DYS/COURSE

News of familial dysautonomia (FD) - Winter 2013

In memory of Elaine Jamie Lipson, whose love of life will remain forever in our hearts.

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DESPITE GENETIC TESTING, FD CASES ARISE

Despite growing awareness and wider screening for familial dysautonomia (FD), many FD carriers are still going untested and, as a result, new cases are identified every year. Some of those new cases are not diagnosed early enough, even though a definitive diagnostic test is available. A recent case in New York reminds us that more must be done to inform doctors and the at-risk community about FD.

More than a decade since the advent of FD carrier screening, the rate of new cases has decreased significantly, but some new cases, including Caitlin Chance of Long Island, New York, still arise.

After years of battling what seemed to be a mystery illness, five-year-old Caitlin was diagnosed with FD just a few months ago. Her mother, Deborah Chance, is both relieved and frightened.

Caitlin, 5, was diagnosed with FD earlier this year.

"I have this happy little girl who I love more than anything in the world, and I am terrified," Deborah said. "I am scared to death to know how FD is going to affect her. I don't want her to have a hangnail, let alone a disorder."

The diagnosis was only recently made after years spent in and out of the hospital dealing with mysterious symptoms. Her feeding trouble, balance issues, numerous aspiration pneumonias, extreme fluctuations in temperature, and a troubling rash that would come and go without warning were all textbook signs of FD, but none of her doctors could diagnose her underlying problem.

Even though Caitlin had been through a basic genetic test that had come back negative for any indicators, the answer only came when FD was identified after a complete genetic sequencing.

When the news finally arrived, the Chance family was scared, but at least they knew what they were dealing with. "From a selfish standpoint, I wish I didn't know. From an intelligence standpoint, everything makes sense," Deborah said.

Caitlin is the Chances' only child. Even though they reported their Jewish ancestry to their doctors, the couple did not receive screening and counseling until after Deborah was pregnant. "We never thought to have extensive testing done because we were under 35 with no problematic history. I am surprised that FD was never mentioned to us, now knowing it is one of the top things doctors are supposed to test for."

Deborah is grateful that the way forward has been paved by other FD families. "I have spoken with parents and they have been super supportive. It's also amazing how positive and strong the children are."

For other families who might have sick children, Deborah encourages them to take matters into their own hands and to go with their gut. "Doctors are not always right. They don't always know what's going on." And to doctors, "Don't give up on a child. Keep searching until you find out what is wrong with them."

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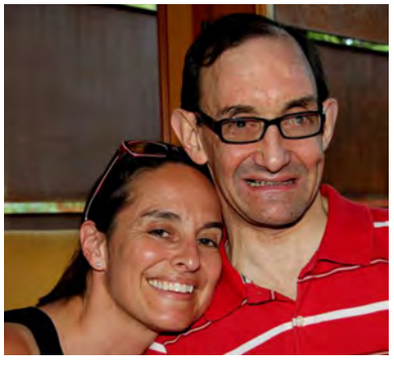
Family Ties Inspire Research

It is not every day that someone dedicates their career to neuroscience in the hope of fixing the medical problems of a family member, but this is exactly what Foundation-funded researcher Frances Lefcort did. Growing up close to two cousins who had familial dysautonomia, Lefcort was fascinated with the nervous system from an early age. Eventually, her young curiosity turned into a lifelong passion. Now she is the Head of Montana State University's Department of Cell Biology and Neuroscience.



Prof. Frances Lefcort with her cousin, Michael Kronick.

"It was inevitable that I would go into studying the nervous system. Though I considered going into medicine, I found I was much more interested in the mechanisms that caused diseases," she said.

(Continued on page 3)

FOUNDATION LEADERSHIP

PRESIDENT'S MESSAGE

Dear Friends:

As we head into the holiday season, we want to express our gratitude for the generous efforts so many of you have provided in support of the Dysautonomia Foundation's many activities.

The most fundamental of these, of course, is the Foundation's underwriting of the superb quality of care for every person with Familial Dysautonomia at the treatment centers in New York and Israel.

In addition to the clinical expertise offered by the doctors at each of these sites, we now work with many scientists and medical researchers who are helping us develop new approaches to FD treatment. These range from simple, effective techniques to improve balance through the use of strategic taping on the legs; to the cutting edge approaches to understand the impact of FD on the optic nerve, generating ideas for interventions that will help preserve vision and quality of life for FD patients as they grow older.

We deeply appreciate your support of our work, and hope you will be thinking of us as you make out your end of the year donations.

Faye Ginsburg, President of the Board of Directors

EXECUTIVE DIRECTOR'S MESSAGE

David Brenner

We never shrink from the great challenges we face at the Dysautonomia Foundation. Our main focus is on medical care and research, but we know that targeting those challenges only addresses one part of the problem of reducing the suffering caused by FD. We also focus on awareness. The at-risk population must be made aware that genetic screening can diminish, if not eliminate, new cases of FD, and doctors must be made aware of FD so they can diagnose the few new cases that do arise as early as possible, allowing for early intervention and the best possible outcomes.

Sadly, a dozen years after the discovery of the FD gene, neither universal testing nor early diagnoses have become the norm.

Our cover story highlights this problem. I'm shocked that a couple with Jewish ancestry in New York could receive basic genetic counseling and never be offered a test for FD. Even worse, I'm alarmed that a child could suffer for five years without being properly diagnosed with FD by pediatricians, geneticists and specialists in the New York metropolitan area. Of course, if it can happen here, it can happen anywhere, and we need to address this problem as aggressively as possible.

With your help, we will begin a new initiative to expand availability and awareness of genetic testing and early detection. Watch for updates about our efforts in these areas in future newsletters, and please support our efforts to face and overcome these challenges as we have, with your help, overcome so many other challenges for the benefit of people with FD.

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20 Years of Healthy Babies

Upper Left: The Baranoff children; Lower Left: The Landaus; Center: The Cohens; Upper Right: The Steiners; Lower Right: The Wexlers

Before the FD gene was identified in 2001, the only way couples could find out if they were both FD carriers was by having a child with FD. For many of these "dual carrier" families, the lack of any available genetic testing led them to decide to avoid having any more children. However, as Foundation-funded research began to close in on finding the gene in the 1990s, linkage analysis became available and enabled FD families to have unaffected children. The process was complicated and expensive, but nonetheless offered great comfort to young families. After the discovery of the gene, the use of a technique called pre-implantation genetic diagnosis provided an additional option for families who wanted to have more children. Thanks to these methods, well over a hundred unaffected siblings and countless others have been born to parents who both carry the FD gene. At the Dysautonomia Foundation, we're proud that the discovery of the FD gene has not only led to a decline in new FD cases, but has also enabled so many families to have so many healthy children.

Family Ties *(Continued from page 1)*

By developing and studying an FD mouse model, Lefcort has recently advanced our knowledge of how FD interferes with cell development and health. She has indicated that people with FD start out life with fewer neurons, or nerve cells, and that apoptosis, or programmed cell death, occurs throughout their lives. "There are two steps to focus on to halt this cell death, and that's what we are working on now," Lefcort said. "The key is to find out why these neurons are dying."

Frances Lefcort, PhD

The findings were published in the journal of Proceedings of the National Academy of Sciences of the United States of America (PNAS). What's more, for the first time that we know of, the well-respected journal broke with tradition and acknowledged the research was in memory of an author's family members. In the acknowledgment section of the article, Lefcort mentions her cousins, Michael Kronick and Barbara Kronick, right alongside mention of the Dysautonomia Foundation and the National Institutes of Health.

"With my cousins and other FD patients I have met, I have noticed that they persevere," Lefcort said. "Those are people you want to help. They have such amazing attitudes in spite of their daily struggles."

Science Conference 2014

The Dysautonomia Foundation is hosting a Research Conference on March 27 and 28, 2014 in New York City. The Conference will bring together Foundation-funded scientists and many other researchers with an interest in FD for presentations, interactions and insights into new areas of research. Past conferences have brought together experts from around the globe, many of whom have formed collaborative teams to help advance FD research and to develop new FD therapies. We are confident that the 2014 Conference will yield equally positive outcomes.

Expanded Diagnostic and Research Capabilities with New Equipment

Thanks to government support, philanthropic organizations and generous private donations, the Dysautonomia Center has now been outfitted with state-of-the-art ophthalmological equipment. When a patient visits the Center for an annual check-up, our resident neuro-ophthalmologist, Dr. Carlos Mendoza-Santiesteban, performs a comprehensive eye exam. The equipment allows Dr. Mendoza-Santiesteban to diagnose progressive vision problems and acquire greater insight into the mechanisms of retinal and optic nerve problems as well as the corneal problems that affect every FD patient. We are confident this new equipment will lead to better vision for FD patients as they grow older.

FD Party

A group of FD teens and adults gather for a party.

The Dysautonomia Foundation recently launched a new program to bring together adults with FD for impromptu fun. Nearly 30 people from the FD community, some from as far away as Florida, gathered at the New York University campus for casino games, music and lunch on Sunday, November 24. This was the first of a series of social gatherings we have planned for FD adults and teens. If you are an FD family and would like more information, please contact the Foundation at (212) 279-1066.

Tour de Foliage, FD Cycle Tour

Organizers (L to R): Melissa Slive, Lisa Rudley, Lisa and Jeff Newman

The 10th Annual Tour de Foliage, FD Cycle Tour in September 2013 was an exciting day for everyone who participated in the event. Riders and sponsors raised $200,000 for treatment and research this year, with $1 million dollars raised over the course of the cycle tour's history. Our gratitude goes to the tireless organizers, Jeff and Lisa Newman, Lisa Rudley and Melissa Slive. Their leadership and dedication throughout the years has made this one of our most successful annual events.

Remembering Harold Newman

Harold Newman

The Dysautonomia Foundation is sad to report the passing of one of our founders, Harold Newman. Harold was the Dysautonomia Association's first vice president in 1954 and our second president in 1957. He was instrumental in starting our first Journal campaign in 1955, which raised $2,000, an impressive figure at the time. He was the father of the late Robert Newman who had FD. Mr. Newman's passing reminds us of all the indomitable parents who formed the organization that would not only grow to become the world's largest source of funding for FD research and treatment, but would also lead the way to longer, healthier lives for all people with FD.

FD Parents' Perspective

Behind our FD heroes stand extraordinary parents who nurture, support and enable their remarkable children. Our new video series takes a look at seven FD parents, providing a rare glimpse into their struggles, joys and experiences as they, too, face life with FD. These brave parents give us an honest look into what it is like to have a child with a mysterious and life-threatening illness. To view this series of videos go to famdys.org/video or youtube.com/famdys.

Join Our Journal Campaign

Fifty-eight years and counting, our FD Journal is a tradition first started by the Dysautonomia Foundation as a humble 28-page booklet in 1955. The publication has grown to contain hundreds of well wishes, photos and tributes every year. Since its inception, millions of dollars have been raised, and thousands of spirits have been uplifted. Time is running out to be part of the treasured tradition this year. Send us your page today by going online at famdys.org/journal or by calling the office at (212) 279-1066.

52 Weeks of Comics

At the beginning of 2013, the Dysautonomia Foundation embarked on a mission to deliver an insightful, and at times funny, FD comic every week for a year. We did not know exactly how the comic would be received, but we hoped our Facebook fans and website followers would be enriched, entertained and amused by our messages, and we were hopeful they would feel that someone out there understands what they are going through. The reception of *No Tears: Life with FD* has exceeded our expectations, reaching much further than the FD community, to the wider disabled and disease communities in the United States and around the world. Here is one of our recent, popular comics. For the complete set, visit: famdys.org/comic or facebook.com/famdys.