6-17-2014

Fear of onset: the inescapable reality of the Huntington's disease gene carrier

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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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TUESDAY, JUNE 17, 2014

Fear of onset: the inescapable reality of the Huntington's disease gene carrier

As a carrier of the devastating and ultimately deadly genetic mutation for Huntington's disease, I have worked hard to live as normally as possible. This blog is replete with examples of coping strategies and ways in which I have strived to balance work, leisure, family, and HD advocacy.

At 54, my HD-stricken mother was rapidly declining, heading towards a troubling and terrible death at the age of 68. Today, at 54, I continue to enjoy the gift of good health – the major reason I can often feel "normal." Scientists are searching to discover the reasons for the wide variability in the age of onset observed in people, like my mother and me, who have the same level of mutation.

Yet my fear of onset often creeps back in.

Recalling a time of innocence

The past few weeks I've been so busy with the "normal" that I've had no time to write in this blog.

At work, I'm transitioning from five-and-a-half years as departmental chair to a year-long sabbatical, during which I aim to write a long-gestating book on the history of former Brazilian radicals now in positions of power. I'm also teaching an intensive, three-week summer session course on the history of Mexico. The next year promises to be an engaging, challenging time.

The transition has required an understandably disruptive move to a new office, but also allowed me to dispose of unneeded books and papers.

As I rummaged through old files and letters, I found myself reminiscing about the seemingly innocent period of my life before Huntington's struck my mother.

It would be great, I thought, not to have to worry about onset. Without the threat of HD, which led me to expand my scholarly endeavors into the history of science, technology, and medicine, I could once again focus exclusively on the history of Brazil.

Watching for early symptoms

I'm also working out the logistics for my upcoming trip to the University of Iowa in Iowa City for my follow-up participation in PREDICT-HD.

An "observational study of the earliest signs of Huntington's disease," PREDICT-HD has aimed at creating key standards for predicting onset and measuring the rate of disease progression.

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I'll be staring onset in the face – and wondering about my performance on the battery of tests.

A visit to Auspex

I discussed my fear of onset and reiterated our community's urgent need for effective treatments in an intense, 80-minute get-acquainted conversation last week with Pratik Shah, Ph.D., the president and CEO of Auspex Pharmaceuticals.

An investor-funded San Diego firm focused exclusively on central nervous system disorders and orphan diseases, Auspex struck me as made-to-order for the fight against HD. It is currently conducting clinical trials for a drug called SD-809, aimed at treating chorea, the involuntary abnormal movements produced by HD.

SD-809 (dutetrabenazine) is a potentially improved version of tetrabenazine, a chorea treatment currently marketed by the pharmaceutical company Lundbeck under the name Xenazine. If SD-809 works as intended, it will require fewer dosages and produce fewer side effects than tetrabenazine.

However, tetrabenazine does not affect the root causes of HD, nor is SD-809 expected to.

Auspex seeks to use SD-809 as a platform to research and develop drugs that would attack those causes.

Dr. Shah and I agreed to schedule soon an interview so that I can write an in-depth article on Auspex's efforts.

I told Dr. Shah about a middle-aged, HD-afflicted man I had met who had maintained much of his cognitive abilities but suffered from strong chorea. However, tetrabenazine controlled the chorea, enabling him to keep driving, something most HD patients have to give up.

Tetrabenazine's approval by the Food and Drug Administration had come too late to benefit my mother, who died of HD in 2006. I told Dr. Shah that she had taken another medication to control her chorea, which was relatively mild, although she had initially had strong chorea in her legs at night. In general, chorea was the least of my mother's problems with HD, which devastated her cognitive abilities and caused serious psychiatric difficulties.

I also related my recent conversation with a former HD support group colleague who has had symptoms for a number of years.

Speaking to a symptomatic individual, I pointed out, provides me a terrifying glimpse of my own future.

A powerful HD dream

As I processed these latest events in my journey with HD, my unconscious mind produced a powerful dream.

I awoke from the dream at 5 o'clock on Sunday morning. Afraid that I would forget its content, I went to my home office to type out the details on my computer, and to outline this article.

In the dream, where I am meeting with other asymptomatic HD gene carriers, I encountered the same HD-affected man whom I had mentioned to Dr. Shah.

The people in our dreams often represent aspects of ourselves. In my interpretation, thinking of a symptomatic man in the context of a group for

the asymptomatic meant that I was wrestling with the inevitable reality of my onset.

Tapping into the soul

As the dream continues, I fly to New York City on HD advocacy business.

In my hotel room, I start to write a blog article describing the recent HD-related aspects of my life. I have my trusty laptop with me but am oblivious to it. Instead, I write in longhand on a white legal pad. It's the way I sometimes wrote in college or now write on airplanes or when I'm revising a draft I've printed out.

There's something pure and primal about this form of writing. It's the way I first learned to write. I'm crossing things out, jotting down ideas, and flipping back and forth through the pages to read and make adjustments. At one point I think that, because I don't have much time before my evening HD meetings, I'll switch to the computer. But I want to first eke out some more lines on the pad.

The dream was compelling me to practice the craft I have enjoyed since childhood, to tap into the soul that defines me.

I later recalled the photograph that an HD-affected man posted of himself illustrating his superb kickboxing skills before the disease struck. He wanted to remember himself at the height of his powers.

The dream, I think, reflected my fear that HD will rob me of my writing skills.

A metaphor for facing HD

Later in the dream, I go to a restaurant along with two other asymptomatic gene carriers and my friend, blog editor, and HD alter ego, Norman. One of the gene carriers, I recognize, is the symptomatic man I'd encountered earlier in the dream, only transformed into a healthy individual. On the way there, I give each gene-positive man a bear hug. I feel deep brotherly love towards these men.

A native New Yorker, Norman describes the restaurant as a very different and unique place. He says it's called Pub Med.

We seem to be on the Upper West Side of Manhattan. Evening is approaching.

The restaurant is made of recently hewn, unpainted pieces of wood, which are also used as furniture: benches and small, round tables. It's outdoors, located in the middle of a square where I can hear kids playing on swings and moms walking their kids. There are small stores on the edges of the square, too.

But there's something very strange: the benches and tables are stacked on top of one another in a pyramid-like fashion. They rise about 30 feet. We climb up and look for a place to sit. Norman is sitting with the first gene-positive person while, at another table, the second gene carrier continues to explain to me the nature of this restaurant-structure and how to sit on it without falling.

I'm still standing. However, as I try to sit down, some of the tables and benches near me shift down or fall off suddenly and unpredictably. I'm afraid that I'll fall off. The second gene carrier seems to know well how to deal with it. He's experienced and seems to take it all in stride.

As I strive to keep my balance on the structure, I gaze at a different kind of Manhattan skyline. On the horizon, I see some burning buildings.

Referring to the restaurant-structure and the buildings ablaze in the distance, I tell the second gene carrier: "I can think of no better metaphor to describe living at risk for Huntington's disease."

Building hope, pondering onset

The dream, I think, represented my fight to continue advocating for the HD cause.

Manhattan is headquarters for three key HD organizations: the CHDI Foundation, Inc., the Hereditary Disease Foundation, and the Huntington's Disease Society of America. Along with other organizations and scientists around the world, they hold the key to finding treatments.

Norman has taken my family and me on a walking tour of Manhattan. He urged me to start this blog. In both the dream and real life, he has acted as a kind of guardian angel in my fight against HD. I believe that the Norman of the dream also symbolizes my own internal editor, who, like the real-life Norman, the author of a richly detailed and public-spirited watchdog blog on Brooklyn's Atlantic Yards project, strives to produce in-depth and understandable reports.

Along those lines, I had told Dr. Shah I would read scientific articles about SD-809 before our planned interview. I believe that the Pub Med restaurant represents my desire to prepare thoroughly for an interview regarding the potentially life-saving work done at Auspex. In reality, PubMed, a well-known research tool, has more than 23 million citations from biomedical literature, life science journals, and online books.

I explained to Dr. Shah that in this blog I seek to provide the HD community with information about potential treatments, breakthroughs, and challenges.

My goal is to provide the community with hope, and advocate for change.

The dream, I believe, also reflected my continued striving for internal equilibrium as I ponder the kind of onset I will experience.

Will I falter like an HD person who can no longer control movements and mind? Will I continue to work and drive? Will I be able to help support my daughter as she studies in college? Will effective treatments arrive in time to at least reduce the severity of symptoms – and prolong my life?

These are the inescapable questions of my reality as a Huntington's disease gene carrier.

Posted by Gene Veritas at 11:43 PM

Labels: advocacy , Auspex Pharmaceuticals , chorea , coping , HD gene carrier , Huntington's disease , mother , Pratik Shah , PREDICT-HD , scientists , SD-809 , symptoms , tetrabenazine , treatments

1 comment:

Albert Counet - Belgium said...

As usual, a very true description of the feelings of a gene positive person. Congratulations!
What a pity that your blog is only in English

2:21 AM, June 30, 2014

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