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What's in a name? How Huntington's disease gene carriers are seen by themselves and others

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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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GENE VERITAS

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HD Links

Huntington's Disease Society of America

WEDNESDAY, MARCH 26, 2014

What's in a name? How Huntington's disease gene carriers are seen by themselves and by others

In 1999 I received the results of a genetic test that showed I had 40 CAG repeats on the huntingtin gene inherited from my mother, who died of Huntington's disease in 2006 after a two-decade struggle with the disorder.

Everybody has this gene, which first appeared 800 million years ago in a species of amoebae. Huntingtin helps our cells function properly.

The gene's CAG repeats refer to the sequence of three nucleotide bases – cytosine, adenine, and guanine, all building blocks of DNA – on the DNA molecule. Most people have 27 or fewer repeats. The gene I inherited from my father had fewer than 20.

My mother's high CAG count caused her to start experiencing HD symptoms – typically manifested as emotional distress, cognitive loss, and involuntary movements – in her late forties.

The term "CAG repeats" and my mother's count of 40 were two of the very first facts I learned about HD after receiving news of her diagnosis in late 1995.

The geneticist used the same terminology when he revealed my test results.

However, as he told me and many other recipients of HD test results, "a positive test result is not a diagnosis." While everybody with 40 or more repeats *will* develop HD in his or her lifetime, scientists cannot yet predict the exact moment and type of disease onset.

According to John Warner, Ph.D., the director of biostatistics for CHDI Management, Inc., which carries out the day-to-day mission of the non-profit, HD drug-discovery biotech CHDI Foundation, Inc., 95 percent of those individuals with 40 CAG repeats will experience disease onset between the ages of 50 and 74. (A future article will explore the statistical meaning of the CAG count in greater detail.)

With an ominous test result at age 39 but no symptoms, I needed to construct a definition of my genetic predicament for both myself and for others.

As I said recently in an interview, unlike treatments for certain kinds of cancer, I cannot irradiate my defective huntingtin gene to destroy it. It's part of me, literally residing in every cell.

Because of its genetic nature, HD also requires a far more nuanced kind of diagnosis. Subtle symptoms can exist for years before the more noticeable symptoms commence.

International Huntington Association
Huntington's Disease Drug Works
Huntington's Disease Lighthouse
Hereditary Disease Foundation
Huntington's Disease Advocacy Center
Thomas Cellini Huntington's Foundation
HDSA Orange County (CA) Affiliate
HD Free with PGD!
Stanford HOPES
Earth Source CoQ10, Inc.

HD Blogs and Individuals

Chris Furbee: Huntingtons Dance
Angela F.: Surviving Huntington's?
Heather's Huntington's Disease Page

'Gene-positive'

For many years, I referred to myself as "gene-positive for Huntington's disease," a term I heard often in HD family and scientific circles. I also used phrases such as "tested positive for HD."

"Gene-positive" echoed the term "HIV-positive" used by the AIDS community. It meant not only that I had tested positive for a condition, but that I inevitably faced its dire consequences.

Thus, "gene-positive" resonated with the deep stigma, discrimination, and alienation suffered by members of both the AIDS and HD communities.

"Gene-positive" further implied an activist stance. As with the early years of the fight against AIDS, we in the HD community needed to tell the world we needed treatments and the resources to find them.

I experienced all of these feelings in the late 1990s and early 2000s, as I immersed myself in advocacy work for the Huntington's Disease Society of America.

They remain with me today as we still await the discovery of an effective treatment.

Changing perceptions

As my knowledge about HD increased, and as I came into ever closer contact with HD researchers in labs and at events such as the annual CHDI-sponsored HD Therapeutics Conference, both my perceptions of HD and the terms I used to describe my situation changed.

As I learned to my first visit to CHDI in 2009, many scientists see gene-positive individuals as genetically and, at least at the cellular level, even functionally compromised from birth.

I started to hear scientists used the word "premanifest" to describe asymptomatic, gene-positive individuals.

Soon I would be introduced to "prodrome" and "prodromal". A precursor or forerunner to the disease, prodrome refers to the period before onset.

However, I could never imagine using such a technical term to describe myself to others.

Scientists and physicians also used "asymptomatic" and especially "presymptomatic" to describe people like me. I have frequently used the former to indicate to people that I face the danger of HD but am fine for now.

Other phrases I have used or heard include: *HD gene carrier; HD gene mutation carrier; asymptomatic HD gene carrier; disease-gene carrier; tested positive for the genetic defect that causes Huntington's disease;* and *carry the gene for Huntington's disease.*

Living with the 'phantom gene'

At the World Congress on Huntington's Disease in Rio de Janeiro last September, HD activist, historian, and author Alice Wexler, Ph.D., noted that much recent scientific discussion has focused on defining when HD actually begins.

During a panel on coping with HD, Dr. Wexler asked how global HD advocate Charles Sabine and I – both gene-positive but asymptomatic –

viewed ourselves as individuals living with the "phantom gene" and in what circumstances would consider ourselves as having HD.

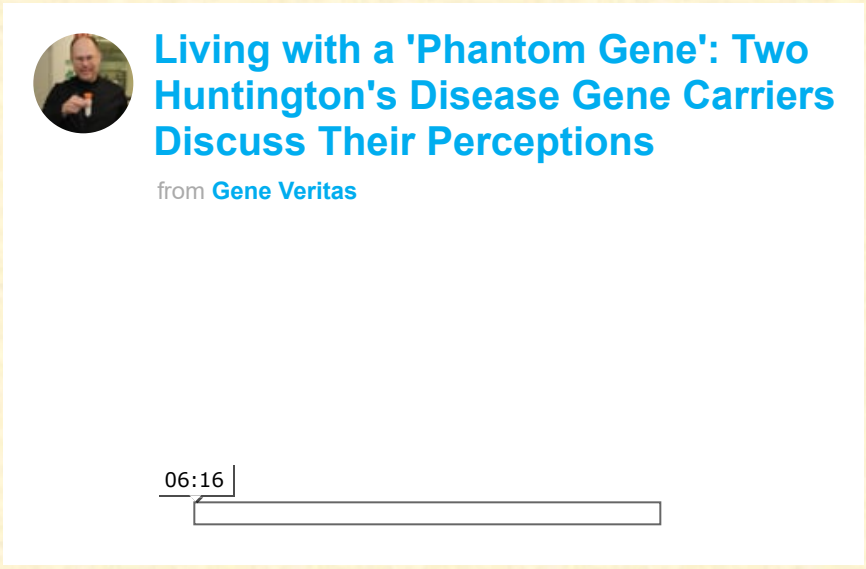
"It changes for me depending on where I am," I replied. "If I'm at a conference like this: 'Oh, my God! I have HD.' Because I see all these studies and brain scans and searches for biomarkers ... and references to me as prodromal.... There's a tendency of the scientific community to see gene carriers as diseased from Day One."

In settings such as my doctor's office, I felt different, I said. "My doctor's telling me: this time you got a clean bill of health."

Charles, agreeing with my outlook and saying that he "treasured" his current good health, answered the question in a "wider, more metaphysical sense."

"We are not just someone who's had a bit of bad luck," Charles said about having inherited the HD mutation. "We are a part of history. I have absolutely not a single shred of doubt in my mind that, whether it's 20, 50, or a 100 years [off], that this disease will be managed just like HIV-AIDS can be now."

You can watch the entire exchange in the video below.



Living with a 'Phantom Gene': Two Huntington's Disease Gene Carriers Discuss Their Perceptions from Gene Veritas on Vimeo.

A new shorthand

The latest conception emerged at the CHDI-sponsored HD therapeutics conference in Palm Springs, CA, last month, where Andrea Varrone, M.D., Ph.D., of the Karolinska Institutet (Sweden) gave a presentation whose title included the phrase "Huntington's disease gene expansion carriers."

That phrase very accurately describes someone like me, because it specifically identifies the cause of the disease: an expansion of the huntingtin gene. However, the term does not by itself identify whether a person is symptomatic or asymptomatic.

Nevertheless, it's good shorthand for the concept of expanded CAG repeats.

However, both the phrase and its acronym, HDGEC, are a mouthful! They might not resonate with the community, and even less so with the general

public, which is more familiar with the idea of a "mutated gene" than with the term "expanded gene."

'You don't look like an HD person'

The abundance of terms to describe asymptomatic HD gene carriers reminds me that those of us in this predicament are undergoing "the new and harrowing human experience of living in the gray zone between a genetic test result and the onset of a disease foretold."

Scientists have demonstrated that changes in the brain occur ten and even 20 years before onset – meaning that my brain may already be seriously compromised, even though I function just fine.

Inexorably, perniciously, but silently, HD attacks the brain.

However, it's not discernible from the outside.

"You don't look like a person who has Huntington's disease," a health professional told me recently as I contemplated him writhing with pain and discomfort from a knee operation that forced him to wear a brace and use crutches.

There is no particular way for a premanifest person to look! Moreover, no "crutch" yet exists to help the presymptomatic HD brain recover from the initial assault on the cells.

As an HD gene carrier and advocate for this orphan neurological disorder, I continually face the challenge of explaining the seriousness of the disease and its many social implications.

Along with other neurological disease communities, we in the HD community are still searching for the right formula to project the urgency and significance of our predicament.

A temporary escape

Often those of us in the gray zone prefer not to deal with HD. Unlike others in the community, we don't yet face the minute-by-minute struggle with symptoms.

At the local HD support group meeting this week, I was the only at-risk individual to appear. Even so, the facilitator and her replacement-in-training for the at-risk section (which normally includes both tested and untested asymptomatic individuals) held a session with me. I wanted to help bring the new person up to speed on the history of the support group and the needs of the at-risk section.

We noted that the support group's caregiver section is usually the largest of the three subdivisions, followed by the section for those already affected.

The at-risk is usually the smallest – even though at-risk individuals outnumber affected individuals nationally by a ratio of at least five to one.

I sympathize completely with the occasional need to "escape" from HD, so I understand why other at-risk people didn't attend the meeting. However, I am hyper-aware of the need for more individuals to participate in research studies and clinical trials to create effective treatments.

The transition to patient status

The facilitators and I also discussed the difficult choice individuals and facilitators must make in transitioning newly affected individuals out of the at-risk section and into the affected section.

I've witnessed this transition for a number of people. I can't imagine how hard it is.

Once the symptoms begin, the terminological ambiguity ends. They are now "affected" or "symptomatic" individuals. They are "HD patients."

I anxiously await the moment when an effective treatment would not only ameliorate these and other patients' symptoms, but also prevent onset in asymptomatic gene carriers.

Posted by Gene Veritas at 8:40 PM

Labels: asymptomatic , CAG repeats , diagnosis , gene expansion carrier , gene-positive , genetic test , HD gene carrier , Huntington's disease , onset , positive test result , premanifest , presymptomatic , prodrome , tested positive

5 comments:

Anonymous said...

Thanks for sharing Gene. You're very talented and I appreciate your insight, knowledge, and research on HD. I'm at-risk and considering the test myself. God bless!

10:46 AM, March 27, 2014

Anonymous said...

Congratulations Gene,
That's exactly how I feel at the age of 69 with a 36 CAG count. I'm a member of the club trying to live "as normally as possible". Sometimes succesfully sometimes with some questions. I know what HD means, having cared for my beloved son Cédric who died at the age of 42. As my geneticist says: if I live long enough I might get HD but seeing my current condition,I have more chances to dye from something else.
How funny...
Take care gene.
Albert

2:26 AM, March 28, 2014

Anonymous said...

Hi, I read your article with great interest. My grandmother and mother both died from HD. I have five siblings of which 2 have been diagnosed and 2 have symptoms but have not been diagnosed. Myself and one brother have been tested and are negative. I have one niece that is 39 years old and has tested positive. She does have symptoms. I lived for 37 years wondering what my fate would be. Last August, at 55 year old, I was tested. This is such a sad disease. I pray for a cure every nite. Thank you for doing what you do.

12:27 PM, April 10, 2014

Nathan Provo said...

Hi Gene. I've enjoyed following your blog over the years. Knowing that you've posted on Isis' HD drug development progress previously, I just wanted to quickly inform you Isis-HTTRx has just been added as a preclinical stage drug on Isis' website.

2:58 PM, April 13, 2014

Anonymous said...

Gene, your writings really mean much to me and makes my life as a HD gene carrier easier. Thank you.

I would like to know the definition of disease onset. Is that when the movements starts to appear or any psychiatric symptoms?
If a person get psychiatric symptoms like anxiety at the age of 35, while the movement symptoms starts at 50, would the registered disease onset be 35 or 50?

Take care,
Anonymous (due to health insurance)

10:48 AM, April 27, 2014

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