

Excerpt from the upcoming book entitled, Why Me? Genetics of Autism.
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MTHFR

Genetics has taken such a huge leap in the past 10 years. Our search for an ultimate cause of disease and illness, our search to understand our history, and ourselves has pushed our understanding of our cookbook, our recipe. Everyday, more research is published, more clinical evidence is obtained, and the more the medical community understands how to help people as individuals, not as diseases.

MTHFR (technically known as methylene tetrahydrofolate reductase, and fondly known as the mother.....something gene) is extremely common in the general population, with approximately 40% of Caucasians having inherited one mutation (termed heterozygous) and an additional 10% having inherited 2 mutations, one from each parent (termed homozygous abnormal). That's a whopping 50% of people that have at least one mutation in this gene. (Some ethnicities have greater odds, while other ethnicities have less odds. What matters more is what mutations do *you* have.)

From a conventional genetic perspective, and when you talk to most conventional geneticists, they disregard these mutations, because they are so common. Most geneticists are looking for something uncommon, something rare, not something found in 1 out of 2 people.

However, in my opinion, this mutation *does* matter. Because this MTHFR gene mutation contributes as a powerful risk factor for cardiovascular disease and emotional imbalances such as anxiety and depression. Those conditions affect a LOT of people, and more importantly, can be easily treated. So what if this is common? What if treating this can help you feel better! Other health conditions are common too, high cholesterol, Vitamin D deficiency. While those aren't examples of genetic abnormalities, they are common health conditions that are treated on a regular basis, so why not treat a common genetic mutation on a regular basis if it can help you live longer and be happier!

Let's take a step back into science class and understand the physiology of MTHFR. As a physician, I don't like to understand a patient's disease; I like to understand their physiology. If they have high blood pressure, I want to know why. Because if I treat the 'why', the patient will get a better outcome compared to just giving a high blood pressure

medication. Understanding you deeper helps find better answers. Understanding the process of the body, the physiology is the key to identifying the best treatments.

Most processes in the body can be thought of as an assembly line. If you're making a car, you start with the frame, and then add the wheels, the doors, the horn, etc. The same happens in the body. We take food, water, and oxygen from the air, and abracadabra, we've got bones, muscles, emotions, hormones, and children. It's quite amazing when you think about how our DNA can convert simple things into amazing living beings such as ourselves.

Part of this conversion from food, water, and oxygen into *amazing* includes the use of folic acid. Both folic acid and folate (very similar and I'll use these terms interchangeably) are converted from the foods we eat into a usable form in the body. They follow an assembly line to be transformed into something we can use. This process is known as 'methylation' and one of the essential enzymes in this process is called MTHFR. Methylation is a scientific term for the addition of a carbon atom onto a molecule, but it is an essential process for the body to take folate and make it useful.

Folic acid is converted into a form known as 'methyl-folate' or '5-MTHF'. (These terms are also used interchangeably). Methylfolate is necessary for the production of serotonin, dopamine, norepinephrine, adrenaline, and nitric oxide. Wow! These are some pretty important chemicals when it comes to brain function and mood, behavior and development.

Imagine what would happen if your body lacked serotonin. Bam! Depression. What if it lacked dopamine? Bam! Apathy. Norepinephrine, Adrenaline deficiency? Lack of focus and anxiety. Do these sound like anyone you know? You might have an MTHFR genetic mutation.

A 2016 meta-analysis of 1978 children with autism and 7257 neurotypical children strongly suggested a significant association between MTHFR C677T polymorphism and autism risk. [Rai V](#). Association of methylenetetrahydrofolate reductase (MTHFR) gene C677T polymorphism with autism: evidence of genetic susceptibility. [Metab Brain Dis](#). 2016 Aug;31(4):727-35.

Do you know your MTHFR status? Testing is easy. It can be done through saliva, cheek swab, or blood. Insurance may cover the test, depending on your individual plan. If a cheek swab is a simple answer for you, we offer the testing at: www.SpectrumAwakening.com

Once you've had a positive genetic test, or you just want to try the treatment without a test results (Yes! You can do that!), the treatment is easy. Remember the assembly line.

MTHFR converts folic acid to methylfolate. Well, methylfolate comes in a pill. If you can't make it, you can take it!

Now, remember the temperament of Goldilocks, not too small, not too big, but just right. That's a lot of medicine. With methylfolate dosing, more is not necessarily better. For those of you statistically inclined, think of a bell curve. A small dose of methylfolate gives you a small response, a larger dose gives you a better response, and too much makes you go crazy!

Unfortunately, there is no magic rule to dosing. It's not age, it's not weight... it's genetics. So there is some trial and error in dosing. The most minimal dose is around 400mcg (0.4mg), while the highest dose recommended is 20,000mcg (20mg). It's also good to pair methylfolate with B12. They go together like peas & carrots.

I formulated 2 all-natural products for my patients in my autism clinic. One is a simple combination of methylfolate and methyl-B12, called B-Licious for MTHFR. If you're looking for a good tasting chewable, where the dose can be increased and decreased easily, this is a nice, simple formula.

If you want to kick it up a notch, try my original formula, Power & Focus. It combines methylfolate and methyl-B12 with amino acids to create serotonin, as well as B6, magnesium, and zinc, that are often beneficial to support normal and healthy brain function.

Both are available on www.SpectrumAwakening.com

What should you expect when you take a methylfolate supplement? I may sound like a weight loss commercial, but results may vary! I have hundreds of stories from my practice, but my favorite was of a boy named Tyler. He was nine years old when I met him, and for the past 3 years, he had been around the country being treated for suicidal ideation. Now, what 6-year-old wants to commit suicide!?!? He had been on a myriad of anti-depressants, anti-psychotics, and anti-anxiety meds without benefit, and his family was supportive and loving, and not the family you would expect to have a child with emotional problems. He tested positive for an MTHFR mutation, and after one week of taking B-Licious for MTHFR, his mother called me in tears, crying, "I have my boy back! I have my boy back!"

Well by the end of our conversation, I was in tears too. His depression had lifted like a blanket from a bed. What medications couldn't do in years, methylfolate did in days. That was 6 years ago, and while he needs to take methylfolate for life, he is a happy, productive kid in high school, doing well academically, playing sports, and enjoying his friends and family.

So that is a great example of the potential of methylfolate and treating MTHFR mutations. But not all people end up with such a great response. Sometimes, too much methylfolate or methyl-B12 can make you *crazy*!

COMT / MAO

The assembly lines of our body are in constant motion. We produce and remove, produce and remove. We breathe in... and out... and in... and out. We take in nutrition, we expel waste. We inhale oxygen, we exhale carbon dioxide. So be it with our emotions. We convert our food into chemicals of feeling, such as serotonin, dopamine, and for this section, adrenaline.

Fight or flight. Feel it coursing through our veins. The rush of adrenaline gives us the power to overcome overwhelming odds! Too much, and we explode! Too little, and we cower in fear. Are we the Warrior? Or the Worrier?

This is the dichotomy of adrenaline. Too much and we start punching and biting kids in school. Too little, and we fear the world and hide behind our parents legs. We must always go back to the metaphor of the assembly line. I often reflect back to a popular 'I Love Lucy' episode when she was in the chocolate factory.

When the machine was slow, she could handle everything. She had time to spare. When the machine sped up, she kept busy, but could handle the flow, and when that machine went into overdrive, she couldn't keep control.

The same thing happens in our brains. What is the speed that we process adrenaline? This is a process known as metabolism. Are we fast metabolizers? Or slow metabolizers?

Fast metabolizers remove adrenaline from the body quickly. Without adrenaline, we become cowards. We stick out heads in the sand like an ostrich. We fear everything; we have anxiety, especially around transitions and unexpected change. (This is why visual schedules work so well with kids with autism. We show them what to expect for their day and reduce anxiety of sudden changes.) With these children, we often have to walk on eggshells.

Slow metabolizers are our warriors. The angry, the fighters. They can't remove the adrenaline from their bodies. Perhaps they have a diagnosis of Oppositional Defiance Disorder. Perhaps you go around trying to please them every second so they don't punch your face when you're not looking.

The genetics of adrenaline metabolism are COMT and MAO. They are two different enzymes that can remove adrenaline from the brain. Do it too fast, and you're anxious. Too slowly, and you're a warrior.

A certain COMT genotype, termed Val/Val, is present in 12% of children with autism versus 20% of neurotypical children. The lower this percentage, the more the overactivity. The prevalence of COMT polymorphisms are also very common in children with 22q11.2 deletion. MAO activity was reduced in 70% of children with autism.

[Guo T¹](#), [Wang W](#), [Liu B](#), [Chen H](#), [Yang C](#). Catechol-O-methyltransferase Val158Met polymorphism and risk of autism spectrum disorders. [J Int Med Res](#). 2013 Jun;41(3): 725-34.

[Gu F¹](#), [Chauhan V¹](#), [Chauhan A¹](#). Monoamine oxidase-A and B activities in the cerebellum and frontal cortex of children and young adults with autism. [J Neurosci Res](#). 2017 Feb 2.

Do you know your COMT/MAO status? Testing is easy. It can be done through saliva, cheek swab, or blood.

Once you've had a positive genetic test, or you just want to try the treatment without a test results (Yes! You can do that!), the treatment is easy. First, identify the anxious versus the aggressive. Now this can be tricky. Some children react defensively to stress. They may lash out to protect themselves, versus someone who is consciously aggressive.

If a child is fine when things are going their way, they may be a fast metabolizer. If they are aggressive without prompting, they may be a slow metabolizer. Remember the chocolate factory! How well can I handle my adrenaline?

Those who are anxious and do not like transition, the walking on eggshell kids, try our sweet syrup, *Temper Tamer*. It tastes like molasses. For the angry, I've developed two supplements, an amino acid combination called *Calm & Cool*, as well as a *Calcium/Magnesium Pear Powder*. These two in combination can lower adrenaline levels and support a normal mood.

These supplements are available at: www.SpectrumAwakening.com

One of my great families had boy named Brain. His mother jokingly called him the 'tumor' because he never left her side. Ever. Let me explain the definitiveness of this word. He was eight, and never went to school, never slept in his own bed, and never let his mother have a moment of piece in the bathroom. Ever. His anxiety was so extreme, he couldn't function. He is the poster child of a fast metabolizer. There's no adrenaline left to have some cajones. You do need some courage. Courage to be brave. Courage to face

our fears. Courage to let mom have some time in the bathroom to herself, or maybe have a night in bed with only her husband!

I gave him a combination of *Temper Tamer* and *Calm & Cool*. And again, within a couple weeks, Brian's mom called me in tears. He was playing downstairs by himself! The boy who never left her side in eight years said, "I'm ok Mom! I'm just playing!" She emailed me a letter, which I hang in my office. Every time I read it, I know there's an answer. There is hope for everyone, recovery is possible, and what I tell every family in my practice, do everything you can for your child.

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