What the MTHRFR?!

This common and potentially dangerous genetic defect can affect your ability to metabolise nutrients. Tara Thorne explains how to find out if you have it.

IN 2003 the human genome was mapped – an incredible feat that now paves the way in nutrigenomics, which is the study of the interaction of nutrition and genes, especially in regard to disease prevention and treatment. This is such an exciting time because we can now take information from our DNA and translate this into individualised health treatment and optimisation plans. And one gene that's garnering a lot of attention is MTHFR.

MTHFR stands for (deep breath) methylenetetrahydrofolate reductase. It's a gene and also an enzyme: genes make enzymes and the MTHFR gene makes the MTHFR enzyme. Enzymes work in the body as catalysts – i.e. to stimulate a reaction. Enzymes require nutrient co-factors in order to work; for example, co-factors that enable the MTHFR enzyme to work include B12, B6, B2, and folate, nutrients primarily and most easily absorbed from animal-based protein. When you don't have enough co-factors, your MTHFR enzyme slows down - but something else that will slow this enzyme is a mutation known as a single-neucleotide-polymorphism or 'snp', for short.

MTHFR mutation

Mutations in the MTHFR gene are common: about 40 to 60 percent of us carry a mutation of some sort, and what type you have determines how well your MTHFR enzyme works. For example, if you have two copies of a variant of MTHFR, (one from each of your parents) you

> Enzymes are like locks, requiring nutrients to 'unlock' them so they can work – but if the enzyme is misshapen, the nutrient key won't fit.

might find yourself with a 75 percent reduction in MTHFR efficiency; but if you only have one copy of a variant of MTHFR that isn't mutated, you may only have a 30 percent reduction in MTHFR enzyme efficiency. So the type and number of copies of an MTHFR variant you have is important.

Why focus on MTHFR when there are 20,000 genes in the human genome? It has become a significant player in the world of nutrigenomics because it affects so many people. It all boils down to the role this enzyme plays in the body. MTHFR is a fundamental player in regulating a process called methylation. Methylation happens in every cell in the body at a billion times per second. Here's a snapshot of just some of the things methylation plays a role in: Eliminating toxins (detoxification); eliminating histamine (think: allergies of all kinds); gene regulation and expression; building and breaking down neurotransmitters (e.g. dopamine, norepinephrine, epinephrine, serotonin, melatonin); processing potentially harmful oestrogens; building immune cells (T-cells, natural killer cells); DNA production and repair; production of glutathione (the body's main antioxidant); production of energy; production of myelin (the protective coating on nerves); and building and maintaining all cell membranes.

The list of conditions associated with MTHFR is similarly extensive: Anxiety and depression; autism; addictions (smoking, drugs, alcohol); Down syndrome; miscarriages (particularly recurrent); schizophrenia; fibromyalgia; chronic fatigue syndrome; chemical sensitivity; Parkinson's; irritable bowel syndrome; pre-eclampsia; stroke; spina bifida; acute lymphoblastic leukaemia; vascular dementia; bipolar disorder; colorectal adenoma; idiopathic male infertility; blood clots; rectal cancer; meningioma; glioma; congenital heart defects; deficits in childhood cognitive development; gastric cancer; migraines with aura; low HDL; high homocysteine; post-menopausal breast cancer; atherosclerosis; oral clefts; type I diabetes; epilepsy; Alzheimer's; potential drug toxicities (methotrexate, anti-epileptics); cervical dysplasia; increased bone fracture risk in post-menopausal women; multiple sclerosis; hypertension; differentiated thyroid carcinoma; prostate cancer; heart attack; heart murmurs; asthma; and bladder cancer.

Getting tested

Getting a DNA test doesn't have to be scary. These tests can be empowering and an extremely helpful way to optimise your health. This information can also be the difference between why a treatment plan works for one person, but not for another. If you suspect you have an MTHFR mutation, get your DNA tested at 23andMe. It's best to work with a health professional that knows what they're doing when it comes to MTHFR, because treatment is complicated, especially when it comes to adding in supplements that can have a big impact on the way MTHFR functions.

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How can you optimise MTHFR?

Remember: your genes are not your fate. When it comes to genetic mutations, genes load the gun - but environment and lifestyle pull the trigger: although you might have an MTHFR mutation, this doesn't automatically mean it's expressed. However, MTHFR mutations are rising, because we reduce MTHFR enzyme function and turn on MTHFR mutation via our diet and lifestyle choices, and our environment. So even if you're not born with the mutation, it doesn't mean you're not susceptible. There is, however, a great deal you can do to support your MTHFR enzyme:

- Eat uncooked dark leafy greens daily, because these are the best source of folate – a key player in healthy MTHFR function
- Avoid synthetic folic acid at all costs; do not eat processed or "enriched" foods
- Employ stress reduction techniques; stress down-regulates the MTHFR enzyme
- Drink clean, filtered water and reduce chemical exposure as much as possible
- Amp up your detoxification efforts with dry skin brushing, rebounding, using saunas or hot yoga and drinking 2 litres of water daily
- Get adequate sleep and exercise
- Eat a whole food, nutrient-dense diet; eliminate all processed foods
- Reduce alcohol consumption and treat candida overgrowth - both alcohol and candida produce a substance called acetaldehyde, which severely impacts MTHFR function
- Manage thyroid function optimally
- Avoid medications, in particular antacids, metformin, statins and nitrous oxide