

First do no harm: A guide to choosing wisely in general practice

For patients – **MTHFR gene testing**



Is this information for you?

This information is for you if you:

- are seeking methylenetetrahydrofolate reductase (*MTHFR*) gene testing
- have already had an *MTHFR* gene test
- have an *MTHFR* gene variant and want to know more about it.

About *MTHFR* gene testing

About the *MTHFR* gene

- *MTHFR* genes are involved in how your body uses folate. There are variations in this gene that can be identified in a blood test.
- Although a variation can reduce the gene's effectiveness, this has very little to no impact on people's health.¹

MTHFR gene variations and medical conditions

- Folate is very important, before and during pregnancy, to prevent malformations in the baby. Variations of the *MTHFR* gene do not change our recommendation to take a folate supplement.
- Some people get an *MTHFR* test for other medical conditions, but even if the test shows that they have a variation, it does not change the treatment for their condition.
- Some people who have a medical condition may also have an *MTHFR* gene variation, but others with the same medical condition will not have the gene variation. Having the gene variation does not mean you will develop the condition.

Why GPs do not recommend *MTHFR* gene testing

- There is no scientific evidence that having an *MTHFR* test will have any effect on your health.
- In addition to you having to pay for the test, an *MTHFR* may generate further testing that may cause harm.

- Whether they have a *MTHFR* gene variation or not, people with similar physical or psychological conditions receive the same treatment.²
- Living a healthy lifestyle that includes a balanced diet and physical activity will be more beneficial than having this test.

Common questions about *MTHFR* gene testing

My naturopath has suggested that I ask my GP for a pathology form for this test so it can be bulk billed. Will my GP do this?

Legally, GPs can request that a test be bulk billed only if it is 'clinically relevant'. Because a variation in your *MTHFR* gene will not affect any treatment of any condition you might have, it cannot be bulk billed.

I've had some pregnancy losses, so I'd like to have the test to see if an *MTHFR* variation is the cause.

It's very distressing to have experienced pregnancy losses, and it is natural to want to know the cause of them, but unfortunately the test won't provide a reason for your losses.²

Alternatives

- Talk to your GP if you have ongoing concerns or are worried that you might have an *MTHFR* variation, and mention what you are specifically worried about (such as pregnancy loss or developing heart disease).
- Talk to your GP about healthy eating and fitting regular exercise into your routine.
- If you are worried that you are developing a particular condition, or that your symptoms are not being adequately investigated, speak with your GP.

More information

- The RACGP, [Appropriate diagnostic testing – Patient information](#)
- NSW Government and Centre for Genetics Education, [MTHFR gene testing](#)

If you still want to go ahead

You may have the test if you choose to. However, remember the test cannot be bulk billed, which means you will have to pay for it.

If you have already had the test

Discuss your results with your GP and tell them if you are:

- taking any medications or supplements
- pregnant or breastfeeding
- worried about the result, especially if you are pregnant or breastfeeding.

References

1. [The Royal College of Pathologists of Australasia. MTHFR genetic testing. New South Wales: RCPA, 2016](#) [Accessed 9 March 2022].
2. [Long S, Goldblatt J. MTHFR genetic testing: Controversy and clinical implications. Aust Fam Physician 2016;45\(4\):237–40](#) [Accessed 3 March 2022]

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