produced after eating asparagus.<br>...more... (hide)<br>Asparagus anosmia research was performed by 23andMe and presented at the 2009 ASHG according to geneticfuture. Asparagus anosmia Asparagus Metabolite Detection [GWAS:Common traits (Other)] |
| rs9938025(A;G)<br>Magnitude: 2<br>Frequency: 45.1%<br>References:0 | Unknown effect...more... (hide)<br>23andme reports that having two copies of an 'A' at rs9938025 is associated with moderately lower odds of dry earwax and having two copies of a 'G' is associated with moderately higher odds in their customers with European ancestry compared to those who have 'AG'. https://www.23andme.com/en-eu/about/factoids/ |
| rs460000(C;C)<br>Magnitude: 2<br>Frequency: 56.6%<br>References:3 | Better response to amphetamine<br>Individuals with the C/C genotype at rs460000 (N = 83) reported approximately twofold higher ratings of stimulation and euphoria relative to the A/A+A/C (N = 69) genotype group, at both the 10 and 20 mg doses.<br>...more... (hide) |
| rs2108622(C;C)<br>Magnitude: 2<br>Frequency: 60.7%<br>References:51 | lower warfarin dosing...more... (hide)<br>rs2108622 is a SNP in the cytochrome P450, family 4, subfamily F, polypeptide 2 CYP4F2 gene. In the males rs2108622(G) was significantly higher in cerebral infarction patients (P = 0.025) A study of Italian patients concluded that rs2108622(T;T) patients require 5.49 mg/day of warfarin versus |

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| | 2.93 mg/day for (C;C) patients. Analysis of variance indicates that about 7% of mean weekly warfarin dose variance is explained by CYP4F2 genotype. A study of 370 Brazilian ('admixed') patients concludes that prospective CYP4F2 genotyping is of little value to these patients, presumably due to ethnic-based differences. [PharmGKB:Curated Risk or phenotype-associated allele: no allelic association. Phenotype: Improved pharmacogenetic algorithm-based warfarin dosing. Study size: 370. Study population/et... |
| rs2281617(C;C)<br>Magnitude: 2<br>Frequency: 69.0%<br>References:0 | Normal (better) response to amphetamine<br>The main finding of the study is that subjects with genotype C/C at rs2281617 scored significantly higher on Euphoria and Energy than subjects with genotypes C/T and T/T after 10 mg amphetamine, and trends were also observed with measures of Stimulation and (lower) Sedation.<br>...more... (hide)<br>Some evidence exists that the T;T and C;T genotypes are associated with less euphoria and energy in response to a given dose of a stimulant drug, e.g. dextroamphetamine, than occurs for the C;C genotype. See [especially figure 3 http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3377371/figure/F3/] |
| rs17822931(C;C)<br>Magnitude: 2<br>Frequency: 75.9%<br>References:12 | Wet earwax. Normal body odour. Normal colostrum. This is normal for non-Asians. It makes ear-wax wet, sticky, and honey-coloured. It also produces colostrum for breast feeding. People with this gene need to use deodorant to avoid smelling bad, unlike some Asians.<br>...more... (hide)<br>rs17822931, also known as c.538G>A or G180R, is a SNP in the ATP-binding cassette, sub-family C (CFTR/MRP), member 11 ABCC11 gene. The ABCC11 protein helps transport small molecules across apical membranes such as those in apocrine secretory cells. This SNP determines wet vs dry earwax as well as sweat production, and it is also associated with lipid secretion. It is commonly (T;T) for Asians and (C;C) for Europeans and Africans. A study of deodorant usage in a large (17,000+) population showed that rs17822931(T;T) individuals were at least 5-fold less likely to use deodorant, consistent with them being 'genotypically nonodorous'. Although not statistically significant, there was some evidence for a heterozygote effect, implying that individuals with a (C;C) genotype for this SNP were l... |
| rs5400(C;T)<br>Magnitude: 1.7 | significantly higher sugar consumption?...more... (hide) |

Frequency: 19.1%
References:6

rs5400(T;T) carriers had a significantly higher intake of sugars blog discussion Food Preference

rs1800454(A;A)
Magnitude: 1.5
Frequency: 2.7%
References:1

affected Of interest since it is a variation from omim, but no obvious consequences. ...more... (hide)

Variation in the ATP-binding cassette transporter 2 gene is a separate risk factor for systemic lupus erythematosus within the MHC. [OMIM:PEPTIDE TRANSPORTER PSF2 POLYMORPHISM]

rs1042725(C;C)
Magnitude: 1.5
Frequency: 28.3%
References:23

~0.8cm taller...more... (hide)

nature SNP rs1042725 is associated with height (P = 4E-8) in a study involving over 20,000 individuals. The gene harboring this SNP, HMGA2, is a strong biological candidate for having an influence on height, since rare, severe mutations in this gene are known to alter body size in mice and humans. rs1042725 is estimated to explain approx 0.3% of population variation in height in both adults and children (approx 0.4 cm increased adult height per C allele). rs1042725 is associated with increased height (0.36 cm 95% IC[0.12-0.61] per C allele, P=0.004). in men rs1042725 may explain 3% of height variability [PharmGKB:Non-Curated GWAS Results: A common variant of HMGA2 is associated with adult and childhood height in the general population (Initial Sample Size: 4,921 individuals; Replication S...

rs1799782(C;T)
Magnitude: 1.3
Frequency: 11.7%
References:55

1.3x increased risk for oral cancer among Asians ...more... (hide)

rs1799782, a SNP also known as Arg194Trp located in the DNA-repair gene XRCC1, is associated with a reduced risk for certain types of cancer. The (T) allele encodes the Trp amino acid. A study of ~200 Korean cases of skin cancer indicated that the rs1799782(T;T) genotype was inversely associated with squamous cell cancer risk (adjusted odds ratio 0.06, CI: 0.006-0.63). A meta-analysis of 8 case-control studies concluded that significant association was found between the rs1799782 polymorphism and oral cancer risk among Asians, showing an odds ratio of 1.347 (CI: 1.0 - 1.81) for allele comparison, 1.378 (CI: 1.070 - 1.775) for (T;T) homozygotes vs (C;C) homozygotes, and 1.420 (CI: 1.041 - 1.936) for comparison under a dominant model. 18067C>T possibly related to non-Hodgkin lymphoma rs20...

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| rs997669(A;A)<br>Magnitude: 1.1<br>Frequency: 38.9%<br>References:2 | 1.18x higher risk for breast cancer...more... (hide)<br><br>rs997669 shows an association with breast cancer in a British study involving ~2300 patients. The odds ratio for the (G;G) vs (A;A) homozygotes is 1.18 (CI: 1.04-1.34, p=0.003). |
| rs4794067(T;T)<br>Magnitude: 1.1<br>Frequency: 56.6%<br>References:2 | Lower risk of Aspirin Induced Asthma. Normal (higher) risk of lupus and intractable Grave's Disease. This is the normal version. It doesn't cause Aspirin Induced Asthma, but it doesn't reduce the risk of lupus or Grave's disease either like the rare version does.<br>...more... (hide)<br>rs4794067(C;C) and rs4794067(C;T) genotypes for the TBX21 gene are calculated to be at 2.15 fold higher risk (CI: 1.26-3.64) for aspirin-induced asthma (AIA) compared to rs4794067(T;T) homozygotes, based on a study of 72 patients vs 640 controls in a study of a Japanese population. Note that in this population, but perhaps not others, linkage disequilibrium exists between this SNP located upstream of the gene, rs4794067, and a common synonymous SNP located in the first exon, rs2074190, with a predictive accuracy of 92%. On the other hand, rs4794067(C;C) may greatly decrease the risk of lupus if rs17250932(C;C) is also present (at least in Chinese people). And those 2 SNPs also reduce the risk of intractable Graves' disease (at least in Japanese people). |
| rs1892534(A;G)<br>Magnitude: 1.05<br>Frequency: 43.8%<br>References:12 | Normal C-Reactive Protein levels. Very slight (x1.05) increase in overall risk for cancers. This causes a median hsCRP Level of 1.88 mg/L in women. Strangely this was associated with a slightly higher overall cancer risk (in Finnish people).<br>...more... (hide)<br>rs1892534 is a SNP that was originally associated with C-reactive protein levels, and given other findings that suggest C-reactive protein levels may be predictive of cancer risk, was thought to be linked to cancer risk. A large study pooling data from 3 Finnish studies totaling over 18,000 individuals concluded that while the rs1892534(A) allele was not likely to be causative (relative to cancer), it is associated with increased risk for cancer overall. The odds ratio for 1 or 2 such alleles was 1.05 (CI: 0.90 - 1.23) and 1.2 (CI: 1.01 - 1.42) overall. Two other CRP SNPs (rs1169300 and rs2464196) were specifically associated with increased lung cancer risk. CRP SNPs were not associated with colorectal, prostate or breast cancer risk. [PharmGKB:Curated GWAS Results: Loci Related to Metabo... |

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| rs3807153(C;T)<br>Magnitude: 1<br>Frequency: 5.3%<br>References:1 | carrier of a benign change...more... (hide)<br><br>[OMIM:RENAL TUBULAR ACIDOSIS, DISTAL, AUTOSOMAL RECESSIVE] |
| rs1835740(A;G)<br>Magnitude: 1<br>Frequency: 37.2%<br>References:5 | unreplicated, higher risk for migraine...more... (hide)<br><br>rs1835740 is a SNP from the 8q22.1 chromosomal region, best known for harboring various genes involved in glutamate metabolism and transport. A large study comprising over 5,000 migraine patients and over 10,000 matched controls, primarily from Europe, found that the rs1835740(A) allele conferred a (slightly) higher risk for the disorder. The reported odds ratio was 1.18, (CI: 1.127–1.244, p = 1.69 × 10e-11).. However this study has failed to replicate in 2 followups. |
| rs1800469(C;C)<br>Magnitude: 1<br>Frequency: 53.1%<br>References:75 | lower TGF-β1 levels, higher risk for COPD? This normal C version significantly reduces TGF-β1 levels by allowing AP1 to downregulate TGF-β1 production.<br>...more... (hide)<br>rs1800469, also known as -509 C>T or −1347 C > T, is an SNP in the promoter region of the transforming growth factor beta1 TGFB1 gene. That means it doesn't change the nature of the TGF-β1 protein, but it does change the amount produced. The T version increases the amount of TGF-β1 produced, by preventing AP1 from binding to this region where in the C version it would normally downregulate production. That increased TGF-β1 production with the T version is probably what causes all the health effects of this SNP. Whether increased TGF-β1 is good or bad probably depends on the disease you are interested in. It has been associated with increased risk for chronic obstructive pulmonary disease (COPD). Although smoking is the main risk factor, smokers with a rs1800469(C) allele (or 2 other ... |
| rs167771(A;A)<br>Magnitude: 1<br>Frequency: 67.3%<br>References:9 | Possibly more 'Insistence on Sameness' on ADI-R test.Possibly increased FTND nicotine dependence. [http://www.ncbi.nlm.nih.gov/pubmed/22309839?dopt=Abstract<br>...more... (hide)<br>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 |

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| | (81 presenting with EPS and 189 without) [PharmGKB:Curated Risk 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.] |
| rs3914132(T;T)<br>Magnitude: 1<br>Frequency: 68.1%<br>References:6 | Normal (not lower) otosclerosis risk...more... (hide)<br><br>Each C allele at rs3914132 decreases the likelihood of developing otosclerosis, which can cause hearing loss. [PharmGKB:Non-Curated In a GWAS done in Belgian-Dutch cases and controls (694 of each, split into a discovery and a replication set) and then in a second replication set of 455 French cases and 480 controls, rs3914132 was found to be associated with otosclerosis.] [OMIM:OTOSCLEROSIS; OTSC1] [GWAS:Otosclerosis] |
| rs2283123(C;C)<br>Magnitude: 1<br>Frequency: 75.5%<br>References:0 | normal risk of schizophrenia in limited study ...more... (hide)<br><br>Associated with schizophrenia in a study of Croatians. Due to the limited number and diversity of the studied population, more study is needed to confirm any link. The T allele was associated with a decreased risk of schizophrenia. |
| rs1800407(G;G)<br>Magnitude: 1<br>Frequency: 85.8%<br>References:13 | blue/gray eyes more possible...more... (hide)<br><br>rs1800407 is located in exon 13 of the OCA2 gene at amino acid position 419, and it is therefore also known as the R419Q variant. The (A) allele encodes the amino acid glutamine (instead of arginine), and it is associated with increased odds of having green/hazel eye color in some Caucasian populations. One copy of of rs1800407(A) is sufficient for this effect. [PMID 12163334, PMID 15889046; OMIM 203200.0012] According to tree scanning analysis, the proportion of eye colour variation explained by this nucleotide position is merely 4%. Thus, additional variation present in the OCA2 gene and perhaps some other pigment related genes must be taken into account in order to explain the high phenotypic variation in iris colour. Among rs12913832(A;A) individuals, the penetrance for green/hazel eye... |
| rs1861046(G;G) | Conduct disorder (CD) is one of the most |

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| Magnitude: 1<br>Frequency: 92.9%<br>References:1 | prevalent childhood psychiatric conditions, and is associated with a number of serious concomitant and future problems. CD symptomatology is known to have a considerable genetic component, with heritability estimates in the range of 50%. Four SNPs reached genome-wide significance with CD symptom count, based on P < 5x10^-8 Two of these SNPs (rs16891867 and rs1861046) were in the gene C1QTNF7 (C1q and tumor necrosis factor-related protein 7). These SNPs were in high LD (r^2=0.97). The other top two SNPs meeting genome-wide significance were intergenic and located on chromosomes 11 and 13. Alleles A/G were found to be associated with CD symptom count or CD case status. [http://www.downstate.edu/hbnl/documents/2010-Dick-Genome-wideassociationstudyof...<br>...more... (hide)<br>[GWAS:Conduct disorder (case status)] |
| rs6994992(C;T)<br>Magnitude: 1<br>References:16 | Carrier for rs6994992(T;T) which one report has associated with both creativity and schizophrenia<br>...more... (hide)<br>also known as SNP8NRG243177 Initial reports of a link to schizophrenia remain inconclusive. paper implicated in schizophrenia effects white matter density and integrity schizophrenia risk-associated The neuregulin 1 promoter polymorphism rs6994992 is "'not"' associated with neurocognition or chronic schizophrenia in a study of 738 patients T allele (in a per allele fashion) significantly associated with lateral ventricle volume in 95 first-episode schizophrenics points out that their result is that (T) was weakly associated with schizophrenia, however this is the opposite of the original association study where the C allele. [OMIM:NEUREGULIN 1; NRG1] |