IU MOLECULAR GENETICS DIAGNOSTIC LABORATORY
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INFORMED CONSENT FOR STATEMENT FOR PRE-SYMPTOMATIC TESTING FOR HUNTINGTON DISEASE

You have requested molecular genetic testing for Huntington disease (HD). In order to perform this test, we will need a blood sample from you. We ask that you donate approximately 3 mL of blood, which will be collected by a needle stick in your arm. This blood sample will be used to test for the presence or absence of the Huntington mutation.

If no one else in your family has been tested and found to carry the HD mutation, we recommend that one closely related relative who has HD-like symptoms, or with HD is also tested in order to make certain that the diagnosis of HD in your family is correct. It is possible that as a result of this testing, we may discover that the diagnosis of HD in your affected family member is not correct. If this fact is discovered, this information will be communicated to you.

The gene for Huntington disease is located on chromosome 4. The genetic mutation, or change, that causes Huntington disease is a variation in the number of a particular genetic sequence made up of three DNA bases—cytosine, adenine and guanine (CAG). Since the discovery of the HD gene in 1993 and the advent of direct gene testing, research has indicated that there are four possible outcomes of this test based on the number of CAG repeats that are found. These are:

CAG REPEAT SIZE	INTERPRETATION
26 and below	Normal
27-35	Non-penetrant but meiotically unstable
36-39	Variable penetrance and meiotically unstable
40 and above	Huntington disease

To clarify this further, any number of CAG repeats that is less than or equal to 26 is considered normal. Within this range, the gene also appears to be stable, i.e.: does not appear prone to expansion. If the number of CAG repeats is within the range 27-35, you yourself are unlikely to develop the symptoms of HD. However, the gene appears to be unstable in the range and the number of CAG repeats may be expanded so that the possibility exists that one or more of your children may have inherited a CAG repeat that is larger than yours and may be in the HD range. If the number of CAG repeats is within the range of 36-39, we cannot predict with any certainty whether or not you will develop the symptoms of HD. Within this range, some people have been found to have symptoms of HD and some people have been found who are very old and do not appear to have any symptoms of HD. The gene is also unstable in this range and may expand so that one or more of your children may have a number of CAG repeats that is clearly within the HD range. If the number of CAG repeats is 40 or greater, it is virtually certain that you yourself will develop the symptoms of HD at some time.

Please remember that although research is progressing rapidly, we have no cure or treatment of HD at this time. The major risks to you from taking this test are psychological problems (depression, anxiety, family problems) that may result if you find that you carry the gene for HD and will develop HD at some time in the future. Every effort will be made to support, assist, and treat you should you experience distress.

The decision whether or not to be tested for HD is a very personal one. Please be aware that testing is strictly voluntary, and you can change your mind about testing at any time. We want to be sure that only those who are very sure that they wish to know this information do so. Please feel free to ask any questions that you might have about testing.

Information regarding the results of your test will be kept in a locked storage in the Department of Medical and Molecular Genetics. A copy of your test results will be given to you. Any other release of this information must be requested in writing.

Consent

I give my consent to have a sample of my blood tested for the presence or absence of the genetic mutation that causes Huntington disease.

Signature _____ Date _____

Signature of Witness _____ Date _____