ARUP LABORATORIES

Frequently Asked Questions About

HUNTINGTON DISEASE (HD)

What is Huntingon Disease?

Huntington disease (HD) is a condition that affects the brain and nervous system. People who have HD will experience gradual physical and mental changes. Physical symptoms may include uncontrolled movements (i.e., chorea), balance and coordination problems, twitching, slurred speech, and problems swallowing. Mental symptoms may include changes in behavior and mood (e.g., becoming more easily irritated or angry), memory and concentration problems, and issues with thinking and judgment. HD is a progressive condition—symptoms gradually get worse over time. Affected individuals typically develop symptoms between 30 and 50 years of age, but the age of symptom onset ranges from 2 to 90 years.

Does HD run in families?

Yes, HD is an autosomal dominant condition that is most often inherited from a parent. People with HD have a 50% likelihood of passing the condition on to each of their children.

How is HD inherited?

HD is caused by a change in the *HTT* gene. This gene controls the production of a protein found in nerve cells in the brain. In the *HTT* gene, there are sections of repeating nucleotides (abbreviated as CAG). Everyone is born with a certain number of CAG repeats. Individuals normally have 26 or fewer CAG repeats and are not at risk of developing HD. HD correlates with the following ranges in CAG repeats:

- Individuals with 27–35 CAG repeats will not develop HD, but males have an estimated 2.5% chance of having children with HD (due to expansion that can happen when males pass on the *HTT* gene).
- Individuals with 36–39 CAG repeats are at risk for developing HD. Some people with repeats in this range develop classic HD symptoms, whereas others never become symptomatic.
- Individuals with 40 or more CAG repeats will develop HD at some point in their life.

How is HD testing performed?

A blood test can determine how many CAG repeats a person has in their *HTT* gene. Because there are several other neurocognitive disorders that may present with symptoms similar to those of HD, testing is often performed to confirm a diagnosis of HD after symptom onset. A genetic test can help confirm whether an individual has this disease. An individual's diagnosis is helpful to guide treatment, plan future care, and assess risk to family members.

Testing may also be performed to determine if a person who does not have symptoms will later develop the disease (i.e., presymptomatic testing). Choosing to undergo presymptomatic testing is a personal decision and should not be made under pressure from a spouse, friends, or family members. Currently, there is no cure for HD and there are no proven treatments to delay the onset or slow the progression of the disease.

People who choose to undergo presymptomatic testing for HD should have detailed psychiatric and genetic counseling both before and after testing to ensure that they are aware of the emotional and practical issues that may accompany either a positive or negative test result. There are many HD testing centers and teams across the country who address patients' questions and concerns about HD testing. One specific recommendation is to have a support person (who is not at risk for HD) attend counseling and informational sessions with an individual who chooses to pursue presymptomatic testing.

Are there medical benefits of presymptomatic testing?

At this time, there are no medical benefits available to those who test positive for HD before they become symptomatic. Presymptomatic testing cannot predict the type or severity of symptoms, age of onset, or how quickly the disease will progress. Additionally, knowing a person will eventually develop HD will not delay the onset or slow the progression of the disease.

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Are there potential risks to presymptomatic testing?

There are recognized social and psychological risks associated with predictive testing for HD, including an increased risk for depression, regardless of the test result, potential changes in the individual's sense of self, feelings of anger, and changes in relationships with friends and family. Another potential risk is the possibility of being denied life, disability, or long-term care insurance in the future.

To address these risks and concerns, it is recommended that people seeking presymptomatic testing do so through an HD testing center or with the support of an HD predictive testing team. Refer to the HD testing guidelines referenced at the end of this document for more information.

Why is a signed informed consent document required by the lab before HD testing can be performed?

ARUP requires that any individual seeking HD testing freely consent to the test and discuss the potential risks and benefits with the ordering healthcare provider. The patient must sign an HD-specific consent form to indicate that they understand the risks and benefits and wish to be tested. The ordering healthcare provider will also sign the form to confirm that appropriate counseling was provided.

A consent form can be found on the ARUP genetics website:
aruplab.com/files/resources/forms/consent_huntington.pdf

Will ARUP perform HD testing on minors?

Children younger than 18 years of age cannot legally consent to HD testing. Additionally, there are no early treatments available and no changes would be made to medical management if they were to test positive. For these reasons, the decision to have predictive testing should belong to the individual and should be made once they reach the age of legal adulthood.

Although finding out that an at-risk child will not develop HD later in life would be a great relief to the parents and family of that child, confirming that they will develop HD puts the child at risk for psychological and social harm, as well as an increased risk for discrimination at school, at work, and in insurance matters.

For these reasons, ARUP will not perform HD testing on minors.

Healthcare providers offering genetic testing for HD should be familiar with the HD testing guidelines published by the Huntington's Disease Society of America (HDSA):
hdsa.org/wp-content/uploads/2015/03/GeneticTesting-for-HD.pdf

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