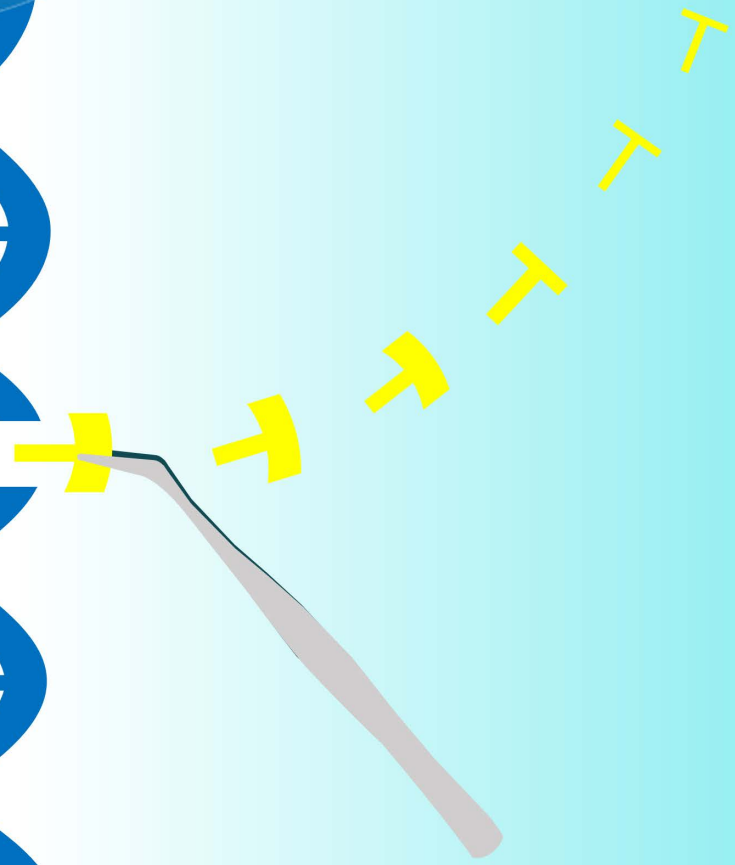
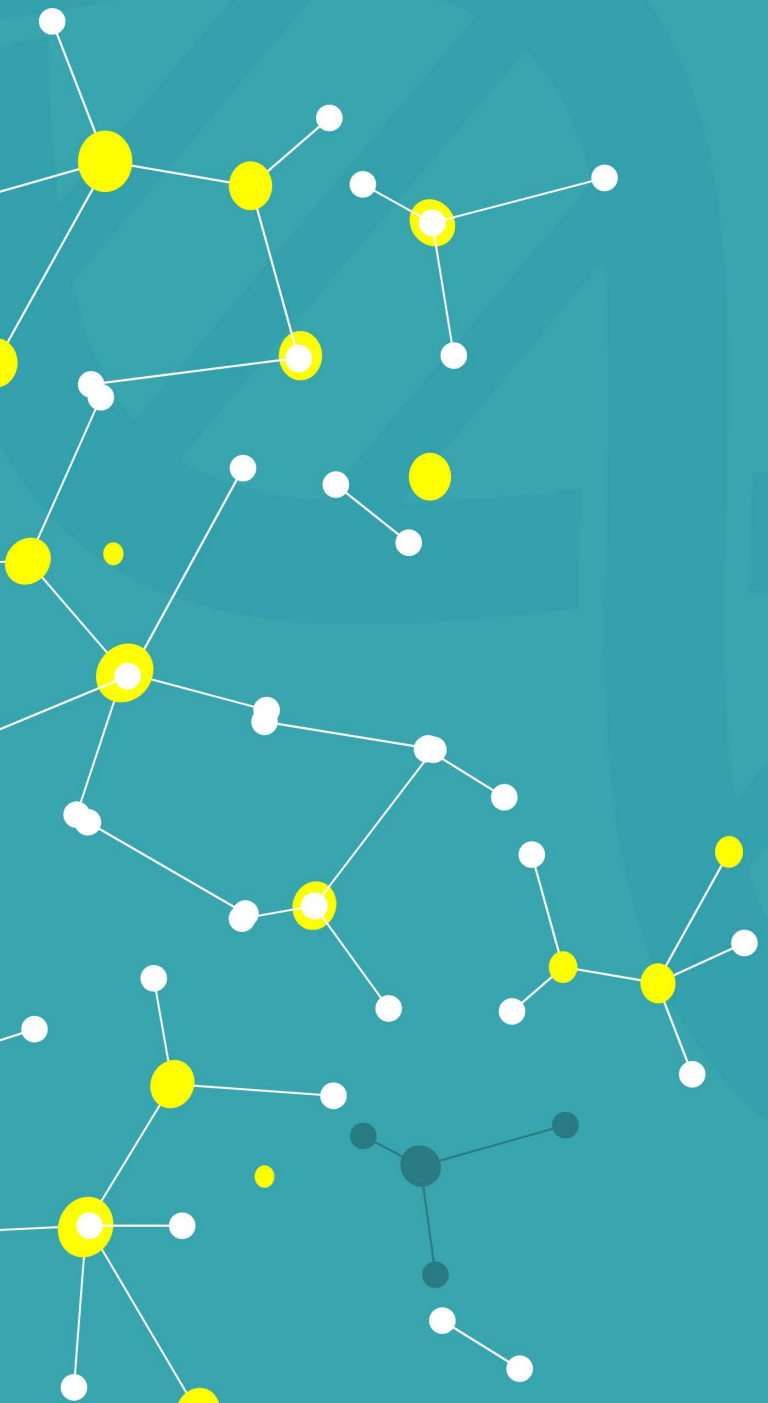
FAMILIAL DYSAUTONOMIA FOUNDATION, INC

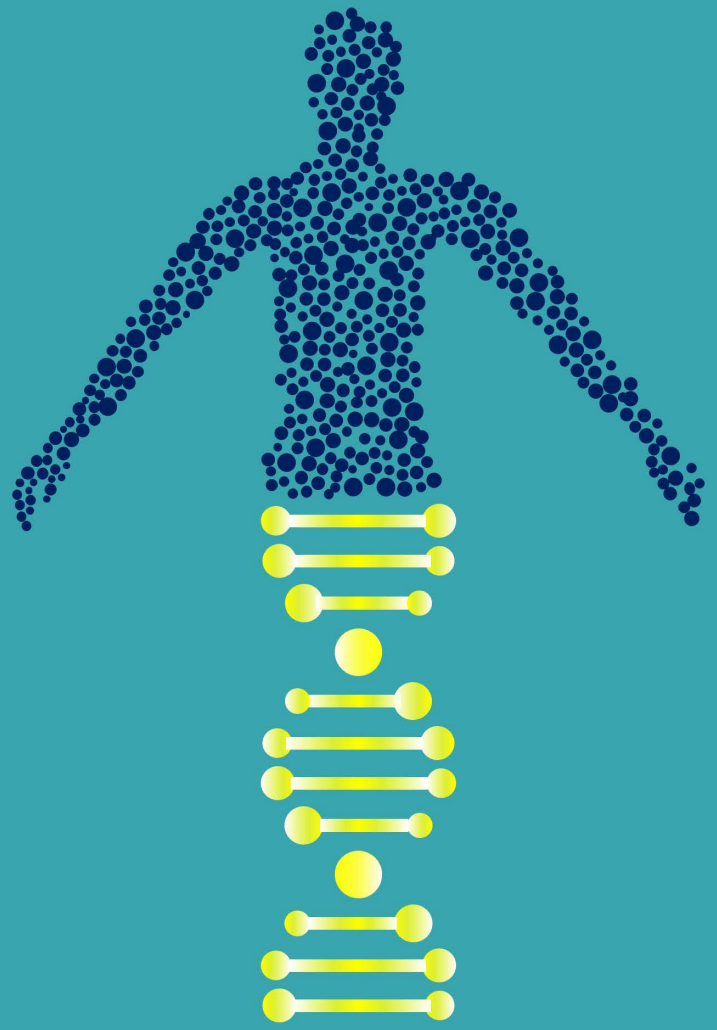


About Familial Dysautonomia



- Genetic disorder affecting the autonomic and sensory nervous systems.
- Found almost exclusively in people with Ashkenazi Jewish heritage (about 1 in 26 are carriers).
- Rare disease – fewer than 350 people currently living with the disorder worldwide (a rare disease is defined as fewer than 20,000 people affected worldwide).

Every day is a challenge for people who have FD and those who love and care for them



Symptoms begin in infancy, including inability to suck. Many need a feeding tube.

Affects all involuntary bodily functions such as:

- **breathing**
- **swallowing**
- **blood pressure regulation**
- **temperature control**
- **muscle control**
- **producing tears**
- **balance**

Symptoms increase in number and severity with age, which can lead to:

- **kidney & heart problems**
- **lung infections**
- **severely impaired vision**
- **Scoliosis**
- **poor growth & weak muscle tone**
- **learning difficulties**

What is **Autonomic Crisis?**

Most people with FD tell us that Crisis is the worst part of having FD

During a crisis, they might experience symptoms like excessive sweating, reddish blotting of the skin, rapid changes in blood pressure and heart rate, and vomiting episodes.

Attacks are unpredictable and sometimes without obvious cause, although some common triggers include emotions, infection/illness, or surgery.

Autonomic crises can last for hours, or for several days.

Many of the treatments for crisis have negative side effects, but doctors at the Dysautonomia Center are finding encouraging outcomes with carpidopa, an inhibitor which blocks dopamine synthesis.

FAMILIAL DYSAUTONOMIA FOUNDATION, INC

Our Mission

To raise funds and operate programs to pursue the best possible medical treatment, scientific research, public education and social services for the benefit of those affected with or at risk for familial dysautonomia.

NORD 2020 MEMBER PLATINUM

SEAL OF TRANSPARENCY 2019 PLATINUM GUIDESTAR

★★★★ CHARITY NAVIGATOR Four Star Charity

A SMALL FOUNDATION

We serve and advocate for fewer than 350 people currently living with this disorder and their families.

WITH A LARGE IMPACT

Research funded by the FD Foundation identified the FD gene in 2001.
Genetic testing, now available to all, means **thousands of healthy babies born** every year to carrier families.

A **Game Changer**

In 2001, scientists funded by the Foundation, in partnership with National Institute of Health, discovered the FD gene mutation, IKBKAP/ELP1.

- This led to the development of genetic testing for FD.
- Testing resulted in countless healthy babies born to at-risk carrier couples.
- Understanding the mutation is vital to emerging gene-targeting therapies.

What programs do contributions to the Foundation support?

Telemedicine

The Center's newly developed telemedicine program ensures continued access to medical care and expands the reach of the Natural History Study.

FD Natural History Study

Managed by the Dysautonomia Center, the NHS tracks the progression of the disease in people over time. It is the key to all current and future scientific and clinical research in FD.

Dysautonomia Center at NYU Langone

The world's pre-eminent center for care, treatment and research on FD, the Foundation funds an interdisciplinary team including physicians, nurse practitioners and scientists

Education, Outreach & Community Building

The Foundation hosts an annual international conference for FD patients, families, health care providers, scientists and other stakeholders. Education and outreach initiatives include a website, newsletter and active social media presence.

Portable Oxygen Concentrator Loan Program

To prevent hypoxemia which can result in brain injury and organ damage, the Foundation lends, free of charge, POC machines for patients to use during air travel.

Mental Health & Well-Being

Virtual mental health counseling and virtual "hang-out" social gatherings are among programs funded by the Foundation that promote mental health and well-being for people with FD.

Timeline

	1950's - 60's	1970's	1980's	1990's	2000's	2010's	2020's VISION
	FD Foundation is established in 1951 by parents of children with FD who had nowhere to turn for information and support	Foundation joins forces with NYU Medical Center to create a dedicated treatment center for FD	Foundation strengthens its partnership with Israel's FD community by supporting establishment of a second Israeli FD Center, at Hadassah Hospital at Mt. Scopus	Foundation prioritizes identifying the gene that causes FD and pledges to invest millions	With identification of the FD gene and the development of carrier testing, FD is added to the Jewish Genetic Screening Panel, resulting in dramatically fewer FD births worldwide	Advances in medical care leads to longer and better-quality lives for people with FD	Foundation envisions major investments in research for treatments to slow or stop degenerative effects of FD
Treatment Center		In 1970 the FD Foundation establishes the Dysautonomia Treatment Center at NYU Medical Center, under the direction of Felicia Axelrod, MD	The Center institutes supportive treatments and distributes Patient Care Manual for parents	Foundation names its first permanently endowed chair at NYU, the Carl Seaman Family Professorship	Clinical studies explore therapeutic potential of treatments aimed specifically at modifying the FD splicing mutation	World-renowned neurologist Horacio Kaufmann, MD becomes Director of the Center upon the retirement of Dr. Axelrod	Center envisions expanded access for patients through telemedicine; and continued building of robust Natural History Study as basis for all clinical studies
Research		FD patient registry is established to collect and store key medical and demographic data	In clinical studies, supportive treatments are shown to significantly increase patient survival	A decade of genetic research collaboration between NYU and Mass General results in identifying the FD gene located on Chromosome 9Q	SUCCESS IN 2001! Identification of the specific gene mutation inspires search for therapeutic drugs to correct splicing error	Scientists create FD animal models to facilitate research aimed at enhancing and extending lives for people with FD	SAB envisions collaborating with an international team of researchers to advance promising scientific breakthroughs in FD treatment

NYU Langone Dysautonomia Center

Established at NYU Langone in 1970 through a unique partnership with the FD Foundation, the Dysautonomia Center is dedicated to improving the quality of life for people with autonomic disorders.

In addition to providing patients with cutting-edge medical care, the Center also comprises a research program and laboratory, equipped with state-of-the-art diagnostic equipment.

The Center, housed in Suite 9Q (named for the location of the gene mutation), is also home to the FD Natural History Study.

The Dysautonomia Center team has made a number of discoveries that have shaped our understanding of autonomic disorders, allowing us to bring new treatment options to people living with this condition.

The only treatment center in the country dedicated to providing medical care and clinical research for FD

The Center is staffed by a multidisciplinary team of highly trained physicians, nurse practitioners and researchers who manage daily patient care, perform annual physicals, handle patient emergencies 24/7 and oversee clinical studies.

NYU Langone Dysautonomia Center

Led by Horacio Kaufmann, MD, a world-renowned expert in autonomic disorders.

The Dysautonomia Center has transformed the lives of those living with FD from a once fatal childhood disease into a chronic condition.

Thanks to advances in care provided by the Center, people born with FD who had a 50% chance of living to age 5, now have an 80% chance of surviving into adulthood.

NYU Langone Dysautonomia Center

TELEMEDICINE AT THE DYSAUTONOMIA CENTER

With the advent of the coronavirus pandemic, the Center added a telemedicine initiative.

Virtual Check-up Kit contains components to gather data to monitor and track patients' health status.

Benefits of telemedicine:

- Continuity of care: patients do not have to miss their annual check-ups.
- Expanded care: patients who were never physically (or economically) able to travel to New York for medical care can now be seen by the Center's experts.
- Improved access to care: with telemedicine, FD medical professionals are just a videocall away in any emergency.
- Better data: Increased number of check-ups means more data collected for the Natural History Study and thus a more robust database for research.

NYU Langone
Dysautonomia Center

Natural History Study

Tracks the progression of the disease in the patient population

Database is essential for clinical care and research, especially for an ultra-rare disorder.

Enables us to track the incidence of problems, the outcome of treatment and explore how to better manage FD.

Natural History data is vital to planning for future clinical trials

NHS will help us understand whether new therapies can prolong survival and improve symptoms.

NYU Langone Dysautonomia Center

Examples of Research Initiatives underway at the Dysautonomia Center

Gut Flora

Purpose: To better understand micro-organisms living in gut of FD patients.

- Maintaining healthy weight is a persistent problem for many FD patients.
- Compare diets between tube and oral fed patients.
- Will hopefully show whether fungal growth in GI tract is associated with symptoms in absence of known pathogens.

Brainstem Reflexes

Purpose: To understand if dysphagia and dysarthria in FD are due to a reduction in number and/or excitability of afferent trigeminal nerve fibers.

- Achieved through using electrophysiological techniques.

Muscle Atrophy

Purpose: Examine muscle function and hereditary sensory neuropathies in FD patients.

- Incidence of rhabdomyolysis (muscle destruction) is higher in people with FD.
- Small pieces of muscle are obtained during programmed surgery (scoliosis, hip replacement, etc...) and studied.

Scientific Research

There is **no cure** for FD.

Scientists today focus on
alleviating the most debilitating symptoms and
slowing the degenerative effects of the disorder.

Researchers are approaching FD from all angles including:

- **fixing gene splicing**
- **correcting the gene mutation and**
- **supporting the life-cycle of the neurons**

We are encouraged that several potential treatments are
currently in the pipeline and could be ready for the clinic as soon as late 2021/early 2022.

Our "Superstar" Researchers

HORACIO KAUFMANN, MD, FAAN
Felicia B. Axelrod Professor of Dysautonomia Research, Department of Neurology at NYU Grossman School of Medicine
Director, Dysautonomia Center at NYU Langone Medical Center
Professor, Department of Medicine at NYU Grossman School of Medicine
2020 recipient of the Irwin Schatz Award for Autonomic Neurology by American Academy of Neurology
Learn more: https://nyulangone.org/doctors/1134190291/horacio-kaufmann

FRANCES LEFCORT, PHD
Distinguished Professor of Cell Biology and Neuroscience at Montana State University
Co-Chair, FD Foundation Scientific Advisory Board
Invited to speak about the impact of FD Research at the Congressional Neuroscience Caucus Briefing in Washington DC in March 2021
Recipient of 5-year, $2.9 million NIH grant to study the roles of the gut microbiome, metabolism and the nervous system in people with FD
Learn more: https://www.montana.edu/mbi/directory/1524309/frances-lefcort

ADRIAN R. KRAINER, PHD
Professor, Cold Spring Harbor Laboratory
Awarded 2021 Wolf Prize in Medicine by the President of Israel
2020 Senior Scientist Winner of the Innovators in Science Award for his work on Spinraza
2019 Laureate of the Breakthrough Prize in Life Sciences for the development of an effective antisense oligonucleotide therapy for children with the neurodegenerative disease spinal musical atrophy (SMA)
Learn more: https://www.cshl.edu/research/faculty-staff/adrian-r-krainer/

FD Research:

Cross Pollination → Global Impact

Scientists do not yet know the genetic basis for many of the more common neurological diseases like:

- **Alzheimer's disease**
- **Parkinson's disease**
- **ALS (Lou Gehrig's disease)**
- **Multiple Sclerosis**

This makes it very difficult to study these equally devastating diseases. In contrast, we **DO** know the genetic basis for FD.

Neurons die in FD in a very **similar** manner as they do in **Alzheimer's, Parkinson's** and **ALS**
By studying **FD**, scientists are learning about the cellular and molecular pathways that cause the death of neurons in those more prevalent diseases.

Potential cures for FD (including several currently in the pipeline) may be effective in these other diseases.

Role of **Scientific Advisory Board**

The scientists, medical doctors and representatives from the pharmaceutical industry who participate on the scientific advisory board (SAB) guide and advise the Foundation on research initiatives, ensuring that research remains focused on things that matter to those living with FD while at the same time maintaining the highest level of scientific rigor.

Scientific Advisory Board

Co-Chair
Frances Lefcort, PhD
Professor of Neuroscience
Montana State University

Co-Chair
Adrian Gilbert, PhD
VP, Pharmaceutical Development
Neuroderm

Felicia Axelrod, MD
Former Director
NYU Dysautonomia Treatment Center

Bat el Bar Aluma, MD, PhD
Physician
Sheba Medical Center

Alex Gileles-Hillel, MD
Pediatric Pulmonologist
Hadassah Univ. Medical Center

Horacio Kaufmann, MD
Director
NYU Dysautonomia Treatment Center

Adrian Krainer, PhD
St. Giles Foundation Professor
Cold Spring Harbor Laboratory

Ralph Laufer, PhD
Chief Scientific Officer
Lysogene

Adam Sachs, MSE
Senior Pharmaceutical &
Biotech Operations
AstraZeneca

Eric Schon, PhD
Prof. Neurology Genetics & Development
Columbia University

Susan Slaugenhaupt, PhD
Scientific Director
Massachusetts General Hospital

Gail E. Sonenshein, PhD
Prof. Developmental, Molecular & Chemical Biology
Tufts School of Medicine

The FD Foundation has funded research at many prestigious institutions over the past six decades including:

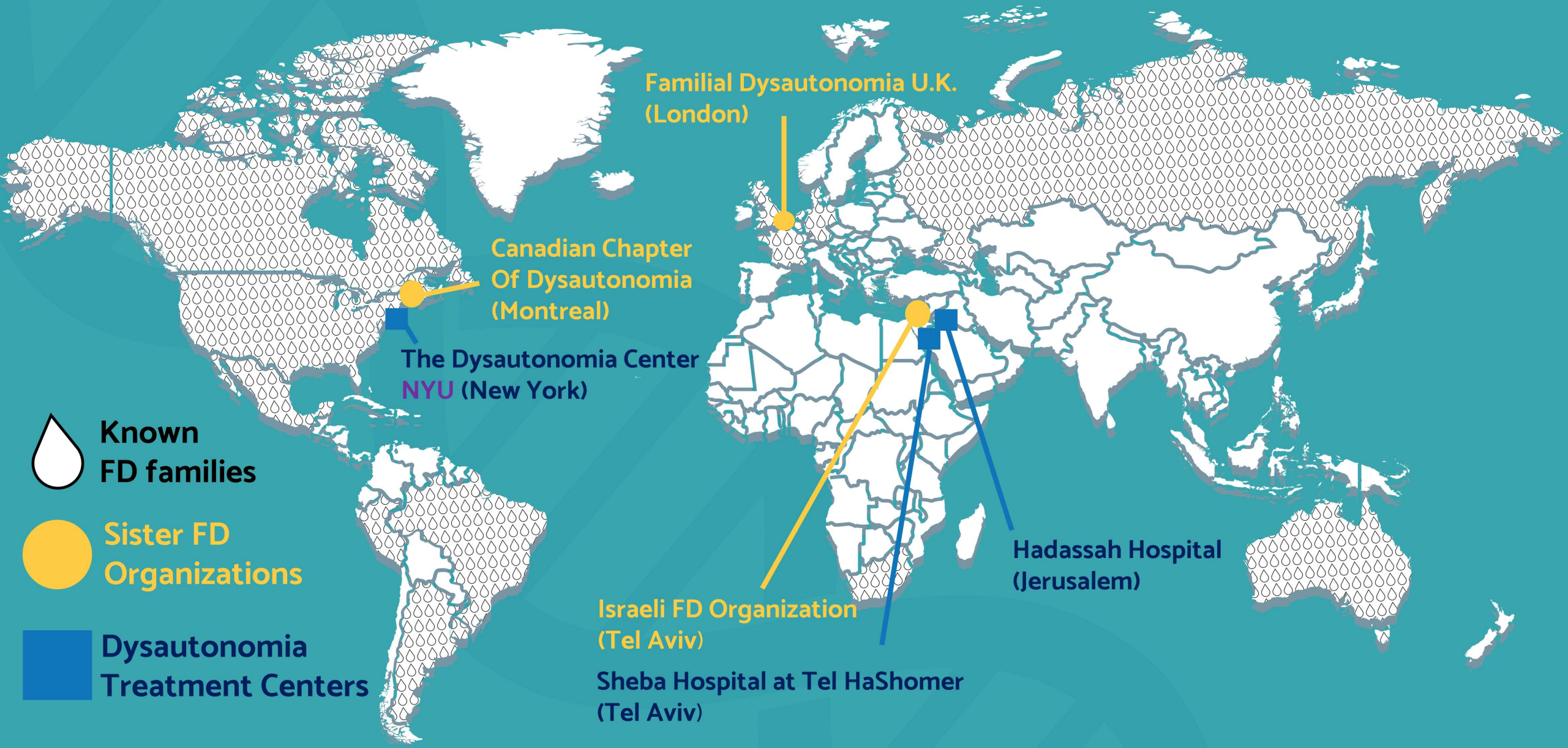
U.S.

- Massachusetts General Hospital
- Harvard Medical School
- The University of Tennessee Health Science Center
- Cold Spring Harbor Laboratory
- Columbia University
- University of Virginia School of Medicine
- Montana State University
- UC San Diego School of Medicine
- Tufts University
- Vanderbilt University Medical Center
- Illinois State University
- Rush University

International

- Danish Cancer Society
- Aix*Marseille université
- Tel Aviv University
- Bar-Ilan University
- The Francis Crick Institute
- Technion Israel Institute of Technology
- Hadassah University Medical Center

International Collaborations



Our FD Partners in Israel

Israel has the second largest community of people living with FD after the US.

Both communities benefit from close collaboration between FD organizations based in the US and Israel.

Sheba Hospital at Tel HaShomer (Tel Aviv)

- Largest and most comprehensive medical center in Israel and the Middle East.

Dr. Ori Efrati
Director
Pediatric Pulmonary

Dr. Bat El Bar Aluma
Physician,
Pediatric Pulmonology
***6-month fellowship at NYU Dysautonomia Center**

Hadassah Hospital (Jerusalem)

- Established 1980.
- Modeled after NYC Dysautonomia Center.

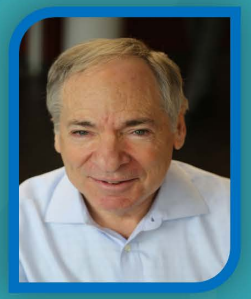
Dr. Alex Gileles-Hillel
Director,
Pulmonology Clinic & Dysautonomia Center.

Leadership

President

Faye Ginsburg

Secretary



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Gregg Meyers

Gerald Adler

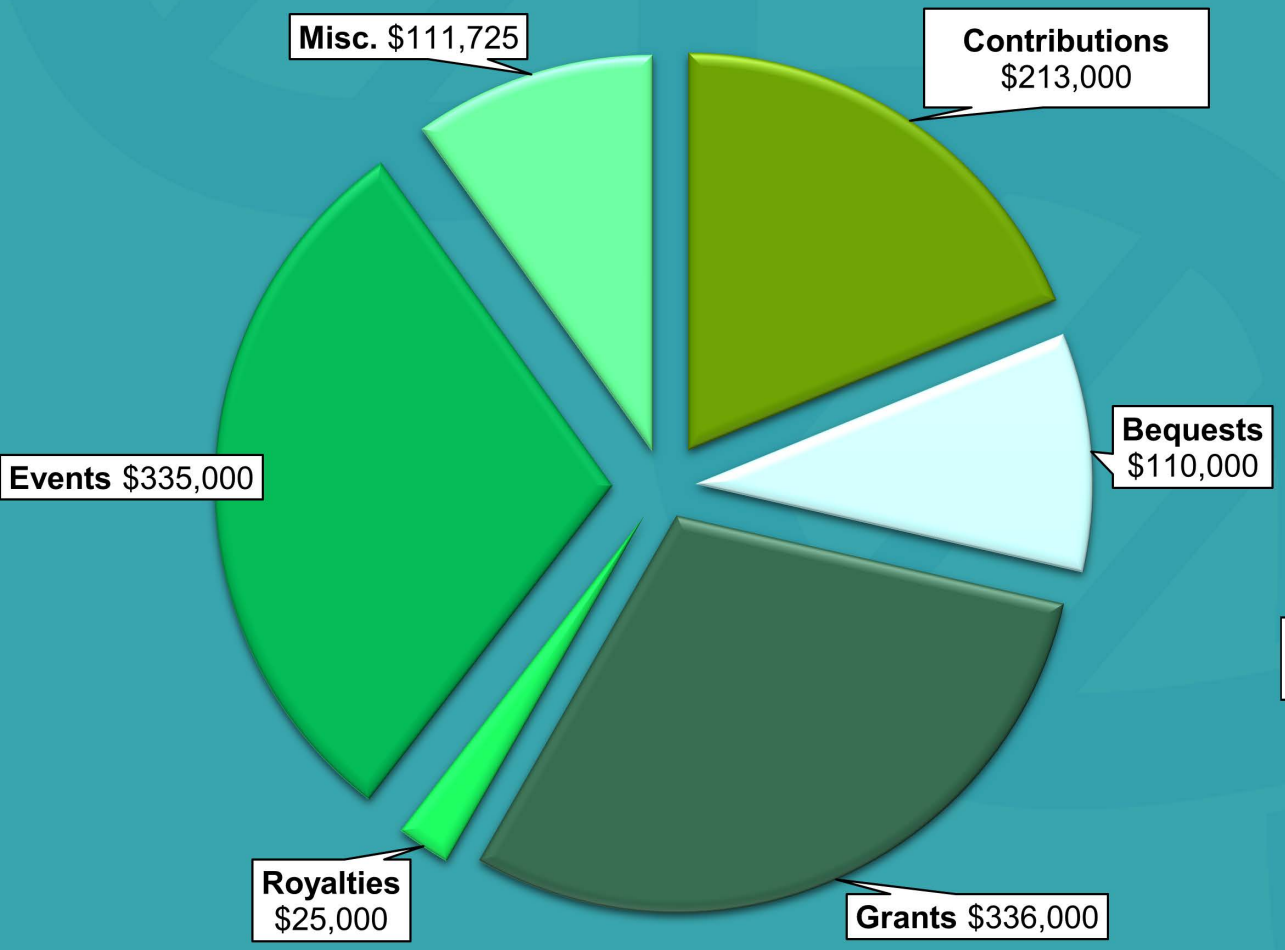
Howard Weiser

Brian Stillman

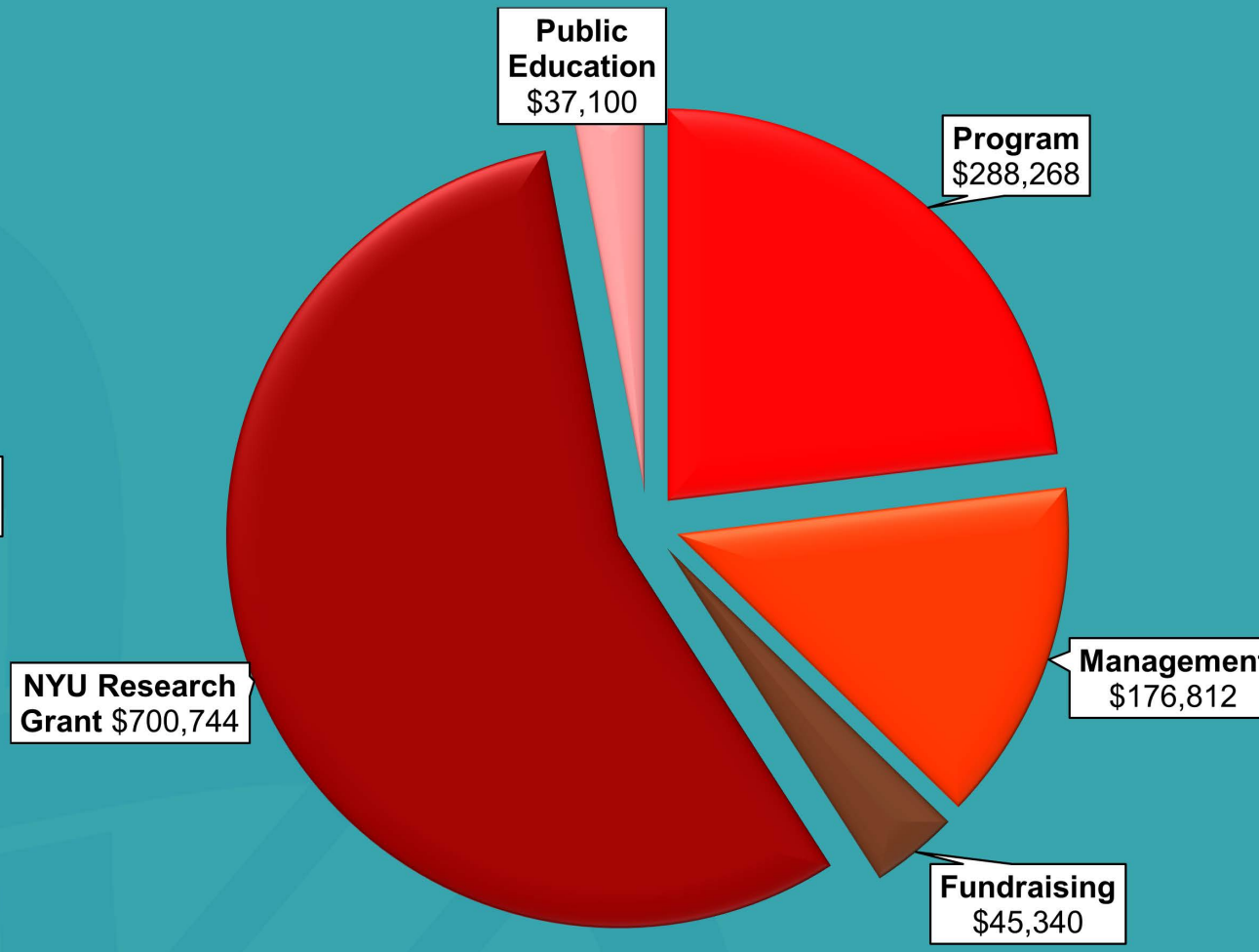
Daniel Landau

Annual Budget

Income: $1,130,725



Expenses: $1,248,264



- 82% of budget goes directly to research & programming.
- Figures reflect 2020 budget.

Where Can I Learn More?

FAMILIAL DYSAUTONOMIA FOUNDATION, INC

Visit our website www.famdys.org

Contact the Foundation at 212-279-1066 or email Lanie Etkind, Executive Director, at letkind@famdys.org

View latest research at https://dysautonomiacenter.com/category/research-papers/

Watch this video in which several young adults share how having FD has affected their lives