

WHAT THE REPORT INCLUDES

- Detailed EXPLANATION of the test performed and recommendations to be followed;
- SUMMARY TABLE showing the metabolic areas investigated and the results obtained from the DNA analysis, in order to have a quick overview of one's general situation and to check for compromised situations;
- BIBLIOGRAPHY providing scientific references for the test.

COLOURS USED

Green indicates that the variants identified in the analysis do not unfavourably alter the enzymatic activity of the proteins they encode and/or the risk associated with certain diseases.

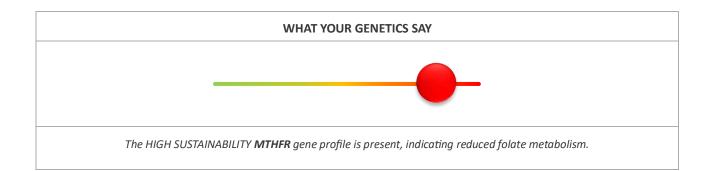
Orange indicates that the variants identified in the analysis slightly unfavourably alter enzyme activity and/or the risk associated with certain disorders or diseases.

Red indicates that the variants identified in the analysis alter enzyme activity in a particularly unfavourable way, resulting in an increased risk of developing certain disorders or associated diseases.

The results shown, as well as the considerations and explanations contained in the following pages of this booklet, should not be regarded as a medical diagnosis. It is important to bear in mind that genetic information is only a part of the total information needed to gain a complete picture of a person's state of health, and the data reported here is therefore a tool available to the treating physician to formulate a correct assessment of the patient's physiological state and suggest an appropriate personalised treatment.

Test result:

Gentras ID	Gene	Allelic variants	Genotype		Predisposition
GTS003	MTHFR	С			
		т	T	Т	HIGH



Explanation of genetic analysis:

The C677T variable gene site on the MTHFR gene was evaluated.

C677T is indicative of the body's ability to metabolise folate because it affects the functionality of a key enzyme in this metabolism, methylen tetrahydrofolate reductase (MTHFR), and is a measure of blood folic acid deficiency. MTHFR is an enzyme that affects the efficiency of folic acid metabolism.

What are Folates

Folate is a form of vitamin B9, a vitamin essential for many biological functions. They occur naturally in various foods, such as green leafy vegetables (spinach, broccoli, asparagus), pulses, fruits (oranges, avocados), and whole grains. They play a crucial role in DNA synthesis, cell division and growth, and red blood cell formation. For this reason, they are particularly important during pregnancy, as adequate intake can prevent neural tube defects in the foetus. Another form of vitamin B9 is folic acid, which is the synthetic version used in food supplements and some fortified foods.

Folate metabolism

Folate metabolism is the process by which our body uses folate (vitamin B9) to perform several essential functions:

- <u>Absorption:</u> When we eat folate-rich foods (such as green leafy vegetables), folates are absorbed in the small intestine.
- <u>Conversion:</u> Once absorbed, folates are converted into an active form called tetrahydrofolate (THF). This is the form that the body can use for its functions.
- <u>DNA synthesis:</u> THF is involved in the production of DNA, which is essential for cell division and growth.
- <u>Red blood cell formation:</u> It also serves to create red blood cells, which carry oxygen in the blood.
- <u>Amino acid metabolism:</u> THF helps transform certain amino acids, which are the building blocks of proteins.
- <u>Methylation:</u> Folate plays a role in a process called methylation, which is like 'switching on' or 'switching off' certain genes, contributing to the proper functioning of cells.

In the presence of low folate levels, problems such as anaemia (few healthy red blood cells) and, during pregnancy, foetal malformations can occur.

The MTHFR (methylenetetrahydrofolate reductase) variant is a genetic mutation that can affect folate metabolism. This mutation can reduce the efficiency with which the body converts folic acid (the synthetic form of vitamin B9) and dietary folate into 5-MTHF (5-methylenetetrahydrofolate), the active form used.

If a person has an unfavourable variant of the MTHFR gene, he or she may be at greater risk of problems such as elevated homocysteine, which is linked to an increased risk of cardiovascular disease, and may not metabolise folic acid well.

Countermeasures to be considered if the MTHFR variant is unfavourable

Here are the recommended countermeasures:

- <u>Supplementation with 5-MTHE</u>: Instead of taking folic acid, it is recommended to take 5-methyltetrahydrofolate (5-MTHF), which is already the active form of vitamin B9, directly. This bypasses the need for efficient conversion by the MTHFR gene.
- <u>Natural folate intake:</u> Consuming foods rich in natural folates, such as green leafy vegetables, pulses and citrus fruits, can be helpful. Folate found in natural foods tends to be better assimilated by the body than synthetic folic acid.
- <u>Monitor homocysteine levels:</u> People with an MTHFR variant may have elevated homocysteine levels in their blood. Monitoring these levels regularly with a doctor is important to manage cardiovascular risk.
- <u>B vitamins:</u> It is often helpful to take other B vitamins (B6 and B12), which work together with folate to lower homocysteine levels.
- <u>Healthy lifestyle:</u> Maintaining a balanced diet, regular physical activity and reducing stress can help improve cardiovascular health and offset any negative effects of the MTHFR variant.

Consulting a doctor or nutritionist is essential for a customised supplementation plan.

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