

The Link Between MTHFR Gene Mutations and Disease, Including Thyroid Health

An Interview With Dr. Ben Lynch, Expert on MTHFR Polymorphisms

By [Mary Shomon](#) - Updated June 13, 2014



Dr. Ben Lynch, expert on MTHFR Gene Mutations and Polymorphisms

The issue of a gene called MTHFR and its link to disease – including [thyroid disease](#) – is becoming an increasingly hot and controversial topic. To learn more about the theories and developments related to MTHFR – which is the abbreviation for the gene called methylenetetrahydrofolate reductase, I turned to Dr. Ben Lynch, an [naturopathic](#) physician who has focused attention on MTHFR polymorphisms and genetic changes, and the linkage to diseases and conditions. Dr. Lynch runs a site, [Mthfr.net](#), that is dedicated to research and awareness of MTHFR, and his book on the subject will be published in 2014.

Question: Can you please explain, for those who don't know or haven't heard, exactly what an MTHFR polymorphism is?

Dr. Ben Lynch: The MTHFR gene produces an enzyme called methylenetetrahydrofolate reductase. The job for the MTHFR enzyme is to convert one form of folate into the most active and usable form of folate in the human body – in every single cell. MTHFR takes folic acid and changes it so the body can use it. Folic acid by itself is worthless – it does absolutely nothing – until it is run through various enzymes in the body and transformed into these forms of folate the body can use – such as methyltetrahydrofolate, or commonly shortened as methylfolate.

Methylfolate is a key compound that performs two critical tasks.

First, methylfolate helps make neurotransmitters in your brain. Neurotransmitters are what allow us to think, sleep, run away, express emotions and learn. When methylfolate levels are low, so are your neurotransmitters. Low production of neurotransmitters may cause conditions of addictive behavior, [depression](#), anxiety, ADHD, mania, irritability, insomnia, learning disorders and others.

Two, methylfolate also allows us to make a critical compound called s-adenosylmethionine -- also known as SAMe. SAMe is critical as it helps regulate more than 200 enzymes in the human body – this is second only to adenosine triphosphate (ATP), which is the body's cellular power unit. Without ATP, life ceases. Without SAMe, life ceases.

With decreased levels of [SAmE](#), you, or your loved ones, are at higher risk of conditions such as cancer, infertility, [miscarriage](#), autism, Down syndrome, [thrombosis](#), [high blood pressure](#), speaking problems and other health issues.

It is very common for the MTHFR gene to not work as well as it should. Why? Because one in two people have an area in the MTHFR gene that has an [incorrect](#) DNA base. This DNA base change is called a polymorphism. Poly meaning 'many' and morphic meaning 'shape'. When the MTHFR gene has a polymorphism, the enzyme it produces has an altered shape. The altered shape reduces the functional ability of the MTHFR enzyme. The altered function causes decreased [neurotransmitter](#) function and decreases SAmE production.

Question: What are the methylation pathways and what do they do?

Dr. Ben Lynch: Let's first define [methylation](#). Methylation is the act of taking a single carbon and three hydrogens, namely a methyl group, and having it attach itself to an enzyme in your body. When this methyl group attaches to an enzyme, the enzyme performs an action. A common action performed by methylation that you appreciate daily is the breakdown of histamine. A methyl group is made by the methylation pathway and it floats around until it finds a specific enzyme to bind to. In this case, the methyl group binds to histamine. When a methyl group binds to histamine, histamine breaks apart and goes away.

You can now figure out what happens if your methylation pathway is not producing enough methyl groups in this case. Your histamine does not break apart and thus your [histamine](#) levels increase, causing your nose to run and eyes to itch.

What is the function of [SAmE](#)? The function of SAmE is to simply take what is called a 'methyl group' and give it away to over 200 enzymes in the body in order to perform various critical functions. Some key functions of this freely donated methyl group are to:

- Protect your DNA. This is very important. For example, if your DNA is not protected, then it is susceptible to damage by viruses, bacteria, heavy metals, solvents and others. Over time, this damage becomes significant and may result in cancerous cell proliferation. -
- Reduces [histamine levels](#)! Repeating this so it sinks in. A methyl group given away by SAmE helps eliminate histamine from the body. Those with allergies or rashes may have higher levels of histamine and decreased methyl groups. -
- Produce a key component for your cell membranes called [phosphatidylcholine](#). The methyl group donated by SAmE helps build phosphatidylcholine which then gets incorporated into the walls of all your cells, known as cell membranes. If these cell membranes become damaged and weak, the cells become fragile, allow toxins and harmful things into the cell, do not carry in useful nutrients and then they die. Excessive cell membrane damage can lead to serious medical conditions such as [multiple sclerosis](#) and [cancer](#) to name a few.

Question: How did you become aware of MTHFR, and what motivated you to make it your professional crusade?

Dr. Ben Lynch: One afternoon, a lady asked me about natural treatment options for [bipolar disorder](#). I began rattling off all the usual natural approaches to stabilizing those with bipolar disorder. For some reason, I felt my answer incomplete, likely because I had been out of medical school for a few years and I wanted to see if research brought forth anything new.

So I hit the research and discovered that researchers were investigating bipolar and genetics, specifically MTHFR. I had no idea what MTHFR was so I entered 'MTHFR' into the medical research database, PubMed, which is the US National Library of Medicine's research database. I was astounded what came back to me. Over 3,000 papers at the time. Now it is over 5,200 papers just on MTHFR alone – and increasing.

As I scanned the research paper titles with MTHFR in it, I was amazed by how many conditions appeared. [Cleft palate](#), [hypertension](#), [thrombosis](#), [schizophrenia](#), [myocardial infarction](#) (heart attack), [acute lymphoblastic leukemia](#), [recurrent pregnancy loss](#) – and this was just on page 1!! ([You can see for yourself!](#))

I was so stunned that I never heard about this that I actually 'tweeted' about MTHFR – and I hardly ever 'tweet'. The next day, I was bombarded by people who were looking for a physician knowledgeable about MTHFR. Needless to say, my knowledge was nil but I promised them that I would learn. To this day, I continue to learn the complexities of MTHFR and pass my knowledge onto physicians.

MTHFR polymorphisms are SO prevalent and so damaging to millions and millions of people. Yet, with awareness, those with MTHFR polymorphisms can increase their odds of a totally healthy life if they just knew that their lifestyle, dietary and environmental exposures really speed up their risk for disease.

Question: Whose health can benefit most from an awareness of MTHFR ?

Dr. Ben Lynch: Simply – those who have the genetic polymorphism in their MTHFR gene – which remember, is one in two of us. Even those who do not have MTHFR polymorphisms benefit knowing about it as their loved ones or friends may have it and if they are made aware of it, they may impact their lives significantly.

It should be mentioned that doctors need to be made aware of how significant MTHFR is because they are the ones managing the health of the patient. Thus, it is up to the doctor to inform the patient and screen them for MTHFR polymorphisms.

Question: Who should get tested for MTHFR and where/how does one get tested?

Dr. Ben Lynch: Everyone should get tested. Why? Because 1 in 2 people are affected and if one knows they have a MTHFR polymorphism, they know they have to be very proactive in taking care of themselves.

Testing for MTHFR needs to be seen as a screening test. If one wants to get pregnant, they test for MTHFR. If one wants to reduce disease risk factors in general and optimize their health, they test for MTHFR. If one wants to identify why their family seems to have more risk of neurological disorders, cardiovascular disease, cancer, depression or autism to name a few, they test for MTHFR.

If you approach your physician asking to screen you for MTHFR, two things will happen. One, they do not know what it is and two, they will state MTHFR is not a big deal and there is nothing to worry about – it is all a fad.

On the off chance you have a progressive, current physician, they will test you for MTHFR polymorphisms. A word of caution here.

Many insurance companies do not cover MTHFR testing and if they say they do, they still may come back and charge you an exorbitant fee.

That said, there are some reliable testing companies out there that do MTHFR testing reliably and affordably and may even be covered by insurance.

The two leading MTHFR testing companies are [Spectracell Labs](#) and [Any Lab Test Now](#).

If you do not have insurance, out of pocket expense is not that bad, especially given the potential life-changing results you may obtain.

For information about MTHFR testing options – along with the ethical considerations – [learn more at the MTHFR site](#).

Question: What are your top recommendations for people with MTHFR polymorphisms?

Dr. Ben Lynch: MTHFR polymorphisms have been around for generations and generations – and they are being [passed on at an even faster rate due to folic acid supplementation](#).)

I believe the poor choices in food, unhealthy environment, fast-paced lifestyle, corporate lobbyists and symptom-based medicine are contributing the expression of MTHFR polymorphisms.

That said, it is critical people snap out of it and stop listening to the drug ads on TV imploring ‘Ask your doctor if this drug is right for you.’ I can tell you right now, it is not.

The top recommendations in bullet form -- and to let you know I practice what I preach -- are the following:

- Eat whole, organic, non-GMO, free range, wild [foods](#).
- Stop ingesting synthetic folic acid supplements and folic acid enriched foods. Use only natural folates such as folic acid and/or methylfolate.
- Eat foods containing natural folates such as uncooked leafy greens. (Yes -- you have to eat salads. Daily.)
- Drink filtered tap water – avoid bottled water in plastic that contains bisphenol A (BPA).
- Avoid gluten and dairy products.
- Limit exposure to chemicals in the environment (i.e. formaldehyde in new clothing, carpets, furniture and new [home construction](#), and fire retardants in clothing, bedding, couches, carpets).
- Belly breathing.
- Positive thinking.
- Laughter.
- Get out in nature. We are, after all, mammals. Don't forget that. We need natural settings.
- Love your occupation – if you don't love it, switch.

- Relationship - a happy marriage or positive relationship,
- Family – participate with your loved ones and enjoy them.
- Friends – share and participate in activities.
- Take vacations.
- Exercise – enjoyable – not excessively. Participate in team sports, clubs.
- Sauna.
- Build a medical team with progressive, forward thinking MDs, NDs, DOs and other health professionals.

Question: Is there a [connection](#) between thyroid health and MTHFR?

Dr. Ben Lynch: Yes there is. We all know how prevalent [hypothyroidism](#) is. If one is hypothyroid, meaning their [thyroid gland](#) is functioning slowly, then the individual will have a sluggish MTHFR enzyme as well – even if this person does not have the MTHFR polymorphism.

Why?

Because the thyroid produces what is called T4, also known as [thyroxine](#). Thyroxine helps produce the body's most active form of vitamin B2, flavin adenine dinucleotide (FAD). Vitamin B2 must be converted into active FAD by thyroxine in order for the body to effectively use vitamin B2.

The connection between FAD and MTHFR is that the MTHFR enzyme must have an ample supply of FAD in order to function. If FAD levels are low due to low levels of thyroxine, then the MTHFR enzyme slows down, causing low methylfolate levels. We now know from above that low methylfolate leads to low neurotransmitters and low SAME.

Question: What can hypothyroid [patients](#) do, i.e. nutrition and lifestyle changes, to cope with MTHFR inhibition?

Dr. Ben Lynch: Hypothyroid patients must have their doctor monitor their [TSH, T4, T3, and thyroid antibodies](#) often in order to optimize thyroid function. If any of these values are off, then the thyroid function will be compromised and thus the MTHFR enzyme will be as well.

It is critical that doctors monitor all these markers and [not just TSH](#). TSH measurement by itself is worthless. It is similar to someone asking you where the car keys are and you reply back, "Over there." 'Where?' you ask again. They reply back, "Over there." Not very helpful unless the person actually points where 'there' is or defines it better for you.

Key nutrients for thyroid function are magnesium, iodine, selenium, zinc and tyrosine - which comes from eating – and absorbing - enough protein. Methylfolate, [produced](#) by the MTHFR enzyme, is also needed to help convert tyrosine into active thyroid hormone.

A lifestyle change for optimizing your thyroid would be to limit exposure to chlorine, bromine (bromide), and [fluoride](#)- basically the other halogens. Why? Because these other halogens mimic the charge of the halogen [iodine](#), and that prevents iodine from binding to the receptor on your thyroid gland.

If you have other halogens on your thyroid receptor, how is iodine supposed to be bound to it? Difficult – especially if one is iodine deficient and [some American women are deficient in iodine](#). Talk with your doctor about how to monitor iodine levels and properly supplement when necessary.

I recommend installing chlorine shower filters for your shower, filtering your drinking water, limiting exposure to fluoride, and limiting or avoiding drinking soda, as many citrus sodas contain brominated vegetable oil.

Environmental changes to protect your thyroid would be avoiding heavy metals such as mercury. Mercury and other heavy metals are very toxic to the thyroid. Mercury amalgams are researched to be an [aggravating factor in causing Hashimoto's disease](#).

A pervasive food that negatively affects your thyroid is gluten. [Removing gluten entirely from your diet may significantly improve your thyroid](#). This means 100% avoidance – not ingestion once a week or once a month – complete avoidance. This is especially important if you have [celiac disease](#).

Question: Can you tell us about your MTHFR.net website?

Dr. Ben Lynch: I do my homework. If I state something, I cite research or I clearly state it is a working theory. I don't pretend I know something I don't. If I don't know something, I simply don't write it or make up some answer, I simply say three words – "I don't know."

If people would stand up and understand that saying "I don't know" is okay and useful, then medicine and other areas of life would be so much better.

I constantly read – and critically analyze - cutting edge research papers on MTHFR, methylation, nutrigenomics, one carbon metabolism, mitochondrial function and epigenetics. I love what I do and am totally engrossed and passionate about it as well. Why? Because I know it is going to change how medicine is practiced.

The medical model today is one that needs to be chucked out the window. The symptom-disease based model is highly ineffective and doctors know this. They are turning to [naturopathic](#) and functional medicine. The problem is these doctors continue to practice the same symptom-disease based model using supplements, herbs and other concoctions instead of drugs and surgery. Yes, this is an improvement most of the time but it still is not addressing the absolute underlying cause of why their patient is ill.

I am educating other physicians about the new model that is emerging from all this and that is biochemical nutrigenomics. When a physician focuses on the patient's lifestyle, diet, surrounding environments, hobbies, emotional status, biochemistry, genetics and genetic expression, then amazing things happen. Just ask the doctors who have attended some of my conferences. The learning curve is steep and frankly a pain, but very worthwhile because once you learn it, you continue to perfect it and your patients get better faster – and faster.

There is nothing better to a physician than having to put a sign out on the front door – "Patients needed. All my other ones are healthy."

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Dr. Ben Lynch is a naturopathic physician who has focused his research and teaching on MTHFR polymorphisms and genetic changes, and the connection to disease. Dr. Lynch's site <http://mthfr.net> features research, articles, links and information related to MTHFR genetic mutations and polymorphisms. His book on this topic will be available during the spring of 2014 at this site, and online bookstores. You can also find Dr. Lynch on [Facebook](#).