MTHFR Test Report

Patient and report summary

Patient name: **John Doe**
Patient date of birth: **1968-08-28**
Collection date: **2020-09-21**
Specimen ID: **2134850158760**
Specimen type: **Buccal swab**
Receive date: **2020-09-21**

Ordering provider: **Sample Doctor**
Ordering facility: **Healthcare Institution**
Product type: **MTHFR**
Report type: **Original**
OneOme report date: **2020-09-21**

Test results and interpretation

MTHFR

Normal activity (677 CC, 1298 AA)

Normal MTHFR activity. The conversion from folic acid to methylated folate (the active form of folate) is predicted to be normal, although other genetic and/or clinical factors may influence the folate cycle.

Variants Interrogated		Result
rs1801131	NM_005957.4:c.1286A>C	AA
rs1801133	NM_005957.4:c.665C>T	CC

Background information

The *MTHFR* gene, residing on the minus strand of chromosome 1, encodes the enzyme methylenetetrahydrofolate reductase. This enzyme is integrally involved in the DNA synthesis pathway, specifically the conversion of homocysteine to methionine through the methylation cycle of folic acid. Common variants in this gene, namely 677C>T (rs1801133) and 1298A>C (rs1801131), can disrupt this pathway, altering folic acid metabolism and/or leading to hyperhomocysteinemia. However, the American College of Medical Genetics and Genomics (ACMG) determined that *MTHFR* genotyping has minimal clinical utility as part of the routine evaluation for thrombophilia.

Methodology and limitations

Analytical results were produced using tests developed and validated by OneOme, LLC, a clinical laboratory located at 807 Broadway Street NE Suite 100, Minneapolis, MN 55413. These tests have not been cleared or approved by the U.S. Food and Drug Administration. OneOme is certified under CLIA-88 and accredited by the College of American Pathologists as qualified to perform high-complexity testing. This test is used for clinical purposes and should not be regarded as investigational or for research.

Genomic DNA was analyzed by PCR using Thermo Fisher TaqMan® and/or LGC Biosearch BHQ® probe-based methods to interrogate the variant locations listed in the table above.

The test does not detect all known and unknown variations in the gene(s) tested, nor does absence of a detectable variant (designated as *1 for genes encoding drug metabolizing enzymes) rule out the presence of other, non-detected variants.

As with other common SNP genotyping techniques, these assays cannot differentiate between the maternal and paternal chromosomes. In cases where observed variants are associated with more than one haplotype, OneOme infers and reports the most likely diplotype based on published allele frequency and/or ethnicity data. Inferences with potential clinical impact are reported in the *Report and laboratory comments* section.

The variant detection methods validated by OneOme provide >99.9% accuracy; however, PCR may be subject to general interference by factors such as reaction inhibitors and low quality or quantity of extracted DNA. When present, these interferents typically yield no result rather than an inaccurate one. Very infrequent variants or polymorphisms occurring in primer- or probe-binding regions may also affect testing and could produce an erroneous result or assay failure. Variant locations tested by the assay but not assigned a genotype call are reported as “No Call.” Test results and clinical interpretation may be inaccurate for individuals who have undergone or are receiving non-autologous blood transfusions, tissue, and/or organ transplant therapies. Although extremely rare, results could also be impacted by other factors not addressed above, such as laboratory error.

Due to the complexity of interpreting some genetic test results, such as those that may carry a probabilistic risk of disease, patients and providers should consider the benefits of consulting with a trained genetic counseling professional, physician, or pharmacogenomic specialist. For additional support, contact OneOme through the website or by calling 844-663-6635.

OneOme liability disclaimer

The interpretations and clinical annotations provided by OneOme are intended solely for use by a medical professional and do not constitute medical advice by OneOme. The treating provider remains ultimately responsible for all diagnosis and treatment decisions for the patient. OneOme disclaims liability for any errors, omissions or ambiguities in any translation or interpretation of a report by a third party, including without limitation direct, indirect, incidental, special, consequential or exemplary damages, whether such damages arise in contract, negligence, tort, under statute, in equity, at law or otherwise. Information included in this report is based upon scientific literature, including information from and guidelines published by professional associations (e.g., CPIC, FDA, DPWG), and does not take into account other genetic variants and environmental or social factors that may affect a patient's response. Other factors not included in this report include, but are not limited to, environmental factors (e.g., smoking), health factors (e.g., diet), social and familial factors, various medical conditions, and drug-to-drug interactions. Administration of any medication, including the ones listed in the OneOme reports, requires careful therapeutic monitoring regardless of the phenotype or genotype-predicted interaction reported. As a matter of practice, OneOme will routinely update its pharmacogenomic database as new information becomes available to the scientific community. Genotype-predicted interactions and annotations found on the patient's RightMed Comprehensive Test Report, Vantage Reports, or RightMed specialty reports are therefore dependent on the date of generation and/or the database version used to generate that report. Providers may access these reports with updated annotations using OneOme's latest released version through the provider portal at portal.oneome.com.