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No time for complacency: get ready for HD clinical trials

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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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In the past few years, scientists have made huge strides towards treating and perhaps even controlling Huntington's disease, whose killer gene inhabits every cell of my body. While scientists scrupulously avoid providing false hope, even the most pessimistic among them now talk of "when" a treatment or treatments will come, not "if," as international HD spokesperson Charles Sabine observed last October at the annual Huntington's Study Group conference in San Diego, CA.

That's enormous progress, compared to a decade ago, when practically no pharmaceutical companies showed interest in HD.

But this is no time for complacency. On the contrary, as labs ramp up for potential clinical trials to test the first group of the 700-plus potential "drug targets" for HD, many daunting challenges and tasks remain.

I plan to deliver this message in a speech this coming Saturday, July 30, at the 2011 Inaugural Clinical Research Symposium of the Northwest Chapter of the Huntington's Disease Society of America (HDSA) at Evergreen Hospital Medical Center in Seattle, WA (click here for the program). The speech will be titled "What HD Families Should Know about Clinical Trials: Initial Thoughts from a Gene-Positive Activist."

The clinical trial administrators will need a large number of symptomatic people to participate, ranging from 20 to as many as a couple thousand people per drug. With only 30,000 people in the entire U.S. affected by HD, it may prove impossible to fill the numerous spots in the trials.

That is why CHDI Foundation, Inc., the so-called "cure Huntington's disease initiative," has inaugurated Enroll-HD, the first-ever worldwide database of at-risk, gene-positive, and HD-affected people, in order to expand the base of possible trial participants.

All must chip in

As advocates like Charles and me have pointed out, those active in organizations such as the Huntington's Disease Society of America (HDSA) can no longer limit our focus to fund-raising. At-risk, gene-positive, and affected people must also collaborate with the researchers and physicians in the process of planning and implementing the trials.

Preparing for potential trials requires that we care for our health as much as possible.

And, as I wrote in 2009, the untested in our community, who constitute a majority of the at-risk, need to muster the courage to learn their status. With the hope of treatments, refusing testing makes less and less sense. And, if people don't get tested, they can't participate in a trial.

Huntington's Disease Society of America
International Huntington Association
Huntington's Disease Drug Works
Huntington's Disease Lighthouse
Hereditary Disease Foundation
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Thomas Cellini Huntington's Foundation
HDSA Orange County (CA) Affiliate
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HD Blogs and Individuals

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In short, if we all don't chip in, treatments won't be found.

The painful barrier of denial

Chances are, if you read this blog, you already agree with this outlook. You're probably active in the HD effort in some way. So, as they say, I'm preaching to the choir.

We need to reach out to those in the community who shun involvement, usually out of fear, and sometimes out of ignorance.

I know all too painfully how denial works.

My mother Carol died of HD in 2006 after battling HD for nearly two decades. Her at-risk, untested brother and his wife hid the truth about her disease from their children and their families by attributing Mom's symptoms to "mental problems."

A split family

I am also estranged from my own sister, the untested mother of three at-risk, untested adult sons whom she conceived before my mother's diagnosis. She prefers to do nothing, because she does not believe, or is unaware, that there is hope. She is completely uninterested in advocacy, research, and trials.

As a gene-positive activist, I personify the knife-edge of HD for my family. My sister became especially uncooperative after the birth of our "miracle baby," who tested negative in the womb. My sister was jealous for two reasons: we had a daughter, and she was HD-free.

In reading this description of my own family, many will knowingly nod in agreement. Denial is powerful, and it is everywhere. Combined with the sorrow, frustration, anger, and fear provoked by HD, it splits families apart.

I have fought back against the threat of HD by participating in numerous observational trials and advocating for the cause.

But I have yet to figure out a way to involve family members so deeply in denial – and so angry at me whenever I raised the issue of HD, directly or indirectly.

Living by example

As Charles observed, it will be truly tragic if people are not ready for clinical trials.

But we must not give up. Through this blog and my activism I have met many people new to HD and looking for ways to help. We need to welcome these individuals and their families with the greatest of attention – and love.

And we must live by example, because, in the end, our actions will carry far more weight than our words.

Defining success together

The HD community also can participate actively in the clinical trial process by helping researchers, physicians, drug makers, and the federal Food and Drug Administration (FDA) define a successful drug and how to measure that success.

HD causes a triad of symptoms: motor (shaking known as chorea and problems with coordination), cognitive (memory and mental abilities), and

psychiatric (emotional disturbances). For a long time, scientists have spoken of the need for a "cocktail" of drugs to combat the triad and their numerous causes in the brain and brain cells.

So far, only one HD drug – tetrabenazine – has received FDA approval. Marketed by Lundbeck as Xenazine, this medication reduces chorea but does not affect the causes of HD.

The new generation of drug targets would indeed attack the causes. Researchers and drug makers theorize and hope that these targets would improve or perhaps even eliminate particular symptoms, but many of these new kinds of drugs represent uncharted scientific territory. Until the trials get underway, nobody will know how patients will respond.

So, even before trials start, patients must help define specific outcomes for the trials.

Maintaining functionality and personality

During the question-and-answer session after my May 17, 2011, speech at Alnylam Pharmaceuticals, I outlined what I thought were signs of success.

"From my standpoint, wow, maybe I will never have symptoms of Huntington's disease," I said in reflecting on the proposed Alnylam RNA interference drug, intended to attack the disease at its genetic roots and possibly ready for a Phase I trial as early as 2012. "That would be my hope as a patient.

"For the longest time, it's like, 'Well, you're going to get sick.' The question is: 'When are you going to get sick? And how sick are you going to get? And what can you take to stop it?'...

That response came from my individual perspective as a gene-positive, asymptomatic individual – what the scientists refer to as the "presymptomatic" stage of HD.

But I also gave my opinion as to what symptomatic HD patients might want from a drug. I thought of my mother, who had been reduced to a "mere shadow of herself."

We need drugs that will help people maintain their "basic functionality" and personalities. A bit of chorea would probably be acceptable as long as a person retains his or her mind and can go to work, I said.

"People want to keep their personality with this disease," I said. "They want to be recognized as individuals. They don't want to be seen as sick; they don't want to be seen as drunk; they don't want to be seen as disabled individuals."

But I'm just one voice. More people need to give their opinions on this still very open question.

Strengthening the patient-researcher bond

As is often the case, I have only just scratched the surface of one of the many issues surrounding Huntington's disease.

It's clear that we all need to educate ourselves about the kinds of medicines under consideration and how they might affect the disease.

I hope to do my small part at the Seattle symposium. Across the country, our community needs more events such as this.

But in addition to speaking, I want to brainstorm with the audience about the definition of pharmaceutical success and absorb the other speakers'

ideas about clinical trials.

The HD community has a long reputation as one in which patients and researchers collaborate effectively. Now, as we get ready for historic clinical trials, we need to further strengthen that crucial bond.

Posted by Gene Veritas at 3:48 PM

Labels: Alnylam , at-risk , Charles Sabine , CHDI , clinical trial , denial , FDA , gene , gene-positive , Huntington's , Lundbeck , RNA interference , symptoms , tetrabenazine , Xenazine

3 comments:

Beverly said...

What an interesting question: How do we define pharmaceutical success for drugs intended to treat or cure HD? I'm a teacher, so let me "grade" some possible outcomes of clinical trials:

A+ Cure!
A Very good cognitive and emotional regulation and minimal motor impairment
A- Good cognitive and emotional regulation and only slight problems with coordination and balance

B Well, I actually don't want to keep moving down the scale, but you can see the way I'm thinking. I guess F would be that the drug actually causes harm, and C would be that it doesn't help but it doesn't hurt. Even finding out that the drug doesn't help will advance science and move us closer to A+.

My fiance recently participated in his first clinical trial, and we both actually enjoyed the experience. It feels empowering to be able to do something, and we have met wonderful people--researchers and patients and their family members--who are involved with these trials. We are signed up for another clinical trial which will begin sometime before year's end, I believe.

I join Gene in encouraging others to take action. Contact your local HD Center of Excellence and ask them to suggest a clinical trial for you to participate in. We all need each other, and we can all help each other and future generations. Get a move on! Call! Sign up!

1:54 PM, July 30, 2011

Eileen H'van said...

Have a HD question. Just found our that our estranged grandfather's sister died of HD. I keep reading conflicting info about who should be genetically tested. Should I be tested? She was my mother's aunt. I have asked my doctors and they never give an answer. If you could point me the right direction it would be so helpful! Thyanks!

1:26 PM, October 19, 2011

Gene Veritas said...

Getting tested is a personal decision that should be made only after considerable thought. I recommend seeking out the nearest HDSA Center of Excellence, an HD support group, and/or a neurology clinic. Take a look at this blog article: http://curehd.blogspot.com/2006/11/handling-news.html.

10:29 AM, October 21, 2011

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