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Gene Variance report

Monica Johnson 57 / F

ALLERGY

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

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CLEFT LIP/CLEFT PALATE

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	IRF6	rs861020	A	AG	+/-
2	PVT1	rs987525	A	CC	-/-
3	RARA	rs7217852	G	AA	-/-
4	RARA	rs9904270	T	CC	-/-
5	TBX22	rs41307258	A	TT	-/-

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CLOTTING FACTORS

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	CETP	rs1800775	C	CC	+/+
2	CYP4V2	rs13146272	C	AA	-/-
3	F10	rs3211719	G	AG	+/-
4	F11	rs2036914	T	CC	-/-
5	F11	rs2289252	T	TT	+/+
6	F12	rs1801020	A	GG	-/-
7	F12	rs2731672	T	CC	-/-
8	F3	rs1324214	A	AA	+/+
9	F5	rs6025	T	CC	-/-
10	F7	rs6046	A	GG	-/-
11	F9	rs6048	G	AG	+/-
12	GP6	rs1613662	G	AA	-/-
13	HRG	rs9898	T	CC	-/-
14	ITGB3	rs5918	C	TT	-/-
15	NR1I2	rs1523127	C	AC	+/-
16	SERPINC1	rs2227589	T	CC	-/-

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DETOX

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	CTH	rs1021737	T	GG	-/-
2	CYP1A1	rs1048943	C	TT	-/-
3	CYP1A1	rs1799814	T	GG	-/-
4	CYP1A1	rs4986883	C	TT	-/-
5	CYP1A2	rs762551	C	CC	+/+
6	CYP1B1	rs10012	C	GG	-/-
7	CYP1B1	rs1056836	C	CG	+/-
8	CYP1B1	rs1800440	C	CT	+/-
9	CYP2A6	rs1801272	T	AA	-/-
10	CYP2C19	rs12248560	T	CT	+/-
11	CYP2C9	rs1057910	C	AA	-/-
12	CYP2C9	rs1799853	T	CC	-/-
13	CYP2D6	rs1065852	A	AG	+/-
14	CYP2D6	rs16947	A	AG	+/-
15	CYP2E1	rs2070676	G	CC	-/-
16	CYP2E1	rs55897648	A	GG	-/-
17	CYP2E1	rs6413419	A	GG	-/-
18	CYP3A4	rs12721627	C	GG	-/-
19	CYP3A4	rs2740574	C	TT	-/-
20	CYP3A4	rs4986910	G	AA	-/-
21	CYP3A4	rs55785340	G	AA	-/-
22	GPX3	rs8177412	C	TT	-/-
23	GSTM1	rs1056806	T	CC	-/-
24	GSTM1	rs2239892	G	AA	-/-
25	GSTM1	rs4147565	A	GG	-/-
26	GSTM1	rs4147567	G	AA	-/-
27	GSTP1	rs1138272	T	CT	+/-
28	GSTP1	rs1695	G	AG	+/-
29	NAT1	rs4986782	A	GG	-/-
30	NAT2	rs1208	G	AA	-/-
31	NAT2	rs1799930	A	AG	+/-
32	NAT2	rs1799931	A	GG	-/-
33	NAT2	rs1801279	A	GG	-/-

	Gene	rsID	Minor Allele	Genotype	Phenotype
34	NAT2	rs1801280	C	TT	-/-

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GLUTEN INTOLERANCE

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

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IGA

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	CFH	rs6677604	A	AG	+/-
2	HLA-DPA1	rs1883414	A	AG	+/-
3	HLA-DQA2	rs9275224	A	GG	-/-
4	HLA-DRB1	rs9275596	C	TT	-/-
5	HLA-DRB5	rs9271366	G	AA	-/-
6	HORMAD2	rs2412971	G	AA	-/-
7	IFIH1	rs1990760	T	CC	-/-
8	IGF1R	rs2229765	A	AG	+/-
9	IRF5	rs4728142	A	AA	+/+
10	MTC03P1	rs2856717	A	GG	-/-
11	PSMB8	rs9357155	A	GG	-/-
12	TRAF1	rs3761847	G	AG	+/-

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IGE

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	ACKR1	rs2814778	C	TT	-/-
2	C3	rs10402876	C	GG	-/-
3	C3	rs366510	G	TT	-/-
4	CD14	rs2569191	C	CC	+/+
5	FCER1A	rs2251746	C	CT	+/-
6	FCER1A	rs2427824	T	CC	-/-
7	FCER1A	rs2427827	-	CC	
8	FCER1A	rs2427837	A	AG	+/-
9	FCER1A	rs2494262	A	AA	+/+
10	FCER1A / OR10J2P	rs10489854	T	CC	-/-
11	IL13	rs1295685	A	GG	-/-
12	IL13	rs1800925	T	CT	+/-
13	IL5	rs2069812	G	GG	+/+
14	RAD50	rs17772565	T	CC	-/-
15	RAD50	rs17772583	G	AA	-/-
16	RAD50	rs2040704	G	AG	+/-
17	RAD50	rs2240032	T	CT	+/-
18	RAD50	rs6884762	T	--	NC
19	RAG1	rs3740955	A	AG	+/-
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	CC	-/-
3	Intergenetic	rs2013111	C	CT	+/-
4	LOC105369210	rs3751987	A	AG	+/-
5	MUC21	rs1634731	G	AA	-/-
6	TNFRSF13B	rs4792800	G	AA	-/-

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METHYLATION

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	ACAT1	rs3741049	A	AG	+/-
2	ACE	rs4343	G	AG	+/-
3	AGT	rs699	A	AA	+/+
4	AHCY-01	rs819147	C	TT	-/-
5	AHCY-02	rs819134	G	AA	-/-
6	AHCY-19	rs819171	C	TT	-/-
7	BHMT	rs3733890	A	AG	+/-
8	BHMT-02	rs567754	T	CT	+/-
9	BHMT-04	rs617219	C	AC	+/-
10	BHMT-08	rs651852	T	CC	-/-
11	C1orf167	rs4846048	G	AA	-/-
12	CBS	rs2851391	T	CT	+/-
13	CBS	rs4920037	A	GG	-/-
14	CBS A360A	rs1801181	A	GG	-/-
15	CBS C699T	rs234706	A	AG	+/-
16	CBS N212N	rs2298758	A	GG	-/-
17	CLCN6	rs13306560	T	CC	-/-
18	CLCN6	rs13306561	G	AA	-/-
19	CLCN6	rs3737964	T	CC	-/-
20	COMT	rs6269	G	GG	+/+
21	COMT H62H	rs4633	T	CC	-/-
22	COMT P199P	rs769224	A	GG	-/-
23	COMT V158M	rs4680	A	AG	+/-
24	DAO	rs2070586	A	AG	+/-
25	DAO	rs2111902	G	GT	+/-
26	DAO	rs3741775	C	AC	+/-
27	DHFR	rs1643649	C	CT	+/-
28	FOLR1	rs2071010	A	GG	-/-
29	FOLR2	rs651933	G	AA	-/-
30	FOLR3	rs7925545	G	AA	-/-
31	FOLR3	rs7926875	A	CC	-/-
32	FUT2	rs492602	G	GG	+/+
33	FUT2	rs601338	A	AA	+/+

	Gene	rsID	Minor Allele	Genotype	Phenotype
34	FUT2	rs602662	A	AA	+/+
35	G6PD	rs1050828	T	CC	-/-
36	G6PD	rs1050829	C	TT	-/-
37	GAD1	rs10432420	A	GG	-/-
38	GAD1	rs12185692	A	AC	+/-
39	GAD1	rs2058725	C	TT	-/-
40	GAD1	rs2241165	C	TT	-/-
41	GAD1	rs3749034	A	GG	-/-
42	GAD1	rs3791850	A	GG	-/-
43	GAD1	rs3791851	C	CT	+/-
44	GAD1	rs3791878	T	GT	+/-
45	GAD1	rs3828275	T	CT	+/-
46	GAD1	rs701492	T	CT	+/-
47	GAD1	rs769395	G	AG	+/-
48	GAD1	rs769407	C	CG	+/-
49	GAD2	rs1805398	T	GG	-/-
50	GAMT	rs17851582	A	AG	+/-
51	GAMT	rs55776826	T	CT	+/-
52	GIF	rs558660	A	GG	-/-
53	MAOA	rs6323	G	TT	-/-
54	MAOB	rs1799836	C	CT	+/-
55	MTHFD1	rs1076991	T	CT	+/-
56	MTHFD1	rs2236225	A	GG	-/-
57	MTHFD1L	rs11754661	A	--	NC
58	MTHFD1L	rs17349743	C	CT	+/-
59	MTHFD1L	rs6922269	A	GG	-/-
60	MTHFD1L	rs803422	A	AG	+/-
61	MTHFR	rs12121543	-	CC	
62	MTHFR	rs1476413	T	CC	-/-
63	MTHFR	rs17037390	A	GG	-/-
64	MTHFR	rs17037396	T	CC	-/-
65	MTHFR	rs17367504	G	AA	-/-
66	MTHFR	rs2066470	A	GG	-/-
67	MTHFR	rs2274976	T	CC	-/-
68	MTHFR	rs4846049	T	GG	-/-
69	MTHFR A1298C	rs1801131	G	TT	-/-
70	MTHFR C677T	rs1801133	A	AG	+/-

	Gene	rsID	Minor Allele	Genotype	Phenotype
71	MTHFS	rs6495446	T	CT	+/-
72	MTR A2756G	rs1805087	G	AA	-/-
73	MTRR	rs10380	T	CC	-/-
74	MTRR	rs1801394	G	GG	+/+
75	MTRR A664A	rs1802059	A	AG	+/-
76	MTRR K350A	rs162036	G	AA	-/-
77	MTRR R415T	rs2287780	T	CC	-/-
78	NOS1	rs3782206	T	CC	-/-
79	NOS2	rs2248814	A	AG	+/-
80	NOS2	rs2274894	T	GT	+/-
81	NOS2	rs2297518	A	GG	-/-
82	NOS3	rs1800779	G	AG	+/-
83	NOS3	rs1800783	A	AT	+/-
84	NOS3	rs2070744	C	CT	+/-
85	NOS3	rs3918188	A	AC	+/-
86	NOS3	rs7830	T	GG	-/-
87	PEMT	rs4244593	T	TT	+/+
88	PEMT	rs4646406	A	TT	-/-
89	SHMT1	rs1979277	A	AG	+/-
90	SLC19A1	rs1888530	C	CT	+/-
91	SLC19A1	rs3788200	A	GG	-/-
92	SOD2	rs2758331	A	AC	+/-
93	SOD2	rs4880	G	AG	+/-
94	SOD3	rs2855262	C	CT	+/-
95	TCN1	rs526934	G	AA	-/-
96	TCN2	rs1801198	G	CG	+/-
97	TYMSOS	rs502396	T	CT	+/-
98	VDR Bsm	rs1544410	T	TT	+/+
99	VDR Taq	rs731236	G	GG	+/+

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MITOCHONDRIAL FUNCTION

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	ATP5C1	rs1244414	T	CC	-/-
2	ATP5C1	rs1244422	T	TT	+/+
3	ATP5C1	rs12770829	T	CC	-/-
4	ATP5C1	rs2778475	A	AA	+/+
5	ATP5C1	rs4655	C	CT	+/-
6	ATP5G3	rs185584	G	AA	-/-
7	ATP5G3	rs36089250	C	TT	-/-
8	CCL2	rs1024611	G	AA	-/-
9	COX5A	rs8042694	G	AG	+/-
10	COX6C	rs1135382	A	AG	+/-
11	COX6C	rs12544943	G	AG	+/-
12	COX6C	rs4510829	A	AG	+/-
13	COX6C	rs4626565	C	CT	+/-
14	COX6C	rs7828241	C	AC	+/-
15	COX6C	rs7844439	A	AC	+/-
16	NDUFS3	rs2233354	C	TT	-/-
17	NDUFS3	rs4147730	A	GG	-/-
18	NDUFS3	rs4147731	A	GG	-/-
19	NDUFS7	rs1142530	T	CT	+/-
20	NDUFS7	rs11666067	A	AC	+/-
21	NDUFS7	rs2074895	A	AC	+/-
22	NDUFS7	rs2332496	A	GG	-/-
23	NDUFS7	rs7254913	G	AA	-/-
24	NDUFS7	rs7258846	T	GT	+/-
25	NDUFS7	rs809359	G	AA	-/-
26	NDUFS8	rs1051806	T	CC	-/-
27	NDUFS8	rs1104739	C	AC	+/-
28	NDUFS8	rs1122731	A	GG	-/-
29	NDUFS8	rs2075626	C	TT	-/-
30	NDUFS8	rs3115546	G	TT	-/-
31	NDUFS8	rs4147776	C	AA	-/-
32	NDUFS8	rs999571	A	GG	-/-
33	SLC19A1	rs1051266	C	CC	+/+

	Gene	rsID	Minor Allele	Genotype	Phenotype
34	UQCRC2	rs11648723	T	GG	-/-
35	UQCRC2	rs4850	A	GG	-/-
36	UQCRC2	rs6497563	C	TT	-/-

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OTHER IMMUNE FACTORS

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	ADD1	rs4961	T	GT	+/-
2	APOE	rs429358	C	TT	-/-
3	ATG16L1	rs10210302	T	CT	+/-
4	HLA-DRB1	rs660895	G	AA	-/-
5	IL13	rs20541	A	GG	-/-
6	IL4R	rs1801275	G	AA	-/-
7	KIAA1109	rs6822844	T	GG	-/-
8	MEFV	rs11466023	A	GG	-/-
9	MEFV	rs3743930	G	CC	-/-
10	STAT4	rs10181656	G	CC	-/-
11	TNF	rs1800629	A	GG	-/-
12	TNF	rs361525	A	GG	-/-
13	TYR	rs28940879	C	GG	-/-

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SULFOTRANSFERASE

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	SULT1A1	rs1042157	A	--	NC
2	SULT1A1	rs1801030	-	TT	
3	SULT1A1	rs4149381	G	TT	-/-
4	SULT1A1	rs6498090	A	GG	-/-
5	SULT1A1	rs7192559	T	CC	-/-
6	SULT1A1	rs7193599	C	AA	-/-
7	SULT1A1	rs8057055	A	CC	-/-
8	SULT1A1	rs9282862	C	TT	-/-
9	SULT2A1	rs11083907	A	GG	-/-
10	SULT2A1	rs11569679	T	CC	-/-
11	SULT2A1	rs2547231	C	AA	-/-
12	SULT2A1	rs2547242	C	TT	-/-
13	SULT2A1	rs2910393	T	CT	+/-
14	SULT2A1	rs296365	C	CG	+/-
15	SULT2A1	rs296366	T	CT	+/-
16	SULT2A1	rs4149448	G	AG	+/-
17	SULT2A1	rs4149449	T	CT	+/-
18	SULT2A1	rs4149452	T	CC	-/-
19	SULT2A1	rs8113396	G	AA	-/-

Monica Johnson 57 / F

THYROID

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

A gene variance report is a graphical representation of genetic raw data, displayed as a color coded chart. Phenotypes are determined based on the presence or absence of variant alleles in your genotype. Alleles are considered variant if they are the minor allele i.e they occur with less frequency (MAF) in the default global population. Having "no variant" alleles (green) is not necessarily "normal" or protective, and having a homozygous phenotype (red) is not always "abnormal". What is a normal or abnormal phenotype should NOT be determined solely based on this variance report. The significance of your phenotypes should be assessed by reviewing related genome wide studies for context and in consultation with a qualified health practitioner or genetics specialist. All alleles are reported in reference to the foward strand. rsIDs and genotype information are obtained from the genetic raw data prepared by your personal genomic serivce. 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 v3.0

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

Gene Definitions

C11orf30 [+/-]

C11ORF30

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.

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]

F10 [+/-]

At least 130 mutations in the F10 gene have been found to cause a rare bleeding disorder called factor X deficiency. This disorder commonly causes nosebleeds, easy bruising, bleeding under the skin, bleeding of the gums, blood in the urine (hematuria), and prolonged or excessive bleeding following surgery or trauma. Some F10 gene mutations that cause factor X deficiency reduce the amount of coagulation factor X in the bloodstream, resulting in a form of the disorder called type I. Other F10 gene mutations result in the production of a coagulation factor X protein with impaired function, leading to type II factor X deficiency. Reduced quantity or function of coagulation factor X prevents blood from clotting normally, causing episodes of abnormal bleeding that can be severe.

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]

F3 [+/+]

This gene encodes coagulation factor III which is a cell surface glycoprotein. This factor enables cells to initiate the blood coagulation cascades, and it functions as the high-affinity receptor for the coagulation factor VII. The resulting complex provides a catalytic event that is responsible for initiation of the coagulation protease cascades by specific limited proteolysis.

F9 [+/-]

Mutations in the F9 gene cause a type of hemophilia called hemophilia B. More than 900 alterations in this gene have been identified. The most common mutations change single DNA building blocks (base pairs) in the gene. A small percentage of mutations delete or insert multiple base pairs or rearrange segments of DNA within the gene. Mutations in the F9 gene lead to the production of an abnormal version of coagulation factor IX or reduce the amount of this protein. The altered or missing protein cannot participate effectively in the blood clotting process. As a result, blood clots cannot form properly in response to injury. These problems with blood clotting lead to excessive bleeding that can be difficult to control. Mutations that completely eliminate the activity of coagulation factor IX result in severe hemophilia. Mutations that reduce but do not eliminate the protein's activity usually cause mild or moderate hemophilia. Several mutations near the beginning of the F9 gene sequence cause an unusual form of hemophilia known as hemophilia B Leyden. People with these mutations are born with very low levels of functional coagulation factor IX, but hormonal changes cause the levels of this protein to increase gradually during puberty. As a result, adults with hemophilia B Leyden rarely experience episodes of abnormal bleeding.

NR1I2 [+/-]

This gene product belongs to the nuclear receptor superfamily, members of which are transcription factors characterized by a ligand-binding domain and a DNA-binding domain. The encoded protein is a transcriptional regulator of the cytochrome P450 gene CYP3A4, binding to the response element of the CYP3A4 promoter as a heterodimer with the 9-cis retinoic acid receptor RXR.

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]

CYP1B1 [+/-]

This gene encodes an estrogen receptor, a ligand-activated transcription factor composed of several domains important for hormone binding, DNA binding, and activation of transcription. The protein localizes to the nucleus where it may form a homodimer or a heterodimer with estrogen receptor 2. Estrogen and its receptors are essential for sexual development and reproductive function, but also play a role in other tissues such as bone. Estrogen receptors are also involved in pathological processes including breast cancer, endometrial cancer, and osteoporosis. Alternative promoter usage and alternative splicing result in dozens of transcript variants, but the full-length nature of many of these variants has not been determined. [provided by RefSeq, Mar 2014]

CYP2C19 [+/-]

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

CYP2D6 [+/-]

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 [+/+]

SS2; BTN7; BTL-II; HSBLMHC1

CFH [+/-]

At least seven mutations in the CFH gene have been identified in people with dense deposit disease. This condition, which was formerly known as membranoproliferative glomerulonephritis type II, is a form of progressive kidney (renal) disease. Most of the CFH gene mutations that cause dense deposit disease change single protein building blocks (amino acids) in complement factor H. These mutations prevent cells from making this protein or lead to the production of a nonfunctional version of the protein. A shortage (deficiency) of complement factor H can cause uncontrolled 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. CFH gene mutations are responsible for only a small percentage of all cases of dense deposit disease. However, several common variations (polymorphisms) in

the CFH gene have been associated with an increased likelihood of developing the condition. The best-studied of these polymorphisms is written as Tyr402His. Complement factor H usually has the amino acid tyrosine (Tyr) at position 402, but sometimes it has the amino acid histidine (His) instead. People with dense deposit disease are more likely than people in the general population to have histidine at this position. The version of complement factor H with histidine at position 402 is less effective at regulating the complement system on cell surfaces than the version with tyrosine at position 402, which may help explain the increased disease risk.

HLA-DPA1 [+/-]

SS2; BTN7; BTL-II; HSBLMHC1

IGF1R [+/-]

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

IRF5 [+/+]

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

TRAF1 [+/-]

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]

CD14 [+/+]

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]

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 [+/-]

Each spring, summer, and fall, trees, weeds, and grasses release tiny pollen grains into the air. Some of the pollen ends up in your nose and throat. This can trigger a type of allergy called hay fever.Symptoms can includeSneezing, often with a runny or clogged noseCoughing and postnasal dripItching eyes, nose and throatRed and watery eyesDark circles under the eyesYour health care provider may diagnose hay fever based on a physical exam and your symptoms. Sometimes skin or blood tests are used. Taking medicines and using nasal sprays can relieve symptoms. You can also rinse out your nose, but be sure to use distilled or sterilized water with saline. Allergy shots can help make you less sensitive to pollen and provide long-term relief.NIH: National Institute of Allergy and Infectious Diseases

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

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 [+/-]

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

Intergenetic [+/-]

INTERGENETIC

LOC105369210 [+/-]

LOC105369210

ACAT1 [+/-]

More than 40 mutations in the ACAT1 gene have been identified in people with beta-ketothiolase deficiency. Some of these genetic changes disrupt the normal function of the enzyme, while other mutations prevent cells from producing any functional enzyme. A shortage of the ACAT1 enzyme prevents the body from processing proteins and fats properly. As a result, chemical byproducts called organic acids can build up to toxic levels in the blood. These substances cause the blood to become too acidic (ketoacidosis), which can damage the body's tissues and organs, particularly in the nervous system. This damage leads to episodes of vomiting, dehydration, and other health problems associated with beta-ketothiolase deficiency.

ACE [+/-]

This gene encodes an enzyme involved in catalyzing the conversion of angiotensin I into a physiologically active peptide angiotensin II. Angiotensin II is a potent vasopressor and aldosterone-stimulating peptide that controls blood pressure and fluid-electrolyte balance. This enzyme plays a key role in the renin-angiotensin system. Many studies have associated the presence or absence of a 287 bp Alu repeat element in this gene with the levels of circulating

enzyme or cardiovascular pathophysiologies. Multiple alternatively spliced transcript variants encoding different isoforms have been identified, and two most abundant spliced variants encode the somatic form and the testicular form, respectively, that are equally active. [provided by RefSeq, May 2010]

AGT [+/+]

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.

BHMT [+/-]

Methionine is an essential amino acid required for protein synthesis and one-carbon metabolism. Its synthesis is catalyzed by the enzyme methionine synthase. Methionine synthase eventually becomes inactive due to the oxidation of its cob(I)alamin cofactor. The protein encoded by this gene regenerates a functional methionine synthase via reductive methylation. It is a member of the ferredoxin-NADP(+) reductase (FNR) family of electron transferases. Patients of the cbl-E complementation group of disorders of folate/cobalamin metabolism are defective in reductive activation of methionine synthase. Alternative splicing of this gene results in multiple transcript variants encoding distinct isoforms. [provided by RefSeq, Jul 2008]

CBS [+/-]

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

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.

COMT [+/+]

The characteristic signs and symptoms of 22q11.2 deletion syndrome result from a deletion of a small piece of chromosome 22. The chromosomal region that is typically deleted contains 30 to 40 genes, including the COMT gene. As a result of the deletion, people with this disorder have only one copy of the COMT gene in each cell instead of the usual two copies. A loss of one copy of the COMT gene in each cell leads to abnormal regulation of catechol-O-methyltransferase levels in the brain. Researchers believe that changes involving this enzyme in the prefrontal cortex may help explain the increased risk of behavioral problems and mental illness associated with 22q11.2 deletion syndrome. Little is known, however, about the relationship between catechol-O-methyltransferase activity and the specific mental and emotional problems characteristic of this condition. People with 22q11.2 deletion syndrome are much more likely than people without the condition to develop schizophrenia, depression, anxiety, and bipolar disorder.

COMT V158M [+/-]

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DAO [+/-]

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

DHFR [+/-]

Dihydrofolate reductase converts dihydrofolate into tetrahydrofolate, a methyl group shuttle required for the de novo synthesis of purines, thymidylic acid, and certain amino acids. While the functional dihydrofolate reductase gene has been mapped to chromosome 5, multiple intronless processed pseudogenes or dihydrofolate reductase-like genes have been identified on separate chromosomes. Dihydrofolate reductase deficiency has been linked to megaloblastic anemia. Several transcript variants encoding different isoforms have been found for this gene. [provided by RefSeq, Mar 2014]

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.

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]

MTHFD1 [+/-]

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

MTHFD1L [+/-]

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

MTHFR C677T [+/-]

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

MTHFS [+/-]

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

MTRR [+/+]

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.

MTRR A664A [+/-]

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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]

SHMT1 [+/-]

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]

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]

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]

TCN2 [+/-]

More than 20 mutations in the TCN2 gene have been found to cause transcobalamin deficiency. This condition impairs the transport of cobalamin from the bloodstream to cells throughout the body. Affected individuals have difficulty gaining weight and growing at the expected rate (failure to thrive), vomiting, diarrhea, a shortage of all types of blood cells, and neurological problems. Many TCN2 gene mutations lead to a complete or near-complete lack (deficiency) of transcobalamin. Other TCN2 gene mutations result in a transcobalamin protein that cannot bind to cobalamin or a protein that cannot bind to the receptor at the surface of cells. The resulting lack of cobalamin within cells interferes with the functioning of certain enzymes, which impacts many cell activities. As a result, a wide range of signs and symptoms characteristic of transcobalamin deficiency can develop.

TYMSOS [+/-]

TYMSOS

VDR Bsm [+/+]

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

VDR Taq [+/+]

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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]

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.

NDUFS8 [+/-]

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ADD1 [+/-]

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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.

SULT2A1 [+/-]

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

CTLA4 [+/-]

Diabetes is a disease in which your blood glucose, or blood sugar, levels are too high. Glucose comes from the foods you eat. Insulin is a hormone that helps the glucose get into your cells to give them energy. With type 1 diabetes, your body does not make insulin. With type 2 diabetes, the more common type, your body does not make or use insulin well. Without enough insulin, the glucose stays in your blood. You can also have prediabetes. This means that your blood sugar is higher than normal but not high enough to be called diabetes. Having prediabetes puts you at a higher risk of

getting type 2 diabetes.Over time, having too much glucose in your blood can cause serious problems. It can damage your eyes, kidneys, and nerves. Diabetes can also cause heart disease, stroke and even the need to remove a limb. Pregnant women can also get diabetes, called gestational diabetes.A blood test can show if you have diabetes. Exercise, weight control and sticking to your meal plan can help control your diabetes. You should also monitor your glucose level and take medicine if prescribed. NIH: National Institute of Diabetes and Digestive and Kidney Diseases