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Braving bioethical challenges: the importance of 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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GENE VERITAS

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Braving bioethical challenges: the importance of Huntington's disease

Huntington's disease, one of the first conditions for which a predictive genetic test was developed, spotlights the psychosocial ramifications of the Genomic Era.

In addition to the profound impact of HD on people's health and social well-being, the difficult decisions involved in genetic testing have created new ethical challenges.

Over the past few decades, the rapid advance of medical and scientific research has caused ethics – our standards of right and wrong and the study of those standards – to expand into *bio*ethics.

Bioethics is a vast topic. Georgetown University, for example, has an entire library dedicated to research on bioethics, and a number of other universities have centers dedicated to the subject.

Biomedical innovation puts bioethics into a seemingly constant state of flux.

The passage of the Genetic Information Nondiscrimination Act of 2008 (GINA) and the Affordable Care Act of 2010 (Obamacare) are two prominent examples of how society has sought to adapt to new biomedical realities and ethical consequences. GINA seeks to protect individuals from new forms of discrimination made possible by advances in genetics, while Obamacare aims to make health care more inclusive as it undergoes profound transformations.

HD families like mine have *lived* on the frontier of bioethics, often constructing new, personal solutions to the predicaments posed by the disease.

Understanding our contribution to this historic process helps us appreciate our part in the overall effort to combat disease.

New tools, new challenges

I addressed the topic of HD and bioethics at the invitation of the graduate program in bioethics at the Centro Universitário São Camilo, a private Catholic college, in São Paulo, Brazil, during a presentation on September 21.

About 50 people attended the event, including at least a dozen members of the HD community and also Dr. William Saad Hossne, the program's founder, described by one writer as "the guardian of bioethics" in Brazil. Started in 2004, the program was the first of its kind to receive official sanction.

View my complete profile

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
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HD Blogs and Individuals

Chris Furbee: Huntingtons Dance
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Heather's Huntington's Disease Page

Gene Veritas speaking at the Centro Universitário São Camilo

Focusing on how the new "tools" of medicine and biotechnology have deepened our understanding of human biology, I explained how my family braved three predictive tests in just five years: my mother's confirming test for HD in 1995, my own gene-positive result in 1999, and our daughter Bianca's negative test while still in the womb shortly afterward.

All of these tests brought potentially fatal news: a positive test for the HD mutation meant a 100 percent chance of developing the untreatable disorder.

"Because Regina and I wanted to have children, I also had to think about whether I wanted to get tested," I told the audience, speaking in Portuguese.

Rather than following my initial impulse to get tested immediately after learning of my mother's results, I waited for several years. As I explained to the audience, my mother's geneticist had warned me of the possibility of discrimination by my employer, health plan, or insurance companies.

As demonstrated by the discussion around GINA, discrimination has become a major concern of bioethics.

The risks in having a family

"I did the test, and, unfortunately, I tested positive for Huntington's," I continued.

I showed the audience slides illustrating the varying number of CAG repeats (part of the "alphabet" of our DNA) on the huntingin gene. People normally have 10-26 CAG repeats on this gene. An expansion of the gene to 40 repeats signals that a person will develop HD. The tests for both my mother and me showed 40 repeats.

Research shows that the higher the number of repeats, the earlier the disease usually starts, with juvenile onset HD becoming possible if the repeats exceed 80, although even fewer repeats have caused this form of the condition.

Because of the instability of the HD-afflicted male's huntingtin gene in the reproductive process, he can pass on a much higher number of repeats and possibly trigger juvenile HD.

"Having a family becomes like the Way of the Cross," I said with pain in my voice. "In our case, because we wanted to have a family – and that's

why I got tested when I did – we faced a third test. First my mom's. Then mine. Then a third one: of our potential child.

"A low number of repeats: no possibility of having the disease. As the number of repeats rises, the possibility of the disease increases.... The more the repeats, the earlier the disease manifests itself, to the point where five to ten percent of the cases are juvenile Huntington's."

I pointed on the slide to a picture of Olivia Ruggiano, a 12-year-old girl who died of juvenile HD in 2012.

"In my case, with 40 repeats, I could pass on to another person 45 or 55," I continued. "There's a case where a father has 50 some repeats and the children have 80 or 90 repeats. That's when juvenile Huntington's happens."

Very serious questions

I then delved into the heart of HD and bioethics as I had not done before in such detail in a public presentation.

"A family that faces that situation is suddenly confronted with two very serious questions," I said. "If they are thinking of the possibility of aborting the fetus, at what number of repeats would they abort? If you're a couple with the father carrying the gene and the mother gets pregnant, and you're afraid that the child could have the gene, you can test the child in the uterus to see what type of gene it has, whether it's normal or abnormal. If it's abnormal, you can know exactly how many repeats it has.

"And that's where a question of bioethics is forced upon people. Are you going to have that child – or not? Are you going to face a situation of death at the age of nine or 12? Or are you going to end the pregnancy?"

I explained that, living in California, Regina and I faced the additional burden of raising a potential child without familial support. My father dedicated himself to caring for my mom back in my home state of Ohio, while Regina's parents lived in far off Rio de Janeiro.

"How would Regina be able to care of me, a sick person in his forties or fifties, and also a child with symptoms or dying early?" I asked, pointing again to the picture of Olivia.

"These were the questions we dealt with and reflected on as we embarked upon the pregnancy," I observed. "Today there is a method for avoiding that question, with the implantation of healthy embryos. In 1999, that technique didn't exist. The only way was to get pregnant, then test."

Fighting on other fronts

The day our geneticist called with the news of Bianca's negative test in the womb was the happiest of our lives to that moment.

The next slide in the presentation showed two pictures: one of Regina, our gene-negative baby Bianca, and I together in the hospital the day of her birth, another of me clutching our "miracle baby" close to my face.

That terribly difficult and drawn-out part period forms just one part of our journey with HD.

As I pointed out to the São Paulo audience, HD families live the reality of bioethics in numerous other ways: by combatting the stigma and discrimination associated with the condition, negotiating intra-family conflicts arising from the disease, advocating for new and controversial treatments like stem cells, struggling to obtain various kinds of insurance,

facing financial ruin, and dealing with the lack of care facilities and personnel specialized in HD.

Sadly, I also reminded that audience of the high rate of suicide among HD-affected people. Euthanasia is another bioethical issue that comes into sharp focus for HD families.

Emotional testimony

After my 85-minute presentation, the audience offered commentary and questions for another 50 minutes. The emotional testimony from members of HD families and the poignant questions from the audience further underscored the seriousness of the bioethical issues surrounding HD and confirmed their global nature.

One man in his 30s cried as he recalled how his sister, who has the involuntary movements typical of HD, was called a "drunk" by the children at her 12-year-old daughter's school.

A middle-aged woman told how her brother, a computer programmer, lost his job after his performance declined significantly. Despite his obvious cognitive difficulties and aggressive behavior, two telltale signs of HD, both a caseworker and government psychiatrist working for the Brazilian social security system denied him public benefits.

"The psychiatrist said he was able to work and had no problems whatsoever," said the woman, who quit her job to care for her brother at home.

The family appealed the decision, but was denied again. They have sued in an attempt to obtain benefits.

At the last hearing in August, held before a federal judge, the caseworker, still unaware of how HD symptoms are manifested, asked whether the HD man drank alcohol.

At my talk, the HD man's sister referred to government doctors handling the request for benefits as "ignorant" and "stupid." The case is still pending.

"I'm angry and worn out," she said, adding that she is attempting to bring the case to the attention of the Brazilian media. "We need help."

I noted that in the U.S., HD advocates are working towards passage of a federal law to oblige the Social Security Administration to remedy a similar situation in which an inaccurate, outdated definition of the disease has kept many afflicted individuals from obtaining assistance.

Proactive involvement and the hope of treatments

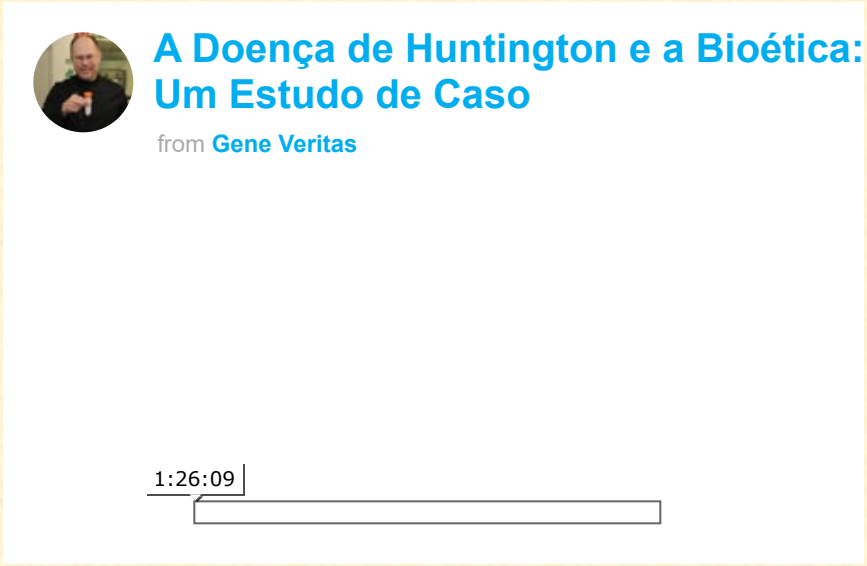
Another, more positive area of bioethics involves participation as subjects in research studies and clinical trials. On this front HD people, gene carriers, untested at-risk individuals, and other family members are taking a proactive approach to contributing to the search for treatments and a cure, usually in a context of high bioethical standards.

Ultimately, allowing HD patients to manage their symptoms with effective remedies, or perhaps someday even curing the disease, will obviate many of the bioethical challenges, although new ones surely will arise – for example, as gene-positive people clamor to try untested drugs.

Our community can and should continue to show leadership on these issues.

For now, as I concluded my presentation, "It's time to conquer Huntington's!"

(The many Brazilian readers of this blog can watch my presentation and the Q & A in the videos below.)



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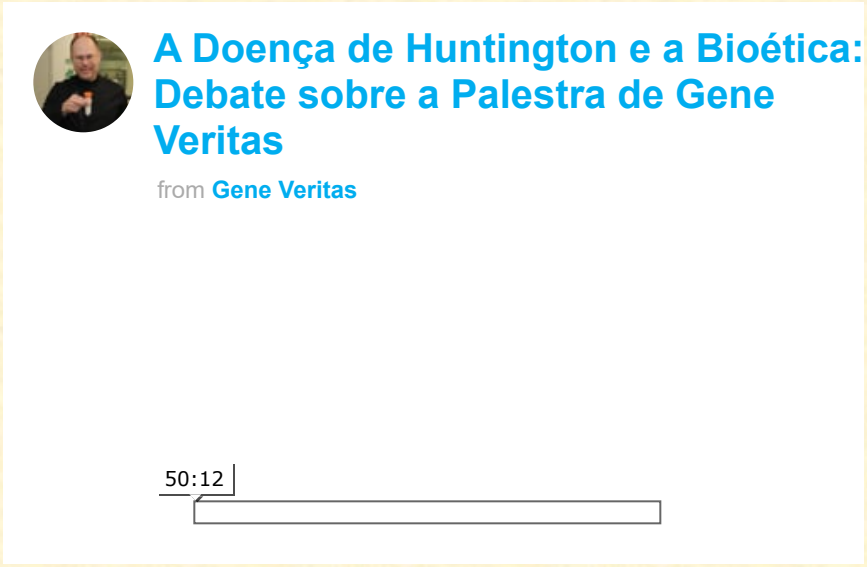

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Posted by Gene Veritas at 2:51 PM

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