News from the Hereditary Disease Foundation

Why We Do It
May is Huntington's Disease Awareness Month. But every month, we aim to increase awareness about the devastating impact of HD and emphasize how research is making a difference. And every moment is a good one to celebrate HD patients and their families. Each has a unique story of heartbreak, resilience, courage, hope, love, and families coming together under the most challenging of circumstances.

Why are we funding scientists in laboratories around the world? Why are brilliant researchers devoting their careers to finding treatments for Huntington's disease? It is for the patients and their families. Their faces are always before us, and their stories inspire us.

Chris O'Brien: I Am the DIFFERENCE
By the time Chris O'Brien was born, his mother was already showing signs of Huntington's disease. He is the youngest of Mrs. Alice and Dr. David O'Brien's seven children. Like his older siblings, he was born with a zeal for life and from an early age, knew that he was at risk for HD.

Chris O'Brien, Mt. Cho Oyu in the Himalayas

In his junior year at Yale University in the early 1990s, Chris and his father attended a medical meeting on HD. It was there that he met Nancy Wexler. In order to isolate the HD gene, Nancy was leading a team of researchers to collect data from the world's largest Huntington's disease family who lived in Venezuela.

Chris worked with Nancy for a few summers at the Hereditary Disease Foundation and accompanied Nancy and her team to Venezuela several times. On his last trip to Venezuela, there was a military coup. Everyone but Nancy left for safety reasons. Chris stayed to help and protect Nancy. "I never really get scared," Chris says. "I've always been unruffled and determined."

Meeting and working with Nancy opened up a whole other world of research to Chris.

"Nancy was my inspiration," Chris says. "That's when I decided I wanted to become an MD/PhD and a neurologist."

Nancy's work in Venezuela led to the discovery of the location of the HD gene, making possible genetic testing for HD. Chris eventually underwent testing and found out he carried the expanded repeat and would inevitably get sick with HD, just like his mother Alice. Knowing his fate didn't change Chris's plans. He would complete his education and training, and along the way, travel the world and climb mountains.

Halfway through medical school, Chris, his father, and his brother Mike climbed Mt. Kilimanjaro in Tanzania. That experience and their passion for seeking new challenges inspired Mike and Chris to hatch a daring plan to raise money for the Hereditary Disease Foundation. They proposed to raise $100,000 by becoming the first pair of American brothers to summit Mt. Everest together. It was Chris's last year of medical school, but he was determined to graduate and meet his fundraising and climbing goal.

By the time the brothers made it to the Everest Base Camp in the spring of 2005, they were still $85,000 short of their fundraising goal, but they were not deterred. They believed that once they reached the summit the money would come along with the publicity. Tragically, while climbing through the Khumbu Icefall, Mike fell into a deep crevasse and was gravely injured. Chris rappelled down to try to save him. Mike was conscious for about 45 minutes, and then started to slip away. Chris and another climber performed CPR on him for two hours until a rescue team arrived from base camp. Three hours after falling, Mike succumbed to his injuries with Chris by his side.

Mike and Chris O'Brien, Mt. Rainer

The New York Times and media worldwide covered the tragedy, raising awareness of their cause and exceeding their fundraising goal. In the end, Chris and Mike's heroic efforts raised $350,000 for the Hereditary Disease Foundation from people all over the world.

Chris graduated from a combined MD/PhD program at Thomas Jefferson University in 2005. He did his internship at Cooper University Hospital in New Jersey and then moved to Texas for his neurology residency, but by his third year, he was no longer able to keep up with the demands of the job. The cognitive symptoms of HD which he had begun to exhibit made it impossible to pass the yearly exams.

"I didn't think it would happen that early. I thought I would practice medicine for a little while," Chris says. But ever the optimist, Chris says that he wouldn't have met his wife, Lily, if he hadn't gone to Texas.

"Five years ago there were no treatments for HD. Now there are options and potential for disease-modifying treatments that go beyond just treating symptoms. It's an exciting time for the HD community!"

Although no longer able to work, Chris continues to exercise every day, running with his three dogs or working out in the gym, and he still goes ice climbing every winter with a group of medical school friends.

Chris also continues to raise awareness for Huntington's disease. He has attended the Huntington Study Group's annual meetings as a neuroscientist for the past few years. He participates in HD research, having taken part in a trial for tetrabenazine and another for coenzyme Q, and he plans to stay involved.

"There's always more to be done."

Chris with his family at the recent HDF gala: (from left) nephew Anthony, sister Kathryn, Chris, Lily, nieces Dagny and Kristina, sister Meghan, nephew Sean

Chris's sister Meghan (O'Brien) Donaldson is the CEO of the Hereditary Disease Foundation.

"My brother Christopher has always lived his life with intelligence, courage, and a passion for helping others and seeking adventure," Meghan says. "Christopher essentially lost his mother, our mother, to the ravages of HD when he was very young, and, like many people at risk, grew up knowing that he might one day share her fate. Christopher never let that loss or that knowledge prevent him from pursuing his dream of becoming a neurologist and HD researcher who might one day help others with HD nor did it deter him from climbing mountains and traveling the world. For the last decade while Christopher has bravely faced his own HD, he has continued to cherish every day and he rarely allows HD to interfere with his adventures. I am honored and proud to call Christopher my brother and I am truly inspired by his courage, grace and love of life."

A Sister's Story

Karen Newman is inspired by her sister Penny who died of complications from Huntington's disease eight years ago. When Penny was diagnosed in the late 1990s, Karen and her family had never heard of HD and did not know that it was in their family. Her father was adopted, showed no signs of the disease, and had no information about his birth family or their medical history.

"It was a lot to come to terms with and the Hereditary Disease Foundation was a lifeline for me. We did not have the same amount of access to information on the Internet back then. I waited eagerly for the HDF newsletter to arrive in the mail so that I could learn about the latest Huntington's disease research."

Karen with her sister Penny circa 1993

Several years ago, Karen decided to get involved. She is now a member of the HDF Board of Directors and serves as Co-Chair of the 2019 HDF Symposium and Dinner that will be held on Monday, October 28, at the Harvard Club.

"The last two to three years have inspired a lot of hope for me. For the first time in my 19-year involvement with this disease I feel real possibilities for treatments and cures – real hope. Thanks to clinical trials, breakthroughs, incredibly dedicated scientists, pharmaceutical companies and organizations working together we are making real progress."

Karen with her other sister Cindy
at the 2017 HDF gala

Karen speaks about the connection between scientists and HD families.

"Through the HDF, I've met a lot of wonderful scientists who have dedicated their life's work to finding treatments and cures for HD, and I am incredibly grateful to them for their commitment and passion. There is this wonderful relationship that exists between the HD families so deeply impacted by this disease and the physicians and scientists who work to treat and develop cures. The HD community is unique. The individuals and families whom I have met over the years have offered me a supportive network, and I am very inspired by their stories. I am honored to be part of the HDF and to offer my contributions to the greater efforts to finding a cure for HD."

Click here to listen to Karen speak about the impact the HDF's work has had on so many lives.

If you are a member of an HD family and would like to increase awareness by sharing your story in our newsletter, please contact Myrna Manners at mmanners@mannersdotson.com.

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