Our inspiration: people impacted by Huntington's disease

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Roche in partnership with Ionis Pharmaceuticals is committed to supporting the needs of the community living with Huntington's disease (HD) and developing treatments for those affected by this devastating disease. We invited journalist and HD patient advocate Charles Sabine to give his perspective at an event organized at Roche in recognition of World Rare Disease Day 2018.

Roche's drive to develop treatments for HD is inspired by the people whose lives are affected by this progressive and fatal inherited disease. Listening to and partnering with patients, family members and advocacy organizations helps us better understand the impact and challenges of the disease for the entire community living with HD. To this end, we invited the Emmy award-winning journalist and HD patient advocate Charles Sabine to speak at an internal Roche event for World Rare Disease Day in February 2018. His powerful story about his journey with HD captivated the audience of more than 350 people. We devote this sponsor's advertorial to Charles' story and his passionate dedication to driving change for the families impacted by HD around the world.

CHARLES' STORY

Nearly 25 years ago, Charles' father was one of the first people in the world to be genetically tested for HD, following identification of the causative CAG-repeat expansion in *HTT* in 1993. Charles was told that his father was suffering from an incurable, untreatable genetic disease, and that he himself would have a 50/50 chance of developing the disease as well, most likely in the next 10–15 years. For Charles' father, worse than death and the lack of any established standard of care was the loss of dignity that came as the once-proud career soldier watched his friends and family wince while his body and mind became unrecognizable. Worse still was the knowledge that he could have passed that nightmare on to his sons. Charles' elder brother, John, studied law at the University of Oxford and went on to be described by a high-court judge as "one of the most brilliant barristers of his generation". Because of the progression of HD, John is no longer able to work and currently requires 24-hour medical care and assistance.

Charles discovered in 2006 that he also has the causative CAG-repeat expansion in *HTT*, meaning that the disease that took his father and is inflicting the same terrible decline on his brother will take him too. His neurologist told him that there was nothing he could do other than to live his life as well as he can. Not an hour went by when Charles did not picture how his quality of life would drain away, or when he did not fear that, however much his friends might promise they would always come and see him, they would not really want to – just as he did not want to see his father when he lost his ability to converse with his family.

Two years after his test, Charles swapped his 26-year career as a television war correspondent on the battlefield for working as an HD advocate to fight against the shame, ignorance, misunderstanding and lack of hope encountered by families living with HD. His work with researchers, clinicians, students, patients, families, carers, pharmaceutical companies, politicians and charities has taught him that, contrary to the original pessimistic advice of his neurologist, there is much that he can do to battle HD. The problem is finding time to do it all.

One stirring example of Charles' empowered action was the 'HDdennomore' campaign event on 18 May 2017, co-organized by Charles and the social non-profit group Factor H. The campaign found a way to bring HD out of the shadows by convening Catholic families from the remotest and poorest parts of South America at the Vatican to meet their Pope, some leaving their villages for the first time. The Pope's physical embrace of people with HD and their families on that day was met with overwhelming emotion and gratitude as he became the first world leader to recognize the plight of people living with and affected by this heavily stigmatized disease.

Charles is one of the many people who have volunteered to further the scientific understanding of HD through participation in clinical trials. He completed the TRACK-HD observational study that confirmed the value of neurofilament light chain as a blood-borne biomarker of progressive neuronal damage in HD and other neurodegenerative disorders[1]. Charles also participated in HDClarity, an international study[2] of cerebrospinal fluid collection, which aimed to facilitate development of therapies for HD.

Charles Sabine with his elder brother John (right) in 2010. Like John, Charles inherited an *HTT* gene with a CAG-repeat expansion from his father and will also develop Huntington's disease (Image: Martin Solyst).

He advocates that dignity can be regained and morale boosted by engaging with scientific research.

Charles has also contributed to efforts to give people living with HD unprecedented access to information about HD research and patient care through websites such as HDBuzz and Huntington's Disease Youth Organization. These activities don't just bring people together, they collate expertise and help to establish a standard of care that Charles found to be absent when his father was alive.

As a journalist reporting from war zones, Charles has known fear, danger and death, and has seen first-hand that humanity perishes when dignity and hope are lost. In spite of this, he believes that the HD community has survived because the very best of humanity surrounds it. The forces of trust and collaboration empower communication; communication leads to understanding; understanding dilutes fear; and the vacuum left by fear can be filled with hope. Compared with the prospect of no treatment at all, even the possibility of a treatment offers an absolute difference: the difference between losing the will to go on and gaining hope.

Charles Sabine and everyone impacted by HD are our inspiration at Roche as we embrace our commitment to, in Charles' words, "serve as the custodians of their voyage". The first step in this journey together is to collaborate with the HD community and engage with global health authorities as we move from early clinical testing to designing a comprehensive clinical development programme in HD. This forms one part of Roche's broader research in neuroscience and rare diseases, such as Alzheimer's disease, multiple sclerosis, spinal muscular atrophy, Duchenne muscular dystrophy and autism.

Acknowledgement

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