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## Hope, cutting-edge science, and poignant moments at the World Congress on Huntington's Disease

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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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## About Me

 GENE VERITAS

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SUNDAY, OCTOBER 06, 2013

## Hope, cutting-edge science, and poignant moments at the World Congress on Huntington’s Disease

The first such HD meeting held in a developing nation, last month’s World Congress on Huntington’s Disease (WCHD) not only highlighted the need for better understanding of the disease in Latin America. It also revealed the growing global importance of the the quest for both better care *everywhere* and the development of treatments.

Featuring activities for both researchers and families, the sixth WCHD featured some 20 panels, a poster session, a satellite symposium, joint meetings of the International Huntington Association (IHA) and the [Associação Brasil Huntington](#) (ABH), entertaining evening wrap-ups by HDBuzz.net editors Dr. Jeff Carroll and Dr. Ed Wild, and a moving presentation by the France-based HD performance group Dingdingdong.

Like most HD conferences, the WCHD stressed advances in the search for treatments. The four-hour-long closing session included an update on clinical trials, a talk on deep brain stimulation and HD, a presentation on cutting-edge RNA-interference-based (RNAi) therapies, and an overview of the efforts to reduce or block the deleterious effects of the faulty protein huntingtin in the brain.

“There is a lot of hope,” said Dr. Doug Macdonald, the director of drug discovery and development for CHDI Management, Inc., which directs the multi-million-dollar effort to defeat HD by the CHDI Foundation, Inc., in his presentation. “There is a rich pipeline of these therapeutics advancing into the clinic. We have direct delivery of huntingtin-lowering agents, the viral delivery of RNAi agents, and viral delivery of zinc finger protein agents.”

Dr. Macdonald provided a clinical-trial timeline for these potential drugs. The first is likely to be the huntingtin-lowering drug under development at Isis Pharmaceuticals, Inc., and Roche, scheduled to enter a Phase I trial by the end of 2014. Other projects may begin trials in the next few years, Dr. Macdonald added.

## 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](#)

For a detailed explanation of these approaches and more on timelines, watch Dr. Macdonald's talk in its entirety in the video below. See also a discussion of other types of trials and general coverage of the WCHD at [www.HDBuzz.net](http://www.HDBuzz.net).

You can view 32 more chronologically ordered conference-related videos by visiting my [2013 World Congress on Huntington's Disease](#) album on Vimeo.



### Modulating Huntingtin Levels to Treat Huntington's Disease: A Talk by Dr. Doug Macdonald

from [Gene Veritas](#)

22:56 |



[Modulating Huntingtin Levels to Treat Huntington's Disease: A Talk by Dr. Doug Macdonald](#) from [Gene Veritas](#) on [Vimeo](#).

## From exciting science to social consequences

The WCHD scientific presenters focused on a panoply of other HD themes, from exciting developments in basic science to current medical treatments of symptoms to the social consequences of HD.

Dr. Elena Cattaneo of Italy presented the latest research on the origins of the huntingtin gene.

“The normal gene is a gene that everyone has,” Dr. Cattaneo explained. “Everyone in the world has that gene. At some point, we started to think that, if we have that gene, it means that gene is important. So about ten years ago my group, but also other groups, started looking for the function of the normal gene. Of course, we know that in the disease the mutant gene causes the loss of the neurons. But in order to understand more about the mutant version, we wanted to understand what the normal version was doing.”

Scientists discovered that the normal gene is important for “keeping the neurons healthy and alive and working properly,” she said.

The huntingtin gene was “born” in an ameba species 800 million years ago, she continued. “This is the first pluricellular organ, and the huntingtin gene is there. Pluricellular means that cells talk to each other to form an organism.... I started thinking of huntingtin as a gene with a social function, because it brings cells together. So let’s assume that huntingtin is such a gene. Huntingtin is a *good* gene. It is not a bad gene.”

You can watch Dr. Cattaneo’s fascinating presentation in the video below.



## The Role of the Huntingtin Gene in Huntington's Disease: A Presentation by Dr. Elena Cattaneo

from [Gene Veritas](#)

22:01 |



[The Role of the Huntingtin Gene in Huntington's Disease: A Presentation by Dr. Elena Cattaneo from Gene Veritas on Vimeo.](#)

Other notable presentations included Portuguese Dr. Joaquim Ferreira’s detailed review of the [various ways in which doctors treat patients](#) and Dr. Anita Goh’s discussion of [genetic discrimination and HD](#).

### **The Latin American perspective**

The WCHD brought a key South American perspective on HD.

“For one thing, the participation of a number of Latin American neurologists, geneticists, and family members offered some new views of Huntington's disease in countries such as Brazil, Peru, Argentina, and Colombia – countries we often do not hear about at international HD meetings,” Dr. Alice Wexler, a historian of HD science, observed in an e-mail after the congress. “Because the extent of Huntington's in these countries is not well known, many of these researchers presented epidemiological and demographic information that was new to most HD researchers, along with clinical and genetic data.”

Dr. Robert Weiser of Venezuela provided [a view of HD in Maracaibo, Venezuela](#), the world's largest known concentration of HD patients, while [Dr. Carlos Cosentino of Peru](#) and [Dr. Laura Jardim of Brazil](#) presented their unique research on other aspects of HD in the region.

The WCHD highlighted a stark contrast between the First World, where many HD patients can consult with physicians in modern clinics, and Latin America, where large pockets of HD-affected individuals get no medical attention and lack even such basics as clean drinking water.

Several participants noted that such conditions must improve dramatically for these families to take part in the studies and clinical trials crucial for finding treatments, including Enroll-HD, discussed below.

[Rodrigo Osorio of Chile](#) and Alice Wexler's sister [Dr. Nancy Wexler](#) of the U.S. gave moving testimony regarding some of the efforts in the region to improve the conditions for patients, while Dr. Ignacio Muñoz-Sanjuan of CHDI advocated for a recently launched charitable, medical, and care initiative serving Latin America called [Factor H: Hope, Huntington's, Humanity](#). In their [interview with me in Spanish](#), activists Aleska González and Vivian Puchi elaborated on the major challenges facing the HD community in their native Venezuela.

You can watch Muñoz-Sanjuan describe Factor H in the video below.



## Factor H: Hope, Huntington's, Humanity

from [Gene Veritas](#)

30:03 |



[Factor H: Hope, Huntington's, Humanity](#) from [Gene Veritas](#) on [Vimeo](#).

### Riveting stories

The WCHD also gave voice to the riveting stories of HD patients, tested and untested at-risk individuals, caregivers,

and family members.

I participated in a plenary session titled “Coping.” The session began with a troubling presentation by Dutch Ph.D. student Marlous Hubers on the topic of “suicidality in Huntington’s disease.”

“In general, suicide occurs two to eight times more often in Huntington’s disease than in the general population,” Hubers stated. “In general, it’s said that 5.7 percent of all deaths in HD are due to suicide, which makes it the third or fourth cause of death in HD.... Screening for suicidal ideation is most important in patients with a depressed mood, as all studies found it as an important predictor.”

Shaken by Hubers’ incisive data, I started my own presentation on coping, which followed hers, by observing that “Marlous’s extremely important research really brings home some issues for me. It brings up lots of memories of how I’ve been trying to cope with living gene-positive for Huntington’s disease. I think she hit the nail right on the head with respect to how gene carriers need help, and gene carriers are kind of on their own, not only with respect to suicide, but other aspects of the disease.”

Collecting myself, I proceeded with a detailed rundown of the many strategies I have adopted to avoid the onset of symptoms.

You can watch my presentation in its entirety in the video below.



## Coping with the Threat of Huntington's Disease: Gene Veritas speaks at the World Congress on HD

from [Gene Veritas](#)

33:17 |



[Coping with the Threat of Huntington's Disease: Gene Veritas speaks at the World Congress on HD](#) from [Gene Veritas](#) on [Vimeo](#).

In one of the most poignant moments of the WCHD, Dr. Carroll and his wife Megan revealed how they conceived

their HD-free twins, a boy and a girl, using PGD, preimplantation genetic diagnosis.

The topic was particularly striking for Latin Americans. Genetic testing and genetic counseling are still a rarity, and it's unclear how much people know about PGD or have used it. In addition, abortion is illegal throughout most of the region, and, at least in the Brazilian case, legislation against genetic discrimination is lacking.

Watch the Carrolls discuss PGD and hear the audience discussion in the video below.



## Preimplantation Embryo Selection: Avoiding the Transmission of Huntington's Disease

from [Gene Veritas](#)

18:30 |



[Preimplantation Embryo Selection: Avoiding the Transmission of Huntington's Disease from Gene Veritas on Vimeo.](#)

### From young people, with love

The WCHD also had a strong youth presence. I interviewed [England-based Matty Ellison](#), the 25-year-old founder of the [Huntington's Disease Youth Organization](#), about his father's fight against HD, his own experience of testing positive for the gene at the age of 19, and his international advocacy.

The most unusual and beautiful moment of the WCHD came with the performance of [Dingdingdong](#), a group of young adult performers representing French HD families and reflecting many themes relevant to young people facing HD.

The group put on *From Huntingtonland with Love*, a three-part presentation in English involving a short play, a video, and a silent dance performance by choreographer and dancer Anne Collod, in which she mimics the chorea, or involuntary, dance-like movements, suffered by most HD patients.

You can watch Collod in the video below.



## A Portrait in Motion of D., a Man Suffering from Huntington's Disease

from [Gene Veritas](#)

04:27 |



[A Portrait in Motion of D., a Man Suffering from Huntington's Disease](#) from [Gene Veritas](#) on [Vimeo](#).

### A costly event

For me, one drawback of the WCHD was the high cost of the venue, the Sheraton Rio Hotel and Resort, a luxury-class facility on the beach of Leblon, one of the world's most expensive neighborhoods. My room cost nearly \$400 per night, the most expensive room I have ever paid for, and out of reach for vast numbers of Latin American families. (And I couldn't even share that expensive room with my wife, since she was back home with our daughter.)

Along with several family advocates and others, I noted how the exclusive facility, as well as the scheduling of the event the same week as the Rock in Rio music festival, had made it extremely difficult for low and even middle-income families – and perhaps also local research students – to take part.

As an experienced [Brazil hand](#), I thought the organizers might have chosen less expensive, more accessible facilities. In my opinion, holding the meeting in less-glamorous but more convenient São Paulo – where ABH headquarters are located – would have been a good option.

Dozens of Brazilians attended the WCHD, but only a handful of people came from other countries, even from the aforementioned world leader in HD patients, Venezuela.

Nevertheless, as I wrote [in my previous article](#), overall I thought the WCHD ran very smoothly, and families and IHA made important new connections.

### A boost to Enroll-HD

Following the WCHD, CHDI ran a seminar to train medical professionals from Argentina, Brazil, Colombia, and Venezuela how to evaluate HD patients and collect medical information on them for Enroll-HD, the recently inaugurated global HD observational study and database.

Enroll-HD aims to improve tools to assess the disease, identify and characterize biomarkers (signs of the disease) necessary for measuring the effectiveness of treatments, recruit participants for other studies and trials, and improve clinical care everywhere.

Featuring Enroll-HD on one of its panels, the WCHD in Rio provided an important moment for publicizing the program and attracting participants.

(I will report on Enroll-HD at the WCHD in a future article.)

Continuing to foster international connections – *and* greater family participation in future conferences at all levels – will increase the chances of success of Enroll-HD and ultimately the defeat of HD.

Posted by [Gene Veritas](#) at 9:07 PM



Labels: [Brazil](#) , [CHDI](#) , [Elena Cattaneo](#) , [genetic discrimination](#) , [huntingtin](#) , [Huntington's disease](#) , [Latin America](#) , [Maracaibo](#) , [researcher](#) , [RNA interference](#) , [science](#) , [treatments](#) , [World Congress on Huntington's Disease](#)

1 comment:

 **Anonymous said...**

Thank you so much Gene...for those of us waiting...and waiting...and waiting. In 1993 we were told that since the gene had been discovered, there would be a CURE within 8-10 years at the most. Then there was "shock and awe" and two wars for which no one is counting the fatalities that have occurred because of all those billions and trillions of dollars going toward fighting them rather than toward research and the finding of cures not only for HD, but cancer, ALS, MS, and ALL the others. Do politicians know the anguish they are causing generations (3 in our case so far plus at-risk kids) of good families by not funding research and development?

8:21 PM, October 08, 2013

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