

Genetic Testing Program for Huntington's Disease

Genetic testing for the Huntington's Disease (HD) gene expansion became possible in 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 HD, 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 HDSA Center of Excellence follows the guidelines for genetic testing recommended by the Huntington's Disease Society of America, the US Huntington's Disease Testing Group, and the National Society of Genetic Counselors. Genetic testing is 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?

Confirmatory testing: If you or a family member currently **have symptoms** of HD, then we recommend that you be seen in our HD Clinic for an initial evaluation. During that visit you will see a neurologist who specializes in HD, a genetic counselor, nurse, and a social worker. You will have a comprehensive evaluation, and our team will discuss diagnosis and develop a plan of care for you. Our team of HD experts can help you and your family member manage the symptoms and challenges associated with HD. A blood test may be drawn at this visit to confirm the diagnosis, and the results will be shared at your follow-up appointment one month later.

Prenatal testing: Testing for an unborn fetus is performed by another department at UC Davis in OB-GYN. Our team can help you access those resources if required.

Pre-Symptomatic Predictive Testing: If you have no symptoms of HD, are at-risk and want to know your gene status, you will be referred for **predictive testing**. Please visit this link for a short video called :

“Predictive testing for Huntington's disease,” discussing basic information about predictive testing: <https://www.youtube.com/watch?v=4HW5YdqM4zs>

The process for predictive testing involves an initial phone consultation and two additional in person clinic visits with our HD Team.

Telephone consultation. Many people have a lot of questions about pre-symptomatic testing that can be discussed in the initial phone consultation. There are many things to consider including: age, insurance and work status, costs, emotional preparedness, timing of testing, how testing may affect your work, life plans and relationships, etc. Review of this document also gives you additional opportunity to consider if this is the right step for you at this time, and to share the information with any other person who is helping you with this process. After the phone interview and review of documents, if you want to proceed, our team will set up the first clinic appointment.

First In-person clinic visit: At the initial in- person visit you will meet with our genetic counselor, social worker and neurologist. A family history and review of your medical history will be taken. Our social worker will ask you questions about your support system, daily activities and mood. The neurologist will do a physical/neurological examination. As you go along, each person on the team will review information with you and answers your questions.

Generally people will have their blood drawn in the laboratory at the end of this visit. Depending on your circumstances, the predictive testing team may recommend that you get additional care (counseling or medical follow up) before the blood is drawn. If that is the case, our team will develop that plan with you. This visit will last 1-2 hours, with additional time for the blood draw.

The DNA blood sample is drawn in the clinical laboratory at UC Davis and sent to a specialized molecular diagnostics laboratory for processing. It takes 3-4 weeks to process the specimen. We will set up your next in-person appointment for results disclosure before you check out from the clinic.

Second in-person visit. The second in- person visit is for results disclosure. 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. Our social worker or genetic counselor will call you within a month after your results to see how you are doing.

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.

We always recommend that people who are considering predictive testing **consider obtaining disability, long term care and life insurance if they are eligible**. The federal Genetic Information Non-discrimination Act (G.I.N.A.) offers protection against discrimination for employment and health insurance on the basis of genetic information, but it does not cover disability or life insurance.

Anonymous testing for HD: using a pseudonym: Some people seeking HD testing prefer to do so anonymously. We offer anonymous testing at UC Davis, but it can only be used when people agree to pay privately for testing. The anonymous testing process cannot be used for people who choose to use their health insurance. The total costs for anonymous testing vary, but are generally about \$1000 for the counseling visits and the blood test.

If you choose to test anonymously, we will take the following steps:

- Create a secure medical record with a unique alpha-numeric code (for example, AA0999) for this testing process.
- Your name or social security number will not be in the electronic medical record.
- Your address and date of birth will be included.
- Your counseling session notes, examinations and test results will be stored in the electronic medical records under your assigned unique alpha-numeric code.
- This medical record has all the same protections that any medical record would within any health system and cannot be shared with any other entity without your express written permission.

Cost

Confirmatory testing is typically billed to the patient's insurance as part of his/her clinic visit.

Predictive Testing can be done using your insurance or through the anonymous testing process described above.

People undergoing predictive testing usually opt to use our anonymous testing protocol. This requires that they agree to pay privately for the costs. If insurance is billed for a predictive test, then the insurance carrier as well as any future insurance carriers would be privy to the information of the test results. We feel the person who chooses to undergo predictive testing should be in control of that information.

Cost varies, but it is generally about \$1,000.00. This includes the clinic visit for evaluation and 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 re-imbusement.

What the results mean

If you have not already viewed the information video “Predictive testing for Huntington’s disease,” please visit this link, which includes a discussion of basic information about the interpretation of predictive testing results:

<https://www.youtube.com/watch?v=4HW5YdgM4zs>

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 or expanded on the HD chromosome than on the normal chromosome and may change in length when it is passed to offspring. Worldwide experience suggests the following interpretations for the results of HD genetic testing:

CAG Repeat length	Interpretation
26 and below	Normal
27 - 35	Normal but potentially unstable
36 - 39	Abnormal, variable penetrance; unstable
40 and above	Huntington’s disease

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.

CAG repeat lengths within the range of 27-35 are also normal, but potentially unstable. This has implications for future generations, and is best discussed with the genetic counselor during your visit.

If the number of CAG repeats is within the range of 36-39, it cannot be predicted with certainty whether or if 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. Our testing center will work with you to assure your privacy to the best of our ability, but of course, we cannot absolutely guarantee electronic security. We also create an additional file *outside* the electronic medical record on a password protected

secure server that can only be accessed by the testing team members. This outside list connects the testing number with the name of the individual in case anyone on the team or someone who has tested would need to obtain the test result and may have forgotten their pseudonym identity.

Our team takes your privacy very seriously. This is how our process works:

Anonymous testing: We create the electronic medical record under a pseudonym (for example AA0999) instead of your name. We also keep a paper “shadow file” locked in our office that DOES have your name and contact information. We have to be able to match who the pseudonym is with the real person for several reasons:

- We must be able to contact you to schedule your visit(s)
- You (and not anyone else) may at some future date request an additional copy of your test result (you receive one at your disclosure visit)
- If you test positive, you may ultimately need care. Your Pseudonym chart could be converted to your real name at that point (with your permission, of course).

It’s important to note that even though we take every precaution to de-identify you through this process, we can’t personally guarantee that a breach of health records would never occur. No health system can ever make that claim.

What is important in this process is if you are paying out of pocket for the test and the visits, no one else will be billed and no one else has the authority to request that information unless YOU give your express written permission. The clinic will not request insurance information from you if you are registering under a pseudonym. As long as you don’t have any other medical records under your name at UC Davis that has insurance information in it, UC Davis would have no one else to bill and the pseudonym record is not released to a third party.

We have tried to make this process as private as we possibly can, but at the end of the day there can’t be a 100% guarantee. Only you can decide if that fits in your comfort zone.

Reminder: Anonymous testing can only be performed when people agree to pay privately, and cannot be carried out with billing to third-party payers.

It is also important to remember that if you choose to undergo anonymous testing your test result will be under your pseudonym. If it is positive and you subsequently receive care here, we can convert it to your real name when it is appropriate to do so with your written permission. If you receive care outside of this system, we cannot make the name conversion which may result in you needing another gene test down the line.

Testing under your insurance: For a variety of reasons some people chose to undergo testing with billing of the costs covered in part or whole their health insurance. The privacy of your health care information is protected by both the federal Health Information Portability and Accountability Act and by the Genetic Information Non-Disclosure Act.

Here are links for more information:

- HIPAA: (see <http://www.hhs.gov/hipaa/for-professionals/privacy/>)
- GINA: (see <http://www.hhs.gov/hipaa/for-professionals/privacy/>)

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, hears the same information and can gain insight into the testing experience. It helps them understand what you are going through and increases their ability to be supportive both during the testing process and after.

We also encourage you to free the entire day you get results. Whether your test result is positive or negative, it will certainly be emotional. Our experience has told us that it is good to have unencumbered time to process the information.

The Team

Vicki Wheelock, MD is a neurologist specialized in Huntington's Disease. She is the director of the HD clinical team, and Director of the HDSA Center of Excellence at UC Davis. You will meet with her or Dr. Duffy at the first visit as part of the evaluation.

Sasha Duffy, DO is a neurologist specialized in Huntington's Disease. She is part of the clinical team for the HDSA Center of Excellence at UC Davis. She and Dr. Wheelock share the responsibility of the neurological evaluation in the testing process.

Lorin Scher, MD is a psychiatrist and is the co-Director of the HDSA Center of Excellence. He is also the HD Clinic Psychiatrist. Although not routinely doing evaluations in the testing process, he works with our Team Social Worker as a consultant.

Mara Sify-Platt, MS is a certified genetic counselor with experience in HD. She is part of the UC Davis Center of Excellence Team and counsels families in clinic as well as individuals going through the testing process.

Terry Tempkin, NP-C, MSN is the nurse practitioner in the HD Clinic. She and the social worker share the responsibility of responding to the initial phone inquiry for the testing process and will work with the clinic admin staff to set up the testing process.

Lisa Mooney, LCSW, is the social worker for the HD Clinic. She performs a mental health assessment as part of the testing process. Lisa can also provide information to families on the testing process

For additional information about genetic testing, or to schedule an appointment, please call the Center of Excellence at (916) 734-6278 or (916) 734-6277.