HDF-Funded Researcher Receives $1.25 Million NIH Grant

Funding the most innovative and promising research is what we do at the Hereditary Disease Foundation. This support can lead to scientists receiving additional funding from other sources for their work to find treatments and cures for Huntington’s disease. How does this happen? The connection between brilliant researchers and visionary philanthropy makes it possible. Perhaps nowhere is this better exemplified than in the story of HDF-funded researcher Anna Pluciennik and HDF Partner in Research Deborah Fine. We invite you to read all about it!

Catalyzing Funding

For Dr. Anna Pluciennik, research assistant professor at Thomas Jefferson University in Philadelphia, HDF funding has been critical in enabling her to advance our understanding of the mechanisms of the DNA repeat expansions that cause Huntington’s disease. “The hope is that as individual factors leading to this DNA
expansion are pieced together, new therapies will be developed to stop the process,” she explains.

“HDF took a risk in supporting me when I was starting out in the field, and I am extremely grateful,” Anna says. “HDF funding is absolutely critical for early-stage investigators who want to enter the Huntington’s disease area, but do not have long track records in the field. This has been crucial in my case by enabling me to secure additional funding to support my research.”

In fact, the National Institutes of Health (NIH) recently awarded Anna a prestigious Research Project Grant (RO1) of grant of $1.25 million over 5 years, which will allow her to continue building her lab and conducting breakthrough HD research.

Anna’s interest in science began when she was growing up on a farm in Poland. “I was driven by an endless curiosity about the animals and plants around me,” she says. “As a kid, I would watch the BBC program Nature every Sunday and that really sparked my lifelong interest in biology and evolution.”

Anna completed her undergraduate and graduate work in Poland at the University of Lodz, where she became interested in the mechanisms of repeat expansions. She came to the United States to conduct postdoctoral research at Texas Medical Center in Houston and then at Duke University. She later joined the lab of Diane Merry, PhD, professor and vice chair of the department of biochemistry and molecular biology at Thomas Jefferson University and a member of the HDF Scientific Advisory Board. Anna was researching another repeat disease, spinal-bulbar muscular atrophy (SBMA), when she became interested in Huntington’s disease.

“My focus on HD has been inspired by recent studies that implicate DNA repair processes in disease manifestations,” Anna explains. She has made several contributions to the understanding of these processes, has established her own lab, and is now taking on new challenges of understanding how these processes drive expansion and disease physiology. Click here to read more about Anna’s research.

Speaking about the importance of HDF Partners in Research, Anna says, “Science is difficult and painstaking. The outcome of investments made in the basic research of human disease can take years to become tangible. Deborah Fine and other supporters deserve the highest praise for their foresight and patience.”
Deborah Lieberman Fine knows all too well how devastating Huntington’s disease is for families. Her grandmother, mother, her two sisters, and several other family members died from the disease. This past year alone, two of Deborah’s nieces, Lizzie and Jody, daughters of each of her sisters, passed away from HD. Both were in their forties.

She recalls her father, Harry Lieberman, a longtime supporter of HDF, sitting her down when she was nineteen and telling her HD was in their family and that she was at risk. “It would be great if this conversation would never have to happen to anybody in the future,” she says.

In the 1970s and 80s, Deborah and her father attended many Hereditary Disease Foundation scientific workshops in California. “It was wonderful to see scientists from labs all around the world talking to each other about their research,” Deborah says. She credits Nancy Wexler, President of the Hereditary Disease Foundation, for making this happen. “Nancy has always been a champion of collaboration. It has made a tremendous difference in the progress of research.”

The Lieberman family established a research prize with the HDF in 1993 to catalyze new efforts following the 1993 HDF-supported discovery of the Huntington’s disease gene. Dr. Gillian Bates, then an up-and-coming junior researcher, received the prize in 1994 and again in 1997. This support was key in enabling Gillian to develop the first mouse model of Huntington’s disease in 1996, a landmark scientific advance which allows researchers to test different approaches to delay or slow the progression of the disease. Dr. Bates is now Professor of Molecular Neuroscience and Co-Director of the Huntington’s Disease Centre at the University College London (UCL) Institute of
Neurology, a Fellow of the Royal Society and a member of the HDF Scientific Advisory Board.

Today, Deborah and her family are supporting the work of Dr. Anna Pluciennik, Research Assistant Professor at Thomas Jefferson University. Deborah, an artist, lives and works in Philadelphia. Supporting a local scientist provides opportunities for her to meet Anna and have a personal connection to the progress of her research.

Deborah values being a Hereditary Disease Foundation Partner in Research because of the broader implications of HDF-funded research. She explains, “The research that HDF has done in the past and continues to do potentially impacts other neurological diseases, such as Alzheimer’s, ALS, and Parkinson’s. Discoveries made by the Hereditary Disease Foundation have affected research in other brain diseases.”

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