

# What GENES

can affect your chances  
of falling pregnant.

# Is MTHFR affecting your chances of falling pregnant and/or staying pregnant?

## About - MTHFR Fertility



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 I can about how and why the MTHFR genetic polymorphisms may affect your ability to fall and stay pregnant. I find it so sad that women are needlessly having multiple miscarriages and suffer the heartache that goes along with that, yet if they had known they had the MTHFR gene mutation before they started out they would have done things differently.

I also see so many couples pushed down the IVF route because they are given the diagnosis of 'unexplained' infertility and yet it's often a simple change that can help them fall pregnant. So stay with me and I will endeavour to teach you all you need to:

1. See if you have the gene mutation.
2. Learn what you need to do if you do have it.

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

I totally understand that many doctors and practitioners just don't have the time or inclination to really delve into the complexities of the MTHFR gene, but what I really get annoyed about is them giving advice that is not based on research or sound clinical advice.

Likewise, people on websites that tell you not to worry, and that everything is fine – when it may not be. I get that they are trying to help, but they shouldn't be giving advice if they don't know what's going on with you.

I'm not saying that everyone with the MTHFR gene will have a problem conceiving, 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 having miscarriages or can't conceive. You know and I know everything is NOT fine.

## 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, 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 and 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.



<https://mthfrgenesupport.com/product-category/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:

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



## What genes can affect your fertility?

The key nutrient that we need for fertility and good DNA for the baby are the folate genes. So any variation in the folate genes that affect how we make active folate could affect our ability to fall pregnant. These genes are:

- Folate Receptor 2
- DHFR
- MTHFD1
- MTHFR



MTHFR is crucial because it is directly involved in creating your active folate for healthy DNA. So that is the gene we are going to concentrate on today.

## 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 is responsible for turning multiple processes within cells "on or off". Proper methylation (adding or removing methyl groups (CH<sub>3</sub>) 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.

**For a more in depth understanding of the importance of methyltransferases click [here](#).**

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.

It is important to know if you have a mutation in the MTHFR gene. Approximately 50-65% of the population has an MTHFR gene mutation. However, if you have a mutation, this DOES NOT automatically mean you will have a problem with your health.

# 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, 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 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

Diet and lifestyle is highly individual. What is right for you, may not be right for the person next to you. View more detailed diet and lifestyle guidelines for MTHFR gene mutations [here](#).

## MTHFR and pregnancy

Because of the lack of folate to help you make hormones and have healthy DNA many people with the MTHFR genetic variant end up having issues falling pregnant and staying pregnant.

This may mean the following:

- hormonal issues from puberty.
- reaction to the oral contraceptive pill
- Family history of estrogen dominance ie: fibrocystic ovaries or breasts, mood swings before your period, irregular menstrual cycle, food cravings, thyroid dysfunction, heavy periods, painful periods, endometriosis, fibroids
- family history of miscarriage
- Family history of heart disease, heart attack
- Trying to fall pregnant but can't
- Falling pregnant but cant keep the pregnancy
- Failed IVF

If you tick yes for any of these, then its essential that you get tested for the MTHFR gene. Here is our quick link to the test kit.

For Australia click [here](#)

For US click [here](#)

## Could MTHFR Be The Reason For Your Pregnancy Complications?

While the list of disorders associated with methylation issues and a lack of methyl folate might be a long one, the one that I feel is crucially important to address is fertility.

I believe if we can optimize folate levels and ensure that anyone who has an MTHFR gene mutation is taking the right form of folate, then we can prevent infertility issues and stop many miscarriages. In my practice many couples come to see me because they have been told they have unexplained infertility and are sent to do IVF. This is where they often discover they have the MTHFR gene mutation and are then sent to me.

I can tell you many of the couples we prepare for IVF actually never have to go because they fall pregnant naturally. So this is important to remember. You can often fall pregnant as long as you are taking the right form and the right amount of folate.

Below I have broken down the latest research on the three most common pregnancy-related issues associated with MTHFR genes, and explain just how these important genetic mutations could be playing a role.

## 1. Recurrent Pregnancy Loss (RPL)

In the world of MTHFR and fertility, it unfortunately is too common to hear many stories of women dealing with recurrent miscarriages.

Homocysteine is an amino acid (small protein) that in excessive amounts can cause blood clots. And while the jury is still out, some scientists have put forward the hypothesis that these blood clots may form close to the placenta, causing a block in the flow of nutrients to the fetus in its early stages of growth.

There are several key genetic mutations that have been associated with an increased risk of clot formation, with the MTHFR genes being some of them. And while studies tend to examine these genes in isolation, one study looked at the effect of these genes in unison and found the cumulative effects of multiple genetic mutations (such as Factor V Leiden and MTHFR) are indeed risk factors for RPL.

This study also found that women suffering from RPL were more likely to carry a homozygous mutation, that is, two copies of a genetic mutation as opposed to one copy, or no mutated copies.

The good news is more and more doctors, specialists, and health practitioners are starting to test women who have either a family history of thrombophilia or who are experiencing RPL for these clotting related genes.

The normal recommendations within the medical field to support these genes is to prescribe folate, B vitamins and anti-clotting medicines like heparin or baby aspirin.



## 2. Preeclampsia

Both variants of the MTHFR gene, A1298C and C677T, have been implicated in preeclampsia, with different studies yielding different results. Preeclampsia is a dangerous high blood pressure in pregnancy.

**Research** has found mutations to the MTHFR A1298C gene to be significant risk factors for several vascular-related pregnancy complications. In this paper, researchers found significantly higher plasma homocysteine levels and increased risk of premature separation of the placenta from the wall of the uterus, smaller fetuses, recurrent pregnancy loss, and preeclampsia among pregnant women with MTHFR A1298C mutations.

**Another study** found women with preeclampsia in their first pregnancy are also more likely to have MTHFR A1298C mutations, higher homocysteine levels, and babies with lower birth weights.

However, the MTHFR C677T variant has also been implicated in preeclampsia, with women suffering from preeclampsia more likely to be carrying a homozygous MTHFR C677T mutation (meaning two copies of the mutated gene).

So while these studies have examined different MTHFR genes and aspects of pregnancy health, both C677T and A1298C variants have been shown to play roles in complications during pregnancy.



### 3. Fetal Developmental Disorders

The foundational reason MTHFR mutations require our acknowledgment and support, whether pregnant or not, is due to their vital role in creating active folate, which we need for cell division and healthy DNA production. Why is MTHFR important in preconception and pregnancy? **Read more about that here.**

Having healthy levels of DNA during pregnancy has **clearly been linked with the prevention of developmental problems** including neural tube defects such as spina bifida and anencephaly.

And while these links are common knowledge within the medical sector, research now also shows:

- The presence of the maternal homozygous MTHFR C677T variant linked with the development of **childhood asthma.**
- An association between MTHFR and **autism spectrum disorders.**
- MTHFR C677T and A1298C linked with orofacial clefts in **Iranian children** and congenital heart disease in **Chinese populations.**



## So what form of folate is best for people with MTHFR mutations?

**Folate**, which is also known as vitamin B9, is required by the cell to ensure efficient nucleic acid synthesis and amino acid metabolism. It should not be confused with folic acid, which is a form of folate. However, folic acid is synthetic and cannot be converted into active folate unless the body has the capacity to do this.

The major process involved in this conversion is a methylation reaction. In some individuals, however, methylation might be affected by various biological and chemical factors. Mutations in the MTHFR gene, which are a major factor influencing methylation, can play a role in low folate levels.

Low levels of folate can result in numerous metabolic defects, such as high levels of homocysteine, amino acid imbalances, and reduced formation of blood cells and platelets. Folic acid, which is introduced into the cells through the consumption of fortified foods and supplements has to be converted into an active folate and this is where your genes may prove to be an issue.

If you have the MTHFR gene mutation then your ability to do this is reduced and you may have a build up of folic acid, which has been speculated to have negative health impacts.



Dietary folate, however, is necessary for the regulation of good health, typically, through consuming dark leafy greens, such as spinach, kale, turnip greens, and romaine lettuce. The consumption of these foods provides an immediate increase in folate levels. Contrary to folic acid, folate found naturally in foods helps reduce the risk of many forms of cancer.

Sufficient blood folate levels also assist in reducing the likelihood of suffering from depression. Naturally occurring folate is better able to be used by the body and does not build up or hide a B12 deficiency like folic acid.

Vitamin B12 is needed to aid in the recycling of homocysteine through conversion to methionine. More importantly though it is required for the uptake of folate into the important methionine cycle so the body can use it for key metabolic processes.

## These processes include:

**1. Creating healthy DNA.** This is the most important reason why knowing your MTHFR gene result is important. Active folate is directly involved in the synthesis of new DNA and while we have a constant demand for the production of new and healthy DNA, you can imagine that demand for this hugely increases during pregnancy to support the growing fetus.

**2. Preventing levels of a substance in the body called homocysteine from climbing too high.** Homocysteine has been connected in the research with blood clots, preeclampsia (high blood pressure in pregnancy that can be fatal to the baby and mother), which can be related to blood clots and increasing risk of blot clot formation during pregnancy.

**3. Creating molecules called 'methyl groups',** which act as instruction manuals for your DNA and cells, telling them the correct way to 'behave', so they do not do anything unwanted (e.g. cause disease or dysfunction within the body). We need healthy levels of these methyl groups to methylate/instruct your DNA, and without it cells are uncontrolled and can start to cause problems.

**4. Forming red blood cells, white blood cells, and platelets,** which are all vital for both the health of the mother during pregnancy and for the health of the child during pregnancy and after birth as they begin to rapidly grow and come into contact with bacteria and pathogens to strengthen their immune system.

As you can see, supporting your MTHFR enzyme during your preconception phase is the best way to support both your body once you fall pregnant, and the growth and development of your new baby.

**That's why you need to test to see if you have the gene mutation.**

## When To Supplement With Folate And What to Supplement With

Folate supplementation should start long before pregnancy begins.

Ensuring proper folate levels during preconception preparation is something everyone hoping to become pregnant should do. It takes time for the body to get the folate cycle back up and running efficiently (at least a couple weeks).

If there is a problem affecting the folate cycle, such as an MTHFR mutation, it may take longer to normalize. Folate supplementation should begin during preconception and continue throughout the entire pregnancy.

You want to prevent your body from having low folate levels long before you reach the periconceptual period of your pregnancy. During the first 3-4 weeks of pregnancy (periconceptual period), **low folate levels play a role in the development of neural tube defects (NTD's) and many other pregnancy complications.**



## Folic Acid (Not Recommended)

The daily recommended intake of folic acid is 400 mcg. Common multivitamins will give you around 400 mcg of folic acid.

Folic acid is a synthetic compound that can be easily stored, transported and has a long shelf life. Other than being less expensive than other forms of folate supplementation, folic acid comes up short for the following reasons:

- 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)

If you have or think you have an MTHFR mutation, folic acid supplementation can create even more problems for your **folate cycle**.



## Folinic Acid (Recommended)

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, making it ideal for people who have DHFR mutations, or people who are exposing themselves to high levels of folic acid by consuming folic acid fortified foods.

Folinic acid directly contributes to physiological processes, unlike folic acid which has no direct physiological function. Folinic acid aids the synthesis of DNA by acting as a cofactor in the metabolic reactions responsible for creating purine and pyrimidine (two of the four building blocks of DNA).

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 body's active form of folate, 5-MTHF. Below is a short recap of the problems linked to low folate status:

- Neural tube defects
- Lower sperm quality
- Blood clotting disorders (thrombophilia)
- Autism
- ADD/ADHD
- Allergies
- Low birth rate



Keep an eye on your folate status if you are trying to become pregnant. During pregnancy, there are high volumes of DNA being produced every day. The child is growing rapidly and has the highest risk of being negatively affected by low folate levels.

The body is a highly complex system, and when you change your diet, your environment, or start supplementing to improve your folate status, it takes a couple months for your body to adjust to healthy folate levels.

## 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.

**Read more on the uses of active folate in the body on our 'What is MTHFR' page.**

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 micograms 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**

# Top Tips For Eliminating Toxins From Your diet and Your Environment

Tips for avoiding and eliminating endocrine disruptors:

1. Eat fresh foods instead of processed, and buy food items sold in bulk over packaged. This will vastly reduce the level of phthalates and BPA the body is exposed to via food packaging.
2. Buy foods and beverages packaged in glass wherever possible.
3. Eat organic wherever possible, reducing your exposure to endocrine disruptors in pesticides. Refer to the **Environmental Working Group's list of the Dirty Dozen (best bought organic) and Clean Fifteen (OK to consume in conventional form)**.
4. Filter all water for cooking or drinking in the household to remove any pesticides, herbicides or hormonal and pharmaceutical residues present in the water supply. A good quality water filter will also remove chlorine, fluoride, microbes and impurities.
5. Avoid all products that use synthetic fragrances e.g. all fragrance personal care products such as perfumes, moisturizers, and body washes, air fresheners, room sprays, car air fresheners, deodorizers and household cleaning products.
6. Avoid using dryer sheets and fabric softener.
7. Use a low chemical, eco-friendly dry cleaner over a commercial one.
8. If you are exposed to a high level of retardants and formaldehydes in the home or car (if they are new or renovated), consider a vacuum cleaner with a HEPA filter, avoid reupholstering old furniture, and allow time for the house to 'settle' if replacing flooring before re-entering. Keep doors and windows open to allow these chemicals to leave the house as soon as possible.
9. Replace all nonstick cookware with eco-friendly options.
10. Never use plastic containers to store hot foods, or food at all if possible. BPA and phthalates easily leach into food when exposed to heat, making heating foods in the microwave in plastic highly unsafe.

11. Convert storage containers over to glass or stainless steel. These containers with plastic lids for ease of transport are still ok.

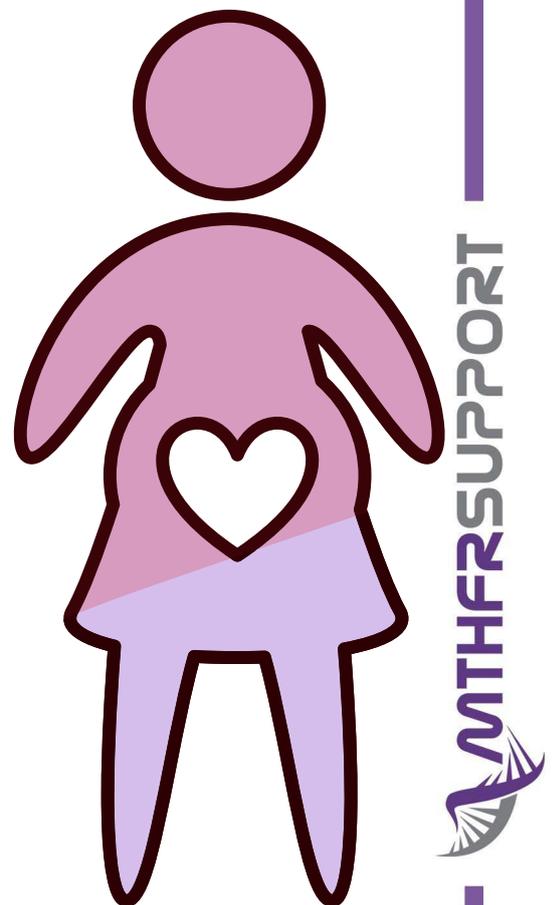
12. Avoid the use of plastic wrap and baking paper where possible.

13. Eliminate all synthetic and chemical based skin care, personal care and makeup from the household. Convert all products over to natural and organic alternatives. With between 60% – 90% of what is put on the skin absorbed into the body, if you wouldn't drink your liquid foundation, why put it on your skin?

14. Convert all household cleaning products over to natural based alternatives.

15. Convert all drink bottles over to stainless steel or glass versions. 'BPA free' bottles and containers still contain xenoestrogens (fake estrogens) and endocrine disruptors, and will begin leaching into the water as they age.

16. Avoid toys high in plastic around the home, choosing more eco-friendly and sustainable materials, such as wood.



## What are your next steps:

1. Take my **FREE 10 day** fertility course
2. Listen to my **FREE Webinar** coming soon
3. Do the MTHFR gene test.

For Australia click [here](#)

For US click [here](#)

4. visit our site to learn more about MTHFR

[www.mthfrsupport.com.au](http://www.mthfrsupport.com.au)

[www.mthfrfertility.com](http://www.mthfrfertility.com)

I wish you all the best in your fertility journey.

## Resources

### Articles referenced in this eBook include:

1. *What is MTHFR?*
2. *How the MTHFR gene mutation affects fertility and pregnancy?*
3. *5 common MTHFR symptoms and how to manage your gene mutation*
4. *Could MTHFR be the reason for your pregnancy complications?*
5. *Taking folic acid when you have an MTHFR mutation | Why it is not recommended?*
6. *Folate supplementation guide*
7. *Top 20 folate containing foods*
8. *Endocrine disruptors, health & fertility: How they're affecting you*

