Genetic testing is available for individuals with hemophilia and their family members. Here are important facts you should know.

» Genetic testing for hemophilia has dramatically changed in the past 5-10 years; therefore, information you received in the past may not be accurate.

» Genetic testing provides information that may be useful for
  - Carrier testing for family members
  - Treatment of the patient with hemophilia
  - The future to determine eligibility for gene therapy

There are different types of genetic testing available. Mutation analysis is the most accurate method of genetic testing. This testing has only been available for about 15 years; therefore, any testing completed before that time may not be accurate. Mutation analysis looks at the gene that produces the clotting factor, either VIII or IX, and identifies the change or mutation that causes the gene to not function as expected. In more than 95% of individuals with hemophilia, the mutation causing the disorder is identified.

**Hemophilia A**

About 40% of individuals with severe hemophilia A have a genetic change called an inversion which is easily detectable. This test is not as labor intensive and is less expensive than full mutation analysis. Therefore, if this mutation is identified in the factor VIII gene, testing is less expensive — currently this costs about $250. The cost to identify the inversion in other family members for the purpose of carrier testing is also about $250.

For individuals who are not found to have an inversion, or for those with mild or moderate hemophilia A, mutation analysis is performed by scanning the entire gene to search for a change. This testing is more labor intensive, and is reflected in the cost. Currently the charge for the first person in a family to be tested is $1,220. Therefore, once the mutation in the family has been identified, testing for other family members is less expensive; it costs about $300.

**Hemophilia B**

There are not any common changes in the factor IX gene similar to the inversion seen in the factor VIII gene. Therefore, factor IX mutation analysis is required to scan the entire gene in search of a change. This testing is available at a cost of about $565; the testing of additional family members once a mutation is identified is about $300.

Additional genetic testing methods are available and appropriate for certain types of disorders, based on family history. Please discuss with the IHTC genetic counselor how these tests might be pertinent to you.

**Paying For Genetic Testing**

Your insurance may cover the cost of genetic testing; check with your insurance provider prior to having testing completed. Some insurance companies will ask for a letter from your physician outlining the benefits of this testing. If this is the case, please contact the IHTC so we will assist you with this requirement.

For more specific information on how genetic testing applies to you and your family, and its benefits, please contact the center at 877-256-8837 and ask to speak to the genetic counselor.