ARUP LABORATORIES

frequently asked questions about

HUNTINGTON DISEASE (HD)

What is Huntingon Disease?

Huntington disease is a neurodegenerative disease present in approximately one in 10,000 individuals in the United States. Symptoms of HD are highly variable, especially in the early stages of the disease. They may include changes in gait, twitching, clumsiness, lapses in judgment and memory, and, sometimes, behavioral changes. Classic later symptoms of HD include chorea (involuntary movements), difficulties with speech and swallowing, and impairment of cognitive abilities. Affected individuals typically develop symptoms sometime between the ages of 30 and 50, but age of onset ranges from 2 to 90 years.

What causes HD?

HD is caused by an abnormal CAG repeat expansion in the *HHT* gene. Individuals normally have 26 or fewer CAG repeats.

- Individuals with 27–35 CAG repeats will not develop HD, but males have an estimated 2.5 percent chance of having children with HD.
- Individuals with 36–39 CAG repeats are at risk for developing HD. Some people with repeats in this range develop classic HD symptoms, while others never become symptomatic.
- Individuals with 40 or more CAG repeats will develop HD at some point in their life.

How is HD inherited?

HD is an autosomal dominant disease; therefore, affected individuals have a 50 percent risk of passing on the condition to each of their offspring.

Why is HD genetic testing performed?

Testing may be performed to confirm a diagnosis of HD in a symptomatic individual. There are several neurocognitive disorders that may present with symptoms similar to those of HD. A genetic test is a way to confirm that an individual does, or does not, have this disease. An individual's diagnosis is helpful in guiding treatment, planning future care, and assessing risk to family members.

Testing may also be performed to determine if an asymptomatic individual will develop the disease. Only 20 percent of at-risk, asymptomatic individuals choose to undergo presymptomatic genetic testing for HD, and they do so for many reasons, including marriage and reproductive planning, financial and career decisions, or simply because they want to know.

Choosing to have presymptomatic testing is a very personal decision and one that should not be made under pressure from friends, spouse, or family members. Currently, there is no cure for HD and no proven treatments to delay the onset or slow the progression of the disease.

Persons seeking presymptomatic testing for HD should have detailed genetic counseling both before and after testing to be sure that they are aware of all the emotional and practical issues that may accompany either a positive or negative test result. They should also have a support person (not at risk for HD) attend these counseling sessions with them.

Are there direct medical benefits to be gained from presymptomatic testing?

At this time, there are no medical benefits available to those who test positive for HD before they become symptomatic. Knowing a person will eventually develop HD will not delay the onset or slow the progression of the disease.

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 that of being denied life, disability, or long-term care insurance in the future.

Persons seeking presymptomatic testing should do so through an HD testing center or with the support of an

HD-predictive testing team. These centers or teams provide information and counseling so that each person inquiring about testing can make a fully informed decision that is right for him or her. The HD testing guidelines referenced at the end of this document detail what a person seeking predictive testing for HD might expect.

Results of HD testing should NEVER be provided over the telephone or electronically. They should be relayed only in person.

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 he/she understands the risks and benefits and wishes to have testing performed. The ordering healthcare provider also signs the form to confirm that appropriate counseling was provided.

A consent form can be found on the ARUP genetics website:
www.aruplab.com/genetics/resources

Will ARUP perform HD testing on minors?

The Huntington's Disease Society of America (HDSA) states that "parental anxiety about a child's risk does not constitute a medically compelling reason for genetic testing." Children under age 18 cannot legally give their consent to have HD testing performed, and most would be predicted to choose not to have presymptomatic testing if the choice were presented to them as an adult. For these reasons, the decision of whether or not to have predictive testing should belong to the individual once he/she reaches the age of majority.

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

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

Healthcare providers offering genetic testing for Huntington disease should be familiar with the HD testing guidelines published by the HDSA:
hdsa.org/wp-content/uploads/2015/03/GeneticTesting-for-HD.pdf

visit our website for more information:
WWW.ARUPLAB.COM

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