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Gene Variance report

Shelby Chambers 17 / F

ALLERGY

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

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CLOTTING FACTORS

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	CETP	rs1800775	C	AC	+/-
2	CYP4V2	rs13146272	C	AC	+/-
3	F10	rs3211719	G	AG	+/-
5	F11	rs2289252	T	CT	+/-
8	F5	rs6025	T	CC	-/-
10	F9	rs6048	G	AA	-/-
11	GP6	rs1613662	G	AA	-/-
12	HRG	rs9898	T	CC	-/-
13	ITGB3	rs5918	C	TT	-/-
14	KNGI598T	rs2731672	T	CC	-/-
15	NR1I2	rs1523127	C	AC	+/-
16	SERPINC1	rs2227589	T	CT	+/-

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DETOX

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	CTH	rs1021737	T	GG	-/-
3	CYP1A1	rs1799814	T	GG	-/-
4	CYP1A1	rs4986883	C	TT	-/-
5	CYP1A2	rs762551	C	AC	+/-
7	CYP1B1 N453S	rs1800440	C	TT	-/-
11	CYP2C9	rs1057910	C	AA	-/-
12	CYP2C9*2 C430T	rs1799853	T	TT	+/+
13	CYP2D6	rs1065852	A	GG	-/-
15	CYP2D6 T2850C	rs16947	A	AG	+/-
17	CYP2E1*1B G9896C	rs2070676	G	CC	-/-
18	CYP2E1*4 A4768G	rs6413419	A	GG	-/-
20	CYP3A4	rs2740574	C	TT	-/-
21	CYP3A4	rs4986910	G	AA	-/-
22	CYP3A4*2 S222P	rs55785340	G	AA	-/-
24	GSTM1	rs1056806	T	CC	-/-
26	GSTM1	rs4147565	A	--	NC
28	GSTP1	rs1138272	T	CC	-/-
29	GSTP1	rs1695	G	GG	+/+
30	NAT1	rs4986782	A	GG	-/-
31	NAT2	rs1208	G	AG	+/-
32	NAT2	rs1799930	A	GG	-/-
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	rs2858331	G	AA	-/-
2	HLA-DQA1	rs2187668	T	CC	-/-

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IGA

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	CFH	rs6677604	A	GG	-/-
2	HLA	rs9271366	G	AA	-/-
3	HLA-DPB2 / COL11A2P	rs1883414	A	AG	+/-
4	HLA-DQA2	rs9275224	A	AG	+/-
5	HORMAD2	rs2412971	A	GG	-/-
6	IFIH1	rs1990760	T	TT	+/+
7	IGF1R	rs2229765	A	AA	+/+
8	IRF5	rs4728142	A	AA	+/+
9	MTC03P1	rs2856717	A	AG	+/-
10	MTC03P1	rs9275596	C	CT	+/-
11	PSMB8	rs9357155	A	GG	-/-
12	TRAF1	rs3761847	G	AG	+/-

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IGE

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	C3	rs10402876	C	CG	+/-
3	CD14	rs2569191	C	CT	+/-
4	DARC	rs2814778	C	TT	-/-
5	FCER1A	rs2251746	C	CT	+/-
6	FCER1A	rs2427824	T	CT	+/-
7	FCER1A	rs2427827	T	CT	+/-
10	FCER1A / OR10J2P	rs10489854	T	CC	-/-
12	IL13	rs1800925	T	CT	+/-
13	IL5	rs2069812	G	AG	+/-
14	RAD50	rs17772565	T	CC	-/-
15	RAD50	rs17772583	G	AG	+/-
16	RAD50	rs2040704	G	AA	-/-
17	RAD50	rs2240032	T	CC	-/-
18	RAD50	rs6884762	T	CC	-/-
19	RAG1	rs3740955	A	AA	+/+
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	CC	-/-
4	MUC21	rs1634731	G	AA	-/-

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METHYLATION

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	ACAT1	rs3741049	A	GG	-/-
2	ACE Del16	rs4343	G	GG	+/+
3	AGT M235T/C4072T	rs699	A	AA	+/+
4	AHCY-01	rs819147	C	TT	-/-
6	AHCY-19	rs819171	C	TT	-/-
8	BHMT R239Q	rs3733890	A	GG	-/-
9	BHMT-02	rs567754	T	CT	+/-
11	BHMT-08	rs651852	T	CT	+/-
13	CBS A13637G	rs2851391	T	CT	+/-
14	CBS A360A	rs1801181	A	AG	+/-
15	CBS C19150T	rs4920037	A	AG	+/-
16	CBS C699T	rs234706	A	AG	+/-
18	CLCN6	rs13306560	T	CC	-/-
21	DAO	rs2070586	A	AG	+/-
23	DAO	rs3741775	C	AA	-/-
24	DHFR	rs1643649	C	TT	-/-
25	FOLR1	rs2071010	A	GG	-/-
26	FOLR2	rs651933	G	AG	+/-
27	FOLR3	rs7925545	G	AA	-/-
29	FUT2	rs492602	G	GG	+/+
30	FUT2	rs601338	A	AA	+/+
31	FUT2	rs602662	A	AA	+/+
32	G6PD	rs1050828	T	CC	-/-
33	G6PD	rs1050829	C	TT	-/-
34	GAD1	rs10432420	A	GG	-/-
35	GAD1	rs12185692	A	CC	-/-
36	GAD1	rs2058725	C	TT	-/-
37	GAD1	rs2241165	C	TT	-/-
38	GAD1	rs3749034	A	GG	-/-
39	GAD1	rs3791850	A	GG	-/-
40	GAD1	rs3791851	C	CT	+/-
41	GAD1	rs3791878	T	TT	+/+
42	GAD1	rs3828275	T	CT	+/-

	Gene	rsID	Minor Allele	Genotype	Phenotype
43	GAD1	rs701492	T	CT	+/-
45	GAD1	rs769407	C	CG	+/-
47	GAMT	rs17851582	A	AG	+/-
48	GAMT	rs55776826	T	CT	+/-
49	GIF	rs558660	A	GG	-/-
50	MAOA R297R	rs6323	G	GT	+/-
51	MAOB	rs1799836	C	CT	+/-
52	MIR4761 (COMT -61 P199P)	rs769224	A	GG	-/-
53	MIR4761 (COMT H62H)	rs4633	T	CT	+/-
54	MIR4761 (COMT V158M)	rs4680	A	AG	+/-
55	MIR4761 (COMT)	rs6269	G	AG	+/-
56	MTHFD1 C105T	rs1076991	T	TT	+/+
57	MTHFD1 G1958A	rs2236225	A	AG	+/-
58	MTHFD1L	rs11754661	A	GG	-/-
59	MTHFD1L	rs17349743	C	CC	+/+
60	MTHFD1L	rs6922269	A	GG	-/-
61	MTHFD1L	rs803422	A	GG	-/-
63	MTHFR	rs1476413	T	CC	-/-
64	MTHFR	rs17037390	A	GG	-/-
66	MTHFR	rs4846049	T	GG	-/-
67	MTHFR (LOC100506310)	rs4846048	G	AA	-/-
68	MTHFR 03 P39P	rs2066470	A	GG	-/-
69	MTHFR A1298C	rs1801131	G	TT	-/-
70	MTHFR A1572G	rs17367504	G	AA	-/-
71	MTHFR C677T	rs1801133	A	AA	+/+
72	MTHFR G1793A (R594Q)	rs2274976	T	CC	-/-
73	MTHFS	rs6495446	T	CC	-/-
74	MTR A2756G	rs1805087	G	GG	+/+
75	MTRR A66G	rs1801394	G	AG	+/-
77	MTRR K350A	rs162036	G	AA	-/-
79	MTRR-11 A664A	rs1802059	A	AG	+/-
81	NOS2	rs2248814	A	AA	+/+
82	NOS2	rs2274894	T	TT	+/+
83	NOS2	rs2297518	A	GG	-/-
84	NOS3	rs1800779	G	AG	+/-
85	NOS3	rs1800783	A	AT	+/-
88	PEMT	rs4244593	T	GT	+/-

	Gene	rsID	Minor Allele	Genotype	Phenotype
93	SOD2	rs2758331	A	AC	+/-
94	SOD2	rs4880	G	AG	+/-
95	SOD3	rs2855262	C	CC	+/+
96	TCN1	rs526934	G	AA	-/-
97	TCN2 C766G	rs1801198	G	CC	-/-
98	VDR Bsm	rs1544410	T	CC	-/-

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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	CT	+/-
16	NDUFS3	rs4147730	A	AA	+/+
19	NDUFS7	rs1142530	T	TT	+/+
22	NDUFS7	rs2332496	A	AG	+/-
24	NDUFS7	rs7258846	T	TT	+/+
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	4q27 Region	rs6822844	T	GG	-/-
2	ADD1 G460W	rs4961	T	GG	-/-
3	APOE	rs429358	C	CT	+/-
4	ATG16L1	rs10210302	T	TT	+/+
5	HLA-DRB1	rs660895	G	AA	-/-
6	IL13	rs20541	A	AG	+/-
7	IL4R	rs1801275	G	AG	+/-
8	MEFV	rs11466023	A	GG	-/-
10	STAT4	rs10181656	G	CC	-/-
11	TNF	rs1800629	A	GG	-/-
12	TNF	rs361525	A	GG	-/-

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SULFOTRANSFERASE

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

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THYROID

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

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TONGUE TIE

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

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Gene Definitions

HLA [+/+]

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 [+/-]

This gene encodes a member of the cytochrome P450 hemethiolate protein superfamily which are involved in oxidizing various substrates in the metabolic pathway. It is implicated in the metabolism of fatty acid precursors into n-3 polyunsaturated fatty acids. Mutations in this gene result in Bietti crystalline corneoretinal dystrophy. [provided by RefSeq, Jul 2008]

F10 [+/-]

Normally, if you get hurt, your body forms a blood clot to stop the bleeding. For blood to clot, your body needs cells called platelets and proteins known as clotting factors. If you have a bleeding disorder, you either do not have enough platelets or clotting factors or they don't work the way they should. Bleeding disorders can be the result of other diseases, such as severe liver disease. They can also be inherited. Hemophilia is an inherited bleeding disorder. Bleeding disorders can also be a side effect of medicines.

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]

NR1I2 [+/-]

NR1I2

SERPINC1 [+/-]

The protein encoded by this gene is a plasma protease inhibitor and a member of the serpin superfamily. This protein inhibits thrombin as well as other activated serine proteases of the coagulation system, and it regulates the blood

coagulation cascade. The protein includes two functional domains: the heparin binding-domain at the N-terminus of the mature protein, and the reactive site domain at the C-terminus. The inhibitory activity is enhanced by the presence of heparin. More than 120 mutations have been identified for this gene, many of which are known to cause antithrombin-III deficiency. [provided by RefSeq, Jul 2009]

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*2 C430T [+/+]

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. This protein localizes to the endoplasmic reticulum and its expression is induced by rifampin. The enzyme is known to metabolize many xenobiotics, including phenytoin, tolbutamide, ibuprofen and S-warfarin. Studies identifying individuals who are poor metabolizers of phenytoin and tolbutamide suggest that this gene is polymorphic. The gene is located within a cluster of cytochrome P450 genes on chromosome 10q24. [provided by RefSeq, Jul 2008]

CYP2D6 T2850C [+/-]

Caffeine is a bitter substance found in coffee, tea, soft drinks, chocolate, kola nuts, and certain medicines. It has many effects on the body's metabolism, including stimulating the central nervous system. This can make you more alert and give you a boost of energy. For most people, the amount of caffeine in two to four cups of coffee a day is not harmful. However, too much caffeine can cause problems. It canMake you jittery and shakyMake it hard to fall asleep or stay asleepCause headaches or dizzinessMake your heart beat faster or cause abnormal heart rhythmsCause dehydrationMake you dependent on it so you need to take more of it. If you stop using caffeine, you could get withdrawal symptoms.Some people are more sensitive to the effects of caffeine than others. They should limit their use of caffeine. So should pregnant and nursing women. Certain drugs and supplements may interact with caffeine. If you have questions about whether caffeine is safe for you, talk with your health care provider.Food and Drug Administration

GSTP1 [+/+]

Glutathione S-transferases (GSTs) are a family of enzymes that play an important role in detoxification by catalyzing the conjugation of many hydrophobic and electrophilic compounds with reduced glutathione. Based on their biochemical, immunologic, and structural properties, the soluble GSTs are categorized into 4 main classes: alpha, mu, pi, and theta. This GST family member is a polymorphic gene encoding active, functionally different GSTP1 variant proteins that are thought to function in xenobiotic metabolism and play a role in susceptibility to cancer, and other diseases. [provided by RefSeq, Jul 2008]

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 [+/-]

This gene belongs to the HLA class II alpha chain family. The encoded protein forms a heterodimer with a class II beta chain. It is located in intracellular vesicles and plays a central role in the peptide loading of MHC class II molecules by helping to release the CLIP molecule from the peptide binding site. Class II molecules are expressed in antigen presenting cells (B lymphocytes, dendritic cells, macrophages) and are used to present antigenic peptides on the cell surface to be recognized by CD4 T-cells. [provided by RefSeq, Jun 2010]

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 receptor binds insulin-like growth factor with a high affinity. It has tyrosine kinase activity. The insulin-like growth factor I receptor plays a critical role in transformation events. Cleavage of the precursor generates alpha and beta subunits. It is highly overexpressed in most malignant tissues where it functions as an anti-apoptotic agent by enhancing cell survival. Alternatively spliced transcript variants encoding distinct isoforms have been found for this gene. [provided by RefSeq, May 2014]

IRF5 [+/+]

This gene encodes a member of the interferon regulatory factor (IRF) family, a group of transcription factors with diverse roles, including virus-mediated activation of interferon, and modulation of cell growth, differentiation, apoptosis, and immune system activity. Members of the IRF family are characterized by a conserved N-terminal DNA-binding domain containing tryptophan (W) repeats. Multiple transcript variants encoding different isoforms have been found for this gene, and a 30-nt indel polymorphism (SNP rs60344245) can result in loss of a 10-aa segment. [provided by RefSeq, Mar 2010]

TRAF1 [+/-]

The protein encoded by this gene is the fifth component of complement, which plays an important role in inflammatory and cell killing processes. This protein is comprised of alpha and beta polypeptide chains that are linked by a disulfide bridge. An activation peptide, C5a, which is an anaphylatoxin that possesses potent spasmogenic and chemotactic activity, is derived from the alpha polypeptide via cleavage with a convertase. The C5b macromolecular cleavage product can form a complex with the C6 complement component, and this complex is the basis for formation of the membrane attack complex, which includes additional complement components. Mutations in this gene cause complement component 5 deficiency, a disease where patients show a propensity for severe recurrent infections. Defects in this gene have also been linked to a susceptibility to liver fibrosis and to rheumatoid arthritis. [provided by

RefSeq, Jul 2008]

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]

FCER1A [+/-]

The immunoglobulin epsilon receptor (IgE receptor) is the initiator of the allergic response. When two or more high-affinity IgE receptors are brought together by allergen-bound IgE molecules, mediators such as histamine that are responsible for allergy symptoms are released. This receptor is comprised of an alpha subunit, a beta subunit, and two gamma subunits. The protein encoded by this gene represents the alpha subunit. [provided by RefSeq, Aug 2011]

IL13 [+/-]

This gene encodes an immunoregulatory cytokine produced primarily by activated Th2 cells. This cytokine is involved in several stages of B-cell maturation and differentiation. It up-regulates CD23 and MHC class II expression, and promotes IgE isotype switching of B cells. This cytokine down-regulates macrophage activity, thereby inhibits the production of pro-inflammatory cytokines and chemokines. This cytokine is found to be critical to the pathogenesis of allergen-induced asthma but operates through mechanisms independent of IgE and eosinophils. This gene, IL3, IL5, IL4, and CSF2 form a cytokine gene cluster on chromosome 5q, with this gene particularly close to IL4. [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]

RAD50 [+/-]

Some research suggests that inherited mutations in the RAD50 gene are associated with an increased risk of developing breast cancer. Other studies have not found this connection, and researchers believe that mutations in the RAD50 gene are not a major genetic risk factor for developing this disease. RAD50 mutations can lead to the production of an abnormally small, nonfunctional version of the RAD50 protein. When this protein is defective or missing, cells may be unable to respond effectively to DNA damage. As defects accumulate in DNA, they can trigger cells to grow and divide uncontrollably and form a tumor.

RAG1 [+/+]

Your immune system is a complex network of cells, tissues, and organs that work together to defend against germs. It helps your body to recognize these "foreign" invaders. Then its job is to keep them out, or if it can't, to find and destroy them.If your immune system cannot do its job, the results can be serious. Disorders of the immune system includeAllergy and asthma - immune responses to substances that are usually not harmfulImmune deficiency diseases - disorders in which the immune system is missing one or more of its partsAutoimmune diseases - diseases causing your immune system to attack your own body's cells and tissues by mistakeNIH: National Institute of Allergy and Infectious Diseases

FCGR2A [+/-]

This gene encodes one member of a family of immunoglobulin Fc receptor genes found on the surface of many immune response cells. The protein encoded by this gene is a cell surface receptor found on phagocytic cells such as macrophages and neutrophils, and is involved in the process of phagocytosis and clearing of immune complexes. Alternative splicing results in multiple transcript variants. [provided by RefSeq, Oct 2008]

ACE Del16 [+/+]

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]

AGT M235T/C4072T [+/+]

At least six mutations in the AGT gene have been found to cause a severe kidney disorder called renal tubular dysgenesis. This condition is characterized by abnormal kidney development before birth, the inability to produce urine (anuria), and severe low blood pressure (hypotension). These problems result in a reduction of amniotic fluid (oligohydramnios), which leads to a set of birth defects known as the Potter sequence. Renal tubular dysgenesis can be caused by mutations in both copies of any of the genes involved in the renin-angiotensin system. Most of the mutations in the AGT gene that cause this disorder change single protein building blocks (amino acids) in the angiotensinogen protein. These changes occur in a region of the protein that is necessary for its conversion to angiotensin I. It is thought that the altered angiotensinogen cannot be converted, leading to a nonfunctional renin-angiotensin system. Without this system, the kidneys cannot control blood pressure. Because of low blood pressure, the flow of blood is reduced (hypoperfusion), and the body does not get enough oxygen during fetal development. As a result, kidney development

is impaired, leading to the features of renal tubular dysgenesis.

CBS A13637G [+/-]

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.

CBS A360A [+/-]

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.

CBS C19150T [+/-]

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.

CBS C699T [+/-]

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.

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 [+/-]

The protein encoded by this gene is a member of the folate receptor (FOLR) family, and these genes exist in a cluster on chromosome 11. Members of this gene family have a high affinity for folic acid and for several reduced folic acid derivatives, and they mediate delivery of 5-methyltetrahydrofolate to the interior of cells. This protein has a 68% and 79% sequence homology with the FOLR1 and FOLR3 proteins, respectively. Although this protein was originally thought to be specific to placenta, it can also exist in other tissues, and it may play a role in the transport of methotrexate in synovial macrophages in rheumatoid arthritis patients. Multiple transcript variants that encode the same protein have been found for this gene. [provided by RefSeq, Jul 2008]

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

GAMT [+/-]

At least 15 mutations in the GAMT gene cause guanidinoacetate methyltransferase deficiency, a disorder that involves intellectual disability and seizures. Most affected individuals of Portuguese ancestry have a particular mutation in which the amino acid tryptophan is replaced by the amino acid serine at position 20 in the enzyme (written as Trp20Ser or W20S). GAMT gene mutations impair the ability of the guanidinoacetate methyltransferase enzyme to participate in creatine synthesis, resulting in a shortage of creatine. The effects of guanidinoacetate methyltransferase deficiency are most severe in organs and tissues that require large amounts of energy, especially the brain.

MAOA R297R [+/-]

Developmental disabilities are severe, long-term problems. They may be physical, such as blindness. They may affect mental ability, such as learning disorders. Or the problem can be both physical and mental, such as Down syndrome. The problems are usually life-long, and can affect everyday living. There are many causes of developmental disabilities, includingGenetic or chromosome abnormalities. These cause conditions such as Down syndrome and Rett syndrome.Prenatal exposure to substances. Drinking alcohol when pregnant can cause fetal alcohol spectrum disorders.Certain viral infections during pregnancyPreterm birthOften there is no cure, but treatment can help the symptoms. Treatments include physical, speech, and occupational therapy. Special education classes and psychological counseling can also help.NIH: National Institute of Child Health and Human Development

MAOB [+/-]

The protein encoded by this gene belongs to the flavin monoamine oxidase family. It is a enzyme located in the mitochondrial outer membrane. It catalyzes the oxidative deamination of biogenic and xenobiotic amines and plays an important role in the metabolism of neuroactive and vasoactive amines in the central nervous sysytem and peripheral tissues. This protein preferentially degrades benzylamine and phenylethylamine. [provided by RefSeq, Jul 2008]

MIR4761 (COMT H62H) [+/-]

Catechol-O-methyltransferase catalyzes the transfer of a methyl group from S-adenosylmethionine to catecholamines, including the neurotransmitters dopamine, epinephrine, and norepinephrine. This O-methylation results in one of the major degradative pathways of the catecholamine transmitters. In addition to its role in the metabolism of endogenous substances, COMT is important in the metabolism of catechol drugs used in the treatment of hypertension, asthma, and Parkinson disease. COMT is found in two forms in tissues, a soluble form (S-COMT) and a membrane-bound form (MB-COMT). The differences between S-COMT and MB-COMT reside within the N-termini. Several transcript variants are formed through the use of alternative translation initiation sites and promoters. [provided by RefSeq, Sep 2008]

MIR4761 (COMT V158M) [+/-]

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MIR4761 (COMT) [+/-]

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MTHFD1 C105T [+/+]

Neural tube defects are birth defects of the brain, spine, or spinal cord. They happen in the first month of pregnancy, often before a woman even knows that she is pregnant. The two most common neural tube defects are spina bifida and anencephaly. In spina bifida, the fetal spinal column doesn't close completely. There is usually nerve damage that causes at least some paralysis of the legs. In anencephaly, most of the brain and skull do not develop. Babies with anencephaly are either stillborn or die shortly after birth. Another type of defect, Chiari malformation, causes the brain tissue to extend into the spinal canal.The exact causes of neural tube defects aren't known. You're at greater risk of having an infant with a neural tube defect if youAre obeseHave poorly controlled diabetesTake certain antiseizure medicinesGetting enough folic acid, a type of B vitamin, before and during pregnancy prevents most neural tube defects.Neural tube defects are usually diagnosed before the infant is born, through lab or imaging tests. There is no cure for neural tube defects. The nerve damage and loss of function that are present at birth are usually permanent. However, a variety of treatments can sometimes prevent further damage and help with complications.NIH: National Institute of Child Health and Human Development

MTHFD1 G1958A [+/-]

Neural tube defects are birth defects of the brain, spine, or spinal cord. They happen in the first month of pregnancy, often before a woman even knows that she is pregnant. The two most common neural tube defects are spina bifida and anencephaly. In spina bifida, the fetal spinal column doesn't close completely. There is usually nerve damage that causes at least some paralysis of the legs. In anencephaly, most of the brain and skull do not develop. Babies with anencephaly are either stillborn or die shortly after birth. Another type of defect, Chiari malformation, causes the brain tissue to extend into the spinal canal.The exact causes of neural tube defects aren't known. You're at greater risk of having an infant with a neural tube defect if youAre obeseHave poorly controlled diabetesTake certain antiseizure medicinesGetting enough folic acid, a type of B vitamin, before and during pregnancy prevents most neural tube defects.Neural tube defects are usually diagnosed before the infant is born, through lab or imaging tests. There is no cure for neural tube defects. The nerve damage and loss of function that are present at birth are usually permanent. However, a variety of treatments can sometimes prevent further damage and help with complications.NIH: National Institute of Child Health and Human Development

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. 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 health problems associated with homocystinuria.

MTR A2756G [+/+]

More than 20 mutations in the MTR gene have been identified in people with homocystinuria. Many of these mutations lead to the production of an abnormally small, nonfunctional version of methionine synthase. Other mutations change single amino acids in the enzyme. One of the most common mutations replaces the amino acid proline with the amino acid leucine at position 1173 (written as Pro1173Leu or P1173L), resulting in an enzyme with reduced function. Without functional methionine synthase, 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 health problems associated with homocystinuria.

MTRR A66G [+/-]

The protein encoded by this gene catalyzes the conversion of 5,10-methylenetetrahydrofolate to 5-methyltetrahydrofolate, a co-substrate for homocysteine remethylation to methionine. Genetic variation in this gene influences susceptibility to occlusive vascular disease, neural tube defects, colon cancer and acute leukemia, and mutations in this gene are associated with methylenetetrahydrofolate reductase deficiency.[provided by RefSeq, Oct 2009]

MTRR-11 A664A [+/-]

The protein encoded by this gene catalyzes the conversion of 5,10-methylenetetrahydrofolate to 5-methyltetrahydrofolate, a co-substrate for homocysteine remethylation to methionine. Genetic variation in this gene influences susceptibility to occlusive vascular disease, neural tube defects, colon cancer and acute leukemia, and mutations in this gene are associated with methylenetetrahydrofolate reductase deficiency.[provided by RefSeq, Oct 2009]

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 a member of the superoxide dismutase (SOD) protein family. SODs are antioxidant enzymes that catalyze the dismutation of two superoxide radicals into hydrogen peroxide and oxygen. The product of this gene is thought to protect the brain, lungs, and other tissues from oxidative stress. The protein is secreted into the extracellular space and forms a glycosylated homotetramer that is anchored to the extracellular matrix (ECM) and cell surfaces through an interaction with heparan sulfate proteoglycan and collagen. A fraction of the protein is cleaved near the C-terminus before secretion to generate circulating tetramers that do not interact with the ECM. [provided by RefSeq, Jul 2008]

COX6C [+/-]

Cytochrome c oxidase, the terminal enzyme of the mitochondrial respiratory chain, catalyzes the electron transfer from reduced cytochrome c to oxygen. It is a heteromeric complex consisting of 3 catalytic subunits encoded by mitochondrial genes and multiple structural subunits encoded by nuclear genes. The mitochondrially-encoded subunits function in electron transfer, and the nuclear-encoded subunits may be involved in the regulation and assembly of the complex. This nuclear gene encodes subunit VIc, which has 77% amino acid sequence identity with mouse subunit VIc. This gene is up-regulated in prostate cancer cells. A pseudogene has been found on chromosomes 16p12. [provided by RefSeq, Jul 2010]

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]

APOE [+/-]

The e4 version of the APOE gene increases an individual's risk for developing late-onset Alzheimer disease. People who inherit one copy of the APOE e4 allele have an increased chance of developing the disease; those who inherit two copies of the allele are at even greater risk. The APOE e4 allele may also be associated with an earlier onset of memory loss and other symptoms. It is not known how the APOE e4 allele is related to the risk of Alzheimer disease. However, researchers have found that this allele is associated with an increased number of protein clumps, called amyloid plaques, in the brain tissue of affected people. A buildup of toxic amyloid beta peptide and amyloid plaques may lead to the death of neurons and the progressive signs and symptoms of this disorder. It is important to note that people with the APOE e4 allele inherit an increased risk of developing Alzheimer disease, not the disease itself. Not all people with Alzheimer disease have the APOE e4 allele, and not all people who have this allele will develop the disease.

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.

IL4R [+/-]

The protein encoded by this gene is a pleiotropic cytokine produced by activated T cells. This cytokine is a ligand for interleukin 4 receptor. The interleukin 4 receptor also binds to IL13, which may contribute to many overlapping functions of this cytokine and IL13. STAT6, a signal transducer and activator of transcription, has been shown to play a central role in mediating the immune regulatory signal of this cytokine. This gene, IL3, IL5, IL13, and CSF2 form a cytokine gene cluster on chromosome 5q, with this gene particularly close to IL13. This gene, IL13 and IL5 are found to be regulated coordinately by several long-range regulatory elements in an over 120 kilobase range on the chromosome. Two alternatively spliced transcript variants of this gene encoding distinct isoforms have been reported. [provided by RefSeq, Jul 2008]

SULT1A1 [+/-]

Sulfotransferase enzymes catalyze the sulfate conjugation of many hormones, neurotransmitters, drugs, and xenobiotic compounds. These cytosolic enzymes are different in their tissue distributions and substrate specificities. The gene structure (number and length of exons) is similar among family members. This gene encodes one of two phenol sulfotransferases with thermostable enzyme activity. Multiple alternatively spliced variants that encode two isoforms have been identified for this gene. [provided by RefSeq, Jul 2008]

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]

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

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