



News from the Hereditary Disease Foundation



Dr. Jean-Paul Vonsattel and Dr. Richard Hickman

A multi-institutional study, led by neuropathologists at Columbia University Medical Center/The New York Brain Bank and funded by the Hereditary Disease Foundation, has gleaned further insight into the developing brain of individuals with Huntington's disease (HD) and points to the possibility that therapies targeting the abnormal *Huntingtin* gene may be of benefit earlier in life. The research was published in the journal [Acta Neuropathologica](#) in January.

In 40 years of studying the of brains of people who have died with HD, Drs. Jean-Paul Vonsattel and Richard Hickman noticed that certain developmental malformations are found in a subset of HD brains and are up to eight times more frequent in HD than in brains from individuals who do not carry the gene causing HD. These malformations were

abnormal collections of neurons that had failed to migrate during early development and were found en route to their target destination. They were found in isolation and were often very small, needing a microscope to see them. While these malformations typically do not contribute to the disease itself, this novel finding gives important insights into how the brain develops in HD.

The abnormal *Huntingtin* gene that causes HD is present from the moment of conception and persists throughout life. Importantly, the *Huntingtin* gene is critical for many cellular functions, including healthy development. Recently, several scientific publications have drawn attention to the alterations in the usual brain development in individuals with the abnormal gene. These suggest that although the disease typically presents in mid-life, the disease process likely starts much earlier before symptoms begin. The malformations described in this new study suggest that the abnormal gene may be affecting the normal migration of nerve cells in the brain. If developmental changes are more widespread and actively contribute to the disease over many years before symptom onset, then therapies that target the abnormal gene may be beneficial earlier in life.

This important study is made possible by Huntington's disease patients who have generously donated their brain tissue post-mortem to advance scientific research. Drs. Vonsattel and Hickman have an HDF research grant to conduct human tissue banking for the Huntington's disease community. Their work provides researchers with insights into the disease mechanisms of HD and is paving the way towards better treatments and therapies for HD patients.

Thank you to Kurt Fischbeck

Dr. Kenneth "Kurt" Fischbeck, Chief of the Neurogenetics Branch at the National Institute of Neurological Disorders and Stroke, has been a dedicated member of our Scientific Advisory Board for decades. As a neurologist, he played an essential part of our research team in Venezuela on multiple trips in the late 1990s. In scaling back his many career roles and responsibilities, Kurt is stepping down from our Board. Speaking of Kurt's extraordinary contributions to the HDF, Dr. Nancy Wexler, President of the Hereditary Disease Foundation, said, "Our Scientific Advisory Board has benefited tremendously from Kurt's scientific brilliance, amazing wisdom and sharp intellect. We will miss his willingness to advise with objectivity and thoughtfulness and his wonderful sense of humor. We thank him for his leadership and guidance over so many years."

Stay Tuned!



We're excited to announce our first webinar series will start soon. It will feature brilliant scientists working to identify treatments and cures for Huntington's disease and other brain disorders. Details to come!

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Published February 2021