ARUP LABORATORIES

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

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THIS IS NOT A TEST REQUEST FORM. Please complete and submit with the test request form or electronic packing list.

INFORMED CONSENT FOR HUNTINGTON DISEASE (HD) DNA TESTING

Patient Name: _____ **Date of Birth:** _____
Sex Assigned at Birth: ☐Female ☐Male ☐Intersex **Gender Identity (optional):** ☐Female ☐Male ☐_____
Medical Record Number: _____
Does the patient have symptoms of HD? ☐ Yes ☐ No If yes, 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? ☐ Yes ☐ No

- Participation in genetic testing is completely voluntary. Genetic counseling is highly recommended before and following genetic testing for HD. Refer to nsgc.org or acmg.net to find a medical genetics professional. The ordering healthcare 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 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 samples.

Test results typically fall into the following categories:

1. Unaffected: Both HD genes have a normal number of CAG repeats (<27). This individual is neither at risk for developing HD nor for 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 nonpaternity or that other family members may be affected with or at risk for developing HD. When testing a prenatal sample, information about the parental HD status will be included in the prenatal report.
- 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 (genome.gov/10002328). All test results are released to the ordering healthcare 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 educational purposes after personal identifiers are removed. You may request disposal of your sample by calling ARUP Laboratories at 800-242-2787 ext. 3301.

Note: All New York samples will not be used for research or quality assurance and will be destroyed automatically withing 60 days of collection.

Patient, Legal Guardian, Power of Attorney (POA): I have the legal authority to request that ARUP Laboratories test this sample for HD. I am the patient, the patient's 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 myself and my family.

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

Relationship to patient: ☐ Self ☐ Guardian ☐ POA

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

_____ _____ _____
Health Provider/Genetic Counselor Printed Name Signature Date

_____ _____ _____
Specialty Phone Number Fax