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This Thanksgiving, let's show gratitude for disease researchers and drug hunters

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.

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TUESDAY, NOVEMBER 22, 2016

This Thanksgiving, let's show gratitude for disease researchers and drug hunters

This Thanksgiving, let's pause to show gratitude for the many researchers and drug developers in America and around the world who strive to treat diseases and improve the quality of health for all.

As we await the first effective treatment for one of the world's cruelest maladies, we in the Huntington's disease community feel especially grateful for the efforts of the scientific, medical, and biotech communities to bring relief from HD's devastating cognitive, motor, and behavioral symptoms.

Our cause is urgent: HD strikes in the prime of life and is ultimately fatal. Stopping HD would mark a historic step in the quest to conquer brain diseases.

As a presymptomatic carrier of the HD gene, I found inspiration for Thanksgiving 2016 in the powerful speech last February by HD advocates Astri Arnesen and Svein Olaf Olsen at the 11th Annual HD Therapeutics Conference, sponsored by CHDI Foundation, Inc.

Leaders of the HD cause in their native Norway and in the European Huntington Association (EHA), this indomitable couple bared their souls about marital commitment, denial, genetic testing, and raising a family.

Their goal: to motivate – and demonstrate appreciation for – the audience of more than 200 world-class HD researchers, physicians, and drug company executives.

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Svein Olaf Olsen (left) and Asti Arnesen at the 11 Annual HD Therapeutics Conference. Astri is president of the EHA and Svein Olaf a member at large of its board (photo by Gene Veritas, aka Kenneth P. Serbin)

'No questions asked'

Astri and Svein Olaf titled their presentation "HD – more than a disease!"

"HD is much more than a disease," Astri said leading off. "It affects every part of your life, and it's really a part of who you are."

Astri's family traced back HD to a maternal great-grandmother. Her maternal grandfather and her mother also developed the disease.

However, as is so often the case in HD, nobody in Astri's family discussed the condition, even though they witnessed relatives' debilitating symptoms each day.

"You just sensed: no questions should be asked," Astri explained. "My mother grew up in this family."

A special education teacher, Astri in the 1980s fell in love with a co-worker, Svein Olaf. She divorced her husband – a brief marriage she had entered to escape caring for her HD-stricken mother – and started a relationship with Svein Olaf.

'The greatest gift of all'

Svein Olaf knew of Astri's 50-50 chance of carrying the HD gene. However, the young couple didn't discuss HD much. Without

Astri getting tested, they had two daughters in the early 1990s, before the discovery of the huntingtin gene in 1993 and the development of a definitive genetic test.

After their older daughter Jannike turned 18 in 2009, she wanted to get tested for HD.

"I said to her, 'That's not possible,'" Astri said. "'I could not by any means let you go through that. I will do the test [first].' So the day before Christmas, I called the doctor."

The process took weeks. The waiting and uncertainty were so traumatic, Svein Olaf told the audience, that afterwards he could not "remember anything about the tests. Nothing."

When the couple visited the clinic to obtain the results, the doctor practically came running to Astri. "You don't have the gene!" she exclaimed.

Svein Olaf crashed his hand on a table. At that moment, he vowed to marry Astri, something they had planned for years but always put off.

"It was really the most amazing thing to come home and tell our daughters that there is no risk anymore," Astri remembered. "That was really for me the most important thing. You can handle your own situation, but knowing that I hadn't passed it on to them was really the greatest gift of all."

The hope of science

Since then the couple have dedicated themselves to the HD cause – in part because Astri's youngest sibling, Arne Dag, was stricken with HD.

"He was a brilliant young man," Astri said. "He was really teaching my father, an engineer, how to fix the car when he was six years old. So he was a future engineer coming up. But in his early 20s, he's starting to have trouble. He doesn't manage to finish his studies."

In a typical symptom of HD, Arne Dag became deeply depressed. He was formally diagnosed in 2005.

At the 2010 meeting of the European Medicines Agency (the counterpart of the U.S. Food and Drug Administration) in London, Astri showed a video of a short interview she did with her brother.

In the clip, Arne Dag struggles to speak, and his body twists and turns because of chorea, the involuntary movements caused by HD.

Arne Dag tells Astri that he wants to participate in a clinical trial as soon as possible. She asks him about his hopes for the future.

"I hope that science is on my side," he says, laughing.

Participants at the 11th Annual HD Therapeutics Conference, Palm Springs, CA, February 2016 (photo by Gene Veritas)

Thanking the researchers

Arne Dag died in April 2015 at the age of 46.

"But he hoped," Astri told the conference attendees. "And that was really important in his life. He hoped for the hard work you [scientists] are doing to give results for him and for us."

As a slide showing infants appeared in the background, she added: "And that's what we are fighting for, and that's why we are so involved in this work. Because now, it's not about him. It's not about me. It's about all these lovely children and grown-ups living with a risk, living with the gene, and who really put their hope on you and your work.

"And no matter how long it takes, that is so important in our daily lives. Knowing that you go to work really makes our day easier. For me, hope has really been my way of coping and dealing with it, and it is for a lot of us. We are many, many out there who support you, who need you, and who are waiting for results.

"So we want to thank you so much. It's amazing to see how many fantastic, brilliant researchers that are in this field. We are so grateful for that."

Happy Thanksgiving!

You can watch Astri and Svein Olaf's presentation in the video below.

'HD - more than a disease!' A presentation by Astri Arnesen and Svein Olaf Olsen

from **Gene Veritas**

1:10:32

Posted by Gene Veritas at 12:07 PM

Labels: Astri Arnesen , biotech , brain , CHDI , cure , drug development , genetic testing , gratitude , HD gene carrier , hope , Huntington's disease , love , researchers , scientists , Svein Olaf Olsen , symptoms , Thanksgiving , treatments

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