

MTHFR Informed Consent

1. **What is MTHFR?** MTHFR is an abbreviation for the enzyme called. "5, 10- methylenetetrahydrofolate reductase". This enzyme converts a substance in the blood called homocysteine into another substance, methionine. Certain mutations in the MTHFR gene cause this reaction to occur more slowly. The resulting increased homocysteine in the blood is a risk factor for arteriosclerotic vascular disease and deep vein thrombosis.

2. **What is the purpose of this test and what are its limitations?** Everyone has two copies of the MTHFR gene. An individual may have two normal copies, two abnormal copies, or one normal and one abnormal. This test detects the presence of two specific genetic changes (mutations) in the MTHFR gene. The purpose of this test is to determine if you have two, one, or no copies of either of two mutations in the MTHFR gene. If you have two mutations, your risk for arteriosclerotic vascular disease and deep vein thrombosis is increased. If you have no or only one mutation, your risk is not increased by the results of this test.

3. **What is required to perform this test?** You will be asked to provide 5 mL of blood, which is equal to about one tablespoon. DNA will be extracted from this blood sample and tested. The only discomfort that you may feel is the stick of the needle in your arm. You may also experience a small bruise at the site of the needle puncture. You will also be asked to provide information regarding your medical history, which is necessary for proper interpretation of your test result. In the unlikely event that you should be injured in the course of being tested, your physician will provide any necessary medical care. However, you would be expected to bear the cost of such medical care. Compensation will not be provided in the event of any injury.

4. **Is there a cost for this test?** This is a routine clinical laboratory test and the results may aid in your diagnosis; thus, you or your health insurer will be billed for this procedure.

5. **What will happen to the DNA once the test is complete?** If you consent by initialing below, your sample might be used for laboratory quality control purposes after identifiers are removed and will be stored for longer than 60 days. If you do not initial the box below, the sample will be destroyed within 60 days of collection and not used for any other purposes.

I consent to my DNA sample being used for laboratory quality control activities and storage of my DNA past 60 days.
Please initial.

6. **How will I obtain results from this test?** DNA testing and interpretation of results are complex. The information from this test will be provided in the form of a written report to your physician who will inform you of the results. The laboratory will not provide results directly to patients. Your physician may suggest genetic counseling prior to performing this test or if your results are abnormal. To the extent permitted by law, all of your laboratory records and results are confidential and shall not be disclosed without your written authorization.

Patient Attestation of Informed Consent:

My signature below indicates that I have received information about this test, **MTHFR**, and that I have read and understood the material in this document. I have been given a full opportunity to ask questions that I may have about the testing procedure and related issues. I agree to undergo this testing.

Patient Signature

Date

Signature of Parent/Guardian if Patient is a minor

Print Name of Parent/Guardian

For the Physician:

As the referring physician, I understand the benefits and limitations of this study and have requested that the above-named patient be tested. I attest to the fact that I have provided the patient with the information contained above and fully answered any questions. I believe that the patient understands the information and is voluntarily signing this informed consent.

Signature of Physician/Health Care Professional

Print Name of Physician/Health Care Professional