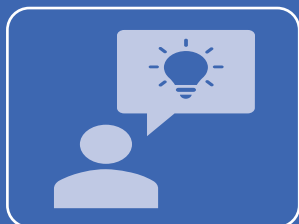


Genetic Testing Program

Mara Sifry, LCGC
Lisa Mooney, LCSW

Genetic testing for the Huntington's Disease (HD) gene expansion became possible in 1993. The UC Davis HDSA Center of Excellence follows the guidelines for genetic testing recommended by HDSA, the US Huntington's Disease Testing Group, and the National Society of Genetic Counselors. Genetic testing is a process and 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 other family members 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.

PREDICTIVE TESTING



This testing is completed IF:

*the person has no symptoms of HD,
is at-risk for HD,
AND wants to know their gene status.*

Testing can be an emotional process and many aspects should be considered before engaging in predictive testing.

RESOURCE: 11 minute video that explores the basic information about predictive testing:
<https://www.youtube.com/watch?v=4HW5YdgM4zs>

PRENATAL TESTING

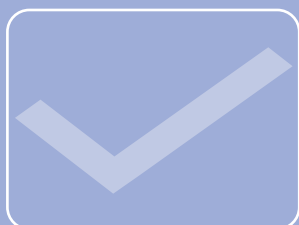


Testing for an:

unborn fetus to determine if it has inherited the HD gene expansion.

This can be a very complex and emotional process so we encourage you to discuss the procedure, issues, and other things to consider with a medical professional knowledgeable about HD.

CONFIRMATORY TESTING



This testing completed IF:

*symptoms of HD are suspected,
includes a comprehensive medical evaluation and exam
will discuss diagnosis and develop a plan of care for you.
blood test may be drawn at this visit to confirm the diagnosis, and the results will
be shared at your follow-up appointment.*

This is often an emotional process. The HD Professionals can help guide you through and provide support and you maneuver through the process.

Things to consider **BEFORE** testing:

<h3 style="text-align: center;">Your Feelings/Needs</h3> <p>You will likely experience an array of emotions, which is NORMAL and COMMON when considering testing for something that will change your life.</p> <p>Common feelings: sadness, depression, anxiety, anger, fear, overwhelm, frustration, urgency, guilt, regret, confusion, etc</p>	<h3 style="text-align: center;">The Impact on Your Life</h3> <p>Consider your age, insurance needs, work/school status, costs, emotional preparedness, timing of testing, & how testing may impact your life</p> <p>Take time to evaluate if this is the RIGHT time for you to start this process.</p> <p>Consider obtaining disability, long term care and life insurance as results may hinder ability for coverage after testing</p>	<h3 style="text-align: center;">Resources</h3> <p>11 minute video that explores the basic information about predictive testing: https://www.youtube.com/watch?v=4HW5YdgM4zs</p> <p>Talk with family, friends, mentors, or other at-risk persons to get why they did or did not test at this time? HDSA, HDYO, HD Social Worker</p>
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Predictive Testing Process:

<i>Phone Consultation & Scheduling</i>	<i>Visit 1 Initial Assessment</i>	<i>Visit 2 Results Disclosure</i>
<ul style="list-style-type: none"> • Discuss process & get general information, HD family history, experience, concerns, etc • Insurance info discussed • Question and answers • Schedule first visit • Watch video before first in person visit 	<p>Meet & participate in assessments from HD Team:</p> <ul style="list-style-type: none"> • Genetic Counselor, Social Worker or Mental Health Professional & Neurologist <p>Assessments include:</p> <ul style="list-style-type: none"> • A family history • medical history • support system • coping & mental health history • physical/neurological examination • provide resources & recommendations for support & coping during this process. • Informed consent obtained • blood drawn is at the end of this visit. • This visit will last 3+/- hours. 	<ul style="list-style-type: none"> • Test results provided • Discussion of what results mean for you • Time to process and ask questions IF you have them. • Follow up options and recommendations • This visit will last 1+/- hour. • 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 shown us that it is good to have unencumbered time to process the information.

Anonymous testing OR testing using insurance and/or legal name

	Anonymous Testing	Testing using Name and/or Insurance
<i>Medical Documentation</i>	<ul style="list-style-type: none"> Legal name, address, phone number, date of birth is collected and stored in writing for the HD team. Information is stored electronically using alpha-numeric code that we assign, birthdate and address. Medical assessment and counseling session notes, and test results will be stored in the electronic medical records under your assigned unique alpha-numeric code. 	<ul style="list-style-type: none"> Legal name, birthdate, SSN, address, phone number, insurance provider Medical assessment and counseling session notes, and test results will be stored in the electronic medical records under your name Insurance provider (if used) will have access to this information
<i>Process</i>	<ul style="list-style-type: none"> 1 telephone consult 2 medical visits 	<ul style="list-style-type: none"> 1 telephone consult May require referral from Primary Care Doctor for insurance authorization 2 medical visits
<i>Time Commitment</i>	<ul style="list-style-type: none"> Depending on the team schedule, holidays and your schedule, on average we can schedule people 2-4 weeks from the date of the phone consultation You may always postpone or wait longer to get your test results 	<ul style="list-style-type: none"> Again, dependent on team schedule, holidays, your schedule AND lab processing, we can schedule Visit 2 no earlier than 4 weeks after visit 1 You may always postpone or wait longer to get your test results.
<i>Costs</i>	<ul style="list-style-type: none"> \$1200 total <ul style="list-style-type: none"> includes counseling, medical assessment & blood test as of March 2021 <ul style="list-style-type: none"> \$200 deposit at visit 1, \$100 at visit 2, and then you are billed the remainder to the address on file. Bill would be sent to your alpha-numeric code as "name" and address on electronic record 	<ul style="list-style-type: none"> \$1200 same as anonymous program, if paying privately (w/out insurance) Insurance costs would be based upon your insurance provider. You will be responsible for any co-pays, premiums or services NOT covered by your insurance provider.
<i>Recommendations</i>	<ul style="list-style-type: none"> Bring a companion (friend/family) to each medical visit for additional support both in the assessments, as well as for results disclosure appointment. 	<ul style="list-style-type: none"> Bring a companion (friend/family) to each medical visit for additional support both in the assessments, as well as for results disclosure appointment.
<i>Privacy/Confidentiality</i>	<ul style="list-style-type: none"> 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. 	<ul style="list-style-type: none"> 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.

	Anonymous Testing	Testing using Name and/or Insurance
	<ul style="list-style-type: none"> ○ Health Information Portability and Accountability Act (HIPAA): http://www.hhs.gov/hipaa/for-professionals/privacy/ ○ Genetic Information Non-Disclosure Act (GINA) GINA: http://www.hhs.gov/hipaa/for-professionals/privacy/ ● We do have the written file that connects your name, DOB to your electronic record for the purpose of: <ul style="list-style-type: none"> ○ contact you to schedule your visit(s) ○ IF you need access to the information at some future date 	<ul style="list-style-type: none"> ○ Health Information Portability and Accountability Act (HIPAA) and by the Genetic Information Non-Disclosure Act (GINA). ○ HIPAA: http://www.hhs.gov/hipaa/for-professionals/privacy/ ○ GINA: http://www.hhs.gov/hipaa/for-professionals/privacy/
<i>Things to consider:</i>	<ul style="list-style-type: none"> ● Required to pay privately as we CANNOT bill to third party payers. ● Test results will list ONLY your unique alpha-numeric code as the “name”, therefore, in the event you test positive and for future HD care may require a test under your legal name. <ul style="list-style-type: none"> ○ <i>For example for social security or supplemental insurance you would NOT be able to submit this test as proof of disease or no disease as your legal name is not listed.</i> ○ <i>In some cases, depending on need you can request a formal letter from UC Davis HDSA Center of Excellence to link your identity to the alpha-numerical code.</i> 	<ul style="list-style-type: none"> ● 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.
<i>Testing Delays</i>	<ul style="list-style-type: none"> ● On RARE occasion 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 identified. However, this is a discussion not an “order” between you and our team to postpone 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. 	

Interpreting Results

The gene is known as the IT15, HTT, 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 causing gene than on the normal gene 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 and Interpretation

- 26 & Below -- Normal
- 27 - 35 Normal, but potentially unstable
- 36-39 Abnormal, variable penetrance; unstable
- 40 & Above, abnormal, fully penetrant, unstable

Gene Negative

26 or less is normal. In this range, the size of the CAG repeat segment also appears to be stable.

Will NOT get HD or pass to future generations

Gene Positive/Expanded

40 CAG or greater are associated with the development of HD symptoms at some time during a normal life span.

Each child would have 50/50 chance of inheriting the gene expansion

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.

The "Gray" area:

27-35 CAG are called Intermediate

Considered normal, but potentially unstable.

This has implications for future generations, and is best discussed with a HD genetic counselor.

The "Gray" area:

36-39 CAG are called reduced penetrance

Considered unstable, and cannot be predicted with certainty 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.

This has implications for future generations and is best discussed with a HD genetic counselor



Our Team

Dr. Wheelock

- Neurologist specializing in HD
- Director of the UC Davis HDSA Center of Excellence

Dr. Duffy

- Neurologist specializing in HD

Mara Sifry

- Licensed Genetic Counselor
- HD experience and knowledge

Lisa Mooney

- Social Worker
- HD experience and knowledge

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



Frequently Asked Questions

What are my testing options?	<ol style="list-style-type: none"> 1. Not test – only about 10% of those at-risk choose to test 2. Anonymous Test 3. Test using insurance or private pay, but test under legal name
I know about HD, can I skip the counseling and just get the blood test?	<p>While we are sensitive to the feeling of urgency and we know it takes courage to start this process, our testing process follows the best practices guidelines for HD genetic testing that have been identified as important for the overall experience and support of the person requesting testing.</p> <ul style="list-style-type: none"> • Most people find the process helpful and learn things they hadn't considered prior to beginning the testing process. • Multiple visits allow you to gather information you need to make an informed decision about testing. • This process allows you to get to 'know' our team, so when results are reviewed, it is from providers who are familiar with you and your story.
How is the blood test processed?	<p>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 several weeks to process the specimen and schedule you for your results visit.</p>
If I change my mind and don't want to continue the process, what do I do?	<p>Testing is a personal decision and you can STOP the process at any time.</p> <p>IF you have scheduled an appointment and do not wish to proceed, please call the HD team to cancel.</p> <p>If you have not scheduled, yet did have the phone consultation, you are NOT obligated to inform us of your decision unless you want to.</p> <p>Our team does not reach out to at-risk persons unless they have contacted us first.</p>
How much does the anonymous test cost?	\$1200 total
Can I do this process over video?	<ul style="list-style-type: none"> • Maybe – please discuss your needs and concerns directly with the HD team. When possible and appropriate we will do our best to accommodate your needs. • Those testing anonymously, must have their blood draw at a UC Davis lab.
Can I get my results over the phone?	No, results are provided in-person or via video.
Can someone else get my results?	Results are never disclosed to a third party

Do I have to have a companion?

- This is a highly emotional process, that those wanting to test can often underestimate how they may feel during the process, therefore we strongly encourage you to identify a companion (such as a spouse, relative or trusted friend) to accompany you through the testing process. By having a companion physically present during the counseling and evaluation sessions, they hear 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 do NOT recommend bringing children (of any age to this process as they have a vested interest in the outcome) thus may not be able to be present for your needs. In the same vein we don't recommend bringing untested siblings because again they have their own gene status to process and may NOT be able to be present for you and your needs during the process.

Who can I call for support during this process?

- YOU are always welcome to reach out to our HD team at any time for support, coping, questions or needs.
- Lisa Mooney, HD Social Worker @ 916-734-6277 or e-mail lmooney@ucdavis.edu
- Other resources are:
 - Support groups
 - HD social workers
 - HDSA.org
 - HDYO
 - Help4HD
 - Faith-based support
 - Mental Health Provider through Employer Assistance Program or Private Pay



Additional Resources

[UC Davis HDSA Center of Excellence](#)

[HDSA.org](#)

[HDYO](#)

[HD Support Groups](#)

[HD Social Worker](#)