

SUNY Upstate Medical University
Authorization for Genetic Test for Hereditary Hemochromatosis

1. **What is hereditary hemochromatosis?** Hereditary hemochromatosis (HH) is an inherited disease that results from the accumulation of iron in organs and eventually, if untreated, in serious illness (organ failure). If diagnosed early enough it can be treated.
2. **What is the purpose of the test and what are its limitations?** Everyone has two copies of the hemochromatosis gene (HFE). An individual may have two normal copies, two abnormal copies or one normal and one abnormal copy. Two abnormal copies have been found in the majority but not all individuals with HH.

This test will detect the presence of two specific mutations, or changes, (C282Y or H63D) associated with a diagnosis of HH. If these mutations are not found, it does not rule out the possibility that other mutations could be present. Furthermore, the absence of these two mutations does not mean that the risk of carrying or developing HH is not present. It is also possible that you may have secondary hemochromatosis, due to non-hereditary causes, which would not be detected by this test.

If these mutations are found by the testing procedure, you may be predisposed to or may have hereditary hemochromatosis. The significance of the results of this test will depend on which mutations are found and what other risk factors or symptoms are also present. For example, the finding of two mutations in an individual with symptoms is highly suggestive of HH, but the finding of only one mutation in an individual without symptoms is of uncertain significance. A positive result by itself should not be used as the sole criteria for diagnosis. Rare (less than 1% of the time) errors may occur, for example due to sample mix-ups, or due to technical errors such as rare genetic variants that mimic or mask mutations being tested. 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?** A 10 ml sample of blood, approximately two teaspoons, is necessary. 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 the test for HH. Residual DNA may be stored 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 any 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, phone number, and address (if known) of all health care professionals, physicians (in addition to the referring physician), or other individuals/organizations (such as a health insurer) to whom you authorize the release of the HH 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 hereditary hemochromatosis testing.

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 genetic test indicated above be performed.

Printed name of Physician/ Health Care Professional

Signature of Physician/Health Care Professional