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The Huntington's community rising and converging

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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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The Huntington's community rising and converging

In the quest to end one of humanity's cruelest conditions, the Huntington's disease community is coalescing as never before.

As one individual commented on this blog last year, our community is "rising and converging."

We have traveled a long road.

Before the founding of the Huntington's Disease Society of America (HDSA) in 1967, nobody advocated for the well-being of HD patients and their families. Huntington's disease was shrouded in a combination of stigma and ignorance.

Today *four* organizations dedicate themselves to the cause: HDSA, the Hereditary Disease Foundation (HDF), the Huntington's Disease Drug Works program (HDDW), and the CHDI Foundation, Inc., informally known as the "cure Huntington's disease initiative." They have brought great hope for treatments and a cure. Britain, Canada, and many other foreign countries also have important HD associations.

HDSA, solidarity, and the Web

Thanks especially to HDSA's work over the decades, public awareness of this killer disease has increased, although HD is still far from being a household word.

HDSA has also promoted community solidarity, from the very first local support groups of the late 1960s to the organization's 26th national convention, scheduled for June 24-26 in Minneapolis, MN. In late 1995, after receiving the shocking news that my mother had HD, one of my first phone calls was to the local HDSA chapter. The chapter president compassionately explained the potential implications of my mother's diagnosis for my own future and invited me to attend the support group.

With the power of scientific knowledge and the reach of the Internet, members of the HD community have strengthened their ties and promoted the cause. People today can learn instantly about the many aspects of

HD Links

Huntington's Disease Society of America
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

Huntington's disease from numerous informational websites, YouTube, and the portals of the primary HD organizations.

I frequently refer to Stanford University's Hopes site, the Huntington's Disease Advocacy Center, and HD Buzz. Communication also takes place through social networking sites and chat rooms. Thousands of people interested in HD belong to Facebook, where they can find support and information.

HDDW even conducted a clinical trial via the Web. I logged in regularly to perform cognitive tests on my computer keyboard. Because I am gene-positive for HD, HDDW founder Dr. LaVonne Goodman reviewed my performance to check for possible signs of symptoms. So far, I remain free of HD's classic symptoms, although the scientific research indicates that the genetic defect most likely is already damaging my brain.

A chapter and a major stem-cell project

For many in the HD community, the lifeblood of the movement is their local HDSA chapter and support group. HDSA has more than 30 chapters and affiliates around the country, many tied to an HDSA Center of Excellence for Family Services and Research.

On May 6 and 7. I paid an emotion-filled visit to the Northern California Chapter (which includes Sacramento, San Francisco, and the northern part of the state) and observed a critical mass of HD-related activities.

On May 6, I spent the day interviewing Jan Nolta, Ph.D., and observing the work of the stem-cell research facility she directs, the Institute for Regenerative Cures, co-funded by the University of California, Davis (UCD), and the California Institute for Regenerative Medicine (CIRM). This 109,000 square-foot facility supports the work of 145 faculty researchers organized into 15 disease teams, including heart disease, blood disorders, HIV, Alzheimer's, Parkinson's, Lou Gehrig's, and Huntington's.

Dr. Nolta and her HD team have used a well-known type of stem cells, called "mesenchymal stem cells" (MSC), to develop two potential treatments for HD.

Dr. Nolta refers to the MSC as "paramedics" because of the way they congregate around and repair damaged cells. Injected into the brain of an HD patient, the MSC might be able to repair damaged neurons (brain cells) and restore the vital connections between them.

Dr. Nolta at the HD bench at the Institute for Regenerative Cures (photo by Gene Veritas)

Dr. Nolta and her HD team have also reengineered MSC to deliver small interfering RNA molecules directly into cells in order to stop the mutant huntingtin protein from causing damage. She has obtained a patent for this technology, hoping to later negotiate an agreement with a pharmaceutical company that would produce and market an MSC drug. If successful, this approach would stop the cause of HD at its genetic roots.

At the Institute for Regenerative Cures, I felt excited as I took a step into the future of medical treatments. I will provide a detailed report on the HD research efforts in a future article.

A record turnout

With nurse practitioner Teresa Tempkin as my guide, I also toured the HDSA Center of Excellence at the UC Davis Medical Center. I viewed the consultation rooms where Teresa, center director Dr. Vicki Wheelock, and a team of other physicians and healthcare specialists attend to the roughly two hundred HD patients living in the region.

By introducing Dr. Nolta to the HD community, Dr. Wheelock helped make HD research a priority of the Institute for Regenerative Cures.

Dr. Vicki Wheelock, director of the UC Davis Center for Excellence (photo by Gene Veritas).

In the evening, I dined at a Sacramento restaurant with Dr. Wheelock, Teresa, chapter president Judy Roberson, family services chair and former president Les Pue, and other participants in the annual Northern California Chapter convention, held the next day and coinciding with HD Awareness Month.

The chapter's annual meetings date back to at least 1991. In 2000, the chapter upgraded them to convention status, now held at the UC Davis Medical Center campus. Starting in 2009, the chapter has received financial assistance from Lundbeck, a Denmark-based pharmaceutical firm that markets Xenazine (tetrabenazine), the first-ever FDA-approved drug for chorea, the shaking and trembling that occurs in many HD patients.

This year's convention attracted a record number of attendees, with 197 official registrants and about eight or more other participants – yet another indication of the rising and convergence of the HD community. People from Lundbeck, Stanford Hopes, and other HD-related initiatives staffed information tables.

The audience at the 2011 HDSA Northern California Chapter convention (photo by Gene Veritas)

HD-free with PGD

The convention featured eight morning workshops divided into two sessions. All were compelling.

The first session I attended, "All About Preimplantation Genetic Diagnosis for HD Families," was presented by Stacy Brookhyser, the gene-positive, 35-year-old mother of twins free from HD after she and her husband opted for the preimplantation procedure, known as PGD.

"When I started this journey, I was newly married, and at risk," Stacy told the audience, which included her HD-stricken mother. "I hadn't tested, and I didn't know what to expect, but I knew about the at-risk status. I knew that if my husband and I went ahead had children naturally, that my children, of course, would have a chance of inheriting Huntington's."

The decisions about her HD test and PGD were "daunting," Stacy added. During the rest of the presentation she recounted how they researched the various options for starting a family, considered their personal and moral beliefs, and went through the medical procedures necessary in PGD.

You can watch Stacy's presentation in the video below, and you can learn more about PGD by visiting her website, www.HDFreeWithPGD.com.

All About Preimplantation Genetic Diagnosis for Huntington's Disease Families

from Gene Veritas

43:43

All About Preimplantation Genetic Diagnosis for Huntington's Disease Families from Gene Veritas on Vimeo.

I was deeply moved by Stacy's presentation. Listening to her, I was transported back to the harrowing moments that my wife and I experienced as we pondered our own extremely difficult decision (in late 1999 and early 2000) to have our child tested in the womb. This was *before* the availability of PGD. Our "miracle baby" tested negative.

I am glad that families today have access to PGD, although it continues to be a highly expensive procedure that is not always covered by insurance. Stacy's PGD cost $30,000, with insurance picking up most of the cost. However, as she pointed out, that sum pales before the cost to the health system of caring for an individual with HD – not to mention the lost income and suffering experienced by the family.

Emotional and behavioral symptoms

I was anxious to attend the talk by psychiatrist Raheel Khan on "Dealing with Emotional and Behavioral Changes in HD."

I have read on many occasions that these symptoms often appear first. My mother's first apparent symptoms, including seemingly inexplicable mood swings and crying, fell into this category.

As a gene-positive individual, I wanted to know what might lie in my not-too-distant future. I want to prepare effectively by learning what to anticipate and how to help my family understand how they might need to assist.

Defining HD as a "classic 'neuropsychiatric' disorder," Dr. Khan outlined the many conditions faced by HD patients, including depression, delirium, loss of interest in daily activities, apathy, anxiety, irritability, and many psychotic symptoms such as hallucinations, delusions, and disorganization.

Dr. Raheel Khan speaks on emotional and behavioral problems in HD (photo by Gene Veritas).

A depressing scenario

Dr. Khan spent a good part of the time on depression, which, he said, occurs in 30 to 50 percent of all HD patients. To paraphrase him, this is a very high percentage. When you have depression, it's tougher to cope with

the underlying disease. This worsens the course of the HD, so this aspect must be treated. Depression is insidious. The person rarely experiences their situation as a symptom. It's up to the caregiver or family member to point out the changes to the patient.

Indeed, Dr. Khan made it very clear that assistance from family members and caregivers is crucial in dealing with this aspect of HD.

One caregiver in the audience wanted to know how to assist an affected loved one who refused to take psychiatric medication.

One solution is to pose the psychiatric condition as a part of the disease and tell the patient of the need for help, Dr. Khan responded. This, he added, was a very common problem.

"People still call me a shrink all the time," he said, noting the stigma that still exists for psychiatric care. Unfortunately, he added, a person can't be forced to see a psychiatrist. The psychiatric problems should be raised during a regular checkup, and the attending doctor should pointed out that *he* or *she* is the one visiting the patient, not the patient visiting the physician, Dr. Khan explained.

Just hearing about such a plethora of potential psychiatric symptoms – and writing about them again now – leaves me depressed. What if *I* suffer from some combination of these symptoms? What if *I* resist medication? I do *not* want to be a burden on my family. As sad and difficult as the task is, I must prepare my family for this scenario.

Connecting to HD people

With multiple emotions flowing after hearing Stacy and Dr. Khan, I wanted to end the threat of HD for my family and for everybody at the convention. Over lunch, I sat with three HD people and their families.

At the start of the afternoon session I watched chapter representatives present HD patient Cheri Harries with the Joseph P. Roberson Foundation's "HD Person of the Year" award for her commitment to the chapter and her exemplary perseverance against the disease. She is the wife of chapter board member Terry Harries.

Accompanied by husband and board member Terry, Cheri Harries approaches podium to receive "HD Person of the Year" award (photo by Gene Veritas).

In our fight I felt connected to all of these individuals – and I wanted to do my part to win it.

'It's time to conquer HD!'

I got my chance to contribute as one of two featured speakers in the afternoon session, held in the main auditorium of the UC Davis MIND Institute. (MIND stands for "Medical Investigation of Neurodevelopmental Disorders," such as autism.)

I titled my speech "A Gene-Positive Activist Copes with the Threat of Huntington's Disease (Fighting Back)." As in my big coming-out keynote address at CHDI's 6th Annual HD Therapeutics Conference on February 7, I once again took off the mask of Gene Veritas to reveal my true self to the audience.

I spoke extemporaneously about my mother's downfall, my father's caregiving as an "HD warrior," my advocacy for the cause, and, finally, pointers on effective activism.

I felt deeply connected to the standing-room-only audience. Many HD people and their families looked on, and people responded with great passion and enthusiasm. When I announced that our daughter had tested negative, the audience applauded. They applauded again when I recalled how I had removed the mask for the first time at the CHDI conference. Afterwards John, a shaking HD man in his 60s, hugged me several times like a long-lost brother.

You can watch my speech in the video below.

Gene Veritas: Fighting Back Against Huntington's Disease

from **Gene Veritas**

33:21

Gene Veritas: Fighting Back Against Huntington's Disease from Gene Veritas on Vimeo.

"And so what are we, as we take off our masks and become this new community?" I asked at the end of my speech. "We are a community rising and converging. We're rising up for the first time. We're fighting against discrimination. We're getting tested. We're advocating for bills. We're doing new and wonderful things as we rise in this community. And as we rise, we are all converging together on the final point. And what is that final point?

"It's time to conquer Huntington's disease!"

Stopping the culprit

Doug Macdonald, Ph.D., CHDI's director of drug discovery, closed the conference with a presentation titled "Huntingtin Suppression: An Exciting New Therapy Being Developed at CHDI.

Dr. Macdonald explained the efforts to reduce the actions of the defective huntingtin gene and its resultant, harmful protein, the culprit in HD.

Dr. Macdonald brought hope: Isis Pharmaceuticals, Inc., of Carlsbad, CA, and Alnylam Pharmaceuticals of Cambridge, MA, are both preparing to test potential huntingtin suppression drugs in humans within the next couple years.

Like Dr. Nolta's approach, the Isis and Alnylam drugs would represent a revolutionary advance in the treatment of HD by attacking the genetic roots of the disease. Currently these approaches come the closest to a "cure," although HD patients most likely would have to take such drugs their entire lives.

You can watch Dr. Macdonald's presentation in the video below.

Stopping the culprit in Huntington's disease: talk by Dr. Doug Macdonald

from **Gene Veritas**

43:07

Stopping the culprit in Huntington's disease: talk by Dr. Doug Macdonald from Gene Veritas on Vimeo.

Inspiration

I came away deeply inspired by the HD movement in Northern California and reenergized in my advocacy. The annual conventions provide a fine model for other chapters and disease organizations, as does the powerful convergence of efforts by the Center of Excellence, stem-cell researchers, the HDSA chapter, Lundbeck, and the HD community.

I also felt gratitude for Lundbeck's sponsoring of this event and the many other HD-related activities it has supported in recent years.

I'm looking forward to my next step: a speech at Alnylam on May 17 and interviews with the scientists on its HD team.

Inching towards our goal

At the end of the day, as we chewed the fat about HD matters and shared a bit of our life stories, Les Pue graciously drove me into San Francisco. There I met up with my wife and our child. Turning 11 next month, our daughter had crossed the Golden Gate Bridge earlier in the day for her Girl Scout "bridging ceremony," which symbolizes a new level of commitment to the organization.

I was thrilled to see my family after such an intense weekend. Sitting with my wife at a restaurant on Fisherman's Wharf, I thoroughly enjoyed eating clam chowder in a sourdough bread bowl and sipping a tall glass of Stella Artois draft beer.

The battle was not yet won, but I felt our community had inched a bit closer to our final goal.

Posted by Gene Veritas at 6:47 PM

Labels: chorea PGD psychiatric activism at-risk Xenazine Huntington's

depression cure Lundbeck symptoms gene-positive stem cell RNA FDA

2 comments:

Anonymous said...

"Gene,"

No matter what your topic within the HD constellation, you reinvigorate me! Thanks!

Jimmy Pollard

5:19 AM, May 12, 2011

Anonymous said...

Gene, You've written a great entry in your blog! Thanks for shining hope into my day. TOGETHER WE WILL SLAY the DRAGON! Lou Alworth

5:29 AM, May 12, 2011

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