Presymptomatic Testing Program for Huntington Disease

Informed Consent

Testing Program Description and Requirements: I am willingly providing a sample of blood for genetic analysis for the purpose of learning whether or not I have inherited the gene (genetic factor) that causes Huntington Disease (HD). I understand that this is a clinical test and is not a research study. I understand that the gene for HD has been found and it is located on chromosome 4. The gene has been described as having a repeating segment of genetic material called a trinucleotide repeat (CAG). It is the size of this repeating segment which determines if HD will occur in a person. The blood test will determine the size of the CAG repeat. I understand that this test will only be evaluating the gene for Huntington Disease and my blood will not be used for any other testing.

I understand that my blood sample is best studied when a sample of blood or stored DNA from a member of my family who has been diagnosed with Huntington Disease is available. I also understand that in order to obtain the blood test and receive the test results, I must participate in a testing program that provides genetic and psychological counseling as well as neurological evaluations and any necessary follow up care, as recommended by the program.

I have been told and understand the following:

1. I am at risk for developing Huntington Disease because there is a history of this condition in my family. The genetic counselor has discussed the inheritance of this disorder and has discussed my risk with me.

2. This test will evaluate my gene for Huntington Disease (HD).

    A. A Negative Result means that the CAG repeat size is in the usual range (26 or fewer repeats), and that I would not be at risk for developing HD when the test is negative.

    B. A Positive Results means that the CAG repeat size is expanded into the typical HD range (40 or more repeats), and that I will develop HD at some point in my life when the test is positive.

    C. An Inconclusive Result means that the CAG repeat size is in the intermediate range (27-39 repeats) and that it is unclear whether I will

or will not develop HD at some point in my life.

I understand that a positive test result cannot tell me when I will begin showing signs of HD. I understand that the diagnosis of the onset of HD can only be made through a neurological examination.

If available, it is recommended that this blood test first be performed on an affected family member in order to confirm the presence of HD in my family.

Description of Procedures: I understand that this test requires me to give 20 cc (two tubes) of my blood. I also understand that the testing procedure requires me to participate in a testing program. The program involves genetic and psychological counseling, a neurological evaluation, and psychological testing. The aim of this program is to determine:

1. that testing is being offered to individuals who fully understand the impact of this test on themselves and their families.

2. that individuals requesting presymptomatic testing do not now have a neurological disease.

3. that appropriate follow up services for medical and psychological care are in place and available to individuals undergoing testing before the results of the test are given.

I agree to participate in the counseling sessions and examinations required for the test. Sessions will last from one to three hours each. Time between sessions will vary depending upon my own choice and the number of other people scheduled for the testing and examinations. I understand that during this time I will take part in psychological evaluations, including an in-depth interview regarding my attitude toward predictive testing, how I could react to various test outcomes, my personal relationships, how I would handle these attitudes and feelings, and other aspects of psychological functioning which have a bearing on the testing procedure.

I am fully aware that my decision to seek testing in the program is completely voluntary and that I may choose to terminate my participation in the program at any time without jeopardy. I also understand that the professionals of the testing program may decide to postpone my testing. The reason for postponing testing will be fully explained to me. I understand that I am encouraged to have a close friend, spouse or other family member not at risk for Huntington Disease, or a member of the clergy accompany me through the testing program. Because the information provided is technical and receiving it may be stressful, the person accompanying me is to help me through the process.

Testing Charges: I understand that I will be responsible for all testing charges. I have been provided with estimates of these fees. In addition, other members of my family who wish to learn whether they have or have not inherited the gene for Huntington Disease will incur the

same costs and must enter the full program in order to receive results. Third party (insurance) coverage may not cover all health costs associated with testing and insurance payment may interfere with the confidentiality of the test results. I understand that the approximate cost for the testing program is $600. The fees are charged through the University of Pittsburgh for laboratory costs ($300) and your examination by a physician ($300). Charges for extensive follow-up are not included in these figures. All charges are subject to change. Advance payment is appreciated if third party carriers such as insurance companies are not to be billed.

Risk and Discomforts: I understand that I will be given lengthy examinations. These may cause me to confront difficult psychological issues. Since I may withdraw from the testing program at any time and the process is a long one, decisions about continued participation may evoke anxiety.

I may also have to confront the fact that I may have inherited the gene that causes Huntington Disease, and so may develop the disease if I live long enough. If I have in fact inherited this gene, I understand that my present and future children will have a 50% chance of also having inherited the gene that causes Huntington Disease. This information may have serious effects on me such as noted below.

A positive result could lead to serious psychological consequences including but not limited to feelings of depression, futility, despair and severe stress. A negative result can produce feelings including but not limited to guilt as well as joy. In addition, I am aware that an inconclusive result can be frustrating and can intensify the ambiguity of my risk situation or can provide relief.

Counseling provided during the test is designed to help me adjust to positive, negative and inconclusive information as best as possible. Counselors will discuss with me other possible risks such as difficulties with confidentiality, employment, or insurance.

Physically, risks usually include the discomfort of blood being drawn from the arm which may result in a black and blue mark that should fade in a few days.

Benefits: I may learn whether or not I have inherited the gene that causes Huntington Disease. This may help me better plan my future and reduce the anxiety of not knowing.

I understand that all information from this clinical testing program will be held strictly confidential. The results of this clinical testing program will be given only to me by personal interview and results will not be released without my written consent or by an order or directive by a court of competent jurisdiction.

Retrospective review of information obtained from this clinical testing program may be used in scientific research publications, but the identity of all persons in the testing program will not be revealed in such publication or in any other report.

Refusal or withdrawal of consent: I may discontinue participation at any time in the testing program without prejudice to my present or future relationships, including medical care, at the University of Pittsburgh or the University of Pittsburgh Medical Center. I also understand that my doctors and counselors may terminate my participation in this testing at any time. The reasons for doing this will be fully explained and I will be helped to obtain any further medical care if that is appropriate.

I understand that the charges for the testing program are payable by me in the amounts in the consent form and I agree to make all such payments in full as required. I understand that release of information pertaining to this testing program could cause future problems for me in the event that such information is received by insurance companies or other third parties who make decisions based upon genetic risk. Therefore, I _____ do not authorize/ _____ authorize (please place an "X" and your initials next to your decision) release of any information pertaining to this clinical testing program to my insurance carrier and other third party only as noted below:

My signature below documents that: 1) I have had the opportunity to read this form or it has been read to me; 2) a physician or physician's representative has explained all the information contained in this form and answered to my satisfaction any questions that I had; and 3) based on my understanding of the testing program, its purposes, possible risks, discomforts and benefits that I may experience; I consent to participate in this clinical testing program.
(1/04)

_____

Patient's Signature Date_____

_____

Patient's Printed Name

_____

Witness Date_____