
INFORMED CONSENT FOR GENETIC TESTING FOR HUNTINGTON DISEASE

I agree to participate in genetic testing for Huntington Disease (HD). This test will analyze the size of the trinucleotide (CAG) repeat within the *HTT* gene in DNA obtained from my blood sample. I have two copies of the *HTT* gene and CAG repeat region, known as alleles. Both alleles will be tested. The size of the CAG repeat determines whether a person will experience symptoms of HD. Although the test is highly accurate, there is a very small possibility of a false negative or false positive result due to sample mix-ups, other DNA mutations, or other sources.

I understand that there are four possible test results:

1. Negative (26 or fewer CAG repeats): I will be told that the size of the CAG repeat is within the normal range, and I am not at risk for developing HD.
2. Normal but mutable (27-35 repeats): I will be told that although the size of the CAG repeat is slightly larger than the normal range, I am not at risk for developing HD. Repeats of this size are unstable and may expand and cause HD if transmitted to any of my children. Each of my children will have a 50% chance of inheriting the unstable allele, but the risk of expansion is not known.
3. Pathogenic, reduced penetrance (36-39 repeats): I will be told that the size of my CAG repeat is usually but not always associated with HD. Each of my children will have a 50% chance of inheriting this allele, and there is significant likelihood that, if transmitted to my child, the CAG repeat will expand to the pathogenic range.
4. Pathogenic, full penetrance (≥ 40 repeats): I will be told that the size of my CAG repeat is always associated with HD. Each of my children has a 50% chance of inheriting this allele and developing HD.

I understand that a positive (pathogenic) test result will not predict when I will begin experiencing symptoms of HD, and that a clinical diagnosis of HD can only be made by neurological examination.

I understand that the risks associated with HD genetic testing are primarily psychological in nature. A negative result can produce feelings of joy as well as guilt. An inconclusive result can cause frustration. A positive result could lead to serious psychological consequences including depression, despair, guilt, and stress. Counseling is available to me.

I understand that all information will be held strictly confidential. My test result will be sent from the laboratory to the ordering provider and disclosed only to me and to no one else without my written consent.

I may give consent for my sample to be stored indefinitely for the possibility of future use in test validation, research, or education. Patient privacy will be maintained at all times. If a response is not checked, consent is implied.

I authorize consent for use of this sample for test validation and education: NO YES

PATIENT/LEGAL GUARDIAN: I have the legal authority to request genetic testing for HD for this sample. I am either the patient or his/her legal guardian. I have been counseled and given an opportunity to ask questions regarding the risks, benefits, and limitations of knowing the test results. I have carefully considered the psychological impact the results may have on the patient and patient's family.

Patient/Legal Guardian Name: _____

Signature: _____ Date: _____

PHYSICIAN/COUNSELOR: I have explained the risks, benefits, and limitations of HD genetic testing to the patient or legal guardian.

Physician/Counselor Name: _____

Signature: _____ Date: _____

COLORADO MOLECULAR CORRELATES LABORATORY

PHONE: 303.724.4754 FAX: 303.724.3096

Ship to: CMOCO Laboratory

BioScience 2, Room 4105

12705 E. Montview Blvd.

Aurora, CO 80045

HUNTINGTON DISEASE TESTING INDICATION FORM

Patient Name (Last, First, MI) _____

DOB (MM/DD/YYYY) _____

Gender (M/F) _____

SYMPTOMATIC PATIENT: GENETIC CONFIRMATION OF CLINICAL SUSPICION OF HD

- Is the patient under 18 years of age? NO YES
- Is there a confirmed family history of HD? NO YES; relationship to patient _____
- Is there a suspected family history of HD? NO YES; relationship to patient _____

Which of the following symptoms are documented in this patient?

- Movement disorder: NO YES; Age of onset: _____
- Behavioral/psychiatric disturbance: NO YES; Age of onset: _____
- Cognitive decline/dementia: NO YES; Age of onset: _____

ASYMPTOMATIC PATIENT: PREDICTIVE GENETIC TESTING FOR HD

Available only to individuals at 50% risk of HD and who are 18 years of age or older

Affected relative(s) (check all that apply): Mother Father Sibling Other: _____

Has an affected relative had genetic testing for HD? NO YES

Relationship to patient: _____

CAG repeat size of affected relative: _____

Instructions: Send consent form, testing indication form, and peripheral blood sample in EDTA on wet ice by overnight delivery to the address above. Samples accepted Tuesday-Friday; do not ship samples for weekend delivery. Result will be returned within 4 weeks.

Physician/Counselor Signature _____ Date: _____

Phone: _____

Fax: _____