

**Informed Consent for Genetic Testing**  
**University of Pennsylvania Health System**  
**Molecular Pathology Laboratory**  
**Department of Pathology and Laboratory Medicine**  
**Phone: 215-662-6121 Fax: 215-614-1986**

**Informed Consent for Genetic Testing**

Please read and sign this form so that we can be sure you understand this genetic testing (also called "DNA testing") and the risks associated with it. Please ask questions about anything on this form you do not understand.

**Purpose**

I understand that a sample of blood will be drawn from me and/or members of my family for the purpose of attempting to determine if I and/or family members are carriers of the disease gene, or are affected with, or at increased risk to some day be affected with this genetic disease.

**Background**

DNA is a chemical that encodes hereditary information. Genes are specific pieces or subunits of DNA that have function in the body. Genes come in pairs, one from our mother and the other from our father. A DNA test can directly detect an abnormality, called a mutation. Mutations are most often found in the gene and result in abnormal gene function, which is associated with disease. Depending on the genetic condition a mutation in one gene may be associated with disease (autosomal dominant) or in other conditions (autosomal recessive) both genes need to have mutations to be associated with disease. The interpretation of my results will be explained to me when they are available.

**Genetic Testing**

This genetic test is specific for \_\_\_\_\_ (specific disease). There is a chance that I will have this genetic condition but the DNA test results will be negative. This is called a false negative. There is a \_\_\_\_\_% chance that this test will detect this condition if I have it. Due to laboratory techniques and capabilities some mutations that might be associated with this disease are not possible to test for at this time. Some individuals who have an altered gene may never develop clinical symptoms associated with the condition. The chance for a gene mutation to cause symptoms has been discussed with me. In some cases, an indirect DNA test called linkage analysis may have to be used. Linkage analysis involves blood samples from other specified family members. If linkage analysis is being used, naturally occurring rearrangements in the DNA, called recombination, may produce an uncertainty in the results. Linkage analysis is not 100% accurate because of the chance for recombination. Results from linkage analysis are reported as a probability. In some families, the DNA markers used in testing may not be informative. If this is the case, this DNA test cannot provide results for that family or for some members of that family. An error in the diagnosis may occur if the true biological relationships of the family members involved are not as I have stated. Also, testing may detect non paternity. Non paternity means that the father of an individual is not the person stated to be the father. DNA analysis is specific only with respect to the Disease. It does not provide me with any information about the current status of my health. It in no way guarantees my health or the health of my unborn child or children.

DNA tests are relatively new and are being improved and expanded continuously. This testing is often complex and utilizes specialized materials so that there is always a small possibility that the test will not work properly or that an error will occur. There is an error rate, although low, even in the best laboratories. In the rare event of a laboratory error, I may be asked to have my blood redrawn. My signature below acknowledges my consent to have my blood re-drawn in the event of laboratory error. In some cases it may be possible for the laboratory to reanalyze leftover DNA samples in the future using new and improved testing methods. However, I understand that this is not a DNA banking facility and my DNA sample may not be available for future clinical studies.

**Use of Specimens**

I understand that any samples obtained for the purpose of this testing become the exclusive property of the University of Pennsylvania Health System and that I relinquish all right, title and interest to such samples. I understand that my blood sample will only be used for DNA testing as authorized by my consent and that my DNA sample will not be made available for future clinical studies or research purposes without my consent.

**Procedure**

The procedure for drawing blood involves placing a needle in a vein in the arm to draw blood. This procedure only takes a few minutes. Occasionally there are minor complications, and I may experience bruising, swelling, black and blue marks, fainting and/or infection at the site. I have disclosed and discussed with the physician any pertinent information or concerns about a personal history of bleeding problems or any other medical conditions before having my blood drawn.

**Alternatives**

There may be other tests that my physician could use to help diagnose my condition. If I am reluctant to have the DNA test, I will discuss this with my physician.

**Psychological Consideration**

Sometimes DNA testing can have an adverse psychological effect on an individual. Some individuals may experience depression, increased anxiety, mood changes and other forms of emotional distress both before and after receiving results. However, some individuals report feeling a sense of relief, closure, and/or decreased anxiety from proceeding with DNA testing and receiving results.

**Confidentiality**

My results are confidential. Because DNA testing is complex and results occasionally difficult to interpret, my results will be reported to me only through a physician or genetic counselor whom I designate. The medical information produced as a result of this testing will become part of my medical record and will only be released to other medical professionals or other parties with my written consent. However, my insurance carrier may receive information about my DNA testing if it is paying for my testing or treatment or if I have authorized the release of my medical records for other reasons. My signature below acknowledges my consent to the disclosure of my genetic testing results by the University of Pennsylvania Health System to the insurance carrier(s) involved or to the individuals or entities listed below or as required by law. This disclosure may include test results, authorization or consent forms, chain of custody forms, or other documentation that may include personal health information.

**Billing**

The billing process for this DNA testing has been explained to me. I will be responsible for covering the costs associated with testing.

**Discrimination Risks**

Some individuals who have elected DNA testing and been found to have the gene leading to the disease have experienced discrimination. I understand that discrimination (insurance, employment, and social) is a risk of genetic testing, and that my health and life insurance rates, my ability to obtain health or life insurance and my employability could be affected.

**Voluntary**

Participation in genetic testing is completely voluntary. When results are available I will be given the opportunity to decline hearing them. If I choose not to receive my results, a record of the results will be kept in my chart for reference should I change my mind. I will receive a copy of this consent form.

**Physician's/Counselor's Statement (SIGNATURE REQUIRED)**

I have explained DNA testing to \_\_\_\_\_(Print Patient Name). I have addressed the procedures involved the possible risks and benefits and the limitations outlined above, and I have answered this person's questions.

**\*\*\*Physician/Counselor's Signature:** \_\_\_\_\_

Date: \_\_\_\_\_

**Patient Consent to Genetic Testing (SIGNATURE REQUIRED)**

I, \_\_\_\_\_ (Print Patient Name), hereby agree to participate in testing for \_\_\_\_\_ (name of disease) using a DNA- based test. This testing will be performed by the University of Pennsylvania Health System.

**\*\*\*Patient Signature:** \_\_\_\_\_

Date: \_\_\_\_\_

**Parent/Guardian Documentation (REQUIRED FOR GENETIC TESTING OF CHILDREN)**

I, \_\_\_\_\_ (print parent/guardian name), hereby give my consent for the participation of minor child, (print name of minor child) \_\_\_\_\_ (Date of Birth: \_\_\_\_\_), in genetic testing using a DNA-based test. This testing will be performed by the University of Pennsylvania Health System. By my signature below, I represent that I have the legal authority to consent to medical procedures performed on the minor child named above. I represent that I am the minor child's natural parent or legal guardian and that my parental rights or guardianship have not been terminated or limited in any way by a court or otherwise by law. If I am a natural parent, and am divorced, unmarried to or legally separated from the other natural parent of the minor child on whose behalf I sign, I represent that I have sole or joint custody of the minor child and that my parental rights have not been terminated or limited in any way by a court or otherwise by law.

**Parent/Guardian Signature:** \_\_\_\_\_  
**Date:** \_\_\_\_\_

**Authorization for Further Disclosure of Genetic Testing Results**

I request that the University of Pennsylvania Health System send copies of my genetic testing results to the following individuals and/or entities at the following addresses. I understand that the test results contain personal health information and that once my results are disclosed consistent with this authorization, federal or state privacy laws may no longer protect the health information contained in the results. I understand that I have the right to revoke this authorization in the future in a separate, signed writing indicating my intent to revoke. This authorization is valid as of the date of my signature below and shall remain valid for a period of twelve months beyond that date.

**Send results to: (Provide full name and address of each individual to whom results should be sent.)**

\_\_\_\_\_  
\_\_\_\_\_

**Patient Signature:** \_\_\_\_\_  
**Date:** \_\_\_\_\_