

**AUTHORIZATION FOR THE  
MOLECULAR TEST FOR  
MTHFR 677C>T**

Patient Name: \_\_\_\_\_ MR#: \_\_\_\_\_

Account #: \_\_\_\_\_ DOB: \_\_\_\_\_ Date: \_\_\_\_\_

**MTHFR 677C>T** Methyl tetrahydrofolate reductase (MTHFR) is an enzyme made by our bodies under the direction of the MTHFR gene. The MTHFR enzyme is part of a metabolic pathway the body uses to make proteins and other important compounds. A change or mutation in the *MTHFR* gene can lead to the body making MTHFR enzyme that does not work like it should. One such genetic change is called MTHFR 677C>T. The MTHFR 677C>T gene change (and its effect on MTHFR enzyme function) may result in a condition called hyperhomocysteinemia when blood folate levels are low. Hyperhomocysteinemia involves build-up of homocysteine in the blood and is treatable by supplementing the diet with folic acid. Everyone has two copies of the MTHFR gene. An individual may have two normal copies, two abnormal copies, or one normal and one abnormal. **Only individuals with two abnormal copies of 677C>T have been found to be at increased risk of hyperhomocysteinemia.** Two abnormal copies of 677C>T have been found in about 10% of the general population. In addition, some studies have found two abnormal copies of 677C>T at higher frequency than is seen in the general population in individuals with the following disorders: thrombosis, coronary heart disease, inflammatory bowel disease, schizophrenia, pre-eclampsia, colon cancer, recurrent fetal loss, having a child with a neural tube defect or IUGR. The precise relationship between the 677C>T mutation and these disorders is unclear.

**Molecular Test for MTHFR 677C>T** You will be required to donate 10 mL of blood, which is equal to about two teaspoons. In addition, you may be asked to provide information regarding your medical history. A correct history is critical for proper interpretation of the data. This is a routine clinical laboratory test so you or your health insurer will be billed for the procedure

**Significance of the Results** The significance of the results of this test will depend on whether 677C>T mutations are identified as well as what other risk factors or symptoms are also present. For example, the finding of two mutations in an individual with symptoms is suggestive that the 677C>T mutations may be associated with a clinical disorder. **To understand your results, you should consult your physician and may wish to consider further independent testing or pursue genetic counseling.**

**Limitations** The MTHFR 677C>T mutation is the only genetic variation that will be tested for. If the 677C>T mutation is not found by the testing procedure, it does not mean that the risk of carrying or developing the above disorders is no longer present. It simply means that this particular mutation has not been found although other mutations could be present in this or other genes. It is also possible that you may be predisposed to have one of the above disorders due to non-hereditary causes, which would also not be detected by this test.

**Results from the test** The test result will be provided to health care professionals directly involved in your care. Genetic counseling may also be appropriate as follow up. To the extent permitted by law, all of the records, findings, and results of this test are confidential and shall not be disclosed without your written consent specifically authorizing to whom such records, findings, and results are to be released. In accordance with NYS law, any remaining DNA will be discarded unless it is retained as a laboratory control, in which case all information identifying the DNA to you will be removed.

**Key:** MTHFR = Methylene tetrahydrofolate reductase, IUGR = Intrauterine growth restriction

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Patient's Name: \_\_\_\_\_ Account #: \_\_\_\_\_ MR#: \_\_\_\_\_

I have read this entire document and understand its contents. In addition, I understand that I am free to withdraw any portion of my consent by crossing off and initialing unacceptable statements. Please note that crossing off the laboratory's option to bill for the testing will result in the testing being cancelled.

If you have any questions about the test to be performed, you may also wish to obtain genetic counseling prior to signing this form. You may also contact the Molecular Diagnostics Laboratory at (315) 464-6806.

Patient's Name (printed): \_\_\_\_\_ Patient's Medical Record Number: \_\_\_\_\_  
(for office use only)

**For the Patient:**

Please print the name, phone number, and address (if known) of all health care professionals, physicians (other than the referring physician), or other individuals/organizations (such as a health insurer) to whom you authorize the release of the MTHFR test result. (Medical results cannot be sent to a patient, a patient's family member or guardian.) Please print legibly.

Name and Title	Address	Phone Number
_____	_____	_____
_____	_____	_____
_____	_____	_____

My signature below indicates that the above information has been explained to me and that I give consent for this MTHFR testing. **I hereby authorize SUNY Upstate Medical University to furnish my designated insurance carrier the information on this form if necessary for reimbursement. I also authorize benefits to be payable to SUNY Upstate Medical University. I understand that I am responsible for any amount not paid by insurance for reasons including, but not limited to, non-covered and non-authorized services. I permit a copy of this authorization to be used in place of the original.**

Date: \_\_\_\_\_ Signature of Patient: \_\_\_\_\_

Name of Parent/Guardian: \_\_\_\_\_

Signature of Parent/Guardian if patient is a minor: \_\_\_\_\_

*As referring physician/health care professional, I understand the benefits and limitations of this clinical assay. I hereby attest to the fact that I have provided the patient or patient's guardian with the information contained above in compliance with the NYS Civil Rights Act, Section 79-l, have answered any questions fully, and have obtained a signed informed consent as appropriate.*

*I request that the above indicated genetic test be performed.*

Signature of Physician/Health Care Professional: \_\_\_\_\_

Printed Name/Stamp/Title: \_\_\_\_\_ Date/Time: \_\_\_\_\_