Methylation Overview and Explanation

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The term methylation describes a biochemical process that is utilized in the body for transport of nutrients, energy production and in gene modulation. In patients with decreased methylation activity (i.e. methylation deficiency), there are significant shortcomings in the ability to execute a variety of important chemical functions in the body. These shortcomings can leave the body unprotected from the everyday assault of environmental and infectious agents, sluggish in neurotransmitter production and slow to recover from inflammatory damage. This shortfall can result in a wide range of medical conditions including neurological issues, such as seizures, migraines, dizziness, depression, anxiety, OCD, ADD/ADHD, developmental delay, autism, dementias and memory issues, chronic persistent infections, auto-immune disease, neural tube defects, infertility and pregnancy complications. Appropriate supplementation with the proper methylated vitamins and nutrients will bypass the genetic and acquired deficits and restore appropriate function of the pathway resulting in improvement in complicated syndromes.

The proper functioning of the methylation cycle is essential for a number of critical reactions in the body. Consequences of genetic weaknesses (mutations) in this pathway are increased risk factors leading to a number of serious health conditions. A central pathway in the body that is particularly amendable to bio-molecular genetic weaknesses is the methionine/folate pathway. There are several sites in this pathway where blocks can occur as a result of genetic differences. In general, single biomarkers are identified as indicators for specific disease states. However, it is possible that for a number of health conditions, including autism and other severe neurological syndromes, it may be necessary to look at the entire methylation pathway as “a biomarker” for underlying genetic susceptibility for a disease state. It may require expanding the view of a “biomarker” beyond the restriction of a mutation in a single gene to a mutation somewhere in an entire pathway of interconnected function.

This does not mean that every individual with mutations in this pathway will have one of the health conditions listed above. It may be a necessary element, but there may not be a sufficient environmental or infectious “trigger”. Most health conditions in society today are multifactorial in nature. In essence, there is an underlying genetically determined risk that requires a significant infectious or environmental “trigger” to initiate the process.

What is MTHFR?

The MTHFR Gene is located on chromosome number one and produces an enzyme that helps convert folic acid (VitB9) in the foods we eat to a usable amino acid used to build DNA and proteins in our body. This enzyme is also important in the process of converting homocysteine into methionine an amino acid in the body needed for growth and metabolism. There are over 50 different mutations of the MTHFR Gene but only two are particularly problematic: mutations on the points C677T and A1298A.

This was one of the first and most publicized genomic single nucleotide polymorphisms (SNPS) identified. MTHFR stands for methylene tetrahydrofolate reductase and is a key enzymatic step in the conversion of dietary folate to activated folate. This key biochemical reaction takes place in every cell of your body and is required for optimal health. But it is NOT the only important enzyme in the methylation cycle that can result in ill health. MTHFR enzyme sits at a critical place within the important methylation cycles. Inefficiency of this and many other enzymes in this pathway can cause these cycles to not work optimally. These genetic alterations can now be identified by DNA analysis and help explain the cause of symptoms.

A genomic SNP like MTHFR is similar to a crack in the foundation of a bridge. What if we could reinforce the cracks and decrease the stress on the body to improve health? By identifying areas of genetic fragility, it is possible to target appropriate nutritional supplementation of these pathways to bypass these blocks and help restore function.

Many patients have their MTHFR enzyme tested only to find that it is completely normal but they still have significant symptoms. This is why more complete testing should be done of the methylation cycle as many genetic SNPS other than MTHFR can result in undermethylation and contribute to disease.

Methylation is a key biochemical process that occurs in every cell of our body over 250 billion times a second. Methylation is simply the addition of a small methyl group to a substrate like an enzyme or protein. Methylation is responsible for activation of the MTHFR enzyme. The four methylation cycles (see diagram below) produce additional methyl donors which are responsible for turning on or off genes like MTHFR- this is known as epigenetic modification of genes.

Methylation is crucial for optimal health. This process controls not only gene activation or silencing, but is responsible for liver detoxification of harmful substances, building neurotransmitters in the brain, processing hormones, building immune cells, DNA and RNA synthesis, producing energy in our cells, and producing the protective coating on nerves.

Under methylation has been linked to various cancers and multiple neuro immune syndromes such as anxiety, depression, ADD/ADHD, dementia, Alzheimer's, chronic fatigue syndrome, tic disorders, coronary artery disease, memory loss, inflammatory autoimmune disorders, celiac disease, gluten sensitivity, eczema, psoriasis, fibromyalgia, rheumatoid arthritis, autistic spectrum disorders, autism, delayed growth, sleep disorders, irritable bowel disease, drug reactions, migraines, POTS disease, diabetes, thyroid disorders, allergies, male and female infertility, developmental delays, and various other complicated conditions that affect the nervous and immune systems. A Hallmark of all of these disorders is that the symptoms vary in intensity daily and thus are very frustrating for the patient and physician. This is why a genomic analysis and epigenetic evaluation is so useful to validate symptoms and create a targeted treatment approach.

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METHYLATION CYCLE SIMPLIFIED