MTHFR SUPPORT

MTHFR

ITS EFFECT ON HEALTH AND DISEASE

What's the effect of MTHFR on health and disease?

Hello,

I'm Carolyn Ledowsky, the founder of MTHFR Fertility, MTHFR Gene Support and MTHFR Support Australia. As an MTHFR researcher, trainer and presenter, I am committed to teaching everyone how and why the MTHFR genetic polymorphisms may affect your health.

One of the things that frustrates me the most is medical practitioners who say it doesn't matter.

I have spent 10 years in clinical practice working with patients with the MTHFR gene and disturbed methylation and I can categorically say that it does. It's the difference between you getting better and not, much of the time.

I'm not saying that everyone with the MTHFR gene will have a problem with their health, but I am saying that you should consider exploring how this may affect you. You can't assume that everything is fine, particularly when you are not progressing with your health.

Cleft Lip and Palate

These are birth defects that occur when a baby's lip or mouth do not form properly during pregnancy. They are called orofacial clefts.

Baby with cleft palate

Cleft palate

Tongue Tie

Tongue-tie (ankyloglossia) is a condition that presents at birth which restricts the tongue's range of motion.

Orofacial clefts and tongue tie are increasing

- Rates are increasing
- They are knowsn within a wider group called Neural Tube Defects
- Folate is key:
- MTHFR
- Folic Acid vs Folate - Big difference

What is MTHFR?

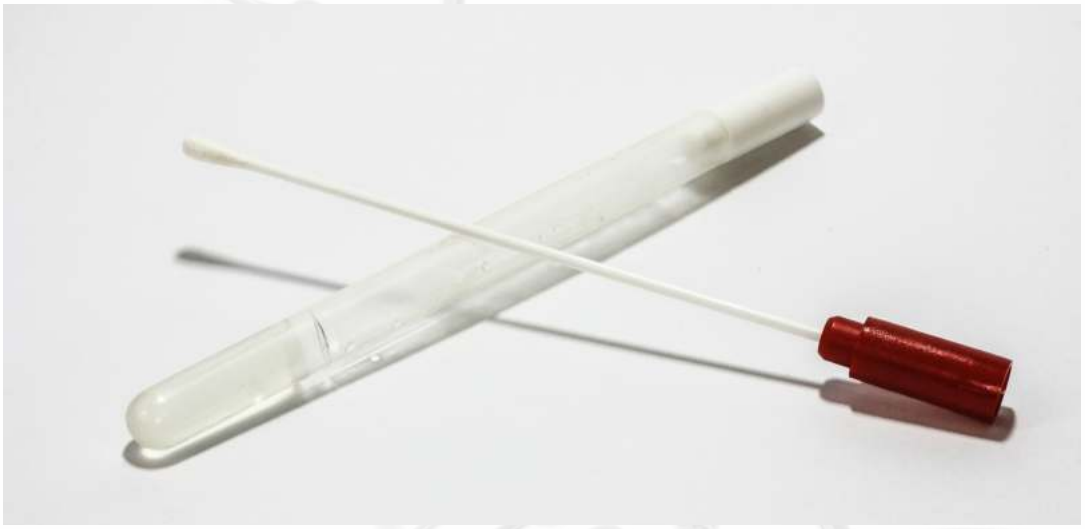
The MTHFR gene encodes for the MTHFR enzyme, which stands for methylenetetrahydrofolate reductase.

The MTHFR enzyme converts folate, in the presence of cofactors, into the active form, 5-methyltetrahydrofolate, for it to be metabolized and used in various biochemical reactions within the human body. This is done through a process called methylation, which is a mechanism of activating certain molecules or mechanisms in the body through adding a methyl group to the target molecule that is required to be activated.

The activation of folate in the form of 5-methyltetrahydrofolate is essential for DNA synthesis, immune function, amino acid metabolism, detoxification, formation of blood cells and platelets, and single carbon metabolism. Therefore, we'll take a deeper look at folate metabolism, how the MTHFR gene is involved in folate metabolism, important concepts in the fields of genomics and epigenetics, and why we care about looking at our genes.

How can I find out if I have an MTHFR Gene mutation?

Know your MTHFR status when you order an at home test kit. This is a buccal swab (which means you collect DNA from the inside of the mouth). Its quick and easy. You send it back to the lab and your results are sent to you via email. Visit our website to learn more about our test kits.



Why is it important to look at your genes?

Your genes are not only essential to understanding what makes you unique, but also vital in determining the cause of any health-related issues such as genetic defects and metabolic disorders. Genes are responsible for encoding the production of proteins in the body.

The production of proteins, and thus enzymes, is necessary for metabolic and other biochemical reactions to occur within the body. A lack of, an abundance of, or mutation of one type of protein could result in a negative impact on your health. This is because a mutation down regulates the amount of methyl folate (active folate) that you can create. This has been associated with many conditions including:

- Autoimmunity
- Chronic fatigue
- Diabetes
- Cancer
- Autism
- Insomnia
- Depression
- Infertility
- Cardiovascular disease

Genomics and Epigenetics

The fields of genomics and epigenetics are crucial to medical research. Genomics focuses on the sequencing of an organism's DNA, whereas epigenetics is the study of changes related to gene expression, but not alteration.

So in other words, you can't do anything about your genes, but you may be able to affect the way they act. Both fields of study are important in the understanding of your genes and how they function in your body.

It's very important to remember that your environment plays a significant factor in how your genes affect your health. Your environment includes what you eat, what you drink, and the toxins you are exposed to at work, including mold, bacteria, viruses, and stress.

Is there one type of MTHFR gene mutation?

Depending on the mutation you have the consequences are slightly different. Each mutation follows a similar trend towards less methylation within the body or less active folate production (5-MTHF). If a mutation is present, the enzyme can have a 20% to 70% loss of function.

Since everyone has two copies of each gene (one from each parent), loss of function depends on whether there are one or two copies of the MTHFR gene mutation present.

One copy of a gene = Heterozygous (C677T= ~40% loss, A1298C=~20% loss) (This means you have one copy from mom OR dad)

Two copies of a gene = Homozygous (C677T=~70% loss, A1298C=~40% loss) (This means you have one copy from both your mom AND dad)

One copy of both C667T and A129C = compound heterozygous = ~50% loss
(This means mom and dad each gave you one copy of C667T or A1298C.)

In general, less methylation occurs in people who have two copies of an MTHFR gene mutation.

MTHFR Mutations = Less Methylation

Methylation and MTHFR

Methylation is responsible for turning multiple processes within cells “on or off”. Proper methylation (adding or removing methyl groups (CH3) from molecules) within the body ensures cells are doing their jobs.

Think of methylation as a master switch. Any biochemical product that ends in MT is a methyltransferase. Methyls act as a switch for methyltransferases, they make them stop and go.

Methyltransferases have important biochemical roles in our bodies. For example, the breaking down of toxic oestrogens through hormone production via COMT, or the health of cellular membranes and energy through choline production via PEMT.

When methylation is not working or is down regulated, your body is not able to produce correct responses to the environment that's damaging your body. Certain processes within cells will be turned on or off for too long, leading to an impaired ability to:

- Get rid of toxins (detoxification)
- Repair and rebuild DNA/RNA
- Produce and process hormones
- Build immune cells
- Repair cell membranes
- Turn the stress response on and off
- Metabolize fat
- Produce energy
- Recycle and build neurotransmitters

When these vital cellular processes are not working correctly, adverse symptoms can arise such as:

- Cardiovascular disease
- Impaired immunity
- Chronic inflammation
- Diabetes
- Anxiety
- Depression
- Chronic fatigue
- Cancer
- Fibromyalgia
- Infertility and miscarriages

Problems with methylation will amplify the symptoms of existing autoimmune and psychiatric conditions.

Managing MTHFR Gene Mutations

Knowing if you are positive for an MTHFR gene mutation is important because it may affect your biochemistry to the point that your energy, hormones, mood, immune system and detoxification systems fail to function properly.

What plays the biggest role with MTHFR mutations causing you problems is your environment. What you eat, the amount of sleep you get, the stress you are under, the toxins you are exposed to, all play a role in the way your genes are expressed.

Your genes are always feeding off of the environment to determine how much a gene should be expressed and what genes to turn on and off. Working to create a stress-free environment is a simple way to begin managing MTHFR gene mutations.

Over the past couple of decades science has been uncovering vast amounts of information in the field of nutritional and biochemical sciences There are some basic guidelines to managing MTHFR mutations through diet and lifestyle. These guidelines revolve around consuming foods that are easy for your body to digest and do not cause inflammation to avoid toxins overloading the body's detoxification systems.

It's recommended that you:

- Avoid cereal grains (because they are fortified with folic acid which is not recommended. Find out why folic acid is not recommended is not recommended in our article here)
- Avoid dairy products (they put extra stress on our immune system)
- Avoid processed foods (they lack nutrients and folic acid is often added)
- Lower alcohol consumption (it depletes all our B Vitamins)
- Quit smoking (puts too many harmful chemicals into our body)
- Reduce or modulate stress (stress responses consume the most methyl groups)
- Reduce environmental toxins (MTHFR mutations impair the ability to detoxify, placing extra stress on the liver)
- Increase vegetable consumption (especially dark leafy greens)
- Maintain a healthy weight

Conditions Associated with a lack of Methylation

- ADD/ADHD
- Addictive Behaviour
- Allergic conditions
- Ageing
- Anorexia
- Alzheimer's Disease
- Anxiety
- Asthma
- Autism
- Autoimmune disease
- Bipolar
- Bulimia
- Cancer
- Chronic degenerative diseases
- Cardiovascular Disease
- Chronic fatigue
- Cleft Palate
- Diabetes
- Down's Syndrome
- Delusions
- Depression
- Poor detoxification
- Fibromyalgia
- Headaches ADD/ADHD
- Infertility
- Joint stiffness, pain swelling
- Insomnia
- Muscle pains
- Low neurotransmitters
- Obesity
- Obsessive Compulsive disease
- Oppositional defiant disorder
- Pain
- Phobias
- PCOS
- Psychosis
- Schizophrenia
- Recurrent Pregnancy Loss/ Miscarriage
- Thyroid Dysfunction

Key Considerations If You Have An MTHFR Mutation

The first steps you need to take are:

- Assess your environmental factors
- Are you getting enough sleep?
- Are you too stressed?
- Are you eating a good diet?
- Are you avoiding folic acid?
- Are you exposed to heavy metals or toxins?

Common Medications That Reduce Methylation

Common medications may be reducing your ability to methylate by lowering B12 or overall folate levels. If less B12 and/or folate is available to use in your body, symptoms may become worse. Medications that could be interfering with your B12 and folate levels include:

- Antacids
- Cholestyramine
- Nitrous oxide – your dentist will often use this. Its known as laughing gas.
- Methotrexate- this may be used in cancer or autoimmune disease
- Niacin (at high doses) – because Niacin uses methyl groups in its metabolism
- Theophylline
- Cyclosporin
- Metformin
- Phenytoin
- Bactrim – an antibiotic
- Sulfasalazine
- Triamterene
- Trimethoprim
- Ethanol
- Oral contraceptive pill
- Antimalarials

Action Steps for Balanced Methylation

1. **Avoid folic acid containing foods** – supplements, bread /baked goods (ALWAYS READ LABELS)
2. **Increase natural folate containing foods** – leafy greens & legumes
3. **Avoid allergenic foods** - gluten, dairy, soy.
4. **Avoid inflammatory foods** e.g. processed foods, refined carbohydrates/sugar, additives.
5. **Reduce exposure to toxins & chemicals** – e.g. plastics/ BPA, synthetic substances. Instead use natural house hold cleaning products, natural make-up / skin care & eat organic where possible.
6. **Increased Colourful vegetables** - antioxidant and detoxification support
7. **Stress management** – save your methyl groups!
8. **Avoid smoking/ passive smoking & reduce or eliminate alcohol**

When To Supplement With Folate And What to Supplement With

So what are some of the things you can do to help?

1. Make sure you are taking the right form of folate
2. Make sure you are taking the right dose of folate
3. Optimize your detoxification pathways to prevent further stress on your thyroid.

Folic Acid (Not Recommended)

- Folic acid is a synthetic compound that has NO physiological function until converted to dihydrofolate by dihydrofolate reductase (DHFR)
- The DHFR enzyme breaks down folic acid much slower than naturally occurring folates which causes a build up of folic acid
- Folic acid has a stronger attraction to the folate receptors, blocking them from pulling natural folates into the cell for metabolic processes (our natural folates like leafy greens are essential for our folate levels)

Essentially that means that you are putting a big brake on your folate pathway. The opposite of what we need to do.

Folinic Acid (One form of active folate)

Folinic acid or 5-formyltetrahydrofolate, enters directly into the middle of the folate cycle. It can be made into any of the possible products of the folate pathway.

There are three main points regarding folinic acid you should take away with you:

- Folinic acid can be made into everything folic acid or natural folates can be made into.
- Folinic acid has direct physiological functions that help create the building blocks of DNA.
- Folinic acid bypasses the DHFR enzyme in the folate cycle, making it an ideal supplementation strategy if you have a DHFR mutation that reduces your ability to metabolize folates and folic acid.

5-MTHF or Active Folate (Recommended)

If you know you have an MTHFR mutation, 5-MTHF is going to be the most important supplement for you to take. 5-MTHF is the active form of folate that allows the body to recycle its methyl donors (the molecules responsible for turning off and on physiological processes within the human body by donating methyl groups).

The folate cycle is designed to produce 5-MTHF. 5-MTHF is the end product of the folate cycle and its production is directly affected by the MTHFR enzyme. Having an MTHFR gene mutation reduces your body's natural ability to produce its active form of folate. When you read about the problems caused by a lack of folate, it's due to a lack of the bodies active form of folate, 5-MTHF.

Consume Folate Containing Foods

While MTHFR gene mutations can inhibit the conversion of folate you eat (dihydrofolate) into the active folate (5-MTHF), is it still vitally important to consume as much natural folate as you can through your diet.

Is it important to remember, a mutation in the MTHFR gene simply means a reduction in function, not that it has shut down completely! So, the more natural folate you consume, the more folate your MTHFR gene will have to convert into the all important active folate.
Consuming your folate through the diet also reduces your reliance on supplementation, allowing your food to provide the nutrition your body needs, just the way nature intended.

And remember, eating these folate-rich foods means you are also receiving the benefits from the many other nutrients and phytochemicals they contain. So eat up, and enjoy!

Top 20 Folate Containing Foods








Content of natural folate in micrograms per 100 gram serving of food)

1. Duck liver (raw)	738 mcg
2. Mung beans (raw)	625 mcg
3. Chickpea /besan flour	437 mcg
4. Leek (freeze-dried)	366 mcg
5. Wheat germ	281 mcg
6. Peanuts (raw)	240 mcg
7. Sunflower seeds (toasted)	238 mcg
8. Red capsicum/peppers (freeze-dried)	229 mcg




9. Spinach (raw)	194 mcg
10. Asparagus (frozen)	191 mcg
11. Mustard greens (raw)	187 mcg
12. Quinoa (uncooked)	184 mcg
13. Lentils (cooked)	181 mcg
14. Kelp seaweed (raw)	180 mcg
15. Collard greens (raw)	166 mcg
16. Lima beans (cooked)	150 mcg
17. Black beans (cooked)	149 mcg
18. Egg yolk (raw)	146 mcg
19. Cos or Romaine lettuce (raw)	136 mcg
20. Kidney beans (cooked)	130 mcg

Information sourced from **NutritionData.Self**

What Next?
- Talk to someone who understands

- **Individualised treatment** - designed specifically to address methylation pathway issues/genetic mutations
- **Ideal for people with long standing health issues** - you know there is something more and you need ongoing assistance and support.
- **You need**- regular appointments, constant support, patient liaison appointments for you to keep in touch.

The level of your ongoing support will be decided by you and your practitioner at your first appointment.

- Patients

Many patients that have MTHFR gene mutations are told in no uncertain terms by many medical practitioners 'it doesn't matter'. Our research and clinical experience tell us, it does, so this information is critical for you to navigate your way back to better health. Whether you are trying to fall pregnant and reduce miscarriages, are suffering from anxiety and depression, chronic fatigue, multiple chemical sensitivity or any other health condition, we know, through 12 years of treating patients just like you that understanding how your genetic susceptibility can influence your health and can change your life. Visit our website to learn more about our Patient Knowledge Centre.

- Practitioners

The MTHFR Support Genomics and Methylation Institute is an accreditation program which aims to train practitioners of all modalities on how to introduce genetics into clinical practice in an easy and uncomplicated fashion. Segmented into Beginner, Intermediate and Advanced, the MTHFR Support Genomics and Methylation Institute is designed to cover practitioners new to Genomics, through to those with prior experience.

The interactive course content is designed to be both engaging and informational with learnings covering a wide variety of topics. Visit our website to learn more about the Institute.