

SUNY Upstate Medical University
Authorization for the Molecular Test for Sickle Cell Disease (HbS & HbC)

1. What is sickle cell disease? Sickle cell disease is a disorder involving the blood that is characterized by a tendency of the red blood cells to become abnormal in shape (“sickled”) when exposed to low oxygen levels. Affected individuals have two abnormal copies of a gene called “beta-globin” and are usually diagnosed early in life with such symptoms as anemia, failure to thrive, an enlarged spleen, repeated infections, and painful swellings of the hands or feet. In the United States, the disease is most commonly observed among African-Americans. Approximately 1 in 12 African Americans have only one abnormal copy of the beta-globin gene and have sickle cell trait. These individuals generally have no adverse symptoms themselves, but are at increased risk of having a child with sickle cell disease or sickle cell trait.

2. What is the purpose of the test and what are its limitations? The most common way to test for sickle cell disease and sickle cell trait is to use a blood sample and analyze the hemoglobin proteins present. This is done routinely to diagnose adults and children. In circumstances when a blood sample is not available, molecular testing can be performed. If parents are at risk of having a child affected with sickle cell disease, prenatal testing may be performed using a molecular analysis of amniotic fluid cells. Molecular analysis is preferable for prenatal testing as the risk to the fetus of amniocentesis is less than the risk of PUBS. Molecular analysis for sickle cell disease is also done in the newborn period. Although blood can be obtained from newborns, these individuals normally have a high level of fetal protein in their blood which makes the protein based blood test ineffective.

The molecular test for sickle cell disease performed at University Hospital detects only 2 variants of the beta-globin gene (beta-globin S and beta-globin C). In some cases, the presence of other globin gene variants (which will not be tested) may result in the same clinical symptoms as sickle cell disease. 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 the mutation being tested.

3. What is required to perform the test? In newborns, a 2 ml blood sample is necessary. For prenatal diagnosis, 10-20 ml (2-4 teaspoons) of amniotic fluid collected by amniocentesis is necessary. To precisely interpret a prenatal diagnosis, the genotypes of both parents must be known or parental blood samples must be submitted for analysis with the prenatal sample. If parental genotypes are not determined, a less precise estimate of the risk of CF in the fetus will be provided. 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 sickle cell disease. 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 sickle cell disease 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 sickle cell 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