

HUNTINGTON DISEASE (HD)

_____ / _____ / _____
 Patient Last Name Patient First Name MI Date of Birth (MM/DD/YY) Biological Sex

INFORMED CONSENT FORM

I would either like to participate in predictive testing for **OR** I am the parent/legal guardian of a symptomatic individual under 18 years of age who would like to participate in testing for the presence of the Huntington disease gene. I understand that the gene for HD has been found and is located on chromosome 4. It has been described as being a trinucleotide (CAG) repeat expansion. It is the size of this trinucleotide CAG repeat which determines whether or not HD will be expressed. The blood test will determine the size of this trinucleotide CAG repeat.

- I. I understand there can be four outcomes to the test:
1. **NEGATIVE:** The CAG repeat size is in the normal range (26 repeats or less). This individual is not at risk for developing HD, and not at risk for passing it on to his/her offspring.
 2. **INTERMEDIATE:** The CAG repeat size is 27 - 35 repeats. This individual is not at risk for developing HD but his/her children could be at risk.
 3. **REDUCED PENETRANCE:** The CAG repeat size is 36 - 39 repeats. It is unclear whether this individual will or will not develop HD at some point in his/her life. His/her children could be at risk.
 4. **POSITIVE (FULL PENETRANCE):** The CAG repeat size is expanded into the HD range (40 repeats and larger). This individual will develop HD at some point in his/her life. Neurological examination is needed to establish the onset of symptoms.
- II. I understand that a positive test cannot tell me when I will begin showing signs of HD. I understand that the diagnosis of HD can only be made through a neurological exam.
- III. The risks of such testing are primarily of a psychological nature. An inconclusive outcome can be frustrating and intensify the ambiguity of the risk situation or can provide relief. A negative result can produce feelings of guilt as well as of joy. A positive result, i.e. the HD gene is present, could lead to serious psychological consequences including feelings of depression, futility, despair, and severe stress. Test results may also reveal that other family members may be affected or at risk for developing HD, or that familial relationships are not as anticipated. The counselor has discussed with me the possible risks of difficulty with employment, insurance, and confidentiality.
- IV. In view of the psychological risks, current standard of care is that HD testing be performed for confirmation of diagnosis in symptomatic patients or for non-symptomatic adults who have undergone appropriate counseling. HD testing is not considered appropriate for non-symptomatic minors.
- V. I understand that all information will be held strictly confidential. The results of the testing will be sent from the lab to my provider and disclosed only to me and to no one else without my written consent.
- VI. I have been given the opportunity to discuss pertinent aspects of the testing program, to ask questions, and hereby consent to presymptomatic testing for HD, or to HD testing for a symptomatic minor.
- VII. I may give consent to allow the sample to be used for test validation, education, and stored indefinitely as long as the patient's privacy is maintained. Refusal to participate in research will not affect the test result. If a response is not checked, consent is implied.

I authorize consent for the use of the above individual's sample for test validation and education: YES NO

VIII. Information obtained from the test may be used in scientific publications, but the identity of all persons in the test will not be revealed in such publications or in any other report.

I have the legal authority to request Baylor Genetics to test this sample for Huntington Disease (HD). I am either the above patient or his/her parent or legal guardian. I have been counseled regarding the risks, benefits, and limitations of knowing the test results and have carefully considered the psychological impact the results may have on the patient and his/her family. My physician or genetic counselor has thoroughly discussed the above information and answered my questions.

Parent/Legal Guardian Signature: _____ Date (MM/DD/YY): _____

Parent/Legal Guardian Name: _____

Physician/Counselor: I have explained HD DNA testing and its limitations to the patient or his/her legal guardian.

Physician/Counselor Signature: _____ Date (MM/DD/YY): _____

Physician/Counselor Name: _____

Phone: _____ Fax: _____