Genomic Results (Buccal Cells)

Companion order =U6030233

Patient:

DOB
Sex:
MRN

Order Number:
Reported: June 16, 2025
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MTHFR	*5,10-methyltetrahydrofolate reductase : METHYLATION*
Location: Chromosome 1 **C677T Your Genotype:** + ↓ | + ↓ **A1298C Your Genotype:** – | –	5,10-methylenetetrahydrofolate reductase (MTHFR) is a key enzyme in folate metabolism, facilitating the formation of methyltetrahydrofolate, a required cofactor in the remethylation of homocysteine (Hcy) to methionine.

Health Implications

- Homozygosity for 677 (+/+) results in 60-70% reduction in MTHFR enzyme activity, which can limit methylation reactions in the body
- Increased risk of high homocysteine, esp. when low levels of B vitamins, mainly folate; several studies also indicate a tendency for lower folate levels
- Most studies suggest increased risk of venous thrombosis, heart disease, hypertension, stroke and diabetic nephropathy; population differences may reflect the influence of B vitamin fortification, which lowers Hcy
- Several studies show moderately increased risks of depression and schizophrenia
- Most studies suggest increased risk of birth defects in the offspring, e.g., neural tube or congenital heart defects, cleft lip and/or palate, and Down syndrome; possible increased risk of recurrent pregnancy loss and male infertility in Asians
- Possible slight increased risk of fracture and/or low bone density; in some studies these associations depend on B vitamin status, while others show no associations
- Increased risk of gastric and esophageal cancer, which may be reversed with adequate folate intake; some studies show higher risk of breast, lung, and cervical cancer in Asians
- Decreased risk of colorectal cancer, but only when high folate status; decreased risk of acute lymphoblastic leukemia in children and Caucasians; and decreased risk of cervical cancer in Caucasians

Clinical Management Considerations

- Ensure adequate intake of dark-green leafy vegetables and other B vitamin-rich foods
- Consider supplementation with folic acid (or 5-methyltetrahydrofolate, which bypasses the MTHFR step), B2, B6 (pyridoxal 5-phosphate), B12 (or methylcobalamin), and betaine (trimethylglycine)
- Most studies suggest easier toxicity from chemotherapy

Key

– – Neither chromosome carries the genetic variation.
+ – One chromosome (of two) carries the genetic variation.
+ + Both chromosomes carry the genetic variation.
(You inherit one chromosome from each parent)

+ ↑ Gene activity increased
+ ↓ Gene activity decreased

This test has been developed and its performance characteristics determined by Genova Diagnostics, Inc. It has not been cleared by the U.S. Food and Drug Administration.

Commentary is provided to the practitioner for educational purposes, and should not be interpreted as diagnostic or treatment recommendations. Diagnosis and treatment decisions are the responsibility of the practitioner.

The accuracy of genetic testing is not 100%. Results of genetic tests should be taken in the context of clinical representation and familial risk. The prevalence and significance of some allelic variations may be population specific.

Any positive findings in your patient's test indicate genetic predisposition that could affect physiologic function and risk of disease. We do not measure every possible genetic variation. Your patient may have additional risk that is not measured by this test. Negative findings do not imply that your patient is risk-free.

DNA sequencing is used to detect polymorphisms in the patient's DNA sample. The sensitivity and specificity of this assay is <100%.