# What You Need to Know About the MTHFR Gene

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## What Is the MTHFR Gene?



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Having two copies of the MTHFR gene mutation may raise your risk for certain health conditions, though more research is needed. You may have the mutation and not find out unless you undergo genetic testing or have a health issue.

You may have seen the abbreviation *MTHFR* pop up in recent health news. It refers to a relatively common genetic mutation.

*MTHFR* stands for methylenetetrahydrofolate reductase. It's getting attention due to a genetic mutation that may lead to high levels of homocysteine in the blood and low levels of folate and other vitamins.

Its main function is to provide your body with instructions in creating the *MTHFR* protein, which helps your body produce folate. This B vitamin is necessary to create DNA.

There's been concern that certain health issues are associated with *MTHFR* mutations, so testing has become more mainstream over the years.

You can have either one or two mutations — or neither — on the *MTHFR* gene. These mutations are often called variants. A variant is a part of a gene's DNA that's commonly different, or varies, from person to person.

Having one variant (heterozygous) is less likely to contribute to health issues. Some experts believe that having two mutations (homozygous) may lead to more serious problems.

There are two variants, or forms, of mutations that can occur on the *MTHFR* gene. The specific variants are:

- **C677T:** About 30 to 40 percent of the American population may have a mutation at gene position C677T. Roughly 25 percent of people of Hispanic descent and 10 to 15 percent of Caucasian descent are homozygous for this variant.
- **A1298C:** There's limited research regarding this variant. However, a 2015 studyTrusted Source found that A1298C is found in 7 to 14 percent in North American, European, and Australian populations. In comparison, the variant is far less common in Hispanic and Asian populations.

It's also possible to acquire both C677T and A1298C mutations, which is one copy of each.

Gene mutations are inherited, which means you acquire them from your parents. At conception, you receive one copy of the *MTHFR* gene from each parent. If both have mutations, your risk of having a homozygous mutation is higher.

However, in cases where a person holds two copies of the C677T mutation or one copy of each mutation, they may be at risk of high homocysteine (an amino acid) levels, which may affect the body's function.

Symptoms vary from person to person and from variant to variant. If you do a quick internet search, you'll likely find many websites claiming *MTHFR* directly causes a number of conditions.

Keep in mind that research around *MTHFR* and its effects is still evolving. Evidence linking most of these health conditions to *MTHFR* is currently lacking or has been disproven.

More than likely, unless you have problems or have testing done, you'll never become aware of your *MTHFR* mutation status.

## Health conditions and MTHFR mutation

It's worth noting that the health conditions linked to *MTHFR* depend on your type of mutation and how many copies you contain.

In cases where you have only one copy of the C677T or A1298C mutation, or two copies of the A1298C mutation, there are typically no health conditions or risks linked.

Conditions that have been proposed to be associated with *MTHFR* include:

- cardiovascular and thromboembolic diseases (specifically blood clots, stroke, embolism, and heart attacks)
- depression
- anxiety
- bipolar disorder
- schizophrenia
- colon cancer
- acute leukemia
- chronic pain and fatigue
- nerve pain
- migraine
- recurrent miscarriages in women of child-bearing age
- pregnancies with neural tube defects, like spina bifida and anencephaly

Learn more about having a successful pregnancy with *MTHFR*.

The risk is possibly increased if a person has two gene variants or is homozygous for the *MTHFR* mutation.

Various health organizations — including the American College of Obstetricians and Gynecologists, College of American Pathologists, American College of Medical Genetics, and American Heart Association — don't recommend testing for variants unless a person also has very high homocysteine levels or other health indications.

Still, you might be curious to discover your individual *MTHFR* status. Consider visiting your doctor and discussing the pros and cons of being tested.

Keep in mind that genetic testing may not be covered by your insurance. Call your insurance provider if you're considering getting tested to ask about costs.

At-home genetic testing kits offer screening for MTHFR as well. The customer's DNA is collected from the inside of the cheek with a sterile cotton swab. The test screens for two of the most common MTHFR mutations. Results are provided 5-6 business days after your sample is received.

Having an *MTHFR* variant doesn't mean that you need medical treatment. It could just mean you need to take a vitamin B supplement.

## High homocysteine levels

Treatment is typically required when you have very high homocysteine levels, almost always above the level attributed to most *MTHFR* variants. Your doctor should rule out other possible causes of increased homocysteine, which can occur with or without *MTHFR* variants.

Other causes of high homocysteine include:

- hypothyroidism
- conditions like diabetes, high cholesterol, and high blood pressure
- obesity and inactivity
- certain medications, such as atorvastatin, fenofibrate, methotrexate, and nicotinic acid

From there, the treatment will depend on the cause and doesn't necessarily take into account *MTHFR*. The exception is when you've been diagnosed with all of the following conditions at the same time:

- high homocysteine levels
- a confirmed *MTHFR* mutation
- vitamin deficiencies in folate, choline, or vitamins B12, B6, or riboflavin

In these cases, your doctor may suggest supplementation to address deficiencies along with medications or treatments to address the specific health condition.

People with *MTHFR* mutations may also wish to take preventive measures to lower their homocysteine levels. One preventive measure is changing certain lifestyle choices, which may help without the use of medications. Examples include:

- stopping smoking, if you smoke
- getting enough exercise
- eating a healthy, balanced diet

Recurrent miscarriages and neural tube defects are potentially associated with *MTHFR*. The Genetic and Rare Diseases Information Center says studies suggest that women who have two C677T variants are at an increased risk of having a child with a neural tube defect.

A 2019 studyTrusted Source examined 246 women who had experienced recurrent miscarriages and held the C677T *MTHFR* gene mutation. While the mutation was linked to these miscarriages, results found that administering folic acid alongside low dose aspirin and enoxaparin was a helpful therapy in helping prevent recurrent miscarriages.

Speak with your doctor about testing if any of the following situations apply to you:

- You've experienced several unexplained miscarriages.
- You've had a child with a neural tube defect.
- You know that you have the *MTHFR* mutation, and you're pregnant.

Though there's little evidence to support it, some doctors suggest taking blood-thinning medications. Extra folate supplementation may also be recommended.

The *MTHFR* gene mutation inhibits the way the body processes folic acid and other important B vitamins. Changing the supplementation of this nutrient is a potential focus in countering its effects.

Folic acid is actually a human-made version of folate, a naturally occurring nutrient found in foods. Taking the bioavailable form of folate — methylated folate — may help your body absorb it more readily.

Most people are encouraged to take a multivitamin that contains at least 0.4 milligrams Trusted Sourceof folic acid each day.

Pregnant people are not encouraged to switch prenatal vitamins or care based on their *MTHFR* status alone. This means taking the standard dose of 0.6 milligramsTrusted Source of folic acid daily.

People with a history of neural tube defects should speak with their doctor for specific recommendations.

Multivitamins containing methylated folate include:

- Thorne Basic Nutrients 2/Day
- Smarty Pants Adult Complete
- Mama Bird Prenatal Vitamins

Speak with your doctor before changing vitamins and supplements. Some may interfere with other medications or treatments you're receiving.

Your doctor may also suggest prescription vitamins that contain folate versus folic acid. Depending on your insurance, the costs of these options may vary in comparison to over-the-counter varieties.

Eating foods rich in folate may help naturally support your levels of this important vitamin. However, supplementation may still be necessary.

People with known *MTHFR* mutations should discuss their nutrition plan in detail, along with any potential supplementation or medical treatments, with a healthcare professional.

Some good food choices may include:

- proteins like cooked beans, peas, and lentils
- veggies like spinach, asparagus, lettuce, beets, broccoli, corn, Brussels sprouts, and bok choy
- fruits like cantaloupe, honeydew, banana, raspberries, grapefruit, and strawberries
- juices like orange, canned pineapple, grapefruit, tomato, or other vegetable juice
- peanut butter
- sunflower seeds

People with *MTHFR* mutations may want to avoid foods that contain the synthetic form of folate, folic acid — though it's unclear whether that's necessary or beneficial.

Be sure to check labels, as this vitamin is added to many enriched grains, like pasta, cereals, breads, and commercially produced flours.

Learn more about the difference between folate and folic acid.

Your *MTHFR* status may or may not be affecting your health. More research is needed to determine the true impact, if any, associated with the variants.

Again, many respected health organizations don't recommend testing for this mutation, especially without other medical indications. Speak with your doctor about the benefits and risks of testing, as well as any other concerns you may have.

Continue to eat well, exercise, and practice other healthy lifestyle habits to support your overall well-being.

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## How we reviewed this article:

Our experts continually monitor the health and wellness space, and we update our articles when new information becomes available.

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