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Gene Variance report

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YASKO'S NUTRIGENOMIC GENES

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	ACAT1-02	rs3741049	A	GG	-/-
2	ACE	rs4343	G	AG	+/-
3	AHCY-01	rs819147	C	CT	+/-
4	AHCY-19	rs819171	C	CT	+/-
5	BHMT-02	rs567754	T	CC	-/-
6	BHMT-08	rs651852	T	CC	-/-
7	CBS A360A	rs1801181	A	GG	-/-
8	CBS C699T	rs234706	A	AG	+/-
9	COMT H62H (MIR4761)	rs4633	T	TT	+/+
10	COMT P199P (MIR4761)	rs769224	A	GG	-/-
11	COMT V158M (MIR4761)	rs4680	A	AA	+/+
12	MAOA R297R	rs6323	T	G	-
13	MTHFR 03 P39P	rs2066470	A	GG	-/-
14	MTHFR A1298C	rs1801131	G	TT	-/-
15	MTHFR C677T	rs1801133	A	AG	+/-
16	MTR A2756G	rs1805087	G	AG	+/-
17	MTRR	rs1801394	G	AG	+/-
18	MTRR A664A	rs1802059	A	AG	+/-
19	MTRR K350A	rs162036	G	AA	-/-
20	VDR Bsm	rs1544410	T	CC	-/-
21	VDR Fok	rs2228570	A	GG	-/-
22	VDR Taq	rs731236	G	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 v3.5

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

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]

AHCY-01 [+/-]

Three mutations in the AHCY gene have been described in people with hypermethioninemia. In a Croatian family, one mutation substitutes the amino acid cysteine for the amino acid tyrosine at protein position 143 (written as Tyr143Cys or Y143C). Another mutation replaces the amino acid tryptophan with a premature stop signal at position 112 (written as Trp112X or W112X), resulting in an enzyme that is abnormally short. A U.S. patient was found to have, in addition to the Y143C mutation, a mutation that substitutes the amino acid valine for the amino acid alanine at position 89 (written as Ala89Val or A89V). These mutations reduce the activity of the S-adenosylhomocysteine hydrolase enzyme, resulting in the signs and symptoms of hypermethioninemia.

AHCY-19 [+/-]

Three mutations in the AHCY gene have been described in people with hypermethioninemia. In a Croatian family, one mutation substitutes the amino acid cysteine for the amino acid tyrosine at protein position 143 (written as Tyr143Cys or Y143C). Another mutation replaces the amino acid tryptophan with a premature stop signal at position 112 (written as Trp112X or W112X), resulting in an enzyme that is abnormally short. A U.S. patient was found to have, in addition to the Y143C mutation, a mutation that substitutes the amino acid valine for the amino acid alanine at position 89 (written as Ala89Val or A89V). These mutations reduce the activity of the S-adenosylhomocysteine hydrolase enzyme, resulting in the signs and symptoms of hypermethioninemia.

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 H62H (MIR4761) [+/+]

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 (MIR4761) [+/+]

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.

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.

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 [+/-]

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 [+/-]

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.

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