UPSTATE
UNIVERSITY HOSPITAL

AUTHORIZATION FOR MTHFR 677C>T GENETIC TESTING

Patient Name: _____ MR#: _____

Account #: _____ DOB: _____ Date: _____

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 as it should. One such genetic change is called *MTHFR* 677C>T. The MTHFR 677C>T variant 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 copies of 677C>T have been found to be at increased risk of hyperhomocysteinemia**. Two 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 intrauterine growth restriction (IUGR). The precise relationship between the 677C>T variant and these disorders is unclear.

Molecular Test for *MTHFR* 677C>T You will be required to donate 10 mL of blood, which is 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 variants 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 MTHFR 677C>T is the only genetic variation that will be tested for. If the 677C>T variant is not found by the testing procedure, it reduces but does not eliminate the risk of carrying or developing the disorder. It means that this particular variant has not been found although other mutations may be present in this or other genes. It is also possible that you may be predisposed to having the disorder due to non-hereditary causes, which would 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 at the end of testing or not more than sixty (60) days after the sample was taken.

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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: _____