

livewello

Gene Variance report

Annabel 6 / F

ALLERGY

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

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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	+/-
4	F11	rs2036914	T	—	NG
5	F11	rs2289252	T	CT	+/-
6	F12	rs1801020	A	—	NG
7	F3	rs1324214	A	—	NG
8	F5	rs6025	T	CC	-/-
10	F9	rs6048	G	AA	-/-
11	GP6	rs1613662	G	AA	-/-
12	HRG	rs9898	T	CT	+/-
13	ITGB3	rs5918	C	TT	-/-
14	KNGI598T	rs2731672	T	CC	-/-
15	NR1I2	rs1523127	C	AA	-/-
16	SERPINC1	rs2227589	T	CC	-/-

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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	AA	-/-
7	CYP1B1 N453S	rs1800440	C	TT	-/-
11	CYP2C9	rs1057910	C	AA	-/-
12	CYP2C9*2 C430T	rs1799853	T	CC	-/-
13	CYP2D6	rs1065852	A	GG	-/-
15	CYP2D6 T2850C	rs16947	A	AG	+/-
17	CYP2E1*1B G9896C	rs2070676	G	CG	+/-
18	CYP2E1*4 A4768G	rs6413419	A	AG	+/-
20	CYP3A4	rs2740574	C	TT	-/-
21	CYP3A4	rs4986910	G	AA	-/-
22	CYP3A4*2 S222P	rs55785340	G	AA	-/-
23	GPX3	rs8177412	C	—	NG
24	GSTM1	rs1056806	T	CC	-/-
25	GSTM1	rs2239892	G	—	NG
26	GSTM1	rs4147565	A	--	NC
27	GSTM1	rs4147567	G	—	NG
28	GSTP1	rs1138272	T	CC	-/-
29	GSTP1	rs1695	G	GG	+/+
30	NAT1	rs4986782	A	GG	-/-
31	NAT2	rs1208	G	AA	-/-
32	NAT2	rs1799930	A	AG	+/-
33	NAT2	rs1799931	A	GG	-/-
34	NAT2	rs1801279	A	GG	-/-
35	NAT2	rs1801280	C	TT	-/-

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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	AG	+/-
3	HLA-DPB2 / COL11A2P	rs1883414	A	AG	+/-
4	HLA-DQA2	rs9275224	A	AG	+/-
5	HORMAD2	rs2412971	A	AG	+/-
6	IFIH1	rs1990760	T	CT	+/-
7	IGF1R	rs2229765	A	AG	+/-
8	IRF5	rs4728142	A	GG	-/-
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	GG	-/-
3	CD14	rs2569191	C	CC	+/+
4	DARC	rs2814778	C	TT	-/-
5	FCER1A	rs2251746	C	TT	-/-
6	FCER1A	rs2427824	T	CC	-/-
7	FCER1A	rs2427827	T	CT	+/-
8	FCER1A	rs2427837	A	—	NG
9	FCER1A	rs2494262	A	—	NG
10	FCER1A / OR10J2P	rs10489854	T	CC	-/-
11	IL13	rs1295685	A	—	NG
12	IL13	rs1800925	T	CC	-/-
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	AC	+/-

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IGG

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	FCGR2A	rs1801274	G	AG	+/-
2	GSTM3	rs7483	T	CT	+/-
3	Intergenic	rs2013111	C	—	NG
4	MUC21	rs1634731	G	AG	+/-
5	TBC1D27	rs3751987	A	—	NG
6	TNFRSF13B	rs4792800	G	—	NG

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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	CT	+/-
5	AHCY-02	rs819134	G	—	NG
6	AHCY-19	rs819171	C	CT	+/-
7	ATG9B (NOS3 G10T)	rs7830	T	—	NG
8	BHMT R239Q	rs3733890	A	AA	+/+
9	BHMT-02	rs567754	T	CC	-/-
10	BHMT-04	rs617219	C	—	NG
11	BHMT-08	rs651852	T	CC	-/-
12	C18orf56 (TYMS)	rs502396	T	—	NG
13	CBS A13637G	rs2851391	T	CT	+/-
14	CBS A360A	rs1801181	A	AG	+/-
15	CBS C19150T	rs4920037	A	GG	-/-
16	CBS C699T	rs234706	A	GG	-/-
17	CBS N212N	rs2298758	A	—	NG
18	CLCN6	rs13306560	T	CC	-/-
19	CLCN6	rs13306561	G	—	NG
21	DAO	rs2070586	A	AG	+/-
22	DAO	rs2111902	G	—	NG
23	DAO	rs3741775	C	AA	-/-
24	DHFR	rs1643649	C	TT	-/-
25	FOLR1	rs2071010	A	GG	-/-
26	FOLR2	rs651933	G	GG	+/+
27	FOLR3	rs7925545	G	AA	-/-
28	FOLR3	rs7926875	A	—	NG
29	FUT2	rs492602	G	AG	+/-
30	FUT2	rs601338	A	AG	+/-
31	FUT2	rs602662	A	AA	+/+
32	G6PD	rs1050828	T	CC	-/-
33	G6PD	rs1050829	C	TT	-/-
34	GAD1	rs10432420	A	AG	+/-

	Gene	rsID	Minor Allele	Genotype	Phenotype
35	GAD1	rs12185692	A	CC	-/-
36	GAD1	rs2058725	C	CT	+/-
37	GAD1	rs2241165	C	CT	+/-
38	GAD1	rs3749034	A	AG	+/-
39	GAD1	rs3791850	A	AG	+/-
40	GAD1	rs3791851	C	CT	+/-
41	GAD1	rs3791878	T	GT	+/-
42	GAD1	rs3828275	T	CC	-/-
43	GAD1	rs701492	T	CT	+/-
44	GAD1	rs769395	G	—	NG
45	GAD1	rs769407	C	CG	+/-
46	GAD2	rs1805398	T	—	NG
47	GAMT	rs17851582	A	GG	-/-
48	GAMT	rs55776826	T	--	NC
49	GIF	rs558660	A	GG	-/-
50	MAOA R297R	rs6323	G	GT	+/-
51	MAOB	rs1799836	C	TT	-/-
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	CT	+/-
57	MTHFD1 G1958A	rs2236225	A	AG	+/-
58	MTHFD1L	rs11754661	A	AG	+/-
59	MTHFD1L	rs17349743	C	CC	+/+
60	MTHFD1L	rs6922269	A	AA	+/+
61	MTHFD1L	rs803422	A	GG	-/-
62	MTHFR	rs12121543	A	—	NG
63	MTHFR	rs1476413	T	CC	-/-
64	MTHFR	rs17037390	A	GG	-/-
65	MTHFR	rs17037396	T	—	NG
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	+/+

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72	MTHFR G1793A (R594Q)	rs2274976	T	CC	-/-
73	MTHFS	rs6495446	T	CT	+/-
74	MTR A2756G	rs1805087	G	AG	+/-
75	MTRR A66G	rs1801394	G	AG	+/-
76	MTRR H595Y	rs10380	T	—	NG
77	MTRR K350A	rs162036	G	AA	-/-
78	MTRR R415T	rs2287780	T	—	NG
79	MTRR-11 A664A	rs1802059	A	GG	-/-
80	NOS1	rs3782206	T	—	NG
81	NOS2	rs2248814	A	GG	-/-
82	NOS2	rs2274894	T	GG	-/-
83	NOS2	rs2297518	A	AG	+/-
84	NOS3	rs1800779	G	GG	+/+
85	NOS3	rs1800783	A	AA	+/+
87	NOS3 T786C	rs2070744	C	—	NG
88	PEMT	rs4244593	T	GT	+/-
90	SHMT1 C1420T	rs1979277	A	—	NG
91	SLC19A1	rs1888530	C	—	NG
92	SLC19A1	rs3788200	A	—	NG
93	SOD2	rs2758331	A	CC	-/-
94	SOD2	rs4880	G	AA	-/-
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	CT	+/-
2	ATP5C1	rs1244422	T	—	NG
3	ATP5C1	rs12770829	T	—	NG
4	ATP5C1	rs2778475	A	—	NG
5	ATP5C1	rs4655	C	—	NG
6	ATP5G3	rs185584	G	—	NG
7	ATP5G3	rs36089250	C	TT	-/-
8	CCL2	rs1024611	G	AA	-/-
9	COX5A	rs8042694	G	AA	-/-
10	COX6C	rs1135382	A	—	NG
11	COX6C	rs12544943	G	—	NG
12	COX6C	rs4510829	A	—	NG
13	COX6C	rs4626565	C	TT	-/-
14	COX6C	rs7828241	C	—	NG
15	COX6C	rs7844439	A	—	NG
16	NDUFS3	rs4147730	A	GG	-/-
17	NDUFS3	rs4147731	A	—	NG
18	NDUFS3 (KBTBD4)	rs2233354	C	—	NG
19	NDUFS7	rs1142530	T	TT	+/+
20	NDUFS7	rs11666067	A	—	NG
21	NDUFS7	rs2074895	A	—	NG
22	NDUFS7	rs2332496	A	AA	+/+
23	NDUFS7	rs7254913	G	—	NG
24	NDUFS7	rs7258846	T	TT	+/+
25	NDUFS7	rs809359	G	AA	-/-
27	NDUFS8	rs1104739	C	—	NG
28	NDUFS8	rs1122731	A	—	NG
29	NDUFS8	rs2075626	C	TT	-/-
30	NDUFS8	rs3115546	G	—	NG
31	NDUFS8	rs4147776	C	—	NG
32	NDUFS8	rs999571	A	GG	-/-
33	SLC19A1	rs1051266	C	CC	+/+
34	UQCRC2	rs11648723	T	GG	-/-

	Gene	rsID	Minor Allele	Genotype	Phenotype
35	UQCRC2	rs4850	A	AG	+/-
36	UQCRC2	rs6497563	C	—	NG

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OTHER IMMUNE FACTORS

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	4q27 Region	rs6822844	T	GG	-/-
2	ADD1 G460W	rs4961	T	GT	+/-
3	APOE	rs429358	C	CT	+/-
4	ATG16L1	rs10210302	T	CC	-/-
5	HLA-DRB1	rs660895	G	AG	+/-
6	IL13	rs20541	A	GG	-/-
7	IL4R	rs1801275	G	AA	-/-
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
2	SULT1A1	rs1801030	C	—	NG
3	SULT1A1	rs4149381	G	TT	-/-
4	SULT1A1	rs6498090	A	—	NG
5	SULT1A1	rs7192559	T	CC	-/-
6	SULT1A1	rs7193599	C	—	NG
7	SULT1A1	rs8057055	A	—	NG
8	SULT1A1	rs9282862	C	TT	-/-
9	SULT2A1	rs11083907	A	GG	-/-
10	SULT2A1	rs11569679	T	CC	-/-
11	SULT2A1	rs2547231	C	AA	-/-
12	SULT2A1	rs2547242	C	—	NG
13	SULT2A1	rs2910393	T	—	NG
15	SULT2A1	rs296366	T	CC	-/-
16	SULT2A1	rs4149448	G	—	NG
17	SULT2A1	rs4149449	T	CC	-/-
18	SULT2A1	rs4149452	T	CT	+/-
19	SULT2A1	rs8113396	G	—	NG

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THYROID

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	CTLA4	rs231775	G	AA	-/-
2	FOXE1	rs10984009	A	AG	+/-
3	FOXE1	rs1867277	A	AG	+/-
4	FOXE1	rs7043516	C	—	NG

A gene variance report is a graphical representation of your genetic raw data, displayed as a color coded chart. Phenotypes are determined based on the presence of known variant alleles in your genotype. Alleles are considered variant if they are the minor allele i.e they occur with less frequency (MAF) in the default global population. In very few instances, the minor allele will not represent a mutation. In such cases, homozygous and/or heterozygous phenotypes may actually be normal. As such, what is normal or abnormal should not be determined solely based on phenotypes displayed in this report. All alleles are reported in reference to the forward strand. rsIDs and genotype information are obtained from the genetic raw data prepared by your personal genomic service. Minor allele frequency (MAF), RefSNP and gene variation/SNP names are obtained directly from dbSNP which is a free public archive for genetic variation maintained by the NCBI <http://www.ncbi.nlm.nih.gov/snp/>.

Report generated by Livewello Gene Variance Software. <http://livewello.com/genetics> v2.0

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 receive your report

Annabel 6 / F

TONGUE TIE

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	IRF6	rs861020	A	GG	-/-
2	IRF6	rs987525	A	AC	+/-
3	RARA	rs7217852	G	AG	+/-
4	RARA	rs9904270	T	—	NG
5	TBX22	rs41307258	A	TT	-/-

A gene variance report is a graphical representation of your genetic raw data, displayed as a color coded chart. Phenotypes are determined based on the presence of known variant alleles in your genotype. Alleles are considered variant if they are the minor allele i.e they occur with less frequency (MAF) in the default global population. In very few instances, the minor allele will not represent a mutation. In such cases, homozygous and/or heterozygous phenotypes may actually be normal. As such, what is normal or abnormal should not be determined solely based on phenotypes displayed in this report. All alleles are reported in reference to the forward strand. rsIDs and genotype information are obtained from the genetic raw data prepared by your personal genomic service. Minor allele frequency (MAF), RefSNP and gene variation/SNP names are obtained directly from dbSNP which is a free public archive for genetic variation maintained by the NCBI <http://www.ncbi.nlm.nih.gov/snp/>.

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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 intrinsic 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]

HRG [+/-]

The protein encoded by this gene was originally identified as a 44-kD glycoprotein that interacts with the NEU/ERBB2 receptor tyrosine kinase to increase its phosphorylation on tyrosine residues. This protein is a signaling protein that mediates cell-cell interactions and plays critical roles in the growth and development of multiple organ systems. It is known that an extraordinary variety of different isoforms are produced from this gene through alternative promoter usage and splicing. These isoforms are tissue-specifically expressed and differ significantly in their structure, and thereby these isoforms are classified into types I, II, III, IV, V and VI. The gene dysregulation has been linked to diseases such as cancer, schizophrenia and bipolar disorder (BPD). [provided by RefSeq, May 2009]

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 can
Make you jittery and shaky
Make it hard to fall asleep or stay asleep
Cause headaches or dizziness
Make your heart beat faster or cause abnormal heart rhythms
Cause dehydration
Make 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

CYP2E1*1B G9896C [+/-]

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 is induced by ethanol, the diabetic state, and starvation. The enzyme metabolizes both endogenous substrates, such as ethanol, acetone, and acetal, as well as exogenous substrates including benzene, carbon tetrachloride, ethylene glycol, and nitrosamines which are premutagens found in cigarette smoke. Due to its many substrates, this enzyme may be involved in such varied processes as gluconeogenesis, hepatic cirrhosis, diabetes, and cancer. [provided by RefSeq, Jul 2008]

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]

HORMAD2 [+/-]

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]

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]

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]

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 interleukin 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 include Allergy and asthma - immune responses to substances that are usually not harmful Immune deficiency diseases - disorders in which the immune system is missing one or more of its parts Autoimmune diseases - diseases causing your immune system to attack your own body's cells and tissues by mistake NIH: National Institute of Allergy and Infectious Diseases

SOCS1 [+/-]

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]

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]

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]

MUC21 [+/-]

MUC-21; KMQK697; C6orf205; bCX31G15.2

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.

AHCY-01 [+/-]

HLA-B belongs to the HLA class I heavy chain paralogues. This class I molecule is a heterodimer consisting of a heavy chain and a light chain (beta-2 microglobulin). The heavy chain is anchored in the membrane. Class I molecules play a central role in the immune system by presenting peptides derived from the endoplasmic reticulum lumen. They are expressed in nearly all cells. The heavy chain is approximately 45 kDa and its gene contains 8 exons. Exon 1 encodes the leader peptide, exon 2 and 3 encode the alpha1 and alpha2 domains, which both bind the peptide, exon 4 encodes

the alpha3 domain, exon 5 encodes the transmembrane region and exons 6 and 7 encode the cytoplasmic tail. Polymorphisms within exon 2 and exon 3 are responsible for the peptide binding specificity of each class one molecule. Typing for these polymorphisms is routinely done for bone marrow and kidney transplantation. Hundreds of HLA-B alleles have been described. [provided by RefSeq, Jul 2008]

BHMT R239Q [+/+]

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 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.

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

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, including Genetic 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 pregnancy. Preterm birth. Often 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

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 you Are obese Have poorly controlled diabetes Take certain antiseizure medicines Getting 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 you are obese, have poorly controlled diabetes, or take certain antiseizure medicines. Getting 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.

MTHFS [+/-]

The protein encoded by this gene is an enzyme that catalyzes the conversion of 5-formyltetrahydrofolate to 5,10-methylenetetrahydrofolate, 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]

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 [+/-]

At least 20 mutations in the MTRR gene have been identified in people with homocystinuria. Some of these mutations change single amino acids in methionine synthase reductase. Other mutations lead to an abnormally small, nonfunctional version of the enzyme. All these mutations prevent the enzyme from functioning normally. Without methionine synthase reductase, methionine synthase cannot convert homocysteine 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.

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 of 119/79 or lower is normal blood pressure 140/90 or higher is high blood pressure Between 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]

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]

ATP5C1 [+/-]

The protein encoded by this gene is a transmembrane glycoprotein that is a member of the protein kinase superfamily. This protein is a receptor for members of the epidermal growth factor family. EGFR is a cell surface protein that binds to epidermal growth factor. Binding of the protein to a ligand induces receptor dimerization and tyrosine autophosphorylation and leads to cell proliferation. Mutations in this gene are associated with lung cancer. Multiple alternatively spliced transcript variants that encode different protein isoforms have been found for this gene. [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 include Leukodystrophies, Phenylketonuria, Tay-Sachs disease, Wilson disease. Many 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]

UQCRC2 [+/-]

Metabolism is the process your body uses to get or make energy from the food you eat. Food is made up of proteins, carbohydrates, and fats. Chemicals in your digestive system break the food parts down into sugars and acids, your body's fuel. Your body can use this fuel right away, or it can store the energy in your body tissues, such as your liver, muscles, and body fat. A metabolic disorder occurs when abnormal chemical reactions in your body disrupt this process. When this happens, you might have too much of some substances or too little of other ones that you need to stay healthy. You can develop a metabolic disorder when some organs, such as your liver or pancreas, become diseased or do not function normally. Diabetes is an example.

ADD1 G460W [+/-]

Adducins are a family of cytoskeleton proteins encoded by three genes (alpha, beta, gamma). Adducin is a heterodimeric protein that consists of related subunits, which are produced from distinct genes but share a similar structure. Alpha- and beta-adducin include a protease-resistant N-terminal region and a protease-sensitive, hydrophilic C-terminal region. Alpha- and gamma-adducins are ubiquitously expressed. In contrast, beta-adducin is expressed at high levels in brain and hematopoietic tissues. Adducin binds with high affinity to Ca(2+)/calmodulin and is a substrate for protein kinases A and C. Alternative splicing results in multiple variants encoding distinct isoforms; however, not all variants have been fully described. [provided by RefSeq, Jul 2008]

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.

HLA-DRB1 [+/-]

Certain variations in the HLA-DRB1 gene have been linked to an increased risk of developing an autoimmune disorder called autoimmune Addison disease. Autoimmune disorders occur when the immune system malfunctions and attacks the body's tissues and organs. In autoimmune Addison disease, the immune system attacks the adrenal glands, which are small hormone-producing glands located on top of each kidney. Loss of hormones produced by the adrenal glands leads to the features of the condition, which include extreme tiredness (fatigue), nausea, low blood pressure (hypotension), and abnormally dark areas of skin (hyperpigmentation), especially in regions that experience a lot of friction such as the armpits, elbows, and knuckles. A particular HLA-DRB1 gene variant called HLA-DRB1*04:04 is the most well-known risk factor for autoimmune Addison disease. Normally, the immune system responds only to proteins made by foreign invaders, not to the body's own proteins. In autoimmune Addison disease, however, an immune response is triggered by a normal adrenal gland protein; in about 85 percent of people with autoimmune Addison disease, the protein is 21-hydroxylase. 21-hydroxylase is found in the adrenal glands where it plays a key role in producing a variety of hormones that regulate many essential functions in the body. The prolonged immune attack triggered by 21-hydroxylase damages the adrenal glands (specifically the outer layers of the glands known, collectively, as the adrenal cortex), preventing hormone production. A shortage of adrenal hormones (adrenal insufficiency) disrupts several normal functions in the body, leading to the diverse features of autoimmune Addison disease. It is not clear how HLA-DRB1*04:04 and other HLA-DRB1 variations are involved in the inappropriate immune response that causes autoimmune Addison disease.

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 include Fatigue, Weight gain, A puffy face, Cold intolerance, Joint and muscle pain, Constipation, Dry skin, Dry, thinning hair, Decreased sweating, Heavy or irregular menstrual periods and fertility problems, Depression, Slowed heart rate. To 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

IRF6 [+/-]

Mutations in the IRF6 gene that cause popliteal pterygium syndrome may change the transcription factor's effects on the activity of certain genes. This affects the development and maturation of tissues in the face, skin, and genitals, resulting in the facial and genital abnormalities, skin webbing, and fusion of the fingers or toes (syndactyly) seen in popliteal pterygium syndrome.

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 α protein, functions differently than the protein products of the normal PML and RARA genes. The PML-RAR α protein binds to DNA and represses gene transcription, like the normal RAR α protein. However, the PML-RAR α 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 α protein does not block proliferation or induce apoptosis. The PML-RAR α 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.

A gene variance report is a graphical representation of your genetic raw data, displayed as a color coded chart. Phenotypes are determined based on the presence of known variant alleles in your genotype. Alleles are considered variant if they are the minor allele i.e they occur with less frequency (MAF) in the default global population. In very few instances, the minor allele will not represent a mutation. In such cases, homozygous and/or heterozygous phenotypes may actually be normal. As such, what is normal or abnormal should not be determined solely based on phenotypes displayed in this report. All alleles are reported in reference to the forward strand. rsIDs and genotype information are obtained from the genetic raw data prepared by your personal genomic service. Minor allele frequency (MAF), RefSNP and gene variation/SNP names are obtained directly from dbSNP which is a free public archive for genetic variation maintained by the NCBI <http://www.ncbi.nlm.nih.gov/snp/>.

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