Lifestyle Genomics Research Centre

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INTRODUCTION (click to read...)	Click on rsids to interpret the meaning of your genotype. Click on gene names to learn more about each gene. Click on category names, to learn about the context, where available.	Your genotype carries stronger risk for the Category (Condition/Pathway) it is listed under	Your genotype carries some risk for the Category (Condition/Pathway) it is listed under

CATEGORY	Gene Name	RSID	Risk Genotype	Your Genotype	SNP INTERPRETATION	EXPRESSION CONTROL	ASSOCIATED SYMPTOMS	ADDITIONAL LAB MARKERS
METHYLATION & TRANS-SULFURATION (CLICK HERE TO EXPLORE)								
Betaine Metabolism	DMGDH	rs121908331	CC	CC	CC= Dimethylglycine dehydrogenase deficiency. This enzyme involved in the catabolism of choline by mediatign formation of sarcosine from dimethylglycine. Sacrosine is then used to manufacture Glycine.	Vitamin D, Testosterone, Estradiol, Selenium, Ursodeoxycholic Acid, Bile acids. Vitamin B2 is a co-factor for this enzyme	fish odor, and unusual muscle fatigue	increased serum creatine kinase
Cobalamin Cycle	MTRR	rs1532268	TT	CC				
Cobalamin Cycle	MTRR A66G	rs1801394	GG	AG	Disorders of Intracellular Cobalamin Metabolism. This may slow down the production of Methylated cobalamin and thus the methylation cycle may be slower too	Epression: Methionine, Choline, Methylfolate, Adenosyl-cobalamine, Methyl-cobalamine, Cofactors: SAMe, B2, B3	Poor feeding and slow growth, microcephaly, encephalopathy, hypotonia, developmental delay, seizures, infantile spasms, infantile maculopathy, degeneration of the spinal cord. Neural tube defects, folate-sensitive, susceptibility to Down syndrome.	High methylmalonic acid (Urine Organic Acid test), Serum methylmalonic acid, Total plasma homocysteine, Plasma amino acid (PAA) analysis, Serum vitamin B12 levels, Plasma acylcarnitine analysis.
Cobalamin Cycle	MTRR H595Y	rs10380	TT	CC				
Cobalamin Cycle	MTRR K350A	rs162036	GG	AA				
Cobalamin Cycle	MTRR R415T	rs2287780	TT	CC				
Cobalamin Cycle	MTRR S257T	rs2303080	TT	TT	Probably non-pathogenic variant but added as an extra marker and it is suggested to b elinked to Disorders of Intracellular Cobalamin Metabolism. This may slow down the production of Methylated cobalamin and thus the methylation cycle may be slower too	Expression: Methionine, Choline, Methylfolate, Adenosyl-cobalamine, Methyl-cobalamine, Cofactors: SAMe, B2, B3	Poor feeding and slow growth, microcephaly, encephalopathy, hypotonia, developmental delay, seizures, infantile spasms, infantile maculopathy, degeneration of the spinal cord. Neural tube defects, folate-sensitive, susceptibility to Down syndrome.	High methylmalonic acid (Urine Organic Acid test), Serum methylmalonic acid, Total plasma homocysteine, Plasma amino acid (PAA) analysis, Serum vitamin B12 levels, Plasma acylcarnitine analysis.
Cobalamin Cycle	MTRR A664A	rs1802059	AA	GG				
Folate Cycle	FOLR1	rs144637717	CC	TT				
Folate Cycle	FOLR1	rs121918405	TT	CC				
Folate Cycle (add folate deficiency symptoms)	DHFR	rs70991108	DD	II				
Folate Cycle	DHFR	rs387906619	AA	GG				
Folate Cycle	MTHFD1	rs2236225	AA	AA	AG = Possible increased risk of birth defects, AA = Slightly (~1.5x) higher risk for Caucasian mothers to give birth to Neural Tube Defect children. Please click on the rsid number to learn more	Coumestrol, Resveratrol, Methylfolate, Vitamin K 3, Choline, Redox potential needs to be balanced as NAD+ acts as a co-factor/substrate.	Symptoms associated with Neural Tube Defects	
Folate Cycle	MTHFR A1298C	rs1801131	GG	TT	Homozygous for the C677T variant and no mutations in the A1298C variant. Your homozygous C677T variant reduces the activity of the MTHFR enzyme by 70 percent	Please click on the relevant rsid number link to learn more about controlling expression of this gene.	Please click on the relevant rsid number link to learn more about possible symptoms.	Please click on the relevant rsid number link to learn more about controlling expression of this gene.
Folate Cycle	MTHFR C677T	rs1801133	AA	AA				
Folate Cycle	MTHFR P39P	rs2066470.1	AA	GG				
Folate Cycle	MTHFS	rs6495446	CC	CC	The C allele, despite being the major one has been associated with increased risk for chronic kidney disease. Each T allele lowers this risk	Increased activity may cause a higher folate turnover rate and folate depletion.	Weight loss, poor appetite, swelling of ankles, feet, and hands, shortness of breath, tiredness, blood in urine, need to urinate more, insomnia, itchy skin, muscle cramps, nausea, headaches. There may be more important contributors to a kidney disease	If symptoms are present, check levels of creatinine in blood and/or Albumin to Creatinine Ratio. A glomerular filtration rate (GFR) is a blood test that checks how well your kidneys are working

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Folate Cycle	SHMT-1 C1420T	rs1979277	AA	GG	**SHMT shifts methylation cycle efforts towards new DNA synthesis. The A allele has been associated with Gastrointestinal stromal tumor . This SNP, on its' own is highly unlikely to cause this disease. Click on rsid link to learn more**			
Folate Cycle	SLC19A1	rs1051266	CC	CC	**The c allele is associated with higher plasma folate and at the same time, women with CT and TT had higher RBC folate levels not associated with serum folate or homocysteine levels. This suggests that the SNP affects the transport of folate into cells**	Methionine, Flavonoids, Estradiol, Choline, Genistein	Symptoms associated with RBC (Red Blood Cell) Folate deficiency. Click on the category title to read more.	**Red blood cell folate is an indicator of long-term status. It's worth noting that RBC folate measures all types of folates under one name. It's advisable to choose a test which breaks dwon these different types.**
Homocysteine Metabolism	AHCY-01	rs819147	CC	TC	**AHCY regulates Homocysteine by catalizing it to Adenosine - this may regulate Methylation cycle. A SNP here may contribute to too much pressure on the CBS enzyme and the Trans-sulfuration pathway. This variant is not connected to any clinical issues**	Addressing should be considered in the context of the entire Methylation Cycle because upregulation of this gene may create too much SAH and thus inhibit SAM - the main methyl donor. AHCY is slowed down by Sarin, Thimerosal, Formaldehyde and Valporic Acid	This SNP on its' own is unlikely to cause any symptoms	This SNP on its' own is unlikely to be the reason for further lab testing. Monitor Homocysteine if the Methylation Cycle flow is a suspected problem.
Homocysteine Metabolism	AHCY-19	rs819171	CC	TC	**AHCY regulates Homocysteine by catalizing it to Adenosine - this helps regulating Methylation cycle. A SNP here may contribute to too much pressure on the CBS enzyme and the Trans-sulfuration pathway. This variant is not connected to any clinical issues**	Addressing should be considered in the context of the entire Methylation Cycle because upregulation of this gene may create too much SAH and thus inhibit SAM - the main methyl donor. AHCY is slowed down by Sarin, Thimerosal, Formaldehyde and Valporic Acid	This SNP on its' own is unlikely to cause any symptoms	This SNP on its' own is unlikely to be the reason for further lab testing. Monitor Homocysteine if the Methylation Cycle flow is a suspected problem.
Homocysteine Metabolism	BHMT	rs3733890	AA	AG	**A allele may increase risk of Neural Tube Defects in folate rich environments and possibly in conjunction with MTHFR rs1801133 SNP. It also may mean DECREASED mortality among breast cancer patients (a protective role)**	Betaine Hydrochloride, Palm Oil, Zinc, Dimethyl Glycine. Cortisol/ Stress can inhibit the action of this enzyme. TMG can supply the end product of this gene but consider the entire Methylation Cycle. Zinc is a co-factor	The symptoms associated with Neural Tube Defects vary depending on the specific type of defect. Symptoms include physical problems (such as paralysis and urinary and bowel control problems), blindness, deafness, intellectual disability,	Homocysteine, SAM to SAH ratio. BHMT converts Homocysteine to Methione which is SAM precursor. Neural tube defects may be diagnosed during the ultrasound scan that around week 12 of the pregnancy or during the anomaly scan that around weeks 18 to 20.
Homocysteine Metabolism	BHMT-02	rs567754	TT	TC	**This SNP may slow down the conversion of Homocysteine to Methionine. Dr. Yasko believes that BHMT-02 plays a role in the gut environmet. The T allele is associated with decrease in Toenail and blood Selenium levels.**	Betaine Hydrochloride, Palm Oil, Zinc, Dimethyl Glycine. Cortisol/ Stress can inhibit the action of this enzyme. TMG can supply the end product of this gene but consider the entire Methylation Cycle	Dry mouth, insulin resistance, skin irritation, Homocysteinuria symptoms. Symptoms associated with Selenium Deficiency include; infertility in men and women, muscle weakness, fatigue, mental fog, hair loss, weakened immune system.	Homocysteine, SAM to SAH ratio. BHMT converts Homocysteine to Methione which is SAM precursor. Blood Selenium levels.

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Homocysteine Metabolism	BHMT-04	rs617219	CC	AC	**This SNP may slow down the conversion of Homocysteine to Methionine. Dr. Yasko believes that BHMT-04 plays a role in the gut environmet. The C allele is associated with Plasma Betaine levels and Angiographic Measurement.**	Betaine Hydrochloride, Palm Oil, Zinc, Dimethyl Glycine. Cortisol/ Stress can inhibit the action of this enzyme. TMG can supply the end product of this gene but consider the entire Methylation Cycle. Zinc is a co-factor	Possibly symptoms related to lower stomach acid, dry mouth, insulin resistance, skin irritation, Homocysteinuria symptoms	Homocysteine, SAM to SAH ratio. BHMT converts Homocysteine to Methione which is SAM precursor.
Homocysteine Metabolism	BHMT-08	rs651852	TT	TC	**This SNP may slow down the conversion of Homocysteine to Methionine and therefore the production of SAMe - the main methyl donor.**	Betaine Hydrochloride, Palm Oil, Zinc, Dimethyl Glycine. Cortisol/ Stress can inhibit the action of this enzyme. TMG can supply the end product of this gene but consider the entire Methylation Cycle. Zinc is a co-factor	Dr. Yasko belives that this SNP, in the presence of higher Cortisol levels, can negatively affect attention span and levels	Homocysteine, SAM to SAH ratio. BHMT converts Homocysteine to Methione which is SAM precursor.
Homocysteine Metabolism	CBS I278T	rs5742905	GG	AA				
Homocysteine Metabolism	CBS 307S	rs121964972	AA	GG				
	CBS	rs28934891	TT	CC				
	CBS	rs2851391	TT	TT	**Common genetic loci influencing plasma homocysteine concentrations and their effect on risk of coronary artery disease.**	✔Vitamin B6. Taurine and Cysteine could help normalize protein levels in patients with homocystinuria, Valproic Acid, Indomethacin. Quercitin, Vitamin C, Dihydrotestosterone (DHT) can all slow down CBS	Could include symptoms associated with vitamin b12 or folate deficiency. High homocysteine can lead to atherosclerosis, thrombosis, blood clots, heart attack, coronary artery disease, stroke dementia, Alzheimer's disease	Blood Homocysteine levels, SAM to SAH ratio, RBC Folate, Serum B12, Methylmalonic acid in urine
	CBS	rs121964970	TT	CC	**TT was associated with pyridoxine-responsive Homocystinuria due to CBS deficiency, Thoracic aortic aneurysm and aortic dissection**			
	CBS	rs234709	TT	TC	**Common genetic loci influencing plasma homocysteine concentrations and their effect on risk of coronary artery disease.**	✔Vitamin B6. Taurine and Cysteine could help normalize protein levels in patients with homocystinuria, Valproic Acid, Indomethacin. Quercitin, Vitamin C, Dihydrotestosterone (DHT) can all slow down CBS	Could include symptoms associated with vitamin b12 or folate deficiency. High homocysteine can lead to atherosclerosis, thrombosis, blood clots, heart attack, coronary artery disease, stroke dementia, Alzheimer's disease	Blood Homocysteine levels, SAM to SAH ratio, RBC Folate, Serum B12, Methylmalonic acid in urine
	CBS	rs121964962	TT	CC				
Homocysteine Metabolism	CBS C699T	rs234706	AA	AG	**There are some mixed messages regarding this variant. Please make sure to click on the rsid number link to find out more.**	There are some mixed messages regarding this variant. Please make sure to click on the rsid number link to find out more.	There are some mixed messages regarding this variant. Please make sure to click on the rsid number link to find out more.	There are some mixed messages regarding this variant. Please make sure to click on the rsid number link to find out more.
Homocysteine Metabolism	MTR A2756G	rs1805087	GG	AA				
Homocysteine Metabolism	MTR	rs2275565	TT	GG				
Methionine Metabolism	MAT1A	rs72558181	TT	CC				
Methionine Metabolism	MAT1A	rs118204002	TT	GG				
Methionine Metabolism	MAT1A	rs118204003	AA	GG				
Methionine Metabolism	MAT1A	rs118204006	TT	CC				
Methionine Metabolism	MAT1A	rs72558181	TT	CC				
Methionine Metabolism	MAT1A	rs118204001	GG	AA				
Methionine Metabolism	MAT1A	rs72558181	TT	CC				
Methylation / Choline Pathway	PEMT +5465G-A	rs7946	CC	TT				

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Methylation / Choline Pathway	PEMT +5465G-A	rs7946	TT	TT	**Each T = lower PEMT levels. Higher risk of nonalcoholic fatty liver disease in caucasians, possibly lower levels of HDL cholesterol, higer risk of Alzheimer's disease in Chinese women. Lower risk of having baby with Neural Tube defects**	This is pure speculation but perhaps the lower PEMT levels utilise less methyl groups and this lowers risk of Neural Tube Defects in babies? To increase PEMT: Vitamin E, Estradiol, Progesterone, Zinc, Choline. Phosphatidylcholine can bypass PEMT	With NFLD You may feel tired or have discomfort in the upper right side of your abdomen, an enlarged liver, Signs of cirrhosis, such as jaundice, a condition that causes your skin and whites of your eyes to turn yellow	Abnormal results on liver tests [ALT, AST] that you had for other reasons, imaging tests, sometimes a biopsy
Methylation / Creatine Formation	GAMT	rs55776826	TT	CC				
Trans-sulfuration / Homocysteine Metabolism	CTH	rs1021737	TT	TG	**TT = significantly higher plasma total homocysteine concentration**	Methylfolate, Sulforaphane, Zinc, Quercetin	Could include symptoms associated with vitamin b12 or folate deficiency. High homocysteine can lead to atherosclerosis, thrombosis, blood clots, heart attack, coronary artery disease, stroke dementia, Alzheimer's disease	
Trans-sulfuration	SUOX	rs705702	GG	AG	**The SUOX gene codes the mitochondrial enzyme, sulfite oxidase. Sulfite oxidase is a metallo-enzyme that uses a molybdopterin cofactor and a heme group to oxidize sulfite to sulfate. The G allele is associated with Polycystic Ovary Syndrome**	Molybdenum and vitamin B1 are cofactors. Acetaminophen and Tretinoin [Vitamin A derivative], increase expression. EGCG from green tea, Quercitin, Copper Sulfate, Cyclosporine - can all slow down the gene expression	Sensitivity to Sulfite in foods. Symptoms associated with Polycystic Ovary Syndrome: irregular periods or no periods at all, difficulty getting pregnant, excessive hair growth, weight gain, oily skin or acne, thinning hair and hair loss from the head	The diagnostic workup for PCOS should begin with a thorough history and physical examination
Trans-sulfuration	SUOX R160Q	rs121908007	AA	GG				
Trans-sulfuration	SUOX	rs121908008	AA	CC				
Trans-sulfuration	SUOX GLY473ASP	rs121908009	AA	GG				
Trans-sulfuration	SUOX R76S	rs202085145	TT	GG				
Trans-sulfuration	SUOX A628C	rs7297662	AA	AA				