The Great Plains Laboratory, Inc.

DNA Methylation Pathway Profile

What are SNPs?

SNPs (single nucleotide polymorphisms) are the simplest of genetic mutations. They are errors that cause a single base substitution for the original, like cytosine for thymine. Most SNPs occur in stretches of DNA that are non-coding regions. When the substitution does fall in a coding region, that base may then call for a different amino acid to be placed in the sequence. Most of these do not cause a problem. However, a relative few will cause the resulting protein to be improperly folded, therefore preventing function, like catalyzing a synthesis fully or partially.

The Importance of Identifying SNPs

With the advent of individualized genetic studies and analysis of the "metabolome," an era of personalized preventive medicine is emerging. When SNPs are known, a metabolic "workaround" can often be determined, or increased amounts of the enzyme's substrate or cofactors (vitamins and minerals) can improve the efficiency of the enzyme itself. In the example of MTHFR, an already methylated form of folic acid, methylfolate, can be given to the patient to increase the efficiency of the affected metabolic pathways. Lifestyle changes in nutritional intake and avoidance of toxic exposure also have the promise of avoiding or slowing the development of chronic disease. The goal is to use knowledge of these genetic weaknesses caused by SNPs to prevent the development of disease conditions.

Overview of the Profile

The DNA Methylation Pathway Profile allows clinicians to screen their patients for a variety of genetic changes that may impact the function of important biochemical processes such as methionine metabolism, detoxification, hormone balance and Vitamin D function. The presence or absence of SNPs may modify disease risk. The risks may be reduced by lifestyle changes, and inefficient biochemical processes can be supported by diet and nutritional supplements to maximize the functions of metabolic pathways. The profile includes a variety of SNPs known to influence many aspects of health including those for:

- Insulin sensitivity
- Bone health
- Cancer risks
- Cardiovascular health
- Detoxification processes
- Fertility
- Mitochondrial function and metabolism
- Methylation
- Neurotransmitter balance

SNPs in the DNA Methylation Profile

The DNA Methylation Profile includes a variety of SNPs known to influence many aspects of health including insulin sensitivity, bone health, cancer risk, cardiovascular health, detoxification capacity, fertility, mitochondrial function and metabolism, methylation capacity, and neurotransmitter balance.

Gene	# of SNPs	Description
VDR: (the vitamin D receptor)	2	A nuclear receptor protein that binds 1,25-dihydroxy vitamin D to activate a signaling molecule that is believed to have important roles in a 3rd of the human genome. Some functions that are known are xenobiotic detoxification.
BHMT: (betaine-homocysteine methylatransferase)	4	A transferase enzyme that catalyzes the transfer of a methyl group from betaine to homocysteine, which produces methionine. Another enzymatic role for BHMT is the choline oxidation processes. This enzyme is found in the liver and kidney.
COMT: (catechol-O-methyltransferase)	3	Functions in the nerve cells, liver, kidneys, and red blood cells. Its role is to help inactivate 2- and 4-hydroxyestradiols, and catecholamine hormones prior to excretion of bile. COMT is also found in the CNS where its role is the degradation of catecholamine neurotransmitters.
MAO A: (monoamine oxidase type A)	1	Main role is to detoxify biological and xenobiotic amines. This enzyme also degrades neurotransmitters in both the central and peripheral nervous system.
AHCY: (adenosylhomocysteinase)	3	An enzyme that breaks down methionine by converting S-adenosylhomocysteine into homocysteine. This reaction regulates the methylation of other compounds.
CBS: (cystathionine beta-synthase)	3	This enzyme is responsible for using vitamin B6 to convert serine and homocysteine into cystothionine which will be later converted into cysteine.
MTHFR: (methylenetetrahydrofolate reductase)	3	Enzyme responsible for the conversion of 5,10-methylenetetrahydrofolate to 5-methyltetrahydrofolate. This reaction allows for the conversion of homocysteine to methionine.
MTR: (methionine synthase)	1	Enzyme that catalyzes the re-methylation of homocysteine to methionine using the methyl-B-12 as a cofactor.
MTRR: (methionine synthase reductase)	6	Responsible for the regeneration of methyl-B-12.
SHMT: (serine hydroxymethyltransferase)	1	Responsible for catalyzing the interconversion of glycine to serine.
SUOX: (sulfite oxidase)	1	Mitochondrial enzyme responsible for oxidizing sulfites to sulfates. Sulfites are produced by the transsulfuration cycle or from diet.

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