

## GUIDELINE

A gene variant report is a graphical representation of genetic raw data, displayed as a color coded chart. Phenotypes are determined based on the presence or absence of variant alleles in your genotype. Alleles are considered variant if they are the minor allele i.e they occur with less frequency (GMAF) in the default global population. Having "no variant" alleles (green) is not necessarily "normal" or protective, and having a homozygous phenotype (red) is not always "abnormal". What is a normal or abnormal phenotype should NOT be determined solely based on this variant report. Please refer to the table below to explore this issue further.

The significance of your phenotypes should be assessed by reviewing related genome wide studies for context and in consultation with a qualified health practitioner or nutrigenetics specialist.

	By default, green means that there are no minor alleles or Hemizygous and therefore that there are no risk alleles. Sometimes, however a minor allele can be protective and in these cases it will also be marked as green.
	By default, yellow means that there is one minor allele. Sometimes, two major alleles (common alleles) are yellow if the major allele is the risk allele or Heterozygous.
	Red means that you need to pay attention to this SNP. By default, Red means that there are two minor (less common) alleles and therefore risk alleles or Homozygous. Sometimes, we change a color to Red if there is only one minor allele and one minor allele is associated with negative health outcomes.



# Chemoprotection / Detox

## *Liver Detox Phase I & II*

A lot of toxins – whether eaten, breathed, or created in our bodies – are broken down by the Cytochrome P450 family of enzymes in what is known as phase I of detoxification. Phase I break apart big toxins and makes them into polar molecules. Most of this takes place in the liver; Phase II detoxification involves taking the metabolites of phase I and changing them so that they can be excreted. Phase II binds with the metabolites from Phase I and makes them water-soluble so that they can be excreted. Sometimes the metabolites of phase I are carcinogenic or reactive, so having phase II detox in sync is very important. Understanding your own genes for detoxification can help you prioritize which toxicants are the most harmful to you and which drugs to avoid.

## *Yeast/ Alcohol metabolism*

The alcohol dehydrogenases comprise a group of several isozymes that catalyse the oxidation of primary and secondary alcohols to aldehydes and ketones, respectively. Alcohol dehydrogenases (ADH) are a group of dehydrogenase enzymes that occur in humans and many other animals and facilitate the interconversion between alcohols and aldehydes or ketones with the reduction of nicotinamide adenine dinucleotide (NAD<sup>+</sup>) to NADH. Thus ADH serve to break down alcohols that otherwise are toxic, and they also participate in generation of useful aldehyde, ketone, or alcohol groups during biosynthesis of various metabolites. In yeast, plants, and many bacteria, some alcohol dehydrogenases catalyze the opposite reaction as part of fermentation to ensure a constant supply of NAD<sup>+</sup>.

## Chemoprotection / Detox

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs2472304	CYP1A2*1F	Liver Detox I	Metabolizes caffeine, Converts estradiol to 2-OH-estradiol. Oxidizes steroids, fatty acids, and xenobiotics.	AA	--
rs762551	CYP1A2*1F C164A	Liver Detox I	Metabolizes caffeine, Converts estradiol to 2-OH-estradiol. Oxidizes steroids, fatty acids, and xenobiotics. AA= upregulation. CC, AC = downregulation	CC	A
rs1056836	CYP1B1 L432V	Liver Detox I	Hydroxylation of Estrogens	CC	C
rs1800440	CYP1B1 N453S	Liver Detox I	Estrogen Metabolism. Converts estradiol to 4-OH-estradiol	TT	T
rs9282671	CYP1B1 T241A	Liver Detox I	Hydroxylation of Estrogens	AA	A
rs1801272	CYP2A6*2 A1799T	Liver Detox I	Xenobiotics, nicotine, drugs metabolism. Works with CYP2B6	TT	A
rs12767583	CYP2C19 C5709T	Liver Detox I	Drugs metabolism	TT	T
rs4986894	CYP2C19 T98C	Liver Detox I	Drugs metabolism	TT	T
rs4986893	CYP2C19*3 G636A	Liver Detox I	Drugs metabolism	AA	G
rs41291556	CYP2C19*8 T358C	Liver Detox I	Drugs metabolism	CC	T
rs9332239	CYP2C9*12 1465C>T	Liver Detox I	Warfarin, ARBs, and NSAIDs metabolism	TT	C
rs1057910	CYP2C9*3	Liver Detox I	Warfarin, ARBs, and NSAIDs metabolism	CC	A
rs9332131	CYP2C9*6 818delA	Liver Detox I	Warfarin, ARBs, and NSAIDs metabolism	DD	I
rs28371706	CYP2D6*17 T107I	Liver Detox I	Drugs metabolism: Codeine, Morphine, Tramadol, Risk = lower activity	AA	G
rs2031920	CYP2E1_-1055C>T G1055T	Liver Detox I	Xenobiotics, drugs, carcinogens metabolism. Works with GST and other CYP genes.	TT	C
rs2740574	CYP3A4*1B A392G	Liver Detox I	Androgens, Xenobiotics, drugs metabolism, testosterone oxidation	GG	T



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SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs1799945	HFE	Liver Detox I	Human Hemochromatosis protein. Regulates iron absorption. This is lower impact gene	GG	C
rs1800562	HFE	Liver Detox I	Human Hemochromatosis protein. Regulates iron absorption. This is the key gene, increasing risk.	AA	G
rs662	PON1 Q192R	Liver Detox I	Organophosphates detox, Oxidation of LDL Cholesterol (CC being risk for LDL oxidation), Homocysteine to Homocysteine Thiolactone	TT	T
rs2472304	rs2472304	Liver Detox I	Metabolizes caffeine, Converts estradiol to 2-OH-estradiol. Oxidizes steroids, fatty acids, and xenobiotics.	AA	--
rs1049793	ABP1/DAO C1933G	Liver Detox II	Extracellular and GI Histamine deactivation, removal of Di-amino acids (associated with aging), NSAID sensitivity.	GG	--
rs3741049	ACAT1 G22670A	Liver Detox II	Dietary fat and protein breakdown	AA	A
rs73598374	ADA G22A	Liver Detox II	Adenosine to Inosine, Purine metabolism	TT	--
rs1042026	ADH1B	Liver Detox II	Alcohol and lipid peroxidation products metabolism. Converts alcohol to acetaldehyde. (Zn)	CC	--
rs6413413	ADH1B A178T	Liver Detox II	Alcohol and lipid peroxidation products metabolism. Converts alcohol to acetaldehyde. (Zn)	AA	T
rs2066702	ADH1B A396C	Liver Detox II	Alcohol and lipid peroxidation products metabolism. Converts alcohol to acetaldehyde. (Zn)	AA	--
rs737866	COMT/TXNRD2 A4251G	Liver Detox II	Adrenaline, Dopamine, Estrogen metabolism (Mg, SAME) Mind the CYP1B1 upregulation, glucuronidation and sulfation.	CC	T
rs2020917	COMT/TXNRD2 C4622T	Liver Detox II	Adrenaline, Dopamine, Estrogen metabolism (Mg, SAME) Mind the CYP1B1 upregulation, glucuronidation and sulfation.	TT	--
rs3741775	DAO	Liver Detox II	Extracellular and GI Histamine deactivation, removal of Di-amino acids (associated with aging), NSAID sensitivity.	CC	--
rs3918347	DAO	Liver Detox II	Extracellular and GI Histamine deactivation, removal of Di-amino acids (associated with aging), NSAID sensitivity.	GG	A
rs2273684	GSS	Liver Detox II	Glutathione Synthesis	GG	T
rs6060124	GSS	Liver Detox II	Glutathione Synthesis	AA	A

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SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs6088659	GSS A5997G	Liver Detox II	Glutathione Synthesis	TT	C
rs1056806	GSTM1	Liver Detox II	Conjugation of Glutathione and toxins	TT	--
rs7483	GSTM3	Liver Detox II	GSTM3 encodes glutathione S-transferase that functions in the detoxification of carcinogens, drugs, toxins, and products of oxidative stress. Mutations of this gene can lead to various cancers. minor "T" allele = lower levels of GSTM3 protein.Reduced antioxidant defense causing oxidative stress in the body. Alzheimer's. Higher levels of pepsinogen, a precursor for pepsin which is used to digest proteins in the stomach.	TT	T
rs1138272	GSTP1	Liver Detox II	conjugation of Glutathione and toxins	TT	C
rs1695	GSTP1	Liver Detox II	conjugation of Glutathione and toxins.Persons having the alleles AA orAG had an increase ininflammatory interleukin-6 (IL-6)upon supplementingalpha-tocopherol (the mostcommon form of Vitamin E in aNorth American diet) while thosewith GG saw a decrease	GG	A
rs2073440	HDC	Liver Detox II	Histidine to Histamine	GG	--
rs901865	HRH1	Liver Detox II	Histamine Receptor	TT	T
rs11662595	HRH4	Liver Detox II	Histamine Receptor	GG	A
rs16940765	HRH4	Liver Detox II	Histamine Receptor	CC	T
rs4800573	HRH4	Liver Detox II	Histamine Receptor	AA	G
rs1041983	NAT2	Liver Detox II	Acetylation, xenobiotic metabolizing enzyme involved in the biotransformation of many aromatic amines and heterocyclicamines.	TT	C
rs1208	NAT2	Liver Detox II	Acetylation, xenobiotic metabolizing enzyme involved in the biotransformation of many aromatic amines and heterocyclicamines.	GG	A
rs1799929	NAT2	Liver Detox II	Acetylation, xenobiotic metabolizing enzyme involved in the biotransformation of many aromatic amines and heterocyclicamines.	TT	C
rs1799930	NAT2	Liver Detox II	Acetylation, xenobiotic metabolizing enzyme involved in the biotransformation of many aromatic amines and heterocyclicamines.	AA	G

## Chemoprotection / Detox

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs1799931	NAT2	Liver Detox II	Acetylation, xenobiotic metabolizing enzyme involved in the biotransformation of many aromatic amines and heterocyclicamines.	AA	G
rs1801279	NAT2	Liver Detox II	Acetylation, xenobiotic metabolizing enzyme involved in the biotransformation of many aromatic amines and heterocyclicamines.	AA	G
rs10183914	NFE2L2 or Nrf2	Liver Detox II	Nuclear factor (erythroid-derived 2)-like 2, also known as NFE2L2 or Nrf2, is a protein (transcription factor). Nrf2 increases the expression of antioxidant proteins that protect against oxidative damage triggered by injury and inflammation. Substances that stimulate the Nrf2 pathway are being studied for the treatment of diseases that are caused by oxidative stress. It responds to injury and inflammation. <a href="http://www.ncbi.nlm.nih.gov/gene/4780">http://www.ncbi.nlm.nih.gov/gene/4780</a> Transcription activator that binds to antioxidant response (ARE) elements in the promoter regions of target genes. Important for the coordinated up-regulation of genes in response to oxidative stress. May be involved in the transcriptional activation of genes of the beta-globin cluster by mediating enhancer activity of hypersensitive site 2 of the beta-globin locus control region. Alternative names: NF-E2-related factor 2 NFE2-related factor 2 Nuclear factor, erythroid derived 2, like 2 It's better to have this gene increased most of the time.	CC	--
rs1962142	NRF2	Liver Detox II	Promoter region, Stimulation of anti - oxidative enzymes	AA	G
rs35652124	NRF2	Liver Detox II	Promoter region, Stimulation of anti - oxidative enzymes	TT	--
rs6726395	NRF2	Liver Detox II	Promoter region, Stimulation of anti - oxidative enzymes	AA	G
rs2547231	SULT2A1	Liver Detox II	Sulfation, catalyses the sulfation of steroids in adrenal glands and bile acids in the liver.	CC	A
rs887829	UGT1A1	Liver Detox II	Glucuronidation, conversion of unconjugated bilirubin(toxic) to conjugated bilirubin. Xenobiotics metabolism/removal.	TT	T
rs6742078	UGT1A1 G179250T	Liver Detox II	Glucuronidation, conversion of unconjugated bilirubin(toxic) to conjugated bilirubin. Xenobiotics metabolism/removal.	TT	T
rs4148323	UGT1A1 G211A	Liver Detox II	Glucuronidation, conversion of unconjugated bilirubin(toxic) to conjugated bilirubin. Xenobiotics metabolism/removal.	AA	G
rs4244593	PEMT	Membranes permeability	phosphatidylethanolamine to phosphatidylcholine	TT	T



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SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs7946	PEMT	Membranes permeability	phosphatidylethanolamine to phosphatidylcholine	TT	T
rs480575	CAT A12175G	Oxidative Stress	H2O2 reduction	GG	A
rs2300181	CAT C21068T	Oxidative Stress	H2O2 reduction	TT	T
rs4626565	COX6C	Oxidative Stress	Last enzyme in Electron Transport Chain. ROS scavenging.	CC	T
rs1050828	G6PD	Oxidative Stress	makes substrate for GSR, important for oxidised Glutathione reduction.	TT	C
rs1050829	G6PD	Oxidative Stress	makes substrate for GSR, important for oxidised Glutathione reduction.	CC	T
rs5030868	G6PD	Oxidative Stress	makes substrate for GSR, important for oxidised Glutathione reduction.	AA	G
rs8007267	GCH1 C36378991T	Oxidative Stress	GCH1 haplotype determines vascular and plasma biopterin availability in coronary artery disease effects on vascular superoxide production and endothelial function	TT	C
rs10483639	GCH1 G55306457C	Oxidative Stress	GCH1 haplotype determines vascular and plasma biopterin availability in coronary artery disease effects on vascular superoxide production and endothelial function.	CC	G
rs2551715	GSR	Oxidative Stress	Oxidised Glutathione reduction	TT	T
rs1799836	MAOB	Oxidative Stress	Oxidation (deactivation) Dopamine, Adrenaline, Noradrenaline (less Serotonin). T= upregulation.	TT	T





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SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs10183914	NFE2L2 or Nrf2	Oxidative Stress	Nuclear factor (erythroid-derived 2)-like 2, also known as NFE2L2 or Nrf2, is a protein (transcription factor). Nrf2 increases the expression of antioxidant proteins that protect against oxidative damage triggered by injury and inflammation. Substances that stimulate the Nrf2 pathway are being studied for the treatment of diseases that are caused by oxidative stress. It responds to injury and inflammation. <a href="http://www.ncbi.nlm.nih.gov/gene/4780">http://www.ncbi.nlm.nih.gov/gene/4780</a> Transcription activator that binds to antioxidant response (ARE) elements in the promoter regions of target genes. Important for the coordinated up-regulation of genes in response to oxidative stress. May be involved in the transcriptional activation of genes of the beta-globin cluster by mediating enhancer activity of hypersensitive site 2 of the beta-globin locus control region. Alternative names: NF-E2-related factor 2 NFE2-related factor 2 Nuclear factor, erythroid derived 2, like 2 It's better to have this gene increased most of the time.	CC	--
rs1800566	NQO1	Oxidative Stress	Breaks down superoxides and peroxides. Directly scavenges superoxide. Activates vitamin K.	AA	A
rs1962142	NRF2	Oxidative Stress	Promoter region, Stimulation of anti - oxidative enzymes	AA	G
rs35652124	NRF2	Oxidative Stress	Promoter region, Stimulation of anti - oxidative enzymes	TT	--
rs6726395	NRF2	Oxidative Stress	Promoter region, Stimulation of anti - oxidative enzymes	AA	G
rs854571	PON1	Oxidative Stress	Oxidised lipids removal	TT	C
rs662	PON1 Q192R	Oxidative Stress	Oxidised lipids removal	TT	T
rs4880	SOD2	Oxidative Stress	Superoxide Reduction (Mn, Zn, Cu)	GG	A
rs2273697	ABCC2	Phase III Detox	Intestinal Toxic Conjugates Transport	AA	A
rs717620	ABCC2	Phase III Detox	Intestinal Toxic Conjugates Transport	TT	C
rs8187710	ABCC2	Phase III Detox	Intestinal Toxic Conjugates Transport	AA	G



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SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs3741049	ACAT1 G22670A	Phase III Detox	Cholesterol synthesis to Bile production	AA	A
rs3733890	BHMT	Phase III Detox	Bile production, Methylation	AA	G
rs567754	BHMT-02	Phase III Detox	Bile production, Methylation	TT	T
rs1405655	LXR	Phase III Detox	LXR increases xenobiotic transport proteins, such as MDR1 (ABCB1) and MRP2 (ABCC2)	TT	T
rs17373080	LXR	Phase III Detox	LXR increases xenobiotic transport proteins, such as MDR1 (ABCB1) and MRP2 (ABCC2)	GG	--
rs2695121	LXR	Phase III Detox	LXR increases xenobiotic transport proteins, such as MDR1 (ABCB1) and MRP2 (ABCC2)	TT	--
rs4244593	PEMT	Phase III Detox	Hepatic PC utilisation, secretion of bile into the intestine.	TT	T
rs7946	PEMT	Phase III Detox	Hepatic PC utilisation, secretion of bile into the intestine.	TT	T
rs1544410	VDR Bsm1	Phase III Detox	Bile Production	TT	C
rs1042026	ADH1B	Yeast/Alcohol Metabolism	Members of the alcohol dehydrogenase family break down a wide variety of things, including alcohol, vitamin A/retinol, other aliphatic alcohols, hydroxysteroids, and lipid peroxidation products	CC	--
rs6413413	ADH1B A178T	Yeast/Alcohol Metabolism	Members of the alcohol dehydrogenase family break down a wide variety of things, including alcohol, vitamin A/retinol, other aliphatic alcohols, hydroxysteroids, and lipid peroxidation products. combinations of this SNP and others were shown to affect alcohol metabolism; most of these combinations are represented by the gs211 genoset.	AA	T
rs2066702	ADH1B A396C	Yeast/Alcohol Metabolism	Members of the alcohol dehydrogenase family break down a wide variety of things, including alcohol, vitamin A/retinol, other aliphatic alcohols, hydroxysteroids, and lipid peroxidation products	AA	--
rs2238151	ALDH2	Yeast/Alcohol Metabolism	Joint effects of alcohol consumption and polymorphisms in alcohol and oxidative stress metabolism genes on risk of head and neck cancer	TT	C
rs671	ALDH2	Yeast/Alcohol Metabolism	A allele significant contributor to alcohol flush and increased hangover effects. A allele is most common in Asian populations.	AA	G

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SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs72547566	ALDH3A2	Yeast/Alcohol Metabolism	.	TT	--
rs769214	CAT				--
rs769217	CAT C14185T				T



# Diet & Nutrition



Diet & Nutrition

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs4148211	ABCG8	Cholesterol Status	The G (minor) allele is associated with: Increased total cholesterol levels Increased levels of LDL cholesterol but Decreased risk of developing type 2 diabetesThe A (major) allele is associated with: Increased levels of triglycerides (AA).Low levels of LDL cholesterol (AA).but Increased risk of developing type 2 diabetes (AA).	GG	A
rs3213445	CPT1A	Cholesterol Status	The Minor "C" allele is associated with:Carriers of the rare allele had significantly higher glutamyl transpeptidase (GGT), glutamic oxaloacetic transaminase (GOT) and glutamic pyruvate transaminase (GPT) activities.A higher fatty liver index.Triglycerides and fasting glucose were significantly higher in C allele carriers.Insulin sensitivity was lower in C allele carriers.Total cholesterol and LDL-cholesterol tended to be higher in C allele carriers.	CC	T
rs482548	FADS2	Cholesterol Status	(TT) Lower than common level of FADS enzyme activity	TT	C
rs4846914	GALNT2	Cholesterol Status	The A (minor) allele is associated with increased levels of HDL cholesterol (good cholesterol) but also is associated with higher risk of hypertriglyceridemia.	GG	A
rs1800961	HNF4A	Cholesterol Status	The Minor "T" allele is associated with: Decreased HDL cholesterol levels due to lower HNF4A levels.	TT	C
rs1884614	HNF4A	Cholesterol Status	The C (major) allele is associated with increased HDL cholesterol levels.The Minor "T" allele is associated with lower levels of HFN4A protein (potentially lower HDL cholesterol)	TT	T
rs1137100	LEPR	Cholesterol Status	G = Higher total cholesterol, (highest in GG).The Major "A" allele is associated with decreased overall cholesterol.	GG	--
rs17700633	MC4R	Cholesterol Status	The Minor "A" allele is associated with decreased HDL cholesterol .The G (major) allele is associated with increased HDL cholesterol.	AA	--
rs2229616	MC4R	Cholesterol Status	The C (major) allele is associated with decreased HDL cholesterol levels.	CC	--
rs174537	MYRF (FADS1)	Cholesterol Status	The G (major) allele is associated with increased LDL cholesterol (bad cholesterol) levels.The T (minor) allele is associated with decreased LDL cholesterol (bad cholesterol) levels.	GG	G
rs7946	PEMT	Cholesterol Status	The T (minor) allele is associated with possible decreased levels of HDL cholesterol (good cholesterol).	TT	T
rs8192678	PGC-1a Gly482Ser	Cholesterol Status	The T (minor) allele is associated with increased percentage of small dense-LDL particles.	TT	C

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SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs2016520	PPARD	<b>Cholesterol Status</b>	The T (major) allele is associated with increased cholesterol levels.	TT	T
rs646776	PSRC1	<b>Cholesterol Status</b>	The C (minor) allele is associated with increased levels of LDL (bad cholesterol).	CC	T
rs659366	UCP2	<b>Cholesterol Status</b>	The "T" (minor) allele is associated with:More UCP2 production and higher levels of UCP2 protein Increased levels of HDL-Cholestrol and decreased levels of LDL-Cholestrol in a Chinese population.Reduced LDL particle size levels in a Chinese population.Increased energy expenditure.Decreased lipid oxidation Decreased high-sensitivity Cholesterol reactive protein (hs-CRP), a biomarker of inflammation. This decrease may indicate a lower level of inflammation in the body.More difficulty in losing weight under a very low-calorie program in Korean women.Higher fasting glucose levels in the blood.Higher risk of Diabetes (Type 2).Increased hardening of the arteries in women.Increased heart disease risk. Increased resting energy expenditure in obese children and increased 24-hour energy expenditure in Pima Indians.	CC	C
rs2073658	USF1	<b>Cholesterol Status</b>	The T (minor) allele is associated with increased LDL cholesterol.	TT	C
rs3733890	BHMT	<b>Choline Status</b>	GG (major) = higher chance to die of breast cancer. Choline may help reduce this risk. <a href="http://www.ncbi.nlm.nih.gov/pubmed/19635752?dopt=Abstract">http://www.ncbi.nlm.nih.gov/pubmed/19635752?dopt=Abstract</a>	GG	G
rs2236225	MTHFD1	<b>Choline Status</b>	G = Decreased risk of choline deficiency.	AA	A
rs1805087	MTR	<b>Choline Status</b>	A = Less choline is needed by the body.	GG	A
rs738409	PNPLA3	<b>Choline Status</b>	T (minor) = higher risk of fatty liver - encourage more Choline.Whites with fatty liver - which may be due to low choline - were more likely to carry the T allele.	TT	C
rs738409	PNPLA3	<b>Choline Status</b>	C = Decreased need for choline in the body.	GG	C
rs11605924	CRY2	<b>High Fat Diet, Ketogenic Diet</b>	A (major) = Reduction in energy utilisation in a high fat diet.	AA	A
rs1800562	HFE	<b>Iron Status</b>	G (major) = Decreased levels of Iron in the blood. The A allele has an association with higher risk for hemochromatosis.	AA	G



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SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs3811647	TF	Iron Status	A = Increased Iron levels in the blood.	AA	A
rs855791	TMPRSS6	Iron Status	A = Increased iron levels in blood.	AA	A
rs182549	MCM6	Lactose Tolerance	MCM6 has influence on the lactase LCT gene, rs182549 and 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. The T (minor) allele is associated with increased ability to digest lactose.	CC	C
rs4988235	MCM6	Lactose Tolerance	MCM6 has influence on the lactase LCT gene, rs182549 and 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. AA = Can digest milk .AG = Likely to be able to digest milk as an adult.GG = Likely lactose intolerant	GG	G
rs17300539	ADIPOQ	Monounsaturated Fat	The G (major) allele is associated with:Worse response to monounsaturated fat intake.GG = Higher BMI with increased monounsaturated fat intake (avoid monounsaturated fats)	GG	--
rs174537	MYRF (FADS1)	Omega fatty acids	The G (major) allele is associated with:Possible decreased need for fish oil supplements to maintain healthy omega-3 fatty acid levelsIncreased levels of arachidonic acid in the body [R]. Arachidonic acid is an Omega-6 polyunsaturated fatty acid.TT = Possible increased need for fish oil supplements to maintain healthy omega-3 fatty acid levels.TT = Lower levels of arachidonic acid in the body. Arachidonic acid is an Omega-6 polyunsaturated fatty acid.	TT	G
rs9939609	FTO	Response to Carbohydrates	Each A= increases hunger and caloric intake, but not energy expenditure.Obesity.Less brown fat and more white fat.FTO correlates with galanin, an orexigenic peptide.	AA	A
rs2338104	KCTD10	Response to Carbohydrates	GG (major) = lower LDL (bad cholesterol) if high carb intake.	CC	G
rs894160	PLIN1	Response to Carbohydrates	CC (major) =does worse with a high complex carb diet with regard to weight.People with the "T" or minor allele do better with complex carbs and worse with saturated fat. The T allele helps protects from weight gain from a higher complex carb diet, but causes weight gain on a lower carb complex carb diet and insulin resistance on a higher saturated fat diet.	CC	--
rs1799986	LRP1	Saturated Fat	The C (major) does better with saturated fat (decreased weight gain)The AG genotype is associated with normal saturated fat levels.	TT	C



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SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs744166	STAT3	Saturated Fat	G (minor) does worse with saturated fat (increased risk of abdominal obesity).	GG	A
rs8069645	STAT3	Saturated Fat	AA does better with saturated fat (decreased risk of abdominal obesity)The G (minor) allele is associated with:Increased levels of STAT3 proteinIncreased risk of abdominal obesity	GG	A
rs7579	CCDC152	Selenium Status	TT = Higher levels of Selenium in subjects with a high BMI.	TT	C
rs2072560	APOA5	Triglyceride Status	C = Lower levels of triglycerides.T = Higher levels of triglycerides.	TT	--
rs2075291	APOA5	Triglyceride Status	CC = Decreased risk of high triglyceride levels.AA = Greater than 4x the risk for high triglyceride levels (n=Asian populations) AC = 4x higher risk for high triglyceride levels (n=Asian populations)	AA	C
rs2266788	APOA5	Triglyceride Status	A = Decreased triglyceride levels.	GG	A
rs3135506	APOA5	Triglyceride Status	GG = Decreased triglyceride levels.CC = 34% Increase in triglyceride concentration.	CC	G
rs651821	APOA5	Triglyceride Status	TT = Decreased triglyceride levels.CC = Higher triglycerides.	CC	T
rs662799	APOA5	Triglyceride Status	AA = Decreased triglyceride levelsEach G=higher triglycerides	GG	A
rs3213445	CPT1A	Triglyceride Status	T = Decreased triglyceride levels.Triglycerides and fasting glucose were significantly higher in C allele carriers.	CC	T
rs174570	FADS2	Triglyceride Status	CC = Normal levels of triglycerides.TT = Increased triglycerides.CT = Increased triglycerides.	TT	C
rs13223993	IGFBP3	Triglyceride Status	A = Increased triglyceride levelsG = Decreased triglycerides and increased HDL, lowering the risk of cardiovascular disease and obesity	AA	A
rs6420424	BCMO	Vitamin A metabolism	GG= reduced BCMO1 activity by 59%, which may reduce the amount of retinol being made	GG	G
rs6564851	BCMO	Vitamin A metabolism	TT= reduced BCMO1 activity by 48%.T allele= lower levels of vitamin A.The G allele may affect health negatively by reducing the availability of vitamin A (retinol) but also positively by increasing the plasma concentration of powerful antioxidant molecules. A number of studies have shown that higher plasma concentrations of antioxidant carotenoids are associated in a protective manner against the development of chronic diseases and disability.	TT	T





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SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs4889294	BCMO1	Vitamin A metabolism	Carotene to retinol conversion	CC	T
rs7501331	BCMO1 A379V	Vitamin A metabolism	Females carrying the T variant had a 32% lower ability to convert Beta-carotene.rs7501331(T)+ rs12934922(T)= 69% lower conversion of Beta-carotene to retinyl esters... The BCMO1 gene (rs12934922and rs7501331) is known to influence the conversion of beta-carotene to vitamin A.TT= 57% reduced conversion activity of beta-carotene (In test tubes)	TT	C
rs492602	FUT2	Vitamin B 12 Status	GG (minor) = Normal absorption of vitamin B12. Women homozygous for the G allele had higher B (12) levels in the blood. This is thought to be caused by increased B12 absorption.TT = Possible malabsorption of vitamin B12 from the GI tract.Absorption of B12 requires the secretion of intrinsic factor (IF) from the gastric cells, binding of IF to vitamin B12 and a functional gastrointestinal absorption system.	TT	A
rs492602	FUT2 A12190G	Vitamin B 12 Status	GG (minor) = Normal absorption of vitamin B12. Women homozygous for the G allele had higher B (12) levels in the blood. This is thought to be caused by increased B12 absorption.TT = Possible malabsorption of vitamin B12 from the GI tract.Absorption of B12 requires the secretion of intrinsic factor (IF) from the gastric cells, binding of IF to vitamin B12 and a functional gastrointestinal absorption system.	TT	A



# GI Health

## GI Health

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs231775	CTLA4	<b>Celiac &amp; Gluten Intolerance</b>	This gene encodes a protein which transmits an inhibitory signal to T cells. Mutations in this gene have been associated autoimmune diseases including: diabetes, Graves disease, thyroid problems, celiac disease, and lupus.	GG	A
rs2858331	HLA	<b>Celiac &amp; Gluten Intolerance</b>	Helps to recognise foreign particles when they enter our body and fight them away in an immune response.	GG	G
rs2187668	HLA-DQA1	<b>Celiac &amp; Gluten Intolerance</b>	Helps to recognise foreign particles when they enter our body and fight them away in an immune response.	TT	C
rs9275224	HLA-DQA2	<b>Celiac &amp; Gluten Intolerance</b>	Helps to recognise foreign particles when they enter our body and fight them away in an immune response.	AA	A
rs3741049	ACAT1 G22670A	<b>Disbiosis</b>	possibly higher tendency for gut dysbiosis (particularly clostridia).	AA	A
rs602662	FUT2	<b>H Antigen</b>	Non-secretors have lower levels of bifidobacteria and would benefit from probiotics	AA	G
rs1049793	ABP1/DAO C1933G	<b>Histamine</b>	Extracellular and GI Histamine deactivation, removal of Di-amino acids (associated with aging), NSAID sensitivity.	GG	--
rs3741775	DAO	<b>Histamine</b>	Extracellular and GI Histamine deactivation, removal of Di-amino acids (associated with aging), NSAID sensitivity.	CC	--
rs3918347	DAO	<b>Histamine</b>	Extracellular and GI Histamine deactivation, removal of Di-amino acids (associated with aging), NSAID sensitivity.	GG	A
rs10156191	DAO C47T or AOC1	<b>Histamine</b>	Protein. Catalyses the degradation of compounds such as putrescine, histamine, spermine, and spermidine, substances involved in allergic and immune responses, cell proliferation, tissue differentiation, tumour formation, and possibly apoptosis. Placental DAO is thought to play a role in the regulation of the female reproductive function. Other names: Diamine oxidase Amiloride-binding protein 1 Amine oxidase copper domain-containing protein 1 Histaminase Kidney amine oxidase KAO	TT	T
rs1800871	IL10-819	<b>Inflammation</b>	A=decreased IL-10..susceptibility to IBD	AA	G
rs2243828	MPO	<b>Microbial defense</b>	MPOD Myeloperoxidase deficiency is a disorder characterized by decreased myeloperoxidase activity in neutrophils and monocytes that results in disseminated candidiasis.	AA	A
rs854571	PON1	<b>Microbial defense</b>	PON1 protects against bacterial infection by destroying the bacterial signalling molecules that cause gram negative bacteria to invade human tissue and form colonies, thus PON1 contributes to the bodies innate immunity. Also associated with IBD.	TT	C



GI Health

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs662	PON1 Q192R	Microbial defense	PON1 protects against bacterial infection by destroying the bacterial signalling molecules that cause gram negative bacteria to invade human tissue and form colonies, thus PON1 contributes to the bodies innate immunity. Also associated with IBD.	TT	T
rs5443	GNB3T (5HT2A)	Stress Response	The T allele is thought to increase second messenger signalling. There are benefits and negative effects to this.IBS. People with genes who produced more 5HT2A receptors were more likely to have IBS.	TT	C
rs6311	HTR2A	Stress Response	The T allele is associated with IBS	TT	C



# Health Report



## *Celiac Disease/Gluten Intolerance*

Celiac disease, sometimes called celiac sprue or gluten-sensitive enteropathy, is an immune reaction to eating gluten, a protein found in wheat, barley and rye. If you have celiac disease, eating gluten triggers an immune response in your small intestine. Over time, this reaction damages your small intestine's lining and prevents it from absorbing some nutrients (malabsorption). The intestinal damage often causes diarrhea, fatigue, weight loss, bloating and anemia, and can lead to serious complications.

## *Clotting Factors*

Coagulation, also known as clotting, is the process by which blood changes from a liquid to a gel, forming a blood clot. It potentially results in hemostasis, the cessation of blood loss from a damaged vessel, followed by repair. The mechanism of coagulation involves activation, adhesion and aggregation of platelets, as well as deposition and maturation of fibrin.



Health Report

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs3741049	ACAT1 G22670A	Blood Glucose	ketone body metabolism	AA	A
rs266729	ADIPOQ	Blood Glucose	Adiponectin is released in the bloodstream from fat cells, where it is involved in the control of fat metabolism and regulating glucose levels .CG was associated with type 2 diabetes in the obese group only OR=2.45. P=0.02, and 59% risk of diabetes could be attributed to that.	GG	--
rs10885122	ADRA2A	Blood Glucose	T=lower fasting glucoseG=higher fasting glucose	GG	G
rs553668	ADRA2A	Blood Glucose	A=increased ADRA2 receptors, higher fasting glucose and diabetes, hypertension, reduced insulin secretion and perhaps obesity. Having a C allele may help in sustaining the endurance training regime to attain a high level of maximal aerobic power in elite endurance athletes	AA	A
rs1405655	LXR	Blood Glucose	LXR helps the secretion of insulin, which helps combat diabetes	TT	T
rs17373080	LXR	Blood Glucose	LXR helps the secretion of insulin, which helps combat diabetes	GG	--
rs2695121	LXR	Blood Glucose	LXR helps the secretion of insulin, which helps combat diabetes	TT	--
rs8192678	PGC-1a Gly482Ser	Blood Glucose	PGC-1 increases energy metabolism and weight loss. This protein may be also involved in controlling blood pressure, regulating cellular cholesterol homoeostasis, and the development of obesity. PGC-1a protects against neurodegenerative diseases. PGC-1 decreases inflammation and insulin resistance. T is the less common allele (codes for serine). This variant causes a lower level of PGC-1a protein including in muscles The T allele results a less efficient coactivator of transcription factors, including those that regulate the PPARGC1A gene itself	TT	C
rs2858331	HLA	Celiac & Gluten Intolerance	Helps to recognise foreign particles when they enter our body and fight them away in an immune response.	GG	G
rs2187668	HLA-DQA1	Celiac & Gluten Intolerance	Helps to recognise foreign particles when they enter our body and fight them away in an immune response.	TT	C
rs9275224	HLA-DQA2	Celiac & Gluten Intolerance	Helps to recognise foreign particles when they enter our body and fight them away in an immune response.	AA	A
rs4961	ADD1	Clotting Factors	The risk (T) allele responded better to diuretics and sodium-restricted diets, in that they tended to lower their blood pressure by ~10 mmHg points compared to rs4961(G;G) homozygotes similarly treated.	TT	G
rs1800775	CETP	Clotting Factors	CETP encodes a protein that is involved in the transfer of cholesteryl ester from high density lipoprotein (HDL) to other lipoproteins.	CC	A



Health Report

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs13146272	CYP4V2	Clotting Factors	A study of 453 VTE cases and 1,327 controls was able to replicate the "mild effects" of this SNP on risk for VTE, however they felt that stronger associations were found between increased VTE risk and either the Factor V Leiden mutation (rs6025) or having blood types O or A2	CC	A
rs3211719	F10	Clotting Factors	This gene encodes a protein called coagulation factor X that helps blood clot after an injury. Mutations can cause deficiency of the protein, which leads to excessive bleeding	GG	A
rs2289252	F11,F11-AS1	Clotting Factors	This gene encodes a protein called coagulation factor XI that helps blood clot after an injury. Mutations can cause deficiency of the protein, which leads to excessive bleeding	TT	T
rs2731672	F12	Clotting Factors	This gene encodes a protein called coagulation factor XII that helps blood clot after an injury. It is involved in the inflammatory response. Mutations can cause deficiency of the protein, which leads to excessive bleeding	TT	C
rs6025	F5	Clotting Factors	The F5 gene encodes a protein that is called coagulation factor V. They that form blood clots to stop bleeding and trigger blood vessel repair after an injury. Mutations can cause abnormal bleeding	TT	C
rs6048	F9	Clotting Factors	This gene encodes a protein called coagulation factor IX, which helps form blood clots. Mutations can cause blood clots to be unable to form and lead to excessive bleeding (hemophilia) or warfarin sensitivity, which can cause severe bleeding problems	GG	A
rs1613662	GP6 Pro219Ser	Clotting Factors	The gene encodes a protein that is a receptor for collagen and plays an important role in collagen- induced platelet aggregation and thrombus formation. Mutations of this gene can cause a bleeding disorder	GG	A
rs9898	HRG Pro204Ser	Clotting Factors		TT	--
rs5918	ITGB3	Clotting Factors	C=increased resistance to blood thinning side effect of aspirin	CC	T
rs1523127	NR1I2	Clotting Factors	This gene encodes a protein that is a receptor and has both a ligand-binding domain and a DNA- binding domain	CC	A
rs2227589	SERPINC1	Clotting Factors	Gene variants associated with deep vein thrombosis	TT	C
rs12273363	BDNF	Cognitive	Neurotrophins are chemicals that help to stimulate and control neurogenesis, BDNF being one of the most active. The C allele is associated with more with mood disorders than T. The minor C allele might increase activity in the amygdala (a region in brain). This might increase anxiety and emotional memory.	CC	T





Health Report

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs6994992	NRG1	Cognitive	Neuregulin 1 is important for synaptic plasticity, inhibiting the amygdala (to shut down anxiety), myelination (Schwann cell maturation, survival, and motility), heart function (cardiac growth factor) and tumor suppression. TT= Decreased activation of frontal and temporal lobe regions. TT= Increased risk of Psychosis. TT=reduced white matter density in the anterior limb of the internal capsule and evidence of reduced structural connectivity in the same region.	TT	T
rs3746544	SNAP25	Cognitive	Synaptosomal-associated protein 25 (SNAP-25) plays a crucial role in the release of neurotransmitters (from presynapse- exocytosis). SNP variants that produce less SNAP25 are associated with less activation in areas of the brain that are critical for the regulation of attention and inhibition (prefrontal cortex). T=presumably lower SNAP25.	TT	T
rs3738401	TSNAX-DISC1, DISC1	Cognitive	This gene encodes a protein that helps neurons grow and assist cortical development through its interaction with other proteins. Mutations are associated with higher risk for schizophrenia.	AA	--
rs823162	TSNAX-DISC1, DISC1	Cognitive	This gene encodes a protein that helps neurons grow and assist cortical development through its interaction with other proteins. Mutations are associated with higher risk for schizophrenia.	CC	T
rs7483	GSTM3	GI Health	GST-M3 activity is possibly involved in protection against mucosal atrophy caused by H. pylori as the levels of IgG titer and PGI are linked to mucosal status.	TT	T
rs4343	ACE G2328A	Kidney Health	Angiotensin I to Angiotensin II, ACE stimulates production of the hormone, aldosterone, which triggers the absorption of salt and water by the kidneys. G=deletion at ACE Del16 and increase of ACE enzyme activity.	GG	A
rs4343	ACE G2328A	Stress Response	Angiotensin I to Angiotensin II, ACE stimulates production of the hormone, aldosterone. Anxiety, decreased learning memory, low frustration threshold, higher anxiety. Made worse by MAO A mutation. I G=deletion at ACE Del16 and increase of ACE enzyme activity.	GG	A
rs10885122	ADRA2A	Stress Response	These receptors have a critical role in regulating neurotransmitter release from your fight or flight system (sympathetic nerves and adrenergic neurons) in the brain. When activated, these receptors induce low heart rate, low blood pressure and sedation	TT	G
rs553668	ADRA2A	Stress Response	The ADRA2A encodes the ADRA2A receptor. These receptors have a critical role in regulating neurotransmitter release from your fight or flight system (sympathetic nerves and adrenergic neurons) in the brain. A=increased ADRA2 receptors	AA	A
rs1360780	FKBP5	Stress Response	Binding of FKBP5 to the Cortisol Receptor reduces its cortisol-binding capacity which leads to impaired negative feedback of HPA axis and a prolonged stress response. TT (worst) and CT are associated with emotional reactivity. Childhood trauma=even more FKBP5 in response to HPA activation.	TT	--

## Health Report

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs3800373	FKBP5	Stress Response	Binding of FKBP5 to the Cortisol Receptor reduces its cortisol-binding capacity which leads to impaired negative feedback of HPA axis and a prolonged stress response.GG (worst) and AG are associated with emotional reactivity	GG	A
rs9470080	FKBP5	Stress Response	Binding of FKBP5 to the Cortisol Receptor reduces its cortisol-binding capacity which leads to impaired negative feedback of HPA axis and a prolonged stress response.TT and CT are equally are associated with emotional reactivity	TT	--
rs1978340	GAD1	Stress Response	A=lower GAD1.AA=increased GABA levels.Since GAD1 levels are believed to be lower with the A allele, the study speculates that GABA production by the GAD1 gene is actually reduced in the A allele carriers and that a compensatory change such as excessive production by GAD2 or a decreased breakdown by GABA transaminase may be causing this unpredicted increase in GABA.	AA	--
rs3749034	GAD1	Stress Response	The G allele=reduced GAD1 production	AA	G
rs5443	GNB3T (5HT2A)	Stress Response	The T allele is thought to increase second messenger signalling. There are benefits and negative effects to this.	TT	C
rs6313	HTR2A	Stress Response	The A allele was associated with lower general health and social function scores. This SNP correlates perfectly with rs6311, as in the A allele here will result in the T allele by rs6311. In schizophrenia, GG tend to do worse on working memory tasks than do individuals with a A allele	AA	G
rs6314	HTR2A	Stress Response	AA had significantly greater mental fatigue scores than the AG, AG is associated with ADHD, better response to antidepressants, worse memory.	AA	G
rs1042778	OXTR	Stress Response	People with the G allele had higher oxytocin levels than T carriers	TT	T
rs13316193	OXTR	Stress Response	CC or CT has been associated with empathy, whereas TT (the risky version) has been linked to decreased expression of oxytocin receptors in the brain, depressive mood and greater risk for Autism	TT	--
rs2268491	OXTR	Stress Response	CT had the highest level of empathy (IRI=68.7), followed by TT (65.8) and then CC (60.7)	TT	C
rs2268494	OXTR	Stress Response	TT is the social version. The A allele, which is not common, was associated with risk for Autism, which suggests lower oxytocin or worse processing of it.	AA	--
rs53576	OXTR (MAIN)	Stress Response	GG promotes efficient functioning of the oxytocin system and enhances the effects of oxytocin supplementation. GG take social rejection worse than others	AA	G

Health Report

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs1867277	FOXE1	Thyroid	The FOXE1 protein regulates the transcription of thyroglobulin (Tg) and thyroid peroxidase (TPO). AA= production of FOXE1 gene is increased (by forming a complex with USF1/USF2 transcription factors).The higher level of production of FOXE1 gene associated with development of thyroid cancer.	AA	--



# Immune Factors & Inflammation



## *IgG, IgE and IgA*

Immunoglobulin's, also known as antibodies, comprise a family of proteins that occur in five major forms, also termed classes or isotopes – IgM, IgD, IgG, IgE and IgA. Immunoglobulins are produced by vertebrate animals as part of the normal immune response to microbial, e.g., bacterial or viral, infection.

## Immune Factors &amp; Inflammation

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs231775	CTLA4	Autoimmunity	This gene encodes a protein which transmits an inhibitory signal to T cells. Mutations in this gene have been associated autoimmune diseases including: diabetes, Graves disease, thyroid problems, celiac disease, and lupus	GG	A
rs2858331	HLA	Autoimmunity	Helps to recognise foreign particles when they enter our body and fight them away in an immune response.	GG	G
rs11575839	HLA-C	Autoimmunity	Increases risk for autoimmunity/inflammation to toxins or infections. AG, AA can increase C4 levels.	AA	--
rs10484554	HLA-C/MHCI	Autoimmunity	Increases risk for autoimmunity/inflammation to toxins or infections. Each T allele=very significant increase in autoimmune disease risk.	TT	T
rs2187668	HLA-DQA1	Autoimmunity	Helps to recognise foreign particles when they enter our body and fight them away in an immune response.	TT	C
rs9275224	HLA-DQA2	Autoimmunity	Helps to recognise foreign particles when they enter our body and fight them away in an immune response.	AA	A
rs3135391	HLA-DRB1	Autoimmunity	Increases risk for autoimmunity/inflammation to toxins or infections. Each A allele=very significant increase in autoimmune disease risk.	AA	G
rs3135388	HLA-DRB5/HLA-DRB1/HLA-DRA/HLA-DQB1/HLA-DQA1	Autoimmunity	Increases risk for autoimmunity/inflammation to toxins or infections. Each A allele=very significant increase in autoimmune disease risk.D3 is protective	AA	G
rs10181656	STAT4	Autoimmunity	The STAT4 protein is activated by cytokines, which are part of the inflammatory response to fight infection. It tells immune cells to get rid of foreign invaders in the cell (R).Mutations of this gene can cause arthritis, lupus, and other autoimmune disorders.The G allele has 1.7x risk for lupus compared to the other allele.	GG	--
rs6677604	CFH	IgA	C3, a protein coding gene, is part of a system of proteins that fight against disease.	AA	G
rs9271366	HLA	IgA	Its main function is to express the receptors on cells, especially T-cells (fighter white blood cells) which helps them recognize foreign particles when they enter our body and fight them away in an immune response.This gene helps to express receptors that can identify the dangerous particle or germ (pathogen) in the body and alert our immune system.	GG	A
rs1883414	HLA-DPB2	IgA		AA	A
rs9275224	HLA-DQA2	IgA	This gene encodes a protein that helps with peptide binding	AA	A



Immune Factors & Inflammation

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs1990760	IFIH1	IgA	This gene encodes a protein that is involved with RNA translation initiation, splicing, and assembly. It is also involved in the formation of embryos and sperm and cellular growth and division	TT	C
rs2280714	IRF5	IgA	IFN-regulatory factor 5 gene increases IFN-alpha. Interferon alpha induces the kynurenine pathway and is associated with depression because of NMDA overactivity. The C (minor) allele is associated with: Possible increased risk of depression when paired with STAT4 T allele. Increased levels of IFN-alpha protein.	CC	T
rs9275596	MTC03P1	IgA		CC	--
rs9357155	PSMB8 / TAP1 / TAP2	IgA		AA	G
rs2229765	IGF1R	IgA, Insulin Growth Factor	IGF1R encodes for insulin growth factor receptors; overexpression can stop cell death by enhancing cell survival and is present in malignant tissues. AA= lower levels of IGF-1 protein. Increased longevity in males 15% of people have AA. Most people have AG.	AA	G
rs2814778	ACKR1	IgE	rs2814778 is within the DARC gene, which encodes the Duffy blood group antigen [PMID 7663520]. This SNP shows an almost perfectly fixed difference in frequency between Europeans and those with African ancestry. (One exception appears to be a certain population of Czech gypsies, and certain non-Ashkenazi Jewish populations.) Additionally the Namibian San samples of the CEPH-HGDP are, uncharacteristically for Africans, all AA homozygotes for this SNP. The rs2814778 (G) allele is associated with African populations, while rs2814778 (A) is associated with European populations and southwestern Native American populations.	CC	T
rs2251746	FCER1A	IgE	This gene encodes a protein that initiates the body's allergic response. High levels of serum IgE are associated with allergies, and are mediators of autoimmune diseases.	CC	C
rs1800925	IL13	IgE	This gene encodes a cytokine that inhibits the production of pro-inflammatory cytokines and chemokines. This SNP is in the promoter region of the IL-13 gene, Asthma related.	TT	--
rs2040704	RAD50	IgE		GG	A
rs2240032	RAD50	IgE		TT	--
rs33977706	SOCS-1 - 820G>T	IgE		AA	--

## Immune Factors &amp; Inflammation

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs1801274	FCGR2A	IgG	This gene encodes a protein that plays an important role in the immune system response by fighting off pathogens. It is associated with higher susceptibility to arthritis, lupus, and malaria.	GG	A
rs7483	GSTM3	IgG	GST-M3 activity is possibly involved in protection against mucosal atrophy caused by H. pylori as the levels of IgG titer and PGI are linked to mucosal status.	TT	T
rs1800871	IL10-819	Inflammation	A=decreased IL-10..susceptibility to IBD	AA	G
rs20541	IL13	Inflammation	rs20541 is a C/T polymorphism on the interleukin 13 gene IL13 associated with IgE levels	AA	G
rs1143643	IL1B	Inflammation	CC=Higher IL-1B, Less responsiveness to emotional stimuli.	CC	--
rs16944	IL1B 511	Inflammation	Each G=higher LPS induced IL-1B, more cortisol resistance.	GG	A
rs1801275	IL4R	Inflammation	Anti-inflammatory cytokines	GG	A
rs2069812	IL5	Inflammation	Anti-inflammatory cytokines	GG	G
rs2069837	IL6	Inflammation	A= higher L-6	AA	A
rs2069840	IL6	Inflammation	G= higher L-6	GG	--
rs1800795	IL6-174	Inflammation	G=Common, higher IL-6 (R).	GG	G
rs7529229	IL6R	Inflammation	CC= higher L-6	CC	--
rs1405655	LXR	Inflammation	LXR plays an anti-microbial role and protects against Tuberculosis. LXR decreases inflammation by inhibiting Nf-kB. LXR represses a set of inflammatory genes after activation by bacterial components or cytokines. Loss of LXR function compromised innate immunity in an animal model, which shows the importance of LXR for a normal functioning immune system.	TT	T



## Immune Factors &amp; Inflammation

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs17373080	LXR	Inflammation	LXR plays an anti-microbial role and protects against Tuberculosis. LXR decreases inflammation by inhibiting Nf-kB. LXR represses a set of inflammatory genes after activation by bacterial components or cytokines. Loss of LXR function compromised innate immunity in an animal model, which shows the importance of LXR for a normal functioning immune system.	GG	--
rs2695121	LXR	Inflammation	LXR plays an anti-microbial role and protects against Tuberculosis. LXR decreases inflammation by inhibiting Nf-kB. LXR represses a set of inflammatory genes after activation by bacterial components or cytokines. Loss of LXR function compromised innate immunity in an animal model, which shows the importance of LXR for a normal functioning immune system.	TT	--
rs6503691	STAT3	Inflammation	CC=more STAT3 production in people.C is the more common allele. About 33% of the alleles in a population are T.Can increase Th1 and possibly Th17	CC	C
rs4833095	TLR1	Inflammation	Toll Like Receptors get activated mainly by bacterial, viral and fungal products. Some of our junk protein can also activate TLRs. Once activated, they start a cascade that will produce cytokines and other inflammatory mediators to combat the infection.	GG	C
rs361525	TNF -238	Inflammation	Tumor Necrosis Factor	AA	G
rs1800629	TNF -308	Inflammation	Tumor Necrosis Factor	AA	G
rs3761847	TRAF1	Inflammation	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.	GG	A
rs7975232	VDR Apal	Inflammation	Vitamin D Receptor, CC presumably results in better vitamin D receptor function in some way, given the associations involved.	AA	--
rs731236	VDR Taq	Inflammation	No difference in calcitriol/1,25(OH)(2)D level. AA=increased VDR in stimulated immune cells.AA genotype is associated with increased VDR expression	AA	A
rs854571	PON1	Microbial defense	PON1 protects against bacterial infection by destroying the bacterial signalling molecules that cause gram negative bacteria to invade human tissue and form colonies, thus PON1 contributes to the bodies innate immunity	TT	C
rs662	PON1 Q192R	Microbial defense	PON1 protects against bacterial infection by destroying the bacterial signalling molecules that cause gram negative bacteria to invade human tissue and form colonies, thus PON1 contributes to the bodies innate immunity	TT	T



Immune Factors & Inflammation

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs6822844	4q27 Region			TT	G
rs429358	APOE		One C = Bad version = Less APOE4.CT: >3x increased risk for Alzheimer's; 1.4x increased risk for heart disease	CC	--
rs10210302	ATG16L1		rs10210302 has been reported in a large study to be associated with Crohn's disease.	TT	T
rs7216389	GSDMB		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	CC	T
rs7412	APOE				C



# Methylation

## Methylation & methionine/homocysteine pathways

Homocysteine is a naturally occurring amino acid produced as part of the body's methylation process. The level of homocysteine in the plasma is increasingly being recognised as a risk factor for disease and seen as a predictor of potential health problems such as cardiovascular disease and Alzheimer's. The complex metabolism of homocysteine within the body is highly dependent on vitamin derived cofactors, and deficiencies in vitamin B12, folic acid and vitamin B6 are associated with raised homocysteine levels. Other factors thought to raise levels are poor diet, poor lifestyle - especially smoking and high coffee and alcohol intake, some prescription drugs (such as proton pump inhibitors), diabetes, rheumatoid arthritis and poor thyroid function. Homocysteine levels can, in many cases, be normalised through diet and vitamin supplementation. The most important nutrients that help lower homocysteine levels are folate, the vitamins B12, B6 and B2, zinc and trimethylglycine (TMG).



Methylation

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs17851582	GAMT	Arginine	Arginine to Guanidoacetate to Creatine (SAmE)	AA	A
rs55776826	GAMT	Arginine	Arginine to Guanidoacetate to Creatine (SAmE)	TT	--
rs4244593	PEMT	CDP-choline pathway	phosphatidylethanolamine to phosphatidylcholine	TT	T
rs7946	PEMT	CDP-choline pathway	phosphatidylethanolamine to phosphatidylcholine	TT	T
rs492602	FUT2	Cobalamin	B12 absorption	GG	A
rs602662	FUT2	Cobalamin	B12 absorption	AA	G
rs492602	FUT2 A12190G	Cobalamin	B12 absorption	GG	A
rs1047781	FUT2 A12404T	Cobalamin	B12 absorption	TT	A
rs1532268	MTRR	Cobalamin	Cobalamin II to Methylcobalamin	TT	T
rs1802059	MTRR - 11 A664A	Cobalamin	Cobalamin II to Methylcobalamin	AA	A
rs1801394	MTRR A66G	Cobalamin	Cobalamin II to Methylcobalamin	GG	A
rs162036	MTRR K350A	Cobalamin	Cobalamin II to Methylcobalamin	GG	A
rs526934	TCN1	Cobalamin	B12 transport	GG	A
rs1801198	TCN2	Cobalamin	B12 transport	GG	G
rs9606756	TCN2	Cobalamin	B12 transport	GG	A
rs1643659	DHFR	Folate	FOLR2 to DHF, DHF to THF	CC	--



Methylation

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs1650697	DHFR	Folate	FOLR2 to DHF, DHF to THF	AA	G
rs1677693	DHFR	Folate	FOLR2 to DHF, DHF to THF	TT	--
rs2071010	FOLR1	Folate	Dietary folates to DHF	AA	A
rs651933	FOLR2	Folate	Dietary folates to DHF	GG	A
rs7925545	FOLR3	Folate	Dietary folates to DHF	GG	A
rs3780126	GGH	Folate	Metabolism of folic acid and in the pharmacology of many antifolate drugs.	AA	A
rs3780127	GGH	Folate	Metabolism of folic acid and in the pharmacology of many antifolate drugs.	AA	G
rs4617146	GGH	Folate	Metabolism of folic acid and in the pharmacology of many antifolate drugs.	TT	C
rs2236225	MTHFD1	Folate	THF to Purines (ATP & GTP) & 10-Formyl-THF to 5,10 Methenyl THF to 5,10 Methylene THF	AA	A
rs1801131	MTHFR A1298C	Folate	5,10 Methylene THF to 5-MTHF	GG	T
rs1801133	MTHFR C677T	Folate	5,10 Methylene THF to 5-MTHF	AA	G
rs2733103	MTHFS	Folate	Helps regulate carbon flow through the folate- dependent one-carbon metabolic network that supplies carbon for the biosynthesis of purines, thymidine, and amino acids.	TT	T
rs6495446	ST20	Folate	Helps regulate carbon flow through the folate- dependent one-carbon metabolic network that supplies carbon for the biosynthesis of purines, thymidine, and amino acids.	TT	T
rs6495446	ST20	Folate	Helps regulate carbon flow through the folate- dependent one-carbon metabolic network that supplies carbon for the biosynthesis of purines, thymidine, and amino acids.	TT	T
rs10925235	MTR	Folate, Cobalamin, Methionine	5-MTHF to THF, Homocysteine to Methionine, Methylcobalamin to Cobalamin I (Zn)	TT	--
rs10925257	MTR	Folate, Cobalamin, Methionine	5-MTHF to THF, Homocysteine to Methionine, Methylcobalamin to Cobalamin I (Zn)	GG	A

## Methylation

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs11799670	MTR	Folate, Cobalamin, Methionine	5-MTHF to THF, Homocysteine to Methionine, Methylcobalamin to Cobalamin I (Zn)	GG	A
rs1805087	MTR	Folate, Cobalamin, Methionine	5-MTHF to THF, Homocysteine to Methionine, Methylcobalamin to Cobalamin I (Zn)	GG	A
rs2275565	MTR	Folate, Cobalamin, Methionine	5-MTHF to THF, Homocysteine to Methionine, Methylcobalamin to Cobalamin I (Zn)	TT	T
rs2275568	MTR	Folate, Cobalamin, Methionine	5-MTHF to THF, Homocysteine to Methionine, Methylcobalamin to Cobalamin I (Zn)	TT	C
rs2789352	MTR	Folate, Cobalamin, Methionine	5-MTHF to THF, Homocysteine to Methionine, Methylcobalamin to Cobalamin I (Zn)	AA	A
rs3820571	MTR	Folate, Cobalamin, Methionine	5-MTHF to THF, Homocysteine to Methionine, Methylcobalamin to Cobalamin I (Zn)	GG	T
rs3733890	BHMT	Methionine	Homocysteine to Methionine shortcut	AA	G
rs567754	BHMT-02	Methionine	Homocysteine to Methionine shortcut	TT	T
rs2851391	CBS	Methionine	Homocysteine to Cystathionine	TT	T
rs4920037	CBS	Methionine	Homocysteine to Cystathionine	AA	G
rs234706	CBS C699T	Methionine	Homocysteine to Cystathionine	AA	G



# Mitochondrial Function



## Mitochondrial Function

Mitochondrial DNA contains 37 genes, all of which are essential for normal mitochondrial function. Thirteen of these genes provide instructions for making enzymes involved in oxidative phosphorylation. Oxidative phosphorylation is a process that uses oxygen and simple sugars to create adenosine triphosphate (ATP), the cell's main energy source. The remaining genes provide instructions for making molecules called transfer RNA (tRNA) and ribosomal RNA (rRNA), which are chemical cousins of DNA. These types of RNA help assemble protein building blocks (amino acids) into functioning proteins. The following conditions are associated with changes in the structure of mitochondrial DNA. Age-related hearing loss, Cancers, Cyclic vomiting syndrome, Cytochrome c oxidase deficiency, Kearns-Sayre syndrome, Leber hereditary optic neuropathy, Leigh syndrome, Maternally inherited diabetes and deafness, Mitochondrial complex III deficiency, Mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes, Myoclonic epilepsy with ragged-red fibers, Neuropathy, ataxia, and retinitis pigmentosa, Nonsyndromic hearing loss, Pearson marrow-pancreas syndrome, Progressive external ophthalmoplegia.

## Mitochondrial Function

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs7134594	MMAB	Cobalamin	Conversion of vitamin B-12 into adenosylcobalamin	CC	T
rs1532268	MTRR	Cobalamin	Contribution of B12 to Krebs Cycle	TT	T
rs1802059	MTRR -11 A664A	Cobalamin	Contribution of B12 to Krebs Cycle	AA	A
rs1801394	MTRR A66G	Cobalamin	Contribution of B12 to Krebs Cycle	GG	A
rs162036	MTRR K350A	Cobalamin	Contribution of B12 to Krebs Cycle	GG	A
rs4147730	NDUFS3	Complex I	Electron Transport Chain, ATP Production	AA	--
rs1142530	NDUFS7	Complex I	Electron Transport Chain, ATP Production	TT	T
rs809359	NDUFS7	Complex I	Electron Transport Chain, ATP Production	GG	--
rs2075626	NDUFS8	Complex I	Electron Transport Chain, ATP Production	CC	T
rs3741049	ACAT1 G22670A	Krebs Cycle	Converts 2-methyl-acetoacetyl-CoA into propionyl-CoA and acetyl-CoA.	AA	A
rs4244593	PEMT	Membranes permeability	phosphatidylethanolamine to phosphatidylcholine	TT	T
rs7946	PEMT	Membranes permeability	phosphatidylethanolamine to phosphatidylcholine	TT	T
rs2293054	NOS1		nNOS (Brain)	AA	G
rs7298903	NOS1		nNOS (Brain)	CC	--
rs2248814	NOS2		iNOS, a reactive free radical	AA	--
rs2274894	NOS2		iNOS, a reactive free radical	TT	--



Mitochondrial Function

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs1800779	NOS3		eNOS	GG	A
rs1800783	NOS3		eNOS	AA	--
rs2970869	PGC-1a		PGC-1? makes new mitochondria and improves its function. PGC-1a increases fatty acid burning by increasing the carnitine genes CPT1A.	TT	--
rs2970869	PGC-1a		PGC-1? makes new mitochondria and improves its function. PGC-1? makes new mitochondria and improves its function. PGC-1a increases fatty acid burning by increasing the carnitine genes CPT1A. Carriers of the T allele had significantly more DNA damage than CC	TT	--
rs3774923	PGC-1a		PGC-1? makes new mitochondria and improves its function. PGC-1a increases fatty acid burning by increasing the carnitine genes CPT1A.	TT	C
rs8192678	PGC-1a Gly482Ser		PGC-1? makes new mitochondria and improves its function. PGC-1a increases fatty acid burning by increasing the carnitine genes CPT1A. T is the less common allele (codes for serine). This variant causes a lower level of PGC-1a protein including in muscles The T allele results a less efficient coactivator of transcription factors, including those that regulate the PPARGC1A gene itself	TT	C



# Neurotransmitters

## Neurotransmitter Pathway: Serotonin & Dopamine

Dopamine and serotonin are both neurotransmitters. They are chemicals that are released from the nerve cells and serve to transfer impulses from nerve cells to other body tissues like other nerve cells, muscles, and organs. These chemicals are very important in sensory perceptions and motoric action-reaction mechanisms.

Dopamine plays an important role in controlling motor behavior, the emotional reward, and behavior motivation mechanisms. It regulates emotional responses, hormone secretion, and motoric actions related to reward sensations.

Serotonin regulates the functioning of the cardiovascular and endocrine systems and has an important role in regulating appetite, sleep, memory, mood, body temperature, and muscle contraction. Genetic variants can influence transporter function by various mechanisms, including substrate affinities, transport velocity, transporter expression levels (density), extracellular membrane expression, trafficking and turnover, and neurotransmitter release.

## Neurotransmitters

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs1643659	DHFR	Biopterin	Biopterin Recycling	CC	--
rs1650697	DHFR	Biopterin	Biopterin Recycling	AA	G
rs1677693	DHFR	Biopterin	Biopterin Recycling	TT	--
rs3783637	GCH1	Biopterin	Tetrahydrobiopterin (BH4) biosynthesis.	TT	C
rs3783641	GCH1	Biopterin	Tetrahydrobiopterin (BH4) biosynthesis.	AA	T
rs3783642	GCH1	Biopterin	Tetrahydrobiopterin (BH4) biosynthesis.	CC	T
rs4411417	GCH1	Biopterin	Tetrahydrobiopterin (BH4) biosynthesis.	CC	--
rs752688	GCH1	Biopterin	Tetrahydrobiopterin (BH4) biosynthesis.	TT	--
rs8007267	GCH1 C36378991T	Biopterin	Tetrahydrobiopterin (BH4) biosynthesis.	TT	C
rs10483639	GCH1 G55306457C	Biopterin	Tetrahydrobiopterin (BH4) biosynthesis.	CC	G
rs2293054	NOS1	Biopterin	Biopterin Recycling	AA	G
rs7298903	NOS1	Biopterin	Biopterin Recycling	CC	--
rs2248814	NOS2	Biopterin	Biopterin Recycling	AA	--
rs2274894	NOS2	Biopterin	Biopterin Recycling	TT	--
rs1800779	NOS3	Biopterin	Biopterin Recycling	GG	A
rs1800783	NOS3	Biopterin	Biopterin Recycling	AA	--

## Neurotransmitters

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs5030858	PAH	Biopterin	Phenylalanine to Tyrosine	AA	G
rs2070762	TH	Biopterin	Tyrosine to L-Dopa	GG	G
rs6356	TH	Biopterin	Tyrosine to L-Dopa	TT	T
rs11178997	TPH2	Biopterin	A=lower production of TPH2 and therefore less serotonin	AA	--
rs4565946	TPH2	Biopterin	Each copy of the common G allele was associated with a increase in the number of psychosomatic symptoms.GG=2.58X increased risk for early onset OCDNo association with risk for bipolar disorder or suicidal behavior or autism Association with ADHD	GG	T
rs4570625	TPH2 G-703T	Biopterin	Tryptophan to 5-HTP, GG=Lower serotonin, more anxiety.	GG	G
rs2238151	ALDH2	Catecholamine aldehyde	Stress Hormones metabolism (Noradrenaline), Serotonin & 5-HIA metabolism (NAD, B1)	TT	C
rs671	ALDH2	Catecholamine aldehyde	Stress Hormones metabolism (Noradrenaline), Serotonin & 5-HIA metabolism (NAD, B1)	AA	G
rs72547566	ALDH3A2	Catecholamine aldehyde	Adrenaline metabolism (NAD,B1)	TT	--
rs4633	COMT H62H	Catecholamines	Methylation (deactivation) of Dopamine, Adrenaline	TT	C
rs4680	COMT V158M	Catecholamines	Methylation (deactivation) of Dopamine, Adrenaline	AA	G
rs1108580	DBH	Catecholamines	GG = Increased levels of DBH proteinGG = Lower Dopamine, higher norepinephrine.GG= Increased risk of having Parkinsons DiseaseGG = Increased risk of alcoholism GG = Methamphetamine psychosis, with more than a 2x greater chance of relapsing after going through complete remission. GG = Faster dependence on alcoholAA = Decreased levels of DBH proteinAA = Higher Dopamine, lower norepinephrine.AA = Slightly increased the risk of Schizophrenia.AA = Hyperactive and oppositional characteristic traits. AA = More accurate and speedier decision making when incorrect advice is given.AA = some association with ADHD in males.	GG	A



Neurotransmitters

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs1611115	DBH	Catecholamines	Decreased levels of Dopamine-beta-Hydroxylase (T).Normal levels of Dopamine-beta-Hydroxylase (C).The Minor "T" allele is associated with:Higher dopamine and lower norepinephrine because of lower DBH (Dopamine-beta-hydroxylase), which converts dopamine to norepinephrine (T) .More neuroticism (TT).More novelty seeking (TT).More impulsivity (TT) .More aggression (TT) Increased risk of adult ADHD (TT) Lower conscientiousness (TT)	TT	T
rs1799836	MAOB	Catecholamines	Oxidation (deactivation)Dopamine, Adrenaline, Noradrenaline (less Serotonin). T= upregulation.	TT	T
rs10994336	ANK3	Dopamine	Density of the DRD2 receptor.	TT	C
rs9804190	ANK3	Dopamine	Density of the DRD2 receptor.	TT	C
rs265981	DRD1	Dopamine	Dopamine Receptor	AA	A
rs4532	DRD1	Dopamine	Dopamine Receptor	CC	C
rs686	DRD1	Dopamine	Dopamine Receptor	GG	G
rs1079596	DRD2	Dopamine	Dopamine Receptor	TT	C
rs1079597	DRD2	Dopamine	Dopamine Receptor	TT	C
rs1079727	DRD2	Dopamine	Dopamine Receptor	CC	--
rs1125394	DRD2	Dopamine	Dopamine Receptor	CC	--
rs2283265	DRD2	Dopamine	Dopamine Receptor	AA	--
rs4436578	DRD2	Dopamine	Dopamine Receptor	CC	--
rs4648317	DRD2	Dopamine	Dopamine Receptor	AA	G





Neurotransmitters

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs1800497	DRD2 (MAIN)	Dopamine	AA = Less dopamine D2 receptors = lower functioning of dopamine [R]. A = decreased level of dopamine D2 receptors = lower level of dopamine [R] G = normal dopamine D2 receptor density = normal level of dopamine	AA	G
rs6277	DRD2 (MAIN)	Dopamine	Dopamine Receptor. AA=decreased dopamine signaling.	AA	--
rs1799978	DRD2 A4651G	Dopamine	Dopamine Receptor	CC	T
rs2242592	DRD2 C71572T	Dopamine	Dopamine Receptor	GG	G
rs12364283	DRD2 T4047C	Dopamine	Dopamine Receptor	GG	A
rs167771	DRD3	Dopamine	Dopamine Receptor	GG	A
rs6280	DRD3	Dopamine	Dopamine Receptor	CC	T
rs1018381	DTNBP1	Dopamine	Synaptic vesicle trafficking, neurotransmitter release. Role in regulation of cell surface exposure of Dopamine Receptor 2	AA	--
rs27072	SLC6A3 G56022A	Dopamine	Dopamine Transport, Dopamine Reuptake via DAT is the primary mechanism through which dopamine is cleared from synapses.	CC	T
rs279836	GABRA2	GABA	GABA receptor. AA=Higher stress response	AA	A
rs3219151	GABRA6	GABA	GABA receptor. TT=higher cortisol, CT=Intermediate	TT	T
rs2241165	GAD1	Glutamate, GABA	Conversion of excitatory Glutamate to calming GABA.	CC	T
rs3749034	GAD1	Glutamate, GABA	Conversion of excitatory Glutamate to calming GABA.	AA	G
rs3791878	GAD1	Glutamate, GABA	Conversion of excitatory Glutamate to calming GABA.	TT	--
rs3828275	GAD1	Glutamate, GABA	Conversion of excitatory Glutamate to calming GABA.	TT	T
rs6994992	NRG1	Glutamate, GABA	Neuregulin 1 is important for synaptic plasticity, GABAergic function within the amygdala (to shut down anxiety). TT= Increased risk of Psychosis	TT	T

## Neurotransmitters

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs1049793	ABP1/DAO C1933G	<b>Histamine, Serine, Dopamine</b>	D-serine neuromodulator in the brain, contribution to dopamine synthesis ( via D-dopa)	GG	--
rs3741775	DAO	<b>Histamine, Serine, Dopamine</b>	D-serine neuromodulator in the brain, contribution to dopamine synthesis ( via D-dopa)	CC	--
rs3918347	DAO	<b>Histamine, Serine, Dopamine</b>	D-serine neuromodulator in the brain, contribution to dopamine synthesis ( via D-dopa)	GG	A
rs4244593	PEMT	<b>Methylation</b>	SAMe (cofactor also for COMT, PNMT) needed here first for cell membrane integrity.	TT	T
rs7946	PEMT	<b>Methylation</b>	SAMe (cofactor also for COMT, PNMT) needed here first for cell membrane integrity.	TT	T
rs3785143	SLC6A2	<b>Noradrenaline</b>	Noradrenaline Transporter, reuptake of noradrenaline	TT	--
rs3785152	SLC6A2	<b>Noradrenaline</b>	Noradrenaline Transporter, reuptake of noradrenaline	TT	C
rs40147	SLC6A2	<b>Noradrenaline</b>	Noradrenaline Transporter, reuptake of noradrenaline	AA	A
rs2242446	SLC6A2 C5884T	<b>Noradrenaline</b>	Noradrenaline Transporter, reuptake of noradrenaline	CC	T
rs11077820	AANAT	<b>Serotonin</b>	Serotonin to Melatonin (B5)	CC	T
rs1451375	DDC	<b>Serotonin</b>	Serotonin Synthesis	AA	A
rs2167364	DDC	<b>Serotonin</b>	Serotonin Synthesis	CC	T
rs7997012	HTR2A	<b>Serotonin</b>	Post-synaptic Serotonin receptor	AA	A
rs6323	MAO A R297R	<b>Serotonin</b>	Oxidation (deactivation) Serotonin, Dopamine, Adrenaline, Noradrenaline.	GG	T
rs1137070	MAOA	<b>Serotonin</b>	Oxidation (deactivation) Serotonin, Dopamine, Adrenaline, Noradrenaline.	TT	C
rs216013	CACNA1C		Mediate the entry of calcium ions into excitable cells and are also involved in a variety of calcium-dependent processes.	GG	A



Neurotransmitters

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs2302729	CACNA1C		Mediate the entry of calcium ions into excitable cells and are also involved in a variety of calcium-dependent processes.	TT	T
rs1006737	CACNA1C or SLC36A3		Voltage-sensitive calcium channels (VSCC) mediate the entry of calcium ions into excitable cells and are also involved in a variety of calcium-dependent processes, including muscle contraction, hormone or neurotransmitter release, gene expression, cell motility, cell division and cell death. The isoform alpha-1C gives rise to L-type calcium currents. Long-lasting (L-type) calcium channels belong to the 'high-voltage activated' (HVA) group. They are blocked by dihydropyridines (DHP), phenylalkylamines, benzothiazepines, and by omega-agatoxin-IIIa (omega-Aga-IIIa). They are however insensitive to omega-conotoxin-GVIA (omega-CTx-GVIA) and omega-agatoxin-IVA (omega-Aga-IVA). Calcium channels containing the alpha-1C subunit play an important role in excitation-contraction coupling in the heart. The various isoforms display marked differences in the sensitivity to DHP compounds. Binding of calmodulin or CABP1 at the same regulatory sites results in an opposite effects on the channel function.	AA	A
rs2007044	CACNA1C				A



# Trans-sulfuration

## *Trans-sulfuration pathway*

The trans-sulfuration pathway is a metabolic pathway where transfer of sulfur from homocysteine to cysteine occurs. The pathway leads to the generation of several sulfur metabolites, which include cysteine, GSH and the gaseous signaling molecule hydrogen sulfide ( $H_2S$ ). Precise control of this pathway is critical for maintenance of optimal cellular function and, therefore, the key enzymes of the pathway, cystathionine  $\beta$ -synthase and cystathionine  $\gamma$ -lyase, are regulated at multiple levels. Disruption of the trans-sulfuration pathway contributes to the pathology of several conditions such as vascular dysfunction, Huntington's disease.

## Trans-sulfuration

SNP RSID	GENE & VARIANT	PATHWAY	VARIANT MEANING	RISK ALLELE	GENOTYPE
rs2851391	CBS		It is responsible for using vitamin B6 to convert amino acids called homocysteine and serine to a molecule called cystathionine. Another enzyme converts cystathionine to cysteine, which is used to build proteins or is broken down and excreted in urine	TT	T
rs4920037	CBS		It is responsible for using vitamin B6 to convert amino acids called homocysteine and serine to a molecule called cystathionine. Another enzyme converts cystathionine to cysteine, which is used to build proteins or is broken down and excreted in urine.	AA	G
rs234706	CBS C699T		AA=Increased responsiveness to homocysteine- lowering effects of folic acid. Limits homocysteine into downstream path. People with CBS mutations will need to be careful with sulfur containing supplements. Increased risk for ammonia detoxification issues.	AA	G

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This report is intended to translate your results into an easier to understand form. It is not intended to diagnose or treat. For diagnosis or treatment, please present this to your doctor. Additionally, genetic mutations are flags that something could be wrong and not a guarantee that you are having all or any of the associated issues. Other factors like environment, ethnic background, diet, age, personal history, etc all have a factor in whether a mutation starts to present itself or not and when.

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