Genetic Testing Program for Huntington’s Disease

Thank you for contacting our Center for information about genetic testing for the Huntington’s gene expansion. We know this is a very important decision you are making, and it is our goal to help you with the process as best we can.

There has been a gene test for Huntington’s Disease since 1993. This test can be used to confirm the diagnosis in someone who already has the symptoms of Huntington’s Disease (referred to as confirmatory testing). It can also be used to test people who are “at-risk” for the disease - people who have no symptoms but are “at-risk” because they have a parent or sibling with the disease. This type of testing is referred to as predictive testing. Predictive testing cannot tell you if you currently have the disease - but it can tell you if you have the gene expansion that can lead to the development of the disease. The HD DNA test is also used for prenatal testing. This involves direct gene testing of an existing pregnancy, and pre-implantation testing.

Our HD Center of Excellence follows the guidelines for genetic testing recommended by the Huntington’s Disease Society of America, the United States Huntington’s Disease Testing Group, and the National Society of Genetic Counselors. Genetic testing should be a process. It is more than just finding out the results of a ‘blood test’. There are many issues at stake - financial, emotional, and social issues that involve not only the person seeking testing but his/her immediate and extended family as well. Prior to giving a blood sample, a person seeking testing needs the opportunity to examine these issues thoroughly with people experienced in genetic testing.

What is the process?

If you or your family member currently have symptoms of HD, then you will be scheduled in our Neurology Clinic for an initial evaluation. During that visit you will see a neurologist who specializes in HD, a nurse, and a social worker. You will have a comprehensive evaluation, and follow-up care as indicated. We
have a team of HD experts available through our Center of Excellence Clinical HD Program, and we can help you and your family member manage the symptoms and problems associated with the disease. A blood test may be done at this visit to confirm the diagnosis.

If you have no symptoms of HD or only possible symptoms of HD and are at-risk for the HD gene expansion, you will be referred for predictive testing. The process for predictive testing involves a minimum of 3-4 visits to different members of our testing team. You will have a visit in the Neurology Clinic with Dr. Wheelock and Terry Tempkin, Nurse Practitioner, for a neurological evaluation. You will meet with our genetic counselor, Mara Sifry-Plat. There is also a visit with our psychiatric clinical nurse specialist, Sally Klein, RN, MS who evaluates mood and cognition. These visits are spread out over a few months to allow the participant time to process the information obtained at the visit – and for the simple reason of scheduling the providers’ time. If after all the visits the participant still wants to proceed with testing, and there has been no problem identified indicating that it would be advisable to wait, the participant returns to have the blood sample taken. It takes 3-4 weeks to process the lab specimen, and all results are given in person. Results are never disclosed to a third party and never given over the phone. Follow up is arranged at the time the results are disclosed.

Many people feel very anxious about starting this process and want to proceed immediately to the blood draw, bypassing the counseling and the evaluations. There are several reasons why we cannot accommodate this. The recommended guidelines for genetic testing require the components of evaluation and counseling and cannot be by-passed – no matter how certain the person is that he/she wants to test. Most people find the process helpful and learn things they hadn’t considered prior to beginning the testing process. Multiple visits allow the participant the opportunity to hear information from different specialists – helping them make a more informed decision about testing. The process allows the participant to get to ‘know’ the testing team, so when results are reviewed, it is from providers who have helped them through the process – not strangers.

We are sensitive to people’s sense of urgency, and we acknowledge it takes an enormous degree of courage to pick up the phone and make the first call. That is why we take our responsibility so seriously and adhere strictly to the required elements of the testing process.

Occasionally it is necessary to delay the testing process. If a significant issue with mood, emotional stability, lack of support or an unsafe social situation is revealed, the team will recommend postponing testing until the issue(s) can be
addressed and resolved. The team will make recommendations for follow up and indications to resume the testing process.

Cost

Confirmatory testing is often billed to the patient’s insurance as part of his/her clinic visit. **We do not recommend third party billing for predictive testing.** People undergoing predictive testing usually do this anonymously and pay for it privately. If insurance is billed for a predictive test, then the insurance carrier of that person as well as any future insurance carriers would be privy to the information of the test results. We feel the person testing should be in control of who gets testing information. Testing information has implications for people other than the person testing. It can be used discriminatorily with spouses, children, and siblings as well.

Cost varies, but it is generally about $1,000.00. This includes the evaluations, counseling, the blood test, and the follow up visit for results. We realize that cost may be a barrier for some, and we will work with you to develop a plan. We wish we could offer this for free, but we are unable to use the health system resources without some form of re-imbursement.

**What the results mean**

The gene is known as the IT-15, or huntingtin gene. The abnormal HD gene contains an expanded and unstable DNA segment, which is composed of the genetic code message, ”CAG” repeated a number of times in a row. The repeating CAG fragment is longer on the HD chromosome than on the normal chromosome and may change in length when it is passed to offspring. This trinucleotide, CAG, codes the amino acid glutamine. Worldwide experience suggests the following interpretations for the results of HD genetic testing:

<table>
<thead>
<tr>
<th>CAG REPEAT SIZE</th>
<th>INTERPRETATION</th>
</tr>
</thead>
<tbody>
<tr>
<td>26 and below</td>
<td>Normal</td>
</tr>
<tr>
<td>27-35</td>
<td>Normal but potentially unstable</td>
</tr>
<tr>
<td>36-39</td>
<td>Abnormal, variable penetrance; unstable</td>
</tr>
<tr>
<td>40 and above</td>
<td>Huntington disease</td>
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To clarify this further, any number of CAG repeats that is less than or equal to 26 is considered normal. Within this range, the size of the CAG repeat segment also
appears to be stable, i.e. does not appear prone to expansion. CAG repeat lengths within the range of 27-35 are also normal, in that they are not associated with symptoms of HD. However, the CAG repeat length can be unstable in this range and can increase, so that a parent with a repeat number in this range can have a child whose repeat number is in the HD range. If the number of CAG repeats is within the range of 36-39, it cannot be predicted with certainty whether or when HD symptoms will develop. Within this range, some people have been found to have classic symptoms of HD, while others have lived to be very old without developing the symptoms of HD. The gene is unstable in this range and may expand so that a child may have a number of CAG repeats that is clearly within the HD range. CAG repeat lengths of 40 or greater are virtually always associated with the development of the symptoms of HD at some time during a normal life span.

It is important to remember that finding out you carry the gene expansion does not indicate when you will get the disease. There are many factors that modify the onset and severity of symptoms.

Confidentiality

Confidentiality is of utmost concern to the testing team and the individuals undergoing predictive testing, for whom the untimely release of private genetic information could have serious adverse effects on personal and professional relationships, stature in the community, or self-esteem. Our testing center will work with you to assure your privacy to the best of our ability. Many people choose to test using a pseudonym (a false name). We can work with you to establish this.

Test results will not be divulged to anyone other than the person undergoing testing. Only in exceptional circumstances, such as prolonged coma or death, may information about an individual’s gene test result be released to the next of kin.

A Companion

We strongly encourage you to identify a companion (such as a spouse, relative or trusted friend) to accompany you through the testing process. The companion by being physically present during the counseling and evaluation sessions can gain insight into the person’s testing experience and thus become a uniquely valuable source of support. We also encourage you to free the entire day you get results. Whether your test has negative or positive results, it will certainly be emotional – and our experience has told us that it is good to have unencumbered time to process the information.
The Team

**Vicki Wheelock, MD** is a board certified neurologist who specializes in movement disorders including Huntington’s Disease. She is the director of the HD clinical team, and Director of the HDSA Center of Excellence at UC Davis Medical Center. She is an active investigator with the Huntington’s Study Group.

**Mara Sify-Platt, MS** is a certified genetic counselor with experience in HD. In addition to her work with genetic counseling, she attends the multi-disciplinary HD clinic to evaluate individuals and families.

**Terry Tempkin, RNC, MSN** is the nurse practitioner for the HD clinical and research programs. She is part of the HD Clinical Team and the study coordinator for all the HD research projects at the Center of Excellence. She is often the initial point of contact for the testing process and will be instrumental in managing the testing process.

**Sally Klein, RN, MS** is a psychiatric clinical nurse specialist. She has over 10 years experience as a part of the HD testing team.

For additional information about genetic testing, or to schedule an appointment, please call the Center of Excellence at (916) 734-6278.