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Gene Variance report

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CORONARY HEART DISEASE RISK MULTIPLE GWAS STUDIES

	Gene	rsID	Minor Allele	Genotype	Phenotype
1	ALDH2*2	rs671	A	GG	-/-
2	ATP2B1	rs7136259	T	CT	+/-
3	BTD	rs7651039	T	TT	+/+
4	C6orf10	rs9268402	A	AA	+/+
5	CDH13	rs8055236	T	GG	-/-
6	CDKN2B-AS1	rs10757274	G	AG	+/-
7	CDKN2B-AS1	rs4977574	G	AG	+/-
8	CDKN2B-AS1	rs7865618	G	AG	+/-
9	CELSR2	rs646776	C	TT	-/-
10	CNNM2	rs12413409	A	GG	-/-
11	COL4A1	rs4773144	G	AA	-/-
12	FMN2	rs17672135	C	TT	-/-
13	HHIPL1	rs2895811	C	CT	+/-
14	HNF1A	rs2259816	T	GT	+/-
15	KIAA1462	rs2505083	C	CT	+/-
16	LPA	rs3798220	C	TT	-/-
17	MIA3	rs17465637	A	AC	+/-
18	MRAS	rs2306374	C	TT	-/-
19	MRAS	rs9818870	T	CC	-/-
20	MTHFD1L	rs6922269	A	GG	-/-
21	MYL2	rs3782889	G	AA	-/-
22	PSRC1	rs599839	G	AA	-/-
23	RP3-323P13.2	rs12524865	A	CC	-/-
24	SEZ6L	rs688034	T	TT	+/+
25	SMAD3	rs17228212	C	CT	+/-
26	SMARCA4	rs1122608	T	GT	+/-
27	SNP?	rs11206510	C	CT	+/-
28	SNP?	rs11752643	T	CC	-/-

	Gene	rsID	Minor Allele	Genotype	Phenotype
29	SNP?	rs1333049	C	CG	+/-
30	SNP?	rs1746048	T	CT	+/-
31	SNP?	rs1842896	G	GT	+/-
32	SNP?	rs3853444	C	TT	-/-
33	SNP?	rs3869109	A	GG	-/-
34	SNP?	rs4380028	T	CT	+/-
35	SNP?	rs501120	C	CT	+/-
36	SNP?	rs579459	C	TT	-/-
37	SNP?	rs6905288	G	AA	-/-
38	SNP?	rs9982601	T	CT	+/-
39	WDR12	rs6725887	C	TT	-/-
40	ZC3HC1	rs11556924	T	TT	+/+

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

BTD [+/+]

More than 150 mutations in the BTD gene have been identified in people with biotinidase deficiency. This disorder, if untreated, can affect many parts of the body and cause delayed development. Most of the mutations that cause biotinidase deficiency change single amino acids in the biotinidase enzyme. These changes occur in critical regions of the enzyme and reduce or eliminate the enzyme's activity. Most BTD gene mutations cause profound biotinidase deficiency. This severe form of the disorder results when the activity of biotinidase is reduced to less than 10 percent of normal. Other mutations cause a milder form of the condition called partial biotinidase deficiency. These mutations reduce biotinidase activity to between 10 percent and 30 percent of normal. Without enough of this enzyme, biotin cannot be recycled. The resulting shortage of free biotin impairs the activity of biotin-dependent carboxylases, leading to a buildup of potentially toxic compounds in the body. If the condition is not treated promptly, this buildup damages various cells and tissues, causing the signs and symptoms associated with biotinidase deficiency.

C6orf10 [+/+]

C6ORF10

CDKN2B-AS1 [+/-]

CDKN2B

HHIPL1 [+/-]

HHIPL1

HNF1A [+/-]

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

MIA3 [+/-]

ARNT; D320; TANGO; TANGO1; UNQ6077

SMAD3 [+/-]

At least 11 mutations in the SMAD3 gene have been found to cause Loeys-Dietz syndrome type III. This disorder affects connective tissue, which gives structure and support to blood vessels, the skeleton, and many other parts of the body. Loeys-Dietz syndrome type III is characterized by abnormal blood vessels and skeletal and joint deformities. Some of the mutations that cause this disorder insert or delete small amounts of genetic material in the SMAD3 gene, while other mutations result in a change to single protein building blocks (amino acids) in the SMAD3 protein. These mutations lead to the production of a nonfunctional SMAD3 protein. Despite a reduction in SMAD3 function, the TGF-\u03b2 pathway is overactive. Researchers speculate that the activity of proteins in this signaling pathway is increased to compensate for the lack of SMAD3 activity; however the exact mechanism responsible for the increase in signaling is unclear. The overactive signaling pathway leads to dysregulated cell proliferation and gene activation, specifically affecting blood vessel and cartilage development. These changes lead to the abnormalities typical of Loeys-Dietz syndrome type III.

SMARCA4 [+/-]

At least six mutations in the SMARCA4 gene can cause Coffin-Siris syndrome. This condition is characterized by delayed development, abnormalities of the fifth (pinky) fingers or toes, and characteristic facial features that are described as coarse. The SMARCA4 gene mutations involved in Coffin-Siris syndrome change single protein building blocks (amino acids) in or remove an amino acid from the BRG1 protein. Although it is unclear how these changes affect SWI/SNF complexes, researchers suggest that SMARCA4 gene mutations result in abnormal chromatin remodeling. Disturbance of this process alters the activity of many genes and disrupts several cellular processes, which could explain the diverse signs and symptoms of Coffin-Siris syndrome. People with Coffin-Siris syndrome do not appear to have an increased risk of cancer (see below).

SNP? [+/-]

SNP?