Informed consent for Huntington disease testing

Patient's name: _____ **Patient's ID number:** _____

Patient's date of birth:_____ **Ordering provider's name:** _____

Note to the ordering healthcare provider: Some states require that patients (or their authorized representative(s)) provide written consent prior to receiving genetic testing and that the ordering healthcare provider maintain documentation of the informed consent in the patient's medical record. This form is intended to assist you with obtaining the patient's informed consent in accordance with applicable law.

Huntington disease (HD) is a rare progressive neurological disorder caused by changes in the HTT gene.[1] The specific type of change for HD is known as a repeat expansion and occurs when three specific DNA nucleotides (CAG) are repeated many times. The specific number of repeats indicates the likelihood of disease. The genetic test for HD evaluates the number of CAG repeats and reports a result based on American College of Medical Genetics (ACMG) guidelines[2]:

- **Negative testing:** Two alleles with ≤ 26 CAG repeats were detected. Repeat sizes in this range are considered normal and are not associated with disease. The results from this test should be used in the context of available clinical findings and should not be used as the sole basis for patient management or treatment. Genetic counseling is recommended.
- **Equivocal testing:** An allele with 27 to 39 CAG repeats was detected. The significance of CAG repeats in this range is unclear at this time. Although alleles in this range have been reported in cases of Huntington disease, co-segregation with disease is not always observed and repeat sizes in this range have also been reported in the general population and in apparently healthy controls. CAG repeat numbers in this range may be unstable, and individuals with CAG repeats in this range are at risk for having children with symptomatic repeat expansions. The results from this test should be used in the context of available clinical findings and should not be used as the sole basis for patient management or treatment. Genetic counseling is recommended.
- **Positive testing:** An allele with ≥ 40 CAG repeats was detected. Repeat sizes in this range are reported to be highly penetrant and are generally considered to be pathogenic. The results from this test should be used in the context of available clinical findings and should not be used as the sole basis for patient management or treatment. Genetic counseling is recommended.

Genetic testing is entirely voluntary, and choosing to participate in this testing means that I am aware of the following:

- **Genetic counseling:** Participating in pre-test counseling may be required for testing. The purpose of this is to ensure that the nature of HD and what the results may mean are fully understood. Post-results counseling is also highly encouraged, regardless of result.
- **Test limitations:** This assay does **not** detect point mutations or deletions/duplications in the HTT gene. This assay does not allow for the sizing of alleles greater than 150 repeats. Thus, alleles larger than this are reported as "> 150 repeats". False negative results are rare. Donor DNA from transplants and recent transfusions can lead to inaccurate results. As in any laboratory test, there is a possibility of error.
- **Disclosure of test results:** Results are kept confidential and will be disclosed only to the ordering healthcare provider (or his or her designated representative) unless otherwise authorized by the patient in writing or as required by law.
- **Specimen retention:** The laboratory does not return any remaining sample to individuals or physicians unless requested. No clinical tests other than those authorized shall be performed on the sample. A request for additional testing must be made by my referring physician or other authorized healthcare professional and there will be an additional charge. The sample will be destroyed at the end of the testing process or not more than 60 days after the sample was taken, unless I expressly authorize a longer period of retention.
- **I consent to have my specimens retained** after completion of initial testing (this consent may be withdrawn at any time and the laboratory will destroy any remaining sample). Patient (or parent/guardian) **Initials:**_____
- **Psychological risks:** Given the nature of HD, testing outcomes may have psychological impacts. A negative result may produce feelings of joy or guilt, while a positive result may lead to serious psychological consequences like feelings of depression, futility and severe anxiety. An equivocal result may produce feelings of frustration or anger.
- **Patient or legal guardian:** My signature on this form indicates that I understand the conditions and risks associated with genetic testing for HD and that I am consenting to having this test performed.

_____	_____
Patient printed name	Date of birth
_____	_____
Patient signature	Date
_____	_____
Healthcare provider/genetic counselor signature	Date

References
1. Huntington disease. United States National Library of Science website. https://ghr.nlm.nih.gov/condition/huntington-disease. Updated March 31, 2020. Accessed April 1, 2020.
2. Bean L and Bayrak-Toydemir P. American College of Medical Genetics and Genomics Standards and Guidelines for Clinical Genetics Laboratories, 2014 edition: technical standards and guidelines for Huntington disease. *Genet Med*. 2014, Dec;16(12).

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