

Informed Consent – *Predictive Testing for Huntington Disease*

NOTE: Please obtain patient signature on consent form and provide a signed copy to EGL Genetics to permit testing and processing.

I, (name) _____, voluntarily request EGL Genetics to perform DNA-based testing for Huntington disease in an attempt to determine whether I am at increased risk to be affected by the condition. The following points were explained and I understand that:

Huntington disease is a progressive neurodegenerative disease characterized by chorea and dementia. This disease is inherited in an autosomal dominant fashion and affects approximately 1 per 10,000 individuals. Although patients may develop symptoms at any time from childhood to later in life, the average age of onset is between 35 and 45. With paternal transmission of the gene, there is a tendency for earlier onset to occur. Over 80% of the patients with an onset before the age of 20 inherited the abnormal gene from their father. Mutations may occur spontaneously as well, but this is uncommon.

I agree to participate in predictive testing for Huntington disease (HD) by direct analysis of the trinucleotide repeat (CAG) within the *HTT* gene located on Chromosome 4. The size of the trinucleotide repeat (CAG) determines clinical expression of HD. The blood test will determine the size of the CAG repeat. The test is highly accurate. False positive or false negative results due to sample mix-ups, laboratory methods, or other sources are possible, but are rare.

I understand that there are four possible outcomes to my test:

1. **Negative** (26 or fewer CAG repeats): I will be told that the CAG repeat is in the normal range and I am not at risk for developing HD.
2. **Normal but mutable** (27-35 CAG repeats): I will be told that I do not have the CAG repeat expansion mutation, but that I carry an unstable CAG repeat in the *HTT* gene that may expand, causing HD if transmitted to my children. Each of my children has a 50% chance of inheriting this unstable allele from me, but the risk of expansion is not known.
3. **Borderline** (36-39 CAG repeats): I will be told that CAG repeats of this length are usually associated with HD, though there are older individuals who carry CAG repeats of this length who show no signs of the disease. Each of my children has a 50% chance of inheriting this allele from me and there is a significant risk of this unstable allele expanding to the disease causing range when transmitted to my children.
4. **HD mutation** (40 or higher CAG repeats): I will be told that CAG expansions of this size always cause HD. Each of my children has a 50% chance of inheriting this mutation from me and developing HD.

I agree to participate in pre-test counseling sessions required for the test. I understand that the purpose of these visits is to assure that I fully understand the nature of HD and address all aspects of the decision to pursue predictive testing. Additionally, I agree to maintain at least telephone contact with the testing counselor after receiving test results.

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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.

I understand that the risks of predictive testing are primarily of a psychological nature. An uninformative outcome, even though a remote possibility, may be frustrating and may cause a sense of uncertainty. A negative result can produce feelings of guilt, as well as joy. A positive result, i.e. that the HD CAG expansion is present, may lead to serious psychological consequences including feelings of depression, futility, or despair. Counseling provided during the test is designed to help me adjust as best as possible to uninformative, positive, or negative information.

Physically, risks include the discomfort of a needle prick and the possibility that a bruise may form as a result of blood being drawn, a mark that will fade in a few days. I understand that the drawing of blood sample involves no unusual hazard or risk.

I understand that I will be responsible for the costs of testing, which will be \$400 for DNA analysis if I choose to pay out-of-pocket. I understand that if I do not utilize a third party payer, I will be expected to pay for the DNA analysis in advance, to EGL Genetics.

All information will be held in strictest confidence. The results of testing will be given only to me and to no one else without my written consent; however, I understand that health insurance companies generally have a right to all medical information that concerns the individuals they insure. Information obtained from this test may be used in scientific publications or other report, only after my name and all identifying information is removed.

My signature on this form signifies that I have decided to participate in this testing program after reading the above information. I have been given the opportunity to discuss pertinent aspects of the testing program, to ask questions, and hereby consent to participate in the testing as outline above.

Client's Signature

Date

Testing Counselor's Signature

Date