



The FD Foundation Commemorates Its 70th Anniversary

[A brief look back at the Foundation's 70-year history...](#)

Seventy years ago, a caring and empathetic pediatrician named Dr. Conrad Milton Riley, who with his colleague, Dr. Richard Lawrence Day, had recently identified a rare hereditary syndrome, brought together the parents to form a support group, one of the first parent support groups for a genetic disease in the United States. While the genetic origins were still unknown, the fact that all the families were of Ashkenazi Jewish descent suggested that this group was at risk. With strength in numbers, the parents could compare symptoms, and explore and discuss treatments and care. That year, in 1951, the small group of forward thinking parents formed the Familial Dysautonomia (FD) Foundation.

Today, the FD Foundation works for the benefit of all people afflicted with FD, fewer than 800 ever diagnosed and fewer than 325 currently living worldwide, by supporting medical care and scientific research, as well as conducting social service and public awareness programs. The Foundation continues to be the main source of funding for the Dysautonomia Treatment Center at NYU Langone Medical Center in New York City, the only place in the US dedicated to providing clinical care for people with FD, and the model for two treatment centers in Israel. The Foundation's support helps fund treatments that improve life expectancy and quality of life for people with FD, as well as research seeking a possible cure. In addition, the Foundation sponsors conferences for doctors and scientists to share their knowledge, and since 1985, has hosted FD Day, an annual symposium that brings together families and caregivers for a day of learning and connecting.

In 1969, Dr. Joseph Dancis was treating pediatric patients with rare diseases at Bellevue Hospital in New York City. He asked Dr. Felicia Axelrod, who had just completed her training at New York University (NYU) School of Medicine, to become the attending physician of record for six to 12 FD patients at University Hospital (today called Tisch Hospital and part of NYU Langone Medical Center). Having seen many of these patients and observed their crises during her training, Dr. Axelrod realized the need to collect and systematically organize information about the patients and their medical histories

The following year, Dr. Axelrod established, and the FD Foundation began funding, the Dysautonomia Treatment Center at NYU Medical Center, which she directed until her retirement in 2015, when Dr. Horacio Kaufmann took over as Director of the Center. By the end of the first year, the Center had about 50 patients, many from outside New York. Today, the Center follows about 160 patients and regularly consults with doctors managing the care of those with FD in other locations such as the UK, Israel, Canada and Mexico. A multidisciplinary team of highly trained physicians, nurse practitioners and researchers manages daily patient care, performs annual physicals, handles patient emergencies 24/7 and oversees clinical research. Currently, 12 clinical studies are underway.

The work of our physicians and scientists has had a tremendous impact on prolonging lives. Today, there is a 50 percent chance that an FD patient will live to age 40; the largest number of adults are living with FD since the first diagnosis, the result of the substantially improved care that the Dysautonomia Treatment Center has pioneered over the last half century.

When patients began treatment at the Dysautonomia Center in the 1970s, they completed medical history questionnaires. In the 1980s, Dr. Axelrod took steps to digitize the FD database. Based on the robust data for over 650 patients, in 2017, the Center received a National Institutes of Health (NIH) grant for a natural history study that will help researchers better understand the progression of this disease and develop new therapeutic treatments to treat degenerative FD symptoms that affect older patients such as failing vision, spine curvature, unstable gait and impaired balance and coordination.

In 1990, after consulting with scientists, the Board of Directors determined that the Foundation should direct most of its funds towards research to locate the gene responsible for FD. The Foundation funded the prominent geneticist, Dr. James Gusella, of the Harvard School of Medicine. In 1991, Dr. Susan Slaughaupt joined Gusella's lab as a post-doctoral fellow, bringing a strong background in gene mapping, the process of locating genes on chromosomes.

Thanks to the Foundation's support, collaboration with the Treatment Center, and further funding from the NIH, in 2001, Dr. Slaughaupt discovered the FD gene mutation on Chromosome 9Q. Once the gene was located, potential parents could be tested before conceiving to determine if they carried the gene, and babies already conceived could be screened prenatally. The Foundation advocated for required screening for at-risk families through the American Academy of Obstetrics and Gynecology. FD was added to the Jewish Genetic Screening Panel and is now a routine part of all prenatal carrier screening, resulting in dramatically fewer FD births worldwide.

Dr. Slaughaupt and her team identified the mutation as a genesplicing error in Elongator complex protein 1 (ELP1, also known as IKBKAP); since then, Sue has dedicated her work to discovering how to repair the splicing defect as a route to a therapeutic intervention for those with FD. In 2016, Dr. Slaughaupt's lab generated a phenotypic mouse model (mice with FD) and in 2019 was able to successfully correct the splicing defect in her mice, a treatment that has the potential to improve patients' sensory-motor coordination and intervene in the onset of spinal abnormalities.

The 2020s are an exciting time for the FD Foundation:

- The work of its international Scientific Advisory Board and its support for research has accelerated the search for treatments to slow or arrest FD's degenerative effects; Researchers have been approaching FD from all angles; several potential treatments are currently in the pipeline and could be ready for clinical trials by 2022.
- The NYU Treatment Center is committed to expanding access for patients through annual checkups via telemedicine (begun during 2020's pandemic so that patients would not have to travel to New York) and will continue to build the natural history study for FD, crucial for providing essential knowledge to research on rare diseases, while proceeding with exciting, groundbreaking treatments.