ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

A nonprofit enterprise of the University of Utah and its Department of Pathology

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INFORMED CONSENT FOR HUNTINGTON DISEASE (HD) DNA TESTING

Patient Name _____ Date of Birth _____ Sex ☐ F ☐ M

Does the patient have symptoms of HD? ☐ Y ☐ N If so, describe _____

Who is the patient's closest relative with HD? _____ Age the relative's symptoms began _____

Was this relative's diagnosis confirmed by DNA testing? ☐ Y ☐ N

- Participation in genetic testing is completely voluntary. Genetic counseling is highly recommended prior to and following genetic testing for HD. See www.nsgc.org to find a medical genetics professional. The ordering health care provider or genetic counselor should explain the test results in person and be available for follow-up genetic counseling. Patients undergoing presymptomatic testing should be accompanied by a support person, who is not at risk for HD, when receiving results.
- HD is an inherited, neurodegenerative condition affecting thought processes, movement, personality, and mood. Although treatments are available, there is currently no cure for HD. This blood test determines the number of CAG repeats in the HD gene. An expanded number of CAG repeats is the cause of most cases of HD. The accuracy of an "affected" HD DNA test result is 99%. The certainty of an "unaffected" test result depends on the accuracy of the HD diagnosis in the family. Possible sources of error include: clinical misdiagnosis of a condition, inaccurate information provided regarding family relationships, sample mislabeling or contamination, transfusion, bone marrow transplantation, and maternal cell contamination of prenatal or cord blood samples.
- There are four possible test results:
  1) **Unaffected:** Both of the HD genes have a normal number of CAG repeats (<27). This individual is neither at risk for developing HD nor having affected offspring.
  2) **Intermediate:** One HD gene has 27-35 CAG repeats. Although this individual is not at risk for developing HD there is a small risk for having affected offspring.
  3) **Affected/ Reduced Penetrance:** One HD gene has 36-39 CAG repeats; therefore, this individual may or may not develop HD and may or may not have affected offspring.
  4) **Affected:** One HD gene contains 40 or more CAG repeats; therefore, this individual will develop HD within a normal lifespan. Offspring of this individual have a 50% risk for developing HD. The age of symptom onset and disease progression cannot be precisely predicted. Neurological examination is needed to establish the onset of symptoms.
- Test results may reveal non-paternity or that other family members may be affected with, or at risk for developing, HD.
- There are psychological risks associated with HD testing. A result that indicates an individual will be unaffected can produce feelings of guilt as well as joy. An intermediate test result, indicating the patient may or may not develop symptoms, can be frustrating. A result that indicates an individual will be affected could lead to serious psychological consequences including feelings of depression, futility, and severe stress.
- If a CAG repeat expansion is identified, insurance rates, the ability to obtain disability and life insurance, and employability could be affected. The Genetic Information Nondiscrimination Act of 2008 extends some protections against genetic discrimination (http://www.genome.gov/10002328). All test results are released to the ordering health care provider and those parties entitled to them by state and local laws.
- Because ARUP is not a storage facility, most samples are discarded after testing is completed. Some samples may be stored indefinitely for test validation or education purposes after personal identifiers are removed. All New York samples are discarded 60 days following test completion. You may request disposal of your sample by calling ARUP Laboratories at (800) 242-2787 ext. 3301.

Patient, Legal Guardian, Power of Attorney (POA) I have the legal authority to request ARUP Laboratories to test this sample for HD. I am the patient, his/her legal guardian, or POA. I have been counseled regarding the risks, benefits and limitations of this test and carefully considered the psychological impact the results may have on the patient and his/her family.

_____
Patient/Legal Guardian/POA Printed Name | Signature | Date

Ordering Healthcare Provider or Genetic Counselor: I have explained HD genetic testing, its risks, benefits, limitations, and alternatives to the patient or legal guardian and addressed all their questions.

_____
Healthcare Provider Printed Name | Signature | Date

_____
Specialty | Phone Number | Fax