Huntington Disease: Protocol for Presymptomatic Testing

University of Iowa
Division of Medical Genetics
Testing Protocols

The University of Iowa has developed a protocol for presymptomatic testing for Huntington Disease (HD). HD is a genetic condition which results in progressive loss of ability to think clearly and uncontrollable movements. Symptoms most commonly appear in mid-adulthood (30-40 years old). There is no specific treatment available at this time. The gene for HD has been identified. By studying the gene, we are usually able to tell people with a family history of HD whether or not they will develop the disease. The testing does have limitations. First, it is not known if everyone who is affected with HD has the same change in the gene causing the disease. We only look for the common change. Second, occasionally, the result of gene analysis cannot be interpreted with confidence. In this case, the test results will not tell you more than you know now. Because of these limitations, testing of a sample from an affected family member is desirable for confirmation of the gene change and the HD diagnosis. Huntington Disease presymptomatic gene testing is offered so you may make informed plans for your future regarding marriage, reproduction, career and finances. It may also be done to relieve the psychological burden of not knowing if you will develop the disease. Some people prefer to know, other people prefer not to know. The decision to do this test should always be an informed, carefully considered and freely chosen personal decision. Individuals should not be persuaded into testing by family, friends, health care providers, employers, or insurance companies. Presymptomatic testing is available for individuals 18 years of age or older.

It is important to do this testing at a time when you have a low stress level in other areas of your life. It is important to have emotional support throughout the testing procedure and we encourage you to have a friend, family member, or clergy person with you at your appointments and available to you throughout the next few months for support. Another at risk individual, such as a brother, sister, or child would not be a good choice. We are happy to assist you in finding professional support either locally or through the University of Iowa.
Test results will not be given to anyone except yourself unless you give written consent to do so. The blood test will only be used for HD testing and not any research purposes.

A team approach is used by our service, including professionals in neurology, genetics, psychiatry and psychology. With rare exceptions, we will request patients undergoing testing follow the protocol outlined below. This protocol was developed based on the international experience with presymptomatic diagnosis of HD. It attempts to ensure each person makes the best decision for them. Individuals may withdraw from the protocol at any time. The protocol requires a minimum of three in person visits (# 2, #4, and #5). In some cases visits #1 and #2 will be combined. Visit #3 can be combined with visit #2 or #4.

Visit #1 Phone Contact or Clinic Visit

Family history, medical history and diagnosis verification will be obtained, insurance implications reviewed, importance of support person discussed and protocol received

Visit #2 Clinic Visit

Review: general information, testing procedure, consequences of testing and importance of psychological support

Receive consent forms for reading at your convenience

Arrange for blood draw to be at least one month after visit #2

Support person should be present

Visit #3 Clinic Visit

Neurological evaluation

Psychological evaluation may be recommended
Visit #4 Clinic Visit

Review of general information, testing procedure and consequences of testing. Review of testing outcomes.

Consent(s) signed

Blood drawn

Support person should be present

Set up next visit at least one month after blood draw. No results will be given over the phone.

Visit #5 Clinic Visit

Support person should be present

Receive results of testing and set up follow-up contact

Visit #6 Phone Contact

Genetic counselor will be calling within one week of results

Visit #7 Phone Contact or Clinic Visit (one month, one year, and then as needed)

Follow up counseling, medical follow-up as needed

The goal of the evaluations and counseling is to assure:

1. individuals requesting testing are aware of their options for testing and the accuracy and limitations of the test.

2. testing is being offered to individuals who fully understand the impact of this test on themselves and their families.
3. individuals requesting presymptomatic testing do not currently have a neurological disease.

4. 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.

You will be responsible for the costs of the laboratory testing and counseling. In some cases, medical insurance will not pay for this testing. Careful consideration should be given to insurance matters. The Huntington Disease Society of America has information to assist you with this.

Please call if you have any questions after reading this information and discussing it with appropriate family members and/or support persons. You may contact:

NAME: _________________________________________ at PHONE NUMBER: ___________________________.

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