



## ❖ TESTING FOR GENETIC MARKERS FOR INHERITED DISORDERS

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Due to the increasing availability of testing to identify genetic markers for inherited disorders, Allina Medical Laboratories has developed a system to manage requests for this testing in a manner that will ensure appropriate coordination of the various issues raised by these requests.

Some testing for relatively common disorders, such as carrier risk for Cystic Fibrosis in pregnant females, is regarded as routine and is covered by guidelines and reference materials from major obstetrical societies. This testing is readily available from most major reference laboratories, along with background statistics regarding ethnic frequencies and variety of markers/mutations tested.

Testing for less common disorders with complex management issues can cause problems if the test results are received before a plan has been established, or the potential issues have been reviewed before initiation of the testing. Knowledge that a disorder exists on a genetic basis may help identify the cause for a clinical disorder, but may offer little in the subsequent patient management. The demonstration of an inherited disorder in a family member raises a variety of issues for the rest of the members of the family, including insurability and questions regarding the Subsequent testing of family members. These issues should be considered prior to initial testing of the potential patient/individual.

Insurance reimbursement for these less frequently requested disorders can vary depending on the insurance carrier involved. Testing cannot be assumed to be covered by the carrier unless prior authorization has been obtained. These tests can be expensive and some are only performed in research settings and are not readily available from standard reference lab sources. These research laboratories may not be willing to bill insurance, and payment guarantee may be required prior to testing. If the patient desires insurance billing, prior authorization from the carrier must be obtained. When billing issues have not been completely clarified, or the insurance carrier has refused to cover all or part of the cost of testing, the patient will be asked to remit payment for any residual costs.

Turnaround times for this specialized testing can be weeks to months, and results, when received, can be complicated and difficult to interpret.

The complex issues raised by this type of testing makes it vital to address them prior to initiation of testing. Genetic counseling prior to the collection of the test sample can be invaluable, since it raises these issues and gives some insight into the likelihood of a disorder in a particular setting. If genetic testing is obtained, the patient and their family will have an understanding of the impact that a positive test will have. Negative results for a particular test may not indicate the absence of an inherited abnormality. Genetic counseling will clarify when an inherited disorder is suggested by the family history, but cannot be demonstrated by the particular tests available, and may also identify other family members who may be at risk, and may expedite testing by directing and narrowing the scope of testing sought. Many laboratories doing this type of testing request that the results of family history and genetic counseling accompany the testing request.

Although the testing listed below is readily available through the Allina Medical Laboratories and its Reference Laboratories, *the patient's insurance carrier may not always cover these tests*. If coverage is in doubt, it is recommended that before the testing is ordered, the insurance carrier is contacted to ensure the nature of the clinical circumstance and the extent of coverage apply.

The following tests can be done at, or initiated by, the Allina Medical Laboratories Cytogenetics or Molecular Laboratories, and do not require that the Genetic Testing Worksheet be completed.

**Cytogenetics:**

- Chromosome Analysis, Amniotic Fluid
- Chromosome Analysis, Blood
- Chromosome Analysis, Chorionic Villus Sample
- Chromosome Analysis, Tissue
- Chromosome-Specific Probe, FISH
- Microdeletion Syndrome, FISH

**Molecular Genetics:**

- Apolipoprotein E Genotyping
- Canavan Disease, Mutation Analysis
- Cystic Fibrosis, DNA Probe Analysis
- Factor V (Leiden) Mutation Analysis
- Fragile X, DNA Probe Analysis
- Gaucher Disease, Mutation Analysis
- Hereditary Hemochromatosis, DNA Mutation Analysis
- Methylenetetrahydrofolate Reductase, DNA Mutation Analysis
- Prothrombin (Factor II) 20210G A Mutation Analysis
- Sickle Cell Anemia, DNA Probe Analysis
- Tay Sachs Disease, Mutation Analysis

**If genetic test requested does not appear on this list, please contact the Sendout Department (612-863-4356) for clarification PRIOR to collection. Patients may need to be rescheduled for this collection until all information is available.**

The following are included with this Update to better manage the issues raised by this type of testing:

- ❑ Genetic Testing Worksheet outlining important information and steps to complete prior to obtaining specimen. Please complete this worksheet and submit with specimen. No testing will be submitted without this completed worksheet.
  
- ❑ Genetic counseling resources within the metro Allina Health System. Genetic counseling is available through *Minnesota Perinatal Physicians* located at Abbott Northwestern Hospital (612-863-4502), Mercy/Unity Hospitals (763-236-8438) and United Hospital (651-220-6270).
  
- ❑ Advanced Beneficiary Notice (ABN) forms