6-24-2009

The time is ripe

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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.

Blog Archive

- ► 2021 (12)
- ► 2020 (16)
- ► 2019 (19)
- ► 2018 (16)
- ► 2017 (14)
- ► 2016 (13)
- ► 2015 (24)
- ► 2014 (24)
- ► 2013 (30)
- ► 2012 (26)
- ► 2011 (33)
- ► 2010 (26)
- ▼ 2009 (21)
  - ► December (1)
  - ► November (2)
  - ► October (3)
  - ► September (2)
  - ► August (3)
  - ► July (3)
  - ▼ June (1)
    - The time is ripe
  - ► May (2)
  - ► April (4)
- ► 2008 (7)
- ► 2007 (7)
- ► 2006 (4)
- ► 2005 (17)

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WEDNESDAY, JUNE 24, 2009

The time is ripe

Although I am gene-positive for Huntington's disease and could very soon develop its dehumanizing symptoms, I feel immensely privileged to live in a time and in a country in which a treatment has become possible.

Huntington's is still a relatively unknown disease; in the HD community, many people are still shrouded in ignorance, fear, and denial. I have touched on these themes frequently in this blog, as in the story of the woman who changed her identity and fled after her symptoms began (*click here* to read more).

However, scientific research has given us an increasingly detailed understanding of the cause of Huntington's disease, its symptoms, and its social impact. And new revelations are emerging.

The path of research

In 1993, the huntingtin gene was discovered. Three years later, the huntingtin protein was found. A plethora of projects carried out by scientists around the world have sought to illustrate how the defective protein wreaks havoc in brain cells.

Meanwhile, neuroscientists and psychologists have worked diligently to trace the earliest, most subtle symptoms of the disease – for example, changes in the sense of smell – in order to prepare the way for drugs designed to stop HD symptoms early.

Several MRI studies have mapped how HD devastates specific areas of the brain. I have taken part in two of these studies. Last year, for instance, I spent most of a day lying inside functional MRI machines at a local university. Sometimes I simply slept, while other times I was required to play a kind of video game so that the machine could check my brain's reactions.

Another discovery

On June 5, scientists made yet another startling revelation that further unravels the mystery of HD: a second protein, called Rhes, interacts with huntingtin in a way that may lead to the death of cells in the striatum, the main area of the brain affected by HD.

Everybody has the huntingtin gene and its protein product of the same name, but only people with an elongated form of the gene develop HD. Huntingtin is present in every cell of the body, so why does HD kill only brain cells? The new research indicates that the presence of Rhes, which is located in the striatum, could be one of the keys to cell death.

Scientists immediately speculated that now Rhes, and not just huntingtin, could become a target for potential drugs against HD.

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The new research was carried out at Johns Hopkins University (*click here* to read more). Rhes itself was first identified several years ago at the Scripps Research Institute in La Jolla.

Nobody can predict when or even if an actual treatment for HD will be developed, because HD involves many factors. Further surprises such as the Rhes discovery could occur – either slowing progress by creating new puzzles or speeding it up by indicating other possible drug targets.

Other signs of hope

On June 15, I made a follow-up trip to Isis Pharmaceuticals in Carlsbad, California, to receive an update on the search for a drug to partially block the effects of the huntingtin gene – the first such attempt of its kind. As I did after my visit there last year, I came away with a feeling of optimism about the quest for a Huntington's disease treatment.

Stay tuned – I will be blogging on my Isis visit very soon. (*Click here* to read my previous posting.)

Two days later I met with two scientists and a businessman who have started a new early-stage drug discovery company (CalAsia Pharmaceuticals) that uses a cutting-edge technology known as DFS, Differential Fragment-Based Screening, a way of identifying molecules that could serve as drugs. They are considering using their technology to help identify possible treatments for HD.

The other advances and programs taking place in HD research are too numerous to discuss here. Indeed, covering the Huntington's disease beat properly would require a full-time, professional science writer. Years ago I used to write a tri-annual research "roundup," summarizing all of the important findings in a single article. In the last few years I have found it too big a task.

That is good news, because it means that scientists are building a critical mass of knowledge about HD that is bringing us closer to treatments and maybe even the cure.

It's a historic moment.

A fighting chance

When my mother conceived me in 1959, she had no inkling that she would someday develop Huntington's disease or that she had passed on a defective huntingtin gene to me. In 1999 – ten years ago this month – I was able to take a definitive predictive test.

Since then I have proactively worked to care for my health and contribute to the search for treatments and a cure. I have participated in a support group, become an activist for the Huntington's Disease Society of America, and taken the supplements recommended by the Huntington's Disease Drug Works program.

My test result was positive. But today I feel that we in the Huntington's disease community have a fighting chance to see this disease beaten in our lifetimes. We owe this chance in part to our greatness as a nation and to a generation of researchers who have passionately pursued the understanding of HD and other neurological diseases.

Posted by Gene Veritas at 4:09 PM

2 comments:

Anonymous said...

Thankyou Gene, It definatly is a great time to be involved. I can only imagine being a part of this process.
I am always impressed with your
insperational point of view.

8:58 PM, June 26, 2009

Unknown said...

Today is the first day I have read your blog. I read it all, and I'd just like to make the following comment.

My grandfather passed away of HD when I was just a few years old, my uncle passed away a decade later, and now my father is in the final stages of Huntington's Disease. I've seen it my entire life (they say that just after I was born my grandfather was able to control his erratic movements just long enough to hold his first born grandson). I'm currently coming to grips with my own mortality, and your blog has helped me today. I will continue to read and respond... but, for now I'd simply like to say... Thank you!

10:40 PM, June 27, 2009

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