

SUNY Upstate Medical University
Authorization for the Molecular Test for MTHFR 677C→T

1. **What is MTHFR 677C→T?** Various disorders including 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 can be caused by different factors including inheriting genetic risk factors. It has been suggested that one of these inherited risk factors may be a mutation in the MTHFR gene, called 677C→T. The usual function of the MTHFR gene product is to methylate homocysteine to methionine. This variant, which shows about half the usual activity, is associated with hyperhomocysteinemia (increased homocysteine plasma levels) when folate status is low. This condition is treatable with folic acid supplementation of the diet.
2. **What is the purpose of the 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 is designed to detect the presence of a specific mutation, or change, called 677C→T, which has been shown to predispose people to hyperhomocysteinemia. Two abnormal copies of 677C→T have been found in about 5-15% of the general population, and at higher frequencies in some studies on individuals with the above conditions. If this 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 hyperhomocysteinemia, due to non-hereditary causes, which would also not be detected by this test. If this mutation is found by the testing procedure, there is an indication that you may be predisposed to hyperhomocysteinemia. The significance of the results of this test will depend on what other risk factors or symptoms are also present. For example, the finding of two mutations in an individual with symptoms is suggestive but the finding of only one mutation in an individual without symptoms is probably not significant. **A positive result by itself should not be used as the sole criteria for diagnosis.** You should consult your physician and may wish to consider further independent testing or pursue genetic counseling.
3. **What is required to perform the test?** 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.
4. **Is there a cost for the test?** This is a routine clinical laboratory test and the results from it may aid in diagnosis, so you or your health insurer will be billed for the procedure.
5. **What will happen to the DNA once the test is complete?** The only testing that will be performed on this sample is to test for MTHFR genetic variation. The residual DNA may be kept indefinitely (this does not constitute DNA banking) to be used as a laboratory control, in which case all identifying information will be removed.
6. **How will I obtain results from the test?** The test result will be provided to your physician who will discuss it with you. 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.

If you have questions about the test that will be performed, you may 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, address (if known) and phone number 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 of licensed physician	Address (if known)	Phone Number
_____	_____	_____
_____	_____	_____
_____	_____	_____
_____	_____	_____

My signature below indicates that the above information has been explained to me and that I give consent for this MTHFR testing.

Date: _____

Signature of Patient

Name of Parent/Guardian

Signature of Parent/Guardian if patient is a minor

For the Physician:

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 genetic test indicated above be performed.

Printed name of Physician/Health Care Professional

Signature of Physician/ Health Care Professional