

The Dysautonomia Foundation: Who We Are & What We Do

Familial dysautonomia (FD) is a rare genetic disease that affects the autonomic and sensory nervous systems of children from birth. The most striking symptoms of FD are reduced sensitivity to pain and temperature, and the inability to produce tears. But FD is much more than “no pain and no tears”, it affects every major system of the body, causing severe respiratory, cardiac, orthopedic, digestive and vision problems.

FD was once thought of as a fatal childhood disease, with most children expected to live, on average, only to five years of age. Advances in treatment have dramatically extended life expectancy, but children with FD still suffer from chronic and often debilitating symptoms that prevent them from leading normal lives.

The Dysautonomia Foundation, founded in 1951, is a 501(c)3 nonprofit public charity that raises funds for FD treatment and research. The Foundation funds the world’s only two FD treatment centers. As a result of the work of these centers, quality of life for people with FD has improved significantly.

In 2001, research funded by the Foundation resulted in the discovery of the gene that causes FD. Now we are focusing on research for new genetic therapies to help treat FD, and, hopefully, one day discover a cure. Through our research efforts, vital supportive treatments have been developed and we have been able to identify the genetic mutation that causes FD, resulting in a general population carrier test. Through carrier screening and our awareness campaigns, we have been able to dramatically reduce the frequency of new cases of FD. Even so, new cases do arise, and people with FD continue to face very difficult challenges just to get through each day.

Children with FD **lack the most basic reflexes and instincts that we take for granted.** As a result, their bodies cannot function normally. They cannot control their blood pressure or heart rate, and they lack the ability to suck at birth and the ability to swallow properly. Because they often swallow into their lungs rather than their stomachs, they are prone to pneumonia. As a result, **most FD patients have a feeding tube**, so they can be fed directly into their stomachs and reduce the risk of pneumonia.

In addition to chronic, life-threatening symptoms, FD causes **a mysterious syndrome called “autonomic crisis”** in which patients experience extreme swings in blood pressure and heart rate, along with dramatic personality changes, and a complete shut down of the digestive system. Once FD patients go into crisis, **they cannot engage in any normal activity until hours or days later**, and they may require hospitalization for observation, sedation and hydration until the crisis abates. These crises are often triggered by physical stress, anxiousness or anticipation. Sadly, something as ordinary as looking forward to a birthday party or a vacation can trigger a crisis.

In order to fund our mission, we rely primarily on the donations of private individuals. We run a variety of fundraisers every year, including golf outings, a cycle tour, a bowl-a-thon, benefit dinners, and a commemorative journal. We are extremely proud that such a small organization has been able to achieve significant advances in treatment and research, but we realize that we still face huge hurdles before we can achieve our ultimate goal of a cure for FD. In the mean time, we continue to work toward the best quality of life for people with FD.

Please join us in our efforts to improve the lives of people with FD and raise awareness of FD in the general population and the medical community.

For more information and to find out how you can help people with FD, please contact the Dysautonomia Foundation at 212-279-1066 or info@famdys.org.

The Dysautonomia Foundation, Inc.

56 Years of Treatment, Research and Awareness for FD

Our Major Accomplishments

We have funded research that has led to:

- the understanding of the disease as a neurological condition.
- the discovery of the FD gene.
- a general population carrier screening test, resulting in a significant reduction in the birthrate of new FD cases.

We have established:

- the world's only treatment centers dedicated exclusively to FD treatment and clinical research.

We have successfully advocated for:

- government recognition of FD as a developmental disability.
- social services for FD adults.
- ACOG (a medical standards organization) to require doctors to inform patients of the risk of FD and the need for genetic testing.

Our Major Initiatives:

- **Funding Clinical Care:** We are the primary source of funding for the FD Treatment Centers in NY and Israel, including staff salaries, equipment budget and clinical research projects.
- **Funding Scientific Research:** Basic scientific research and clinical medical research regarding FD, and the FD gene, is currently being funded by the Dysautonomia Foundation at a number of prestigious hospitals and universities, including:
 - Development of animal models of FD - Harvard/MGH and Univ. of Tennessee
 - Clinical investigation of a new medical therapy - NYU Medical Center
 - Genetic analysis of the FD mutation - Harvard/MGH
 - Sleep factors contributing to sudden death - Univ. of Erlangen, Germany
 - Study of gastrointestinal bleeding in FD – Hadassah Hospital, Israel
- **Supplying Informative Material:** The Dysautonomia Foundation provides a continual flow of information to families, the medical community and other lay and professional persons. We work with the medical community to educate doctors about FD, to let them know that they can call on our experts for assistance in treating FD patients.
- **Advocating for the FD Population:** We encourage medical governing bodies to require doctors to inform patients about the existence, risk and treatment of FD. We also advocate on the government level to try to get states to recognize FD as a developmental disability so that FD patients can more easily obtain assistance for FD treatment, and to require insurance companies to provide coverage for the cost of genetic testing and medical care for FD.

The Familial Dysautonomia Treatment Centers

The Foundation funds the world's only two FD treatment centers at NYU Medical Center, established in 1969, and at Hadassah Hospital in Israel, established in 1981. These treatment centers have increased life expectancy and improved the quality of life for people with FD around the world and serve as resources for all patients and physicians in assessing and treating FD.

The centers continue to grow to this day; construction for an expanded treatment facility and research lab at NYU will begin in Fall of 2007. In addition, the Center also serves as a referral center for other hereditary sensory and autonomic neuropathies (HSAN) and for other pediatric disorders with autonomic dysfunction.

Familial Dysautonomia Fact Sheet

Overview:

In individuals with FD, a progressive neurogenetic disorder, the autonomic and sensory nervous systems malfunction. Symptoms vary, and may include insensitivity to pain, unstable blood pressure and body temperature, absence of overflow tears, frequent pneumonia, and poor growth. FD is often associated with a shortened lifespan. Individuals with FD suffer from episodes of cyclical vomiting accompanied by high blood pressure and increased heart rate, sweating and fever. These “autonomic crises” are one of the most devastating symptoms of this disease.

Carrier rate in Ashkenazi Jews: 1 in 27 / Carrier rate in the general population: Unknown

Age of onset: Birth

Symptoms and severity vary in each patient; symptoms include:

- Absence of overflow tears / corneal drying
- Poor suck at birth
- Drooling
- Swallowing & feeding problems
- Hypotonia / poor muscle tone
- Short stature
- Delayed developmental milestones: motor, language, social
- Inappropriate temperature controls
- Wide swings in blood pressure
- Gastro-esophageal reflux
- Frequent lung infections or pneumonias
- Episodic vomiting
- Decreased or no reaction to pain and temperature
- Excessive sweating
- Blotchy reddening of skin with excitement and/or feeding
- Smooth tongue / lack of taste buds
- Spinal curvature
- Poor weight gain and growth
- Impaired renal function
- Osteoporosis and osteopenia
- Fainting and cardiac arrhythmias
- Sleep apnea
- Restrictive lung disease
- Intelligence is usually normal; however, learning disabilities are common

Average lifespan: Currently, the mean age of the FD population is approximately 15 years. By statistical projection, babies born with FD in 2007 have a 50% chance of surviving to 40 years of age.

What causes the disease: Inefficient gene splicing results in decreased production of a vital protein called IKAP.

Treatment: There is no cure for FD. Treatments are supportive and preventative. Supportive therapies include medications to maintain and regulate cardiovascular, respiratory, and gastrointestinal function. Surgical interventions include fundoplication, gastrostomy, spinal fusion, and tear duct cauterization.

Carrier testing: In January 2001, the two most common mutations that cause the disease were identified. A carrier test is now available. The reliability of the blood test is greater than 99%. One mutation is responsible for over 99% of all cases of FD. A second mutation accounts for all other cases except one. A third mutation is responsible for a single case of FD. Carrier testing is available for the two most common mutations. Carrier screening is based on DNA analysis.

Current research: Research focuses on modifying the splicing defect in order to increase production of IKAP as well as to further understand the role of this vital protein. Researchers are also constructing an FD mouse model to further this aim. Clinical investigations include assessment of sleep physiology and assessment of molecular and functional biomarkers.