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Gene Variance report

Joshua Jaramillo 19 / M

ALLERGY

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	C11orf30	rs2155219	G	TT	-/-
2	HLA-DQB1	rs7775228	C	CC	+/+

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CLOTTING FACTORS

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	CETP	rs1800775	C	CC	+/+
2	CYP4V2	rs13146272	C	CC	+/+
3	F10	rs3211719	G	AA	-/-
5	F11	rs2289252	T	CT	+/-
7	F12	rs2731672	T	TT	+/+
9	F5	rs6025	T	CC	-/-
11	F9	rs6048	-	A	
12	GP6	rs1613662	G	AA	-/-
13	HRG	rs9898	T	CC	-/-
14	ITGB3	rs5918	C	TT	-/-
15	NR1I2	rs1523127	C	AA	-/-
16	SERPINC1	rs2227589	T	CT	+/-

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DETOX

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	CTH	rs1021737	T	TT	+/+
3	CYP1A1	rs1799814	T	GG	-/-
4	CYP1A1	rs4986883	C	TT	-/-
5	CYP1A2	rs762551	C	AC	+/-
8	CYP1B1	rs1800440	C	TT	-/-
11	CYP2C9	rs1057910	C	AA	-/-
12	CYP2C9	rs1799853	T	CT	+/-
13	CYP2D6	rs1065852	A	GG	-/-
15	CYP2D6	rs16947	A	GG	-/-
16	CYP2E1	rs2070676	G	GG	+/+
17	CYP2E1	rs55897648	A	GG	-/-
18	CYP2E1	rs6413419	A	GG	-/-
20	CYP3A4	rs2740574	C	TT	-/-
21	CYP3A4	rs4986910	G	AA	-/-
22	CYP3A4	rs55785340	G	AA	-/-
24	GSTM1	rs1056806	T	CC	-/-
26	GSTM1	rs4147565	A	--	NC
28	GSTP1	rs1138272	T	CC	-/-
29	GSTP1	rs1695	G	AA	-/-
30	NAT1	rs4986782	A	GG	-/-
31	NAT2	rs1208	G	AG	+/-
32	NAT2	rs1799930	A	AG	+/-
33	NAT2	rs1799931	A	GG	-/-
34	NAT2	rs1801279	A	GG	-/-
35	NAT2	rs1801280	C	CT	+/-

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GLUTEN INTOLERANCE

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	HLA-DQA1	rs2187668	T	CC	-/-
2	HLA-DQA2	rs2858331	G	AG	+/-

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IGA

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	CFH	rs6677604	A	GG	-/-
2	HLA-DPA1	rs1883414	A	GG	-/-
3	HLA-DQA2	rs9275224	A	AG	+/-
4	HLA-DRB1	rs9275596	C	TT	-/-
5	HORMAD2	rs2412971	G	AA	-/-
6	IFIH1	rs1990760	T	CT	+/-
7	IGF1R	rs2229765	A	AG	+/-
8	IRF5	rs4728142	A	AG	+/-
9	MHC	rs9271366	G	AA	-/-
10	MTC03P1	rs2856717	A	GG	-/-
11	PSMB8	rs9357155	A	GG	-/-
12	TRAF1	rs3761847	G	AA	-/-

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IGE

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	C3	rs10402876	C	CC	+/+
3	CD14	rs2569191	C	CT	+/-
4	DARC	rs2814778	C	CT	+/-
5	FCER1A	rs2251746	C	TT	-/-
6	FCER1A	rs2427824	T	CC	-/-
7	FCER1A	rs2427827	-	CT	
10	FCER1A / OR10J2P	rs10489854	T	CC	-/-
12	IL13	rs1800925	T	CC	-/-
13	IL5	rs2069812	G	GG	+/+
14	RAD50	rs17772565	T	CC	-/-
15	RAD50	rs17772583	G	AA	-/-
16	RAD50	rs2040704	G	AA	-/-
17	RAD50	rs2240032	T	CC	-/-
18	RAD50	rs6884762	T	CC	-/-
19	RAG1	rs3740955	A	GG	-/-
20	SOCS1	rs33977706	A	CC	-/-

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IGG

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	FCGR2A	rs1801274	G	AG	+/-
2	GSTM3	rs7483	T	CT	+/-
4	MUC21	rs1634731	G	AA	-/-

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METHYLATION

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	ACAT1	rs3741049	A	GG	-/-
2	ACE	rs4343	G	AG	+/-
3	AGT	rs699	A	GG	-/-
4	AHCY-01	rs819147	C	TT	-/-
6	AHCY-19	rs819171	C	TT	-/-
7	BHMT	rs3733890	A	AG	+/-
8	BHMT-02	rs567754	T	CC	-/-
10	BHMT-08	rs651852	T	CT	+/-
11	C1orf167	rs4846048	G	AA	-/-
12	CBS	rs2851391	T	TT	+/+
13	CBS	rs4920037	A	GG	-/-
14	CBS A360A	rs1801181	A	GG	-/-
15	CBS C699T	rs234706	A	GG	-/-
17	CLCN6	rs13306560	T	CC	-/-
20	COMT	rs6269	G	AA	-/-
21	COMT H62H	rs4633	T	CT	+/-
22	COMT P199P	rs769224	A	GG	-/-
23	COMT V158M	rs4680	A	AG	+/-
24	DAO	rs2070586	A	AA	+/+
26	DAO	rs3741775	C	AA	-/-
27	DHFR	rs1643649	C	TT	-/-
28	FOLR1	rs2071010	A	GG	-/-
29	FOLR2	rs651933	G	AG	+/-
30	FOLR3	rs7925545	G	AA	-/-
32	FUT2	rs492602	G	AG	+/-
33	FUT2	rs601338	A	AG	+/-
34	FUT2	rs602662	A	AG	+/-
35	G6PD	rs1050828	-	C	
36	G6PD	rs1050829	-	T	
37	GAD1	rs10432420	A	AG	+/-
38	GAD1	rs12185692	A	CC	-/-
39	GAD1	rs2058725	C	CT	+/-
40	GAD1	rs2241165	C	CT	+/-

	Gene	rsID	Minor Allele	Genotype	Phenotype
41	GAD1	rs3749034	A	AG	+/-
42	GAD1	rs3791850	A	GG	-/-
43	GAD1	rs3791851	C	CT	+/-
44	GAD1	rs3791878	T	GT	+/-
45	GAD1	rs3828275	T	CC	-/-
46	GAD1	rs701492	T	TT	+/+
48	GAD1	rs769407	C	CG	+/-
50	GAMT	rs17851582	A	GG	-/-
51	GAMT	rs55776826	T	CC	-/-
52	GIF	rs558660	A	GG	-/-
53	MAOA	rs6323	-	T	
54	MAOB	rs1799836	-	C	
55	MTHFD1	rs1076991	T	CC	-/-
56	MTHFD1	rs2236225	A	GG	-/-
57	MTHFD1L	rs11754661	A	GG	-/-
58	MTHFD1L	rs17349743	C	CC	+/+
59	MTHFD1L	rs6922269	A	AG	+/-
60	MTHFD1L	rs803422	A	GG	-/-
62	MTHFR	rs1476413	T	CC	-/-
63	MTHFR	rs17037390	A	GG	-/-
65	MTHFR	rs17367504	G	AA	-/-
66	MTHFR	rs2066470	A	GG	-/-
67	MTHFR	rs2274976	T	CC	-/-
68	MTHFR	rs4846049	T	GG	-/-
69	MTHFR A1298C	rs1801131	G	TT	-/-
70	MTHFR C677T	rs1801133	A	AA	+/+
71	MTHFS	rs6495446	T	CT	+/-
72	MTR A2756G	rs1805087	G	AA	-/-
74	MTRR	rs1801394	G	AA	-/-
75	MTRR A664A	rs1802059	A	GG	-/-
76	MTRR K350A	rs162036	G	AA	-/-
79	NOS2	rs2248814	A	AG	+/-
80	NOS2	rs2274894	T	GT	+/-
81	NOS2	rs2297518	A	AG	+/-
82	NOS3	rs1800779	G	AG	+/-
83	NOS3	rs1800783	A	AT	+/-
87	PEMT	rs4244593	T	TT	+/+

	Gene	rsID	Minor Allele	Genotype	Phenotype
92	SOD2	rs2758331	A	AA	+/+
93	SOD2	rs4880	G	GG	+/+
94	SOD3	rs2855262	C	CC	+/+
95	TCN1	rs526934	G	AA	-/-
96	TCN2	rs1801198	G	CC	-/-
98	VDR Bsm	rs1544410	T	CT	+/-
99	VDR Taq	rs731236	G	AA	-/-

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MITOCHONDRIAL FUNCTION

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	ATP5C1	rs1244414	T	CC	-/-
7	ATP5G3	rs36089250	C	TT	-/-
8	CCL2	rs1024611	G	AA	-/-
9	COX5A	rs8042694	G	AG	+/-
13	COX6C	rs4626565	C	TT	-/-
17	NDUFS3	rs4147730	A	AG	+/-
19	NDUFS7	rs1142530	T	CT	+/-
22	NDUFS7	rs2332496	A	AG	+/-
24	NDUFS7	rs7258846	T	--	NC
25	NDUFS7	rs809359	G	AA	-/-
29	NDUFS8	rs2075626	C	TT	-/-
32	NDUFS8	rs999571	A	GG	-/-
33	SLC19A1	rs1051266	C	CT	+/-
34	UQCRC2	rs11648723	T	GG	-/-
35	UQCRC2	rs4850	A	GG	-/-

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OTHER IMMUNE FACTORS

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	ADD1	rs4961	T	GG	-/-
2	APOE	rs429358	C	TT	-/-
3	ATG16L1	rs10210302	T	CT	+/-
4	HLA-DRB1	rs660895	G	AG	+/-
5	IL13	rs20541	A	GG	-/-
6	IL4R	rs1801275	G	AA	-/-
7	KIAA1109	rs6822844	T	GG	-/-
8	MEFV	rs11466023	A	GG	-/-
10	STAT4	rs10181656	G	CG	+/-
11	TNF	rs1800629	A	AG	+/-
12	TNF	rs361525	A	GG	-/-

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SULFOTRANSFERASE

	Gene	rsID	Minor Allele	Genotype	Phenotype
3	SULT1A1	rs4149381	G	TT	-/-
5	SULT1A1	rs7192559	T	CC	-/-
8	SULT1A1	rs9282862	C	TT	-/-
9	SULT2A1	rs11083907	A	GG	-/-
10	SULT2A1	rs11569679	T	CC	-/-
11	SULT2A1	rs2547231	C	AA	-/-
15	SULT2A1	rs296366	T	CC	-/-
17	SULT2A1	rs4149449	T	CC	-/-
18	SULT2A1	rs4149452	T	CT	+/-

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THYROID

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	CTLA4	rs231775	G	AG	+/-
2	FOXE1	rs10984009	A	GG	-/-
3	FOXE1	rs1867277	A	AA	+/+

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TONGUE TIE

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	IRF6	rs861020	A	GG	-/-
2	PVT1	rs987525	A	CC	-/-
3	RARA	rs7217852	G	GG	+/+
5	TBX22	rs41307258	-	T	

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Gene Definitions

HLA-DQB1 [+/+]

SS2; BTN7; BTL-II; HSBLMHC1

CETP [+/+]

The protein encoded by this gene is found in plasma, where it is involved in the transfer of cholesteryl ester from high density lipoprotein (HDL) to other lipoproteins. Defects in this gene are a cause of hyperalphalipoproteinemia 1 (HALP1). Two transcript variants encoding different isoforms have been found for this gene. [provided by RefSeq, Oct 2013]

CYP4V2 [+/+]

At least 42 CYP4V2 gene mutations have been identified in people with Bietti crystalline dystrophy, a disorder in which numerous small, yellow or white crystal-like deposits of fatty (lipid) compounds accumulate in the light-sensitive tissue that lines the back of the eye (the retina). The deposits damage the retina, resulting in progressive vision loss. CYP4V2 gene mutations that cause Bietti crystalline dystrophy are predicted to change the structure of the CYP4V2 enzyme in a way that reduces or eliminates its activity. The mutations likely affect lipid breakdown; however, it is unknown how they lead to the specific signs and symptoms of Bietti crystalline dystrophy. For unknown reasons, the severity of the signs and symptoms differs significantly among individuals with the same CYP4V2 gene mutation.

F11 [+/-]

This gene encodes coagulation factor XI of the blood coagulation cascade. This protein is present in plasma as a zymogen, which is a unique plasma coagulation enzyme because it exists as a homodimer consisting of two identical polypeptide chains linked by disulfide bonds. During activation of the plasma factor XI, an internal peptide bond is cleaved by factor XIIa (or XII) in each of the two chains, resulting in activated factor XIa, a serine protease composed of two heavy and two light chains held together by disulfide bonds. This activated plasma factor XI triggers the middle phase of the intrisic pathway of blood coagulation by activating factor IX. Defects in this factor lead to Rosenthal syndrome, a blood coagulation abnormality. [provided by RefSeq, Jul 2008]

F12 [+/+]

At least two mutations in the F12 gene are associated with hereditary angioedema type III. These mutations change single protein building blocks (amino acids) in factor XII, which increases the activity of the protein. As a result, more bradykinin is produced, which allows additional fluids to leak through blood vessel walls. The accumulation of fluids in body tissues leads to the episodes of swelling in people with hereditary angioedema type III.

SERPINC1 [+/-]

At least 220 mutations in the SERPINC1 gene have been found to cause hereditary antithrombin deficiency. Most of these mutations change single protein building blocks (amino acids) in antithrombin, which disrupts its ability to control blood clotting. Hereditary antithrombin deficiency can be divided into type I and type II based on the mutation in the SERPINC1 gene. Hereditary antithrombin deficiency type I is caused by SERPINC1 gene mutations that prevent the cell from producing antithrombin from the altered gene. Individuals with this type have only one working copy of the

SERPINC1 gene in each cell, which results in approximately half of the normal amount of antithrombin. Affected individuals do not have enough antithrombin to inactivate clotting proteins, which causes the increased risk for abnormal blood clots in hereditary antithrombin deficiency. Mutations that cause hereditary antithrombin deficiency type II result in the production of an altered antithrombin with reduced activity. Individuals with this form of the condition typically have normal levels of antithrombin, but the protein does not function properly. Type II can be further divided based on whether the mutation affects binding to thrombin and other clotting factors (type IIa), heparin (type IIb), or both (type IIc). Individuals with hereditary antithrombin deficiency type IIb have a lower risk of forming an abnormal blood clot than people with other forms of this condition because antithrombin is able to inactivate clotting proteins without heparin.

CTH [+/+]

This gene encodes a cytoplasmic enzyme in the trans-sulfuration pathway that converts cystathione derived from methionine into cysteine. Glutathione synthesis in the liver is dependent upon the availability of cysteine. Mutations in this gene cause cystathioninuria. Alternative splicing of this gene results in three transcript variants encoding different isoforms. [provided by RefSeq, Jun 2010]

CYP1A2 [+/-]

This gene encodes a member of the cytochrome P450 superfamily of enzymes. The cytochrome P450 proteins are monooxygenases which catalyze many reactions involved in drug metabolism and synthesis of cholesterol, steroids and other lipids. The protein encoded by this gene localizes to the endoplasmic reticulum and its expression is induced by some polycyclic aromatic hydrocarbons (PAHs), some of which are found in cigarette smoke. The enzyme's endogenous substrate is unknown; however, it is able to metabolize some PAHs to carcinogenic intermediates. Other xenobiotic substrates for this enzyme include caffeine, aflatoxin B1, and acetaminophen. The transcript from this gene contains four Alu sequences flanked by direct repeats in the 3' untranslated region. [provided by RefSeq, Jul 2008]

CYP2C9 [+/-]

Antidepressants are medicines that treat depression. Your doctor can prescribe them for you. They work to balance some of the natural chemicals in our brains. It may take several weeks for them to help. There are several types of antidepressants. You and your doctor may have to try a few before finding what works best for you.Antidepressants may cause mild side effects that usually do not last long. These may include headache, nausea, sleep problems, restlessness, and sexual problems. Tell your doctor if you have any side effects. You should also let your doctor know if you take any other medicines, vitamins, or herbal supplements.It is important to keep taking your medicines, even if you feel better. Do not stop taking your medicines without talking to your doctor. You often need to stop antidepressants gradually.NIH: National Institute of Mental Health

CYP2E1 [+/+]

This gene encodes a member of the transforming growth factor beta (TGFB) family of cytokines, which are multifunctional peptides that regulate proliferation, differentiation, adhesion, migration, and other functions in many cell types. Many cells have TGFB receptors, and the protein positively and negatively regulates many other growth factors. The secreted protein is cleaved into a latency-associated peptide (LAP) and a mature TGFB1 peptide, and is found in either a latent form composed of a TGFB1 homodimer, a LAP homodimer, and a latent TGFB1-binding protein, or in an active form composed of a TGFB1 homodimer. The mature peptide may also form heterodimers with other TGFB family members. This gene is frequently upregulated in tumor cells, and mutations in this gene result in Camurati-Engelmann disease.[provided by RefSeq, Oct 2009]

NAT2 [+/-]

Note: Loci in other organisms that are functionally homologous to this one are validly referred to as both NAT1 and NAT2; i.e., the functional homologs of NAT1 include mouse and rat Nat2, while the functional homologs of human NAT2 include mouse and rat Nat1. Name:sequence associations are consistent with current use in the field. [27 Apr 2009]

HLA-DQA2 [+/-]

SS2; BTN7; BTL-II; HSBLMHC1

IFIH1 [+/-]

DEAD box proteins, characterized by the conserved motif Asp-Glu-Ala-Asp (DEAD), are putative RNA helicases. They are implicated in a number of cellular processes involving alteration of RNA secondary structure such as translation initiation, nuclear and mitochondrial splicing, and ribosome and spliceosome assembly. Based on their distribution patterns, some members of this family are believed to be involved in embryogenesis, spermatogenesis, and cellular growth and division. This gene encodes a DEAD box protein that is upregulated in response to treatment with beta-interferon and a protein kinase C-activating compound, mezerein. Irreversible reprogramming of melanomas can be achieved by treatment with both these agents; treatment with either agent alone only achieves reversible differentiation. Genetic variation in this gene is associated with diabetes mellitus insulin-dependent type 19. [provided by RefSeq, Jul 2012]

IGF1R [+/-]

This gene encodes a tumor suppressor protein containing transcriptional activation, DNA binding, and oligomerization domains. The encoded protein responds to diverse cellular stresses to regulate expression of target genes, thereby inducing cell cycle arrest, apoptosis, senescence, DNA repair, or changes in metabolism. Mutations in this gene are associated with a variety of human cancers, including hereditary cancers such as Li-Fraumeni syndrome. Alternative splicing of this gene and the use of alternate promoters result in multiple transcript variants and isoforms. Additional isoforms have also been shown to result from the use of alternate translation initiation codons (PMIDs: 12032546, 20937277). [provided by RefSeq, Feb 2013]

IRF5 [+/-]

Studies have associated normal variations in the IRF5 gene with an increased risk of several autoimmune disorders. Autoimmune disorders occur when the immune system malfunctions and attacks the body's tissues and organs. These disorders include systemic lupus erythematosus, Sj\u00f6gren syndrome, and rheumatoid arthritis. There is some evidence that certain variations of the IRF5 gene are associated with increased activity of the gene and elevated cytokines. However, it is unknown what role, if any, these effects play in the increased risk of autoimmune disorders. Researchers believe that a combination of genetic and environmental factors may contribute to the development of these conditions.

C3 [+/+]

At least one mutation in the C3 gene has been found to cause dense deposit disease. This condition, which was formerly known as membranoproliferative glomerulonephritis type II, is a form of progressive kidney (renal) disease. The identified mutation deletes two amino acids from the C3 protein. This genetic change is described as a "gain-of-function" mutation because it leads to abnormal activation of the complement system. The overactive system creates debris that builds up in and damages certain structures in the kidneys. These structures, called glomeruli, are

clusters of tiny blood vessels that help filter waste products from the blood. Damage to glomeruli prevents the kidneys from filtering waste products normally and can lead to end-stage renal disease (ESRD), a life-threatening failure of kidney function. Several normal variants (polymorphisms) in the C3 gene have also been associated with an increased risk of developing dense deposit disease. In particular, the C3F allotype is seen more frequently in people with this condition than in the general population. Researchers are working to determine how the C3F allotype influences disease risk.

CD14 [+/-]

The protein encoded by this gene is a surface antigen that is preferentially expressed on monocytes/macrophages. It cooperates with other proteins to mediate the innate immune response to bacterial lipopolysaccharide. Alternative splicing results in multiple transcript variants encoding the same protein. [provided by RefSeq, Mar 2010]

DARC [+/-]

The protein encoded by this gene is a glycosylated membrane protein and a non-specific receptor for several chemokines. The encoded protein is the receptor for the human malarial parasites Plasmodium vivax and Plasmodium knowlesi. Polymorphisms in this gene are the basis of the Duffy blood group system. Two transcript variants encoding different isoforms have been found for this gene. [provided by RefSeq, Jul 2008]

IL5 [+/+]

This gene encodes a cytokine that acts as a growth and differentiation factor for both B cells and eosinophils. The encoded cytokine plays a major role in the regulation of eosinophil formation, maturation, recruitment and survival. The increased production of this cytokine may be related to pathogenesis of eosinophil-dependent inflammatory diseases. This cytokine functions by binding to its receptor, which is a heterodimer, whose beta subunit is shared with the receptors for interleukine 3 (IL3) and colony stimulating factor 2 (CSF2/GM-CSF). This gene is located on chromosome 5 within a cytokine gene cluster which includes interleukin 4 (IL4), interleukin 13 (IL13), and CSF2 . This gene, IL4, and IL13 may be regulated coordinately by long-range regulatory elements spread over 120 kilobases on chromosome 5q31. [provided by RefSeq, Jul 2013]

FCGR2A [+/-]

Malaria is a serious disease caused by a parasite. You get it when an infected mosquito bites you. Malaria is a major cause of death worldwide, but it is almost wiped out in the United States. The disease is mostly a problem in developing countries with warm climates. If you travel to these countries, you are at risk. There are four different types of malaria caused by four related parasites. The most deadly type occurs in Africa south of the Sahara Desert.Malaria symptoms include chills, flu-like symptoms, fever, vomiting, diarrhea, and jaundice. A blood test can diagnose it. It can be life-threatening. However, you can treat malaria with drugs. The type of drug depends on which kind of malaria you have and where you were infected.Malaria can be prevented. When traveling to malaria-prone regions See your doctor for medicines that protect you Wear insect repellent with DEET Cover up Sleep under mosquito netting Centers for Disease Control and Prevention

GSTM3 [+/-]

Cytosolic and membrane-bound forms of glutathione S-transferase are encoded by two distinct supergene families. At present, eight distinct classes of the soluble cytoplasmic mammalian glutathione S-transferases have been identified: alpha, kappa, mu, omega, pi, sigma, theta and zeta. This gene encodes a glutathione S-transferase that belongs to the mu class. The mu class of enzymes functions in the detoxification of electrophilic compounds, including carcinogens,

therapeutic drugs, environmental toxins and products of oxidative stress, by conjugation with glutathione. The genes encoding the mu class of enzymes are organized in a gene cluster on chromosome 1p13.3 and are known to be highly polymorphic. These genetic variations can change an individual's susceptibility to carcinogens and toxins as well as affect the toxicity and efficacy of certain drugs. Mutations of this class mu gene have been linked with a slight increase in a number of cancers, likely due to exposure with environmental toxins. Alternative splicing results in multiple transcript variants. [provided by RefSeq, Nov 2008]

ACE [+/-]

This gene encodes an enzyme involved in catalyzing the conversion of angiotensin I into a physiologically active peptide angiotensin II. Angiotensin II is a potent vasopressor and aldosterone-stimulating peptide that controls blood pressure and fluid-electrolyte balance. This enzyme plays a key role in the renin-angiotensin system. Many studies have associated the presence or absence of a 287 bp Alu repeat element in this gene with the levels of circulating enzyme or cardiovascular pathophysiologies. Multiple alternatively spliced transcript variants encoding different isoforms have been identified, and two most abundant spliced variants encode the somatic form and the testicular form, respectively, that are equally active. [provided by RefSeq, May 2010]

BHMT [+/-]

This gene encodes a cytosolic enzyme that catalyzes the conversion of betaine and homocysteine to dimethylglycine and methionine, respectively. Defects in this gene could lead to hyperhomocyst(e)inemia, but such a defect has not yet been observed. [provided by RefSeq, Jul 2008]

BHMT-08 [+/-]

This gene encodes a cytosolic enzyme that catalyzes the conversion of betaine and homocysteine to dimethylglycine and methionine, respectively. Defects in this gene could lead to hyperhomocyst(e)inemia, but such a defect has not yet been observed. [provided by RefSeq, Jul 2008]

CBS [+/+]

More than 150 mutations that cause homocystinuria have been identified in the CBS gene. Most of these mutations change single amino acids in cystathionine beta-synthase. The most common mutation substitutes the amino acid threonine for the amino acid isoleucine at position 278 in the enzyme (written as Ile278Thr or I278T). Another common mutation, which is the most frequent cause of homocystinuria in the Irish population, replaces the amino acid glycine with the amino acid serine at position 307 (written as Gly307Ser or G307S). These mutations disrupt the normal function of cystathionine beta-synthase. As a result, homocysteine and other potentially toxic compounds build up in the blood, and homocysteine is excreted in urine. Researchers have not determined how excess homocysteine leads to the signs and symptoms of homocystinuria.

COMT H62H [+/-]

The characteristic signs and symptoms of 22q11.2 deletion syndrome result from a deletion of a small piece of chromosome 22. The chromosomal region that is typically deleted contains 30 to 40 genes, including the COMT gene. As a result of the deletion, people with this disorder have only one copy of the COMT gene in each cell instead of the usual two copies. A loss of one copy of the COMT gene in each cell leads to abnormal regulation of catechol-O-methyltransferase levels in the brain. Researchers believe that changes involving this enzyme in the prefrontal cortex may help explain the increased risk of behavioral problems and mental illness associated with 22q11.2 deletion syndrome. Little is known, however, about the relationship between catechol-O-methyltransferase activity and

the specific mental and emotional problems characteristic of this condition. People with 22q11.2 deletion syndrome are much more likely than people without the condition to develop schizophrenia, depression, anxiety, and bipolar disorder.

COMT V158M [+/-]

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DAO [+/+]

Schizophrenia is a serious brain illness. People who have it may hear voices that aren't there. They may think other people are trying to hurt them. Sometimes they don't make sense when they talk. The disorder makes it hard for them to keep a job or take care of themselves.Symptoms of schizophrenia usually start between ages 16 and 30. Men often develop symptoms at a younger age than women. People usually do not get schizophrenia after age 45. There are three types of symptoms:Psychotic symptoms distort a person's thinking. These include hallucinations (hearing or seeing things that are not there), delusions (beliefs that are not true), trouble organizing thoughts, and strange movements."Negative" symptoms make it difficult to show emotions and to function normally. A person may seem depressed and withdrawn.Cognitive symptoms affect the thought process. These include trouble using information, making decisions, and paying attention.No one is sure what causes schizophrenia. Your genes, environment, and brain chemistry may play a role.There is no cure. Medicine can help control many of the symptoms. You may need to try different medicines to see which works best. You should stay on your medicine for as long as your doctor recommends. Additional treatments can help you deal with your illness from day to day. These include therapy, family education, rehabilitation, and skills training.NIH: National Institute of Mental Health

FOLR2 [+/-]

FOLR2

FUT2 [+/-]

The protein encoded by this gene is a Golgi stack membrane protein that is involved in the creation of a precursor of the H antigen, which is required for the final step in the soluble A and B antigen synthesis pathway. This gene is one of two encoding the galactoside 2-L-fucosyltransferase enzyme. Two transcript variants encoding the same protein have been found for this gene. [provided by RefSeq, Jul 2008]

GAD1 [+/-]

Cerebral palsy is a group of disorders that affect a person's ability to move and to maintain balance and posture. The disorders appear in the first few years of life. Usually they do not get worse over time. People with cerebral palsy may have difficulty walking. They may also have trouble with tasks such as writing or using scissors. Some have other medical conditions, including seizure disorders or mental impairment. Cerebral palsy happens when the areas of the brain that control movement and posture do not develop correctly or get damaged. Early signs of cerebral palsy usually

appear before 3 years of age. Babies with cerebral palsy are often slow to roll over, sit, crawl, smile, or walk. Some babies are born with cerebral palsy; others get it after they are born. There is no cure for cerebral palsy, but treatment can improve the lives of those who have it. Treatment includes medicines, braces, and physical, occupational and speech therapy. NIH: National Institute of Neurological Disorders and Stroke

MTHFD1L [+/+]

The protein encoded by this gene is involved in the synthesis of tetrahydrofolate (THF) in the mitochondrion. THF is important in the de novo synthesis of purines and thymidylate and in the regeneration of methionine from homocysteine. Several transcript variants encoding different isoforms have been found for this gene.[provided by RefSeq, Jun 2011]

MTHFR C677T [+/+]

At least 40 mutations in the MTHFR gene have been identified in people with homocystinuria, a disorder in which the body is unable to process certain amino acids properly. Most of these mutations change single amino acids in methylenetetrahydrofolate reductase. These changes impair the function of the enzyme, and some cause the enzyme to be turned off (inactivated). Other mutations lead to the production of an abnormally small, nonfunctional version of the enzyme. Without functional methylenetetrahydrofolate reductase, homocysteine cannot be converted to methionine. As a result, homocysteine builds up in the bloodstream, and the amount of methionine is reduced. Some of the excess homocysteine is excreted in urine. Researchers have not determined how altered levels of homocysteine and methionine lead to the various health problems affecting multiple parts of the body in people with homocystinuria.

MTHFS [+/-]

The protein encoded by this gene is an enzyme that catalyzes the conversion of 5-formyltetrahydrofolate to 5,10-methenyltetrahydrofolate, a precursor of reduced folates involved in 1-carbon metabolism. An increased activity of the encoded protein can result in an increased folate turnover rate and folate depletion. Three transcript variants encoding two different isoforms have been found for this gene. [provided by RefSeq, Jun 2011]

NOS2 [+/-]

Blood pressure is the force of your blood pushing against the walls of your arteries. Each time your heart beats, it pumps blood into the arteries. Your blood pressure is highest when your heart beats, pumping the blood. This is called systolic pressure. When your heart is at rest, between beats, your blood pressure falls. This is called diastolic pressure. Your blood pressure reading uses these two numbers. Usually the systolic number comes before or above the diastolic number. A reading of119/79 or lower is normal blood pressure140/90 or higher is high blood pressureBetween 120 and 139 for the top number, or between 80 and 89 for the bottom number is called prehypertension. Prehypertension means you may end up with high blood pressure, unless you take steps to prevent it.High blood pressure usually has no symptoms, but it can cause serious problems such as stroke, heart failure, heart attack and kidney failure.You can control high blood pressure through healthy lifestyle habits and taking medicines, if needed. NIH: National Heart, Lung, and Blood Institute

NOS3 [+/-]

Alzheimer's disease (AD) is the most common form of dementia among older people. Dementia is a brain disorder that seriously affects a person's ability to carry out daily activities. AD begins slowly. It first involves the parts of the brain that control thought, memory and language. People with AD may have trouble remembering things that happened recently or names of people they know. A related problem, mild cognitive impairment (MCI), causes more memory problems than normal for people of the same age. Many, but not all, people with MCI will develop AD. In AD, over time,

symptoms get worse. People may not recognize family members or have trouble speaking, reading or writing. They may forget how to brush their teeth or comb their hair. Later on, they may become anxious or aggressive, or wander away from home. Eventually, they need total care. This can cause great stress for family members who must care for them. AD usually begins after age 60. The risk goes up as you get older. Your risk is also higher if a family member has had the disease. No treatment can stop the disease. However, some drugs may help keep symptoms from getting worse for a limited time. NIH: National Institute on Aging

PEMT [+/+]

This gene encodes a membrane-bound protein that is a member of the mucin family. Mucins are O-glycosylated proteins that play an essential role in forming protective mucous barriers on epithelial surfaces. These proteins also play a role in intracellular signaling. This protein is expressed on the apical surface of epithelial cells that line the mucosal surfaces of many different tissues including lung, breast stomach and pancreas. This protein is proteolytically cleaved into alpha and beta subunits that form a heterodimeric complex. The N-terminal alpha subunit functions in cell-adhesion and the C-terminal beta subunit is involved in cell signaling. Overexpression, aberrant intracellular localization, and changes in glycosylation of this protein have been associated with carcinomas. This gene is known to contain a highly polymorphic variable number tandem repeats (VNTR) domain. Alternate splicing results in multiple transcript variants.[provided by RefSeq, Feb 2011]

SOD2 [+/+]

This gene encodes a multifunctional proinflammatory cytokine that belongs to the tumor necrosis factor (TNF) superfamily. This cytokine is mainly secreted by macrophages. It can bind to, and thus functions through its receptors TNFRSF1A/TNFR1 and TNFRSF1B/TNFBR. This cytokine is involved in the regulation of a wide spectrum of biological processes including cell proliferation, differentiation, apoptosis, lipid metabolism, and coagulation. This cytokine has been implicated in a variety of diseases, including autoimmune diseases, insulin resistance, and cancer. Knockout studies in mice also suggested the neuroprotective function of this cytokine. [provided by RefSeq, Jul 2008]

SOD3 [+/+]

This gene encodes an extracellular matrix glycoprotein that is characterized by an N-terminal microfibril interface domain, a coiled-coiled alpha-helical domain, a collagenous domain and a C-terminal globular C1q domain. The encoded protein associates with elastic fibers at the interface between elastin and microfibrils and may play a role in the development of elastic tissues including large blood vessels, dermis, heart and lung. [provided by RefSeq, Sep 2009]

VDR Bsm [+/-]

Mutations in the VDR gene cause vitamin D-dependent rickets type 2 (VDDR2), also known as hereditary vitamin D-resistant rickets (HVDRR). This disorder of bone development is characterized by low levels of calcium (hypocalcemia) and phosphate (hypophosphatemia) in the blood, which lead to soft, weak bones (rickets) that are prone to fracture. A common feature of this condition is bowed legs. The VDR gene mutations that cause this condition prevent the VDR protein from functioning properly. Some changes in the VDR gene lead to an abnormally short version of the VDR protein; others result in the production of an abnormal receptor that cannot bind to calcitriol, to RXR, or to DNA. Despite plenty of calcitriol in the body, the altered VDR cannot stimulate gene activity important for mineral absorption. The lack of calcium and phosphate absorption in the intestines slows deposition of these minerals into developing bone (bone mineralization), which leads to soft, weak bones and other features of VDDR2. Hypocalcemia also causes muscle weakness and seizures in some affected individuals. Most VDR gene mutations impair hair growth, leading to alopecia; however, mutations that block VDR's ability to interact with calcitriol do not cause alopecia, indicating that calcitriol is not necessary for the receptor's role in hair development.

COX5A [+/-]

COX5A

NDUFS3 [+/-]

A genetic brain disorder is caused by a variation or a mutation in a gene. A variation is a different form of a gene. A mutation is a change in a gene. Genetic brain disorders affect the development and function of the brain.Some genetic brain disorders are due to random gene mutations or mutations caused by environmental exposure, such as cigarette smoke. Other disorders are inherited, which means that a mutated gene or group of genes is passed down through a family. They can also be due to a combination of both genetic changes and other outside factors.Some examples of genetic brain disorders includeLeukodystrophiesPhenylketonuriaTay-Sachs diseaseWilson diseaseMany people with genetic brain disorders fail to produce enough of certain proteins that influence brain development and function. These brain disorders can cause serious problems that affect the nervous system. Some have treatments to control symptoms. Some are life-threatening.

NDUFS7 [+/-]

A genetic brain disorder is caused by a variation or a mutation in a gene. A variation is a different form of a gene. A mutation is a change in a gene. Genetic brain disorders affect the development and function of the brain.Some genetic brain disorders are due to random gene mutations or mutations caused by environmental exposure, such as cigarette smoke. Other disorders are inherited, which means that a mutated gene or group of genes is passed down through a family. They can also be due to a combination of both genetic changes and other outside factors.Some examples of genetic brain disorders includeLeukodystrophiesPhenylketonuriaTay-Sachs diseaseWilson diseaseMany people with genetic brain disorders fail to produce enough of certain proteins that influence brain development and function. These brain disorders can cause serious problems that affect the nervous system. Some have treatments to control symptoms. Some are life-threatening.

SLC19A1 [+/-]

The membrane protein encoded by this gene is a transporter of folate and is involved in the regulation of intracellular concentrations of folate. Three transcript variants encoding different isoforms have been found for this gene.[provided by RefSeq, Mar 2011]

ATG16L1 [+/-]

At least one variation in the ATG16L1 gene is associated with an increased risk of Crohn disease, particularly a form of the disorder that affects the lower part of the small intestine (the ileum). This increased risk has been found primarily in Caucasian (white) populations. The identified ATG16L1 variation changes a single protein building block (amino acid) in a critical region of the autophagy related 16-like 1 protein. Specifically, it replaces the amino acid threonine with the amino acid alanine at protein position 300 (written as Thr300Ala or T300A). The effects of variations in the ATG16L1 gene on Crohn disease risk are unclear. Changes in this gene may affect the autophagy process, allowing worn-out cell parts and harmful bacteria to persist when they would otherwise be destroyed. These cell components and bacteria may trigger an inappropriate immune system response, leading to chronic inflammation in the intestinal walls and the digestive problems characteristic of Crohn disease. Researchers continue to study the relationship between changes in the ATG16L1 gene and a person's risk of developing this disorder.

HLA-DRB1 [+/-]

SS2; BTN7; BTL-II; HSBLMHC1

STAT4 [+/-]

Studies have associated a normal variation in the STAT4 gene with an increased risk of several autoimmune disorders. Autoimmune disorders occur when the immune system malfunctions and attacks the body's tissues and organs. These disorders include systemic lupus erythematosus, rheumatoid arthritis, and Sj\u00f6gren syndrome. The variant associated with increased risk of autoimmune disorders changes a single DNA building block (nucleotide) in the STAT4 gene. It is unknown how the gene variation contributes to increased risk of these conditions. Researchers believe that a combination of genetic and environmental factors may play a role in development of autoimmunity.

TNF [+/-]

Asthma is a chronic disease that affects your airways. Your airways are tubes that carry air in and out of your lungs. If you have asthma, the inside walls of your airways become sore and swollen. That makes them very sensitive, and they may react strongly to things that you are allergic to or find irritating. When your airways react, they get narrower and your lungs get less air.Symptoms of asthma includeWheezingCoughing, especially early in the morning or at nightChest tightnessShortness of breathNot all people who have asthma have these symptoms. Having these symptoms doesn't always mean that you have asthma. Your doctor will diagnose asthma based on lung function tests, your medical history, and a physical exam. You may also have allergy tests.When your asthma symptoms become worse than usual, it's called an asthma attack. Severe asthma attacks may require emergency care, and they can be fatal.Asthma is treated with two kinds of medicines: quick-relief medicines to stop asthma symptoms and long-term control medicines to prevent symptoms. NIH: National Heart, Lung, and Blood Institute

SULT2A1 [+/-]

This gene encodes a member of the sulfotransferase family. Sulfotransferases aid in the metabolism of drugs and endogenous compounds by converting these substances into more hydrophilic water-soluble sulfate conjugates that can be easily excreted. This protein catalyzes the sulfation of steroids and bile acids in the liver and adrenal glands, and may have a role in the inherited adrenal androgen excess in women with polycystic ovary syndrome. [provided by RefSeq, Mar 2010]

CTLA4 [+/-]

Diabetes is a disease in which your blood glucose, or blood sugar, levels are too high. Glucose comes from the foods you eat. Insulin is a hormone that helps the glucose get into your cells to give them energy. With type 1 diabetes, your body does not make insulin. With type 2 diabetes, the more common type, your body does not make or use insulin well. Without enough insulin, the glucose stays in your blood. You can also have prediabetes. This means that your blood sugar is higher than normal but not high enough to be called diabetes. Having prediabetes puts you at a higher risk of getting type 2 diabetes.Over time, having too much glucose in your blood can cause serious problems. It can damage your eyes, kidneys, and nerves. Diabetes can also cause heart disease, stroke and even the need to remove a limb. Pregnant women can also get diabetes, called gestational diabetes.A blood test can show if you have diabetes. Exercise, weight control and sticking to your meal plan can help control your diabetes. You should also monitor your glucose level and take medicine if prescribed. NIH: National Institute of Diabetes and Digestive and Kidney Diseases

FOXE1 [+/+]

Your thyroid is a butterfly-shaped gland in your neck, just above your collarbone. It is one of your endocrine glands, which make hormones. Thyroid hormones control the rate of many activities in your body. These include how fast you burn calories and how fast your heart beats. All of these activities are your body's metabolism. If your thyroid gland is not active enough, it does not make enough thyroid hormone to meet your body's needs. This condition is hypothyroidism.Hypothyroidism is more common in women, people with other thyroid problems, and those over 60 years old. Hashimoto's disease, an autoimmune disorder, is the most common cause. Other causes include thyroid nodules, thyroiditis, congenital hypothyroidism, surgical removal of part or all of the thyroid, radiation treatment of the thyroid, and some medicines.The symptoms can vary from person to person. They may includeFatigueWeight gainA puffy faceCold intoleranceJoint and muscle painConstipationDry skinDry, thinning hairDecreased sweatingHeavy or irregular menstrual periods and fertility problemsDepressionSlowed heart rateTo diagnose hypothyroidism, your doctor will look at your symptoms and blood tests. Treatment is with synthetic thyroid hormone, taken every day.NIH: National Institute of Diabetes and Digestive and Kidney Diseases

RARA [+/+]

Gene mutations can be acquired during a person's lifetime and are present only in certain cells. These mutations are called somatic mutations, and they are not inherited. A somatic mutation involving the RARA gene causes acute promyelocytic leukemia, a cancer of the blood forming tissue (bone marrow). Acute promyelocytic leukemia is characterized by an accumulation of promyelocytes in the bone marrow. A rearrangement (translocation) of genetic material between chromosomes 15 and 17, written as t(15;17), fuses part of the RARA gene on chromosome 17 with part of another gene on chromosome 15 called PML . The protein produced from this fused gene, the PML-RAR\u03b1 protein, functions differently than the protein products of the normal PML and RARA genes. The PML-RAR\u03b1 protein binds to DNA and represses gene transcription, like the normal RAR\u03b1 protein. However, the PML-RAR\u03b1 protein does not respond to the signal to induce transcription of genes, so the genes remain repressed. Additionally, the function of the PML protein, the product of the PML gene, is disrupted. The PML protein blocks cell growth and division (proliferation) and induces self-destruction (apoptosis) in combination with other proteins. However, the PML-RAR\u03b1 protein does not block proliferation or induce apoptosis. The PML-RAR\u03b1 protein blocks the differentiation of blood cells at the promyelocyte stage and allows abnormal cell proliferation. As a result, excess promyelocytes accumulate in the bone marrow and normal white blood cells cannot form, leading to acute promyelocytic leukemia.

information for decision making, patients with possible inherited diseases should undergo genetic testing only in the context of genetic counseling.

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Do not make any decisions about your health solely based on the information contained in this report. Always consult with a licensed and experienced health practitioner when you recieve your report

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