gs145 (http://www.snpedia.com/index.php/gs145)

Female Female.

4	Magnitude
20151104	Geno time

rs4244285 (http://www.snpedia.com/index.php/rs4244285)(A;A) (http://www.snpedia.com/index.php/rs4244285(A;A))

poor metabolizer of several popular medicines; patients prescribed Plavix get less benefit, and have higher risk for adverse cardiovascular events

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...

more info (http://www.snpedia.com/index.php/rs4244285)

Bad	Repute
4	Magnitude
Drug response	ClinVar Significance
4.8%	Frequency
0.1983	GMAF
30	References
CYP2C19	Gene
10	Chromosome
94781859	Position
4	Max Magnitude
20160329	Rs time
plus	Stabilized
plus	Orientation

Topics CYP2C19 CYP2D6 Coriell Personalized Medicine Collaborative Drugbank PharmGKB Pharma ADME Pharma DMET

Medicines Clopidogrel Esomeprazole Lansoprazole Losec Nexium Omeprazole Plavix Prevacid Prilosec Proguanil

Medical Conditions Ulcer

ClinVar **Drug response** Mephenytoin Proguanil Clopidogrel response

rs1333049 (http://www.snpedia.com/index.php/rs1333049)(C;C) (http://www.snpedia.com/index.php/rs1333049(C;C))

1.9x increased risk for CAD

rs1333049 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 most likely (C); the odds ratio associated with heterozygotes is 1.47 (CI 1.27-1.70), and for homozygotes, 1.9 (CI 1.61-2.24). This SNP has also been reported to have the highest association of any SNP studied in a subsequent experiment conducted with the resources of the German MI [Myocardial Infarction] Family Study. The initial studies were conducted on Caucasian populations. A subsequent study of Japanese and Korean patients has also found rs1333049 to be associated with increased coronary artery disease risk, with roughly similar odds ratios. '"Further reading (with comments)"' - A long-term study of a cohort of 76...

more info (http://www.snpedia.com/index.php/rs1333049)

Bad	Repute
4	Magnitude
20.4%	Frequency
0.4334	GMAF
97	References
9	Chromosome
22125504	Position
4	Max Magnitude
20160311	Rs time
plus	Stabilized
plus	Orientation
	ambig (http://www.snpedia.com/index.php/Ambiguous_flip)

Topics Coriell Personalized Medicine Collaborative

Medical Conditions Coronary artery calcification | Coronary artery disease | Heart disease | Stroke

gs152 (http://www.snpedia.com/index.php/gs152)

CYP2C19 Poor Metabolizer CYP2C19 Poor Metabolizer. Your body breaks down some medicines more slowly than most people. As a result, you may need a lower than average dosage of the medicines metabolized by CYP2C19 in order to decrease adverse effects, or a higher than average dose to increase efficacy of drugs that require metabolization by CYP2C19 to become active. One example of a drug requiring bioconversion by CYP2C19 is Clopidogrel (Plavix); CYP2C19 poor metabolizers may require higher Clopidogrel dosing or the use of another thienopyridine such as prasugrel. [http://jama.ama-assn.org/cgi/content/full/302/8/849]

Bad	Repute
3.5	Magnitude
20131019	Geno time

Topics CYP2C19

Medicines Clopidogrel Plavix

rs12777823 (http://www.snpedia.com/index.php/rs12777823)(A;A) (http://www.snpedia.com/index.php/rs12777823(A;A))

Avoid Plavix, higher risk for adverse cardiovascular events. CYP2C19 poor metabolizer. poor metabolizer of several popular medicines; Neighboring rs4244285 is a more reliable predictor, but this genotype is linked to being a poor metabolizer of several popular medicines; patients prescribed Plavix (Clopidogrel) have higher risk for adverse cardiovascular events.

JAMA rs12777823 and rs4244285 implicated in platelet response to clopidogrel [PharmGKB:Curated A GWAS found this variant (rs12777823) was the most significantly associated SNP with clopidogrel response. This SNP was in strong linkage disequilibrium with 12 further SNPs within the CYP2C18-CYP2C19-CYP2C9-CYP2C8 cluster (rs12777823; rs10109204; rs2025445; rs1326837; rs717238; rs2860838; rs2860903; rs9332105; rs9332113; rs12572351; rs10509679; rs1934951; rs1934680) and also with the loss-of-function variant CYP2C19*2 (rs4244285). Further findings in this study showed that the CYP2C19*2 genotype accounted for most of all the initially found association with diminished platelet response to clopidogrel.]

more info (http://www.snpedia.com/index.php/rs12777823)

Bad	Repute
3.5	Magnitude
1.8%	Frequency
0.2213	GMAF
3	References
LOC100130970	Gene
10	Chromosome
94645745	Position
3.5	Max Magnitude
20150101	Rs time
plus	Stabilized
plus	Orientation

Topics PharmGKB

Medicines Clopidogrel

rs2243093 (http://www.snpedia.com/index.php/rs2243093)(C;C) (http://www.snpedia.com/index.php/rs2243093(C;C))

A case-control study of Chinese Han patients reported that the rs2243093 (C;C) genotype has an increase risk for coronary heart disease. A meta-analysis was performed and found that there is a strong association with risk of ischemic stroke, however, the authors suggest that this polymorphism may actually be linked with another (unknown) locus that is the culprit responsible for the increase in ischemic stroke risk.

Resides within the platelet glycoprotein (1bα) gene. The rs2243093 variant (C;C) was found to increase the odds of coronary heart disease in a clinical Chinese Han population. The (C;C) genotype is also associated with a risk of ischemic stroke. Rs2243093 is also classified in the literature as the -5T/C, Kozak sequence polymorphism. A case-control study of 246 Chinese Han patients (with preexisting coronary heart disease) reported that in comparison to the '(T;T)' genotype, the '(C;C)' genotype is associated with an increased risk for coronary heart disease with an odds ratio of 3.41 (CI: 1.19-9.75, p<0.029). A meta-analysis was performed in order to test the hypothesis that genetic variation of the platelet glycoprotein 1bα gene(which includes rs2243093 Kozak variant (n=1984...

more info (http://www.snpedia.com/index.php/rs2243093)

Bad	Repute
3.4	Magnitude
0.9%	Frequency
0.1837	GMAF
2	References
GP1BA	Gene
17	Chromosome
4932600	Position
3.4	Max Magnitude
20150103	Rs time
plus	Stabilized
plus	Orientation

Topics PharmGKB

Medical Conditions Coronary artery disease

gs227 (http://www.snpedia.com/index.php/gs227)

fully heterozygous, age related tasting variation You are heterozygous at all 3 of the SNPs which are known to influence the ability to taste bitterness. This means you are better than average at detecting bitter tastes while young, but that this ability will decrease to less than average during adulthood. As a child you will probably hate brussel sprouts, and by early adulthood will discover that olives and brussel sprouts now taste good. A 2010 study shows the change bitter sensitivity which occurs over the lifespan (from bitter sensitive to less so) is more common in people with this genoset. Children with this genotype could perceive a bitter taste at lower PROP concentrations than could heterozygous adults. The threshold for adolescents was intermediate. The 3 SNPs are rs10246939, rs1726866, rs713598 in the gene TAS2R38.

3	Magnitude
20120426	Geno time

Medical Conditions Taste

gs241 (http://www.snpedia.com/index.php/gs241)

lighter green, brown or hazel eye color You have a variant linked to blue eye color, but also less common eye color variations. This is common among western europeans with green, light brown or hazel eye color. This spreadsheet is gathering more information on eye color and genotypes. See gs237 for one version of blue eyes.

3	Magnitude
20150430	Geno time

Topics Eye color

rs3918290 (http://www.snpedia.com/index.php/rs3918290)(A;G) (http://www.snpedia.com/index.php/rs3918290(A;G))

some 5-fluorouracil toxicity You can expect some toxicity from 5-fluorouracil, a common treatment for various cancers. This variation does not influence your risk of cancer.

23andMe reports that the A allele of rs3918290 is associated with the rare recessive disorder dihydropyrimidine dehydrogenase deficiency (DPD), also known as hereditary thymine-uraciluria or familial pyrimidinemia. Note that the terms used in the literature for this gene can be confusing, since the gene is in reverse orientation to the chromosome strand on the reference genome. pharmgkb - defines *2A allele, which has a significantly higher chance of 5-fluorouracil toxicity Fluorouracil Toxicity [PharmGKB:In-Depth Heterozygosity for the A allele of this SNP is associated with mucositis and leukopenia drug toxicity in cancer patients treated with fluorouracil. Heterozygous males were more likely than heterozygous females to develop severe toxicity. In this study, no A/A homozygotes were ob...
more info (http://www.snpedia.com/index.php/rs3918290)

Bad	Repute
3	Magnitude
Pathogenic	ClinVar Significance
0.9%	Frequency
0.002755	GMAF
8	References
DPYD	Gene
1	Chromosome
97450058	Position
3.5	Max Magnitude
20160325	Rs time
minus	Stabilized
minus	Orientation

Topics Drugbank PharmGKB Pharma ADME Pharma DMET

Medicines 5-fluorouracil Fluoropyrimidines

ClinVar Pathogenic Dihydropyrimidine dehydrogenase deficiency Fluorouracil response not provided Hirschsprung disease 1

rs3184504 (http://www.snpedia.com/index.php/rs3184504)(T;T) (http://www.snpedia.com/index.php/rs3184504(T;T))

increased risk for celiac disease increased resistance to bacterial infections

rs3184504 is a nonsynonymous SNP in the SH2B3 gene, and it is also known as R262W. In a recent (2008) study of non-HLA SNP associations of 1600+ celiac disease patients, this SNP was considered one of the most significant. The odds ratio for the minor allele was 1.19 (CI:1.10-1.30, p=1.33x10e-7). rs653178, another SNP in strong linkage disequilibrium (r²>0.99) with rs3184504, was also associated with celiac disease. associated with type-1 diabetes 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 23andMe blog blood pressure Pharmacogenetic implications for eight common blood pressure-associated...

more info (http://www.snpedia.com/index.php/rs3184504)

Bad	Repute
3	Magnitude
21.4%	Frequency
0.2181	GMAF
32	References
SH2B3	Gene
12	Chromosome
111446804	Position
3	Max Magnitude
20151207	Rs time
plus	Stabilized
plus	Orientation

Topics PharmGKB

Medical Conditions Blood pressure | Celiac disease | Coronary artery disease | Hypothyroidism | Myocardial infarction | Rheumatoid Arthritis | Type-1 diabetes