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Wonder if you'll get Huntington's disease? Preparing for the big, 'intensely personal' decision to undergo predictive testing

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At Risk for Huntington's Disease

HD is a genetically caused brain disorder that causes uncontrollable bodily movements and robs people's ability to walk, talk, eat, and think. The final result is a slow, ugly death. Children of parents with HD have a 50-50 chance of inheriting the disease. There is no cure or treatment.

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Wonder if you'll get Huntington's disease? Preparing for the big, 'intensely personal' decision to undergo predictive testing

One of the most daunting challenges facing families affected by Huntington's disease involves genetic testing.

Huntington's is a 100-percent genetically caused disease, and it now can be foreseen – but not yet cured or treated. All humans have the huntingtin gene, which is essential for life. HD's devastating, ultimately deadly symptoms are caused by a specific mutation (called a "CAG repeat expansion") in the gene. Definitive testing for HD became available after the historic discovery of the gene in 1993.

Because every child of an affected HD parent has a 50-50 chance of inheriting the expanded gene, the mere decision to test is often frightful. A positive test result for the expansion means not only that the tested person will develop HD, but carries an added burden: the knowledge that both immediate and extended family members are also at risk of carrying the expansion.

Three scenarios

A person showing no symptoms, or suspecting symptoms, undergoes a *predictive* test, that is, to see whether the individual carries the expansion and therefore might have HD or later develop it. (*Diagnostic* testing confirms whether a person already displaying symptoms has HD. *Prenatal* testing determines whether a fetus or embryo carries the expansion.)

These three scenarios were poignantly portrayed in the July 3 ABC News feature "Living with Huntington's Disease." The 15-minute program focused on the stories of Scott and Kelsey Porter and Justin Furstenberg, who received his test result on camera (starkly reminiscent of the film *The Lion's Mouth Opens*.)

The report's detailed, deeply personal rendering of the genetic testing process also illustrated how HD families rely on supportive genetic counseling and psychological and medical assistance – as well as solid scientific information – to navigate the many challenges involved.

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Scott and Kelsey Porter in a Huntington's Disease Society of America video

According to recommended guidelines, individuals like the at-risk Kelsey must prepare for this procedure by speaking to a genetic counselor and a mental health professional, and should have a support person (such as a spouse or close friend) physically present throughout the process. For testing in the United States, this "protocol" was established by the Huntington's Disease Society of America (HDSA). It was most recently updated in 2016. Testing centers should do the utmost to ensure confidentiality, especially since news of a positive test can risk changing perceptions in the workplace and elsewhere, even if there are new guarantees against genetic discrimination.

Testing centers often intentionally slow the testing process, so that there is time for the individual to reconsider the decision to be tested, to think about the potential downside of testing, and to prepare for the impact of the result. Because of survivor's guilt and other psychological factors, a negative test result can also prove traumatic and disruptive to a person's relationships with family and friends.

In my quarter century of attending the local monthly HDSA support group and advocating for the HD cause, the topic of predictive genetic testing and its many implications has come up regularly. My own family faced all three modes of tests over five years: my mother's positive diagnostic test in 1995, my positive predictive test in 1999, and my daughter's negative prenatal test in late 1999/early 2000. (Click here for details of my family's fight against HD.)

Based on these experiences and my study of the many related issues, this article provides an overview of key steps and resources for people preparing for HD testing, in particular the predictive type.

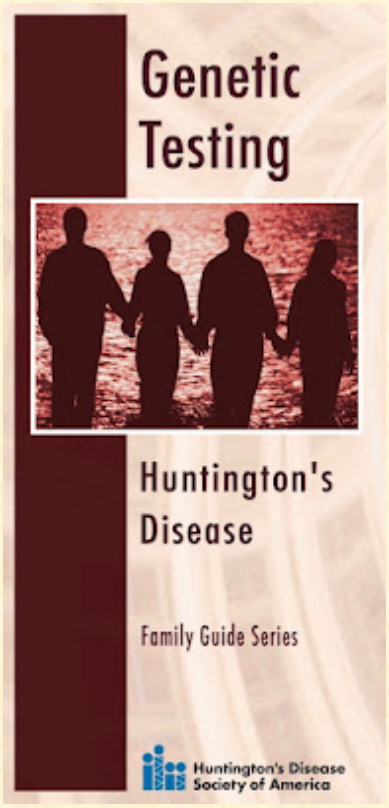
Helpful HDSA resources

HDSA, in addition to its genetic testing protocol, provides a brochure to HD families, *Genetic Testing Huntington's Disease*, that in simple language answers basic questions about the disease, testing procedures, and resources.

The brochure emphasizes a cardinal rule that I learned early in my family's journey with HD, and which I have repeated to other HD family members coming to grips with disease for the first time:

"The decision to undergo genetic testing is an intensely personal one that cannot be taken lightly. Testing should never be forced on an at-risk individual. There are

no 'right' or 'wrong' answers. Each individual will have to take his/her own circumstances into consideration before making the decision."



The HDSA family guide to genetic testing (copyright, HDSA)

The HDSA website furnishes valuable information on "genetic testing and your rights," including the Genetic Information Nondiscrimination Act of 2008 (GINA). As explained on the site, GINA prohibits "health insurance companies and group health plans from denying coverage or charging a higher premium based on genetic information." It also "prohibits employers from using an employee's genetic information to discriminate when making employment decisions about hiring, firing, promotion, or terms of employment."

In chapter 2 of HDSA's *A Physician's Guide to the Management of Huntington's Disease*, leading HD specialist Martha Nance, M.D., provides additional critical information about testing and counseling. The chapter includes a detailed medical discussion of HD genetics.

A diagnosis of HD "affects the entire extended family," Dr. Nance writes. "The person who is diagnosed with HD grieves not only for himself, but also for his at-risk children, and a young adult child caring for an affected parent understands that the parent's disease could one day affect him."

Dr. Nance stresses the importance of "accurate information" necessary for families to make "informed decisions" about genetic testing and family, financial, and life planning. Unfortunately, even decades after the discovery of the gene, "misinformation and misunderstandings" about HD genetics are still common, she notes.

(You can also watch a panel discussion titled "Looking to the Future: Life After Testing," held at HDSA's 35th annual convention, which took place online last month.)

Moving towards 'genetic education'

In 2018, the international Huntington's Disease Youth Organization (HDYO) added to its website a very readable "Genetic Testing Checklist," covering key topics such as motivation for testing, coping with the test results, the testing process, and key things to do *before* testing, such as lining up insurance coverage (discussed below). This resource echoes many of the points made in HDSA materials.

In 2019, veteran University of Washington neurologist Thomas D. Bird, M.D., published *Can You Help Me? Inside the Turbulent World of Huntington Disease*, a book based on his more than 40 years' experience seeing HD patients and their families. It includes detailed discussion of the many issues involved in what Dr. Bird calls the "genetic testing conundrum."

Individuals contemplating genetic testing will find many valuable stories in Dr. Bird's book. He describes the gamut of people's reactions to testing – from individuals who have tested negative but still require a while for it to "sink in," to (sadly) the risk for suicide among people testing positive.

"Suicide represents the cause of death in about 5-6% of persons with HD – five times higher than the national average," Dr. Bird explains. "It can happen at any time but it is most common when a person at risk decides he or she is developing symptoms."

Dr. Bird observes, crucially, that the "genetic test result is not black and white, all or nothing." This reflects the latest genetic research on HD, which has demonstrated that the age of onset of symptoms is driven not just by the severity of the mutation but also by modifier genes (click here to read more).

This is why Dr. Bird stresses a comprehensive understanding of genetic counseling.

"Some people don't like the term *counseling*," he writes. "It sounds too much like psychotherapy, and they are wary of that. In fact, genetic counseling does sometimes have a heavy dose of psychotherapy, but it entails much more. Perhaps the best word would be *education* – genetic education."

(I will review *Can You Help Me?* more fully in a future article.)

Ten key steps

With these resources in mind, I list below ten key steps in preparing for a predictive genetic test and dealing with its short- and long-term consequences. These are my personal thoughts; this list is not meant to be exhaustive or official. Individuals should always consult their physicians. Each individual's situation is unique.

1. Learn as much as you can about HD by studying the resources cited in this article, as well as others.

2. Join a support group, where you can learn from and share ideas with others confronting HD, as well as from facilitators and health professionals.

3. Contact the nearest HDSA Center of Excellence (or other HD or neurology clinic), where you can obtain information about testing and clinical services. You also can become involved in critical efforts towards treatments such as clinical trials and research studies like Enroll-HD.

4. Know your rights regarding genetic testing and healthcare access under federal, state, and local law in your country of residence, and, in the U.S., learn about GINA.

5. Obtain life, disability, and/or long-term care insurance prior to testing. GINA does *not* protect consumers in these areas. In 1999, before testing, I was able to secure a long-term care policy with lifetime coverage. Since then, the long-term care market has gone into crisis, with many fewer policies issued, and far more limited coverage (click here and here to read more). At the time, I found it very helpful to work with an insurance broker recommended by an insurance agent specializing in long-term care who had been a guest speaker at the HD support group.

6. Set up a will, an advanced directive for end-of-life care, and, if appropriate, a living will to help protect assets. Also plan for the potential impact of HD on family finances by consulting a trusted financial advisor.

7. Research and select the testing center for your genetic test, including the cost of the procedure, which can run from a few hundred dollars to more than $1,000. (Some HDSA Centers of Excellence offer free or reduced pricing on testing. One foundation has paid for in vitro fertilization of non-HD-affected embryos but temporarily suspended grants because of the COVID-19 pandemic.) Some HD family members have criticized the quality of guidance provided at some centers. Be your own best advocate, and don't be afraid to ask questions.

8. Find a trusted family member or friend to be your support person.

9. Build a relationship with a trusted psychotherapist.

10. Become active in HDSA and/or other advocacy organizations.

With potential treatments, an expected boom in testing

As the geneticist who revealed my test results in 1999 stated, "a positive test is not a diagnosis." Physicians and scientists underscore this point. Like me, many people live years and even decades after their test before symptoms start.

Currently, no more than ten percent of at-risk individuals choose to be tested. The vast majority fear a potentially depressing result, "and there is no means of prevention," Dr. Bird observes.

However, as clinical trials such as the historic GENERATION HD1 proceed, the potential for the first effective treatments has grown significantly.

Indeed, doctors and HD clinics are preparing for the likely boom in testing for the HD mutation that will occur if GENERATION HD1 or trials of other possible disease-modifying treatments are successful, as people seek to learn their status before starting on a treatment. (Click here and here to read more.)

More than ever, people seeking HD predictive testing and their families will need what Dr. Bird describes as "an experienced, compassionate team to help them through this challenge."

Posted by Gene Veritas at 11:42 PM

Labels: genetic counseling , genetic discrimination , genetic testing , HDSA , huntingtin , Huntington's disease , mutation , predictive testing , Scott & Kelsey Porter , symptoms , tested negative , Thomas D. Bird , treatment

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