5-9-2016

A family united against Huntington's disease

Kenneth P. Serbin
University of San Diego

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)
  - ► November (2)
  - ► October (1)
  - ► September (1)
  - ▼ May (2)
    - Upbeat Huntington's disease film, featuring undaun...
    - A family united against Huntington's disease
  - ► April (2)
  - ► March (1)
  - ► February (2)
  - ► January (2)
- ► 2015 (24)
- ► 2014 (24)
- ► 2013 (30)
- ► 2012 (26)
- ► 2011 (33)
- ► 2010 (26)
- ► 2009 (21)
- ► 2008 (7)
- ► 2007 (7)
- ► 2006 (4)
- ► 2005 (17)

About Me

GENE VERITAS

View my complete profile

HD Links

Huntington's Disease Society of America
International Huntington Association
Huntington's Disease Drug Works

MONDAY, MAY 09, 2016

A family united against Huntington's disease

The overwhelming challenges of Huntington's disease frequently provoke serious conflict – even splits – within families.

The genetic nature of HD, the accompanying stigma, the devastating symptoms, and the heavy caregiving burden often produce emotionally draining conflicts. Then the lack of solidarity impedes the quest for improved care and treatments.

On April 24, during a recent visit to Brazil, I met the extraordinarily united Miranda family. The Mirandas' example provides HD families (and other disease communities) with an alternative to hostility: when adversity hit, they pulled together.

At the invitation of their matriarch, Edília Ferreira Miranda Aded Paz, I met more than a dozen members of the extended family at Edília's spacious home in a prosperous neighborhood on the outskirts of Brasília, the capital.

Edília's father, Marcondes Miranda, died of HD in 1974 at 62. Marcondes left an enormous number of offspring affected by or at risk for the disease. He and his late wife had 14 children, eight of whom inherited the HD genetic defect. Today there are 50 Miranda grandchildren, plus also great-grandchildren and great-great-grandchildren. Some descendants have died from HD, others currently suffer from the disease, and many young ones likely carry the defect.

Edília, who tested negative for HD, took the family's lead on HD in the mid-1990s. At 65, she is the vice president of the União dos Parentes e Amigos dos Doentes de Huntington (UPADH, Association of Relatives and Friends of Huntington's Patients), based in Brasília, and she sits on the board of Brazil's other HD organization, the Associação Brasil Huntington (ABH), headquartered in São Paulo.

"What's interesting about my family is that it united us," Edília, a retired public servant, said of Huntington's. "From the moment we learned that a very large number of people had the gene, we came together."

We spoke in Portuguese during the family meeting, which I recorded so others could listen later. Edília and I had met briefly at the World Congress on Huntington's Disease in Rio de Janeiro in 2013, and she follows this blog. I had long wanted to meet with UPADH members.

"I do everything possible and *im*possible to keep the family united," Edília continued. "Because in my mind, if things are already bad enough with all these people affected, it's even worse if we're alone.

"I don't have the disease, but I never celebrated that fact. Two other sisters who tested negative and I used to say: what's worse – having a disease

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

yourself or seeing siblings die from that disease and knowing that nieces and nephews have the disease? But I took on this mission. I don't do it to show off. I do it because it makes me feel good. I don't have the disease, but I can help."

Edília at her home in Brasília, April 24, 2016 (photo by Gene Veritas)

Unconditional love for the stricken

The meeting at Edília's home was the largest family gathering I have encountered in nearly two decades of HD advocacy. After warm introductions, we sat in a circle in the living room. With Edília's prompt, I began the meeting. I explained that my mother died of HD and that I carried the HD gene.

For the next two hours, as we shared our stories, I witnessed the deep pain that HD has wrought on the Miranda family and their courage to fight back.

Edília's sister-in-law Izaura Maria Soares Miranda lost her husband, Genésio Miranda, to HD. They had three sons and a daughter. All three sons developed HD. The oldest, Marcus Vinícius, died nine months ago, at 46.

"The disease started in him at age 32," Izaura said, still deeply stricken with grief. "I took care of my son for eleven years. I saw him die little by little. I didn't want him to die. I'm still mourning him. A piece of me is missing.

"No mother deserves to go through that, to lose a son. I know he's now at rest. He suffered a lot. But that doesn't diminish the pain."

Seeing her other sons devastated by HD has multiplied her suffering, said Izaura, 65, an attorney who has testified about HD in the Brazilian Congress.

"In the next incarnation I don't want children," she insists. "I don't want to

suffer. I love my children unconditionally. I raised them alone and continue to care for them alone. So I'd kill or die for them. In the next incarnation I don't want to suffer."

Edília's nephew Félix (left), friend Estela, sister-in-law Izaura, and niece Samantha (photo by Gene Veritas)

The other relatives present revealed many other difficult HD challenges: caring for parents and children stricken with the disease, pondering genetic testing, and pooling resources to aid affected relatives unable to earn a living, including one symptomatic woman with young children.

In the words of one niece, Edília has "mobilized the entire family" to assist relatives in dire need because of HD.

In another example of unity, Edília noted the regular collaboration between the UPADH and ABH. The two organizations emerged independently of each other based on the needs of the families they serve in their particular regions of Brazil.

The Mirandas make history

According to Edília and a 2009 news article on the family, in 1995 the Mirandas became the first Brazilian family to undergo genetic testing for Huntington's disease. Scientists identified the disease-causing gene in 1993, making such a test possible.

Advised by a neurologist in Brasília, the extended family pooled its resources and paid about $80,000 for the tests, done in a lab in another city. The family elected Edília to receive the results for the entire family.

Very quickly, most of the family regretted the decision to get tested: HD was untreatable, so what good did it do to know? Edília knew she had tested negative, but at the time nobody else in the family wanted to learn the results. So Edília didn't share the documents with anybody.

Worse, at the time Brazil lacked protocols for genetic testing. Such protocols include a waiting period before collecting the DNA sample, as well as genetic and psychological counseling.

The Brasília neurologist simply handed over the test results with no additional information, Edília recalled.

"As a consequence of that, big changes were made in way test results had to be delivered," Edília said.

In 2008, Edília removed the genetic test documents from the safe in her house and burned the results of those who had tested positive for HD but didn't want to know their status. Edília had memorized the results; at the April meeting, she discussed some of them openly.

"Yes, I burned them, because looking at them brought great suffering," Edília wrote after our encounter in Brasília. Without psychological support for the family or the hope of treatments, "I felt at rock bottom," she recalled.

Anxious to end the 'nightmare'

Today Edília and her family have greater hope.

At the meeting on April 24, the Mirandas eagerly awaited news on the latest HD clinical trials. They were excited to meet an advocate from the United States, where the HD cause is relatively strong and many companies and universities have labs focused on finding treatments.

The Mirandas especially wanted to know about the Ionis Pharmaceuticals Phase I HD gene-silencing clinical trial currently in progress in England, Germany, and Canada. I reported that the first group of gene-silencing volunteers had safely completed their portion of the trial, and that the chief HD drug hunter has expressed confidence that effective HD treatments will eventually appear (click here to read more on these developments).

The family hopes anxiously for a cure to be freed of the "nightmare" of HD, said Jucilene, a niece who struggled terribly with fear before testing negative in 1995.

Noting that I had avoided symptoms into my 57th year, the Mirandas also wanted to discuss my personal strategies for avoiding the inevitable onset. We covered the gamut, from supplements to psychotherapy, exercise to healthy eating.

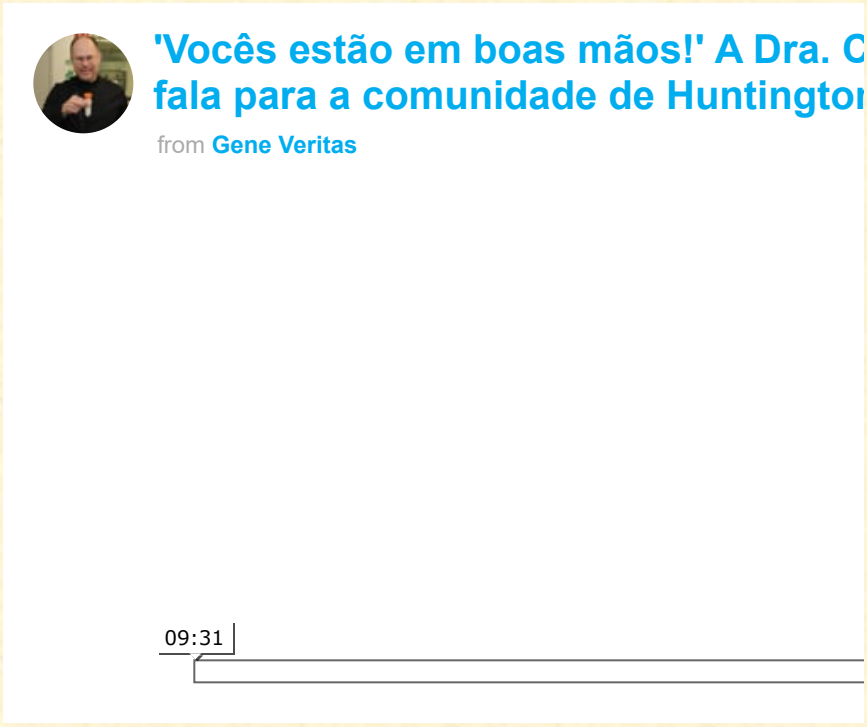
Gene Veritas (seated, center) with members of the Miranda family (personal photo)

I promised further news from two key, Portuguese-speaking members of the effort to develop HD treatments: Celina Zerbinatti, Ph.D., vice president for biology at Evotec, a Germany-based drug discovery company partnering with CHDI Foundation, Inc., the nonprofit virtual HD biotech, and Cristina Sampaio, M.D., Ph.D., CHDI's chief clinical officer and one of the individuals responsible for Enroll-HD, the CHDI-sponsored global patient registry and clinical trial platform.

"You are in good hands!" Dr. Zerbinatti said in an interview aimed at the Brazilian HD community in which she outlined the efforts of CHDI and Evotec.

In another interview, Dr. Sampaio explained the importance of Enroll-HD for Brazil and urged Brazilian advocates to keep pushing for the adoption of the program in their country.

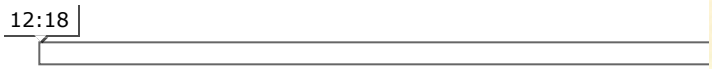
I conducted both interviews at the annual CHDI conference in February. You can watch them in the videos below.



'Vocês estão em boas mãos!' A Dra. Celina Zerbinatti fala para a comunidade de Huntington no Brasil from Gene Veritas on Vimeo.

A Importância do Enroll-HD para a Co Huntington no Brasil

from **Gene Veritas**



A Importância do Enroll-HD para a Comunidade de Huntington no Brasil from Gene Veritas on Vimeo.

Enroll-HD rejected in Brazil

With large HD families such as the Mirandas, Brazil and other Latin American countries could play a pivotal role in defeating HD by providing badly needed volunteers for crucial research studies and clinical trials. The world's sixth largest nation, Brazil has an estimated 20,000 HD-affected individuals.

However, Enroll-HD currently functions in only two countries in the region, Argentina and Chile.

In October 2015, CONEP, Brazil's National Research Ethics Commission, rejected CHDI's proposal to set up Enroll-HD there.

"It never occurred to us that Enroll wouldn't come to Brazil," Edília said. "Enroll is very important for us."

Reconciling different standards

Advocates aim to reverse the commission's decision.

On March 28, Edília, ABH President Vita Aguiar, three former ABH presidents, and other advocates met in São Paulo with the CONEP coordinator, Dr. Jorge Venâncio. Edília's niece Taís, an attorney, also took part. She is at risk but has not tested for the disorder.

"We were very well received," Edília said. "It was a very productive meeting. We wanted to know why the Enroll application hadn't been approved."

According to Edília, Dr. Venâncio explained that the CHDI applicants had not answered all the questions posed in the government paperwork. In addition, the Brazilian officials disagreed with some aspects of the international research study standards included in Enroll-HD.

Edília cited the example of genetic testing. In the Enroll-HD program, which collects participants' blood samples and tests for the HD genetic

defect, the participants can decline to learn their genetic status.

"With the Brazilian government, the patient has to know," Edília said, referring to the country's rules for research studies.

As one of Edília's relatives pointed out, Brazil's requirement will diminish the number of volunteers willing to offer their blood for Enroll-HD. Scientists study aspects of the blood to advance the effort to discover treatments.

For those who would participate and learn their genetic status, either Enroll-HD or the local clinics involved in the program would need to provide genetic and psychological counseling, Edília explained.

As the Miranda family's earlier experience with genetic testing starkly illustrated, Brazil lacks an adequate genetic counseling infrastructure. A recent news report noted that this continent-sized country has only 100 geneticists, for example.

A pledge to resolve issues

Despite this and other disagreements over protocol, Edília remained optimistic that Enroll-HD would enter Brazil. She said that Dr. Venâncio pledged to help resolve all of the pending issues. He guaranteed a response to a new application within three to six months, she said.

The Brazilian advocates also await resumption of the initiative at CHDI, where the long-time Enroll-HD coordinator, Joe Giuliano, recently left to take a position elsewhere.

"The coordinator of CONEP asked us to notify him as soon as we submit the new application so that he can give it priority," Edília said.

Long-term hopes

My trip to Brasília marked milestones in my journey as a college professor, Brazil specialist, and Huntington's disease advocate. I also had the chance to visit my brother-in-law and his family.

From April 25-28, I helped evaluate proposals to establish research in the social sciences and humanities to be funded by Brazil's National Council for Scientific and Technological Development. The prestigious multidisciplinary meeting of some 40 researchers from around the world widened my perspective as a scholar branching into the history of science, technology, and medicine. I hope this endeavor will enhance my ability to interpret the history of the HD cause and advocate even more effectively for it.

On the evening of April 28, for my research on Brazilian politics, I attended a turbulent hearing of the committee installed in the Brazilian Senate to consider the charges of impeachment brought by the Chamber of Deputies against President Dilma Rousseff. On May 6 the committee voted 15-5 to recommend the charges to the full Senate.

I was thrilled to have the health and clarity of mind necessary to witness this historic moment: I had always thought that by now HD would have prevented me from taking the arduous trip to my second home.

Despite Brazilians' current pessimism about their country's immediate political and economic future, I felt a renewed sense of hope for the long term after meeting the Mirandas.

Led by Edília, they will not rest until the day HD no longer threatens their family and the families of so many others.

Gene Veritas (aka Kenneth P. Serbin, Ph.D.) at Brazil's Congresso Nacional building in Brasília, April 28, 2016 (photo by Lucas Souza)

Posted by Gene Veritas at 12:38 PM

Labels: advocacy , at risk , Brazil , Edília Miranda , Enroll-HD , genetic counseling , genetic testing , HD gene carrier , Huntington's disease , Miranda family , psychological counseling , solidarity , stigma , symptoms

1 comment:

Anonymous said...

sent you an e-mail at earthlink, do you still use that acct/

11:47 PM, June 29, 2016

Post a Comment

Newer Post Home Older Post

Subscribe to: Post Comments (Atom)