

A NEW FOCUS ON FD HEALTH

Eyes: A Window to FD

A study on the optic nerve deterioration that begins at a young age for people with FD and progresses over time has offered insight and hope for new FD treatment and therapies, reaching much further than the tiny cells studied.

The health of the eye and vision problems have been a matter of concern to FD patients and their doctors for decades. Initially, therapy centered on problems with the cornea, but recently, researchers have begun to study the retina and optic nerve. In 2012, the Dysautonomia Foundation began an expanded program of funding for optic neuropathy with the hope that chronic vision problems could be mitigated for people with FD.



Carlos Mendoza-Santiesteban, MD

The findings of Neuro-Ophthalmologist Carlos Mendoza-Santiesteban have provided scientists with an understanding of why FD affects vision, and has suggested a new biomarker that could be used to help measure the success of a wide range of FD research into new therapies.

Using a non-invasive technique with light beams from optical coherence tomography (OCT), Mendoza-Santiesteban discovered not only that people with FD present eye problems related to mitochondrial function similar to other hereditary diseases, but also that the cells in the center of the ganglion in the retina are affected, leaving the cells in the periphery healthy and functioning.

Looking ahead, this research could possibly lead to improved vision in people with FD by repairing damaged ganglion. This work could also provide a key indicator of the efficacy of other research that aims to improve not only the eye, but the rest of the body as well, through gene therapy.

The eye is the place where you can best quantify cells, Mendoza-Santiesteban said. The study of the eye provides an opportunity to measure the damage caused by the lack of the protein IKAP, as well as the success of trial therapies, like the administration of kinetin, to increase the presence of this protein. Such therapies hold great promise for FD treatment, but much more work remains to be done. An effective animal model of FD would help accelerate our progress. “In order to do this, we need a good mouse model with the same eye characteristics of FD; that is why we are working not only with the patients, but with other researchers who are making good progress on mouse models,” Mendoza-Santiesteban said.

Researcher Leads the Way to a Better Understanding of FD

Lucy Norcliffe-Kaufmann, PhD, first joined the Dysautonomia Center at NYU Medical Center as a post-doctoral research fellow in 2006. She is now leading the research on several studies focused on improved therapies for people with familial dysautonomia (FD). Her devotion to FD extends beyond the lab and includes initiating FD community activities such as the annual photo exhibiton at FD Day.

Her research has provided the first evidence-based explanation for the dramatic blood pressure volatility often found in people with FD. Part of her success is rooted in constant curiosity as a result of never taking an assumption for granted.

Before taking on FD research, she earned her doctorate in cardiovascular physiology from the University of Leeds in England. The Foundation sat down with Norcliffe-Kaufmann to talk about her work on FD and her involvement with the FD community.



Lucy Norcliffe-Kaufmann, Ph.D.

Q. What part of your FD work makes you the most proud?

A. In November 2010, we finally figured out what causes the blood pressure to be so erratic in FD and how this seems to destroy the kidneys. Based on this, we switched our focus to also treating the high blood pressure, often considered a silent killer.

(Continued on page 3)

INSIDE THIS ISSUE

About the Foundation	2
President’s Vision.....	2
New Research Publications	3
Fundraisers and Community News	4

PRESIDENT'S MESSAGE

Dear Friends,

Despite the fate that many of us share, we are also fortunate to be part of an extraordinary community of people who do whatever we can to enhance the lives of those we love who are struggling with familial dysautonomia (FD). To expand our ranks, we reach out to our relatives and friends, and especially those touched by our children's lives in some way, and ask them to join us as allies in our work.

We are so grateful to you for being part of the FD community, especially at this critical juncture when we have so many new possibilities in development, some of which you will read about in this issue of *DYS/course*. Please accept our profound thanks for joining our cause.

Faye Ginsburg, President of the Board of Directors



EXECUTIVE DIRECTOR'S MESSAGE

David Brenner

We have made tremendous progress, and we are poised for even greater accomplishments as we pursue our mission to provide the best possible medical treatment, scientific research, public education and social services for people afflicted with, or at risk for, FD. This progress is only possible through the incredible support of our donors. You have enabled us to make a profound difference in the lives of those we serve.

Yet we are confronted with great challenges as we try to improve the quality of life and extend life expectancy for people with FD. We are thankful that we no longer consider FD a fatal childhood disease, but we are troubled by the evidence that as they reach adulthood, their cardiovascular, respiratory, renal and ophthalmological health is severely compromised. We are working to further improve the health of FD children so they can grow to become healthier FD adults, and we are redoubling our efforts to help adults live healthier, longer lives.

While we are aware that it is prudent to measure our success by years and decades, we know that for people with FD, every day is precious and every moment is a gift that we should not take for granted. With your help, we will turn those days and moments into lifetimes of health and happiness.

The Dysautonomia Foundation

The Dysautonomia Foundation, Inc., a non-profit public charity established in 1951, is the proud leader of treatment and research for familial dysautonomia (FD). We have established the world's only treatment centers for FD, located in New York and Israel, we are the worldwide leader in FD research funding, and we have created the world's only FD clinical research lab. We have endowed two professorships for the study of FD at NYU School of Medicine.

Through drug trials, genetic research and investigations into blood pressure, gait, eyesight and more, our researchers are constantly working to expand our understanding and to discover improved therapies. Our funding of clinical, translational and basic science research has led to dramatic improvements in quality of life and survival for people with FD.

Our discovery of the FD gene in 2001 has enabled prenatal and carrier screening for FD. The identification of the FD gene has also opened the door to a better understanding of FD and the development of genetic therapies to alleviate the symptoms, slow the progression and ultimately provide the basis for a cure.

Patients who once had an average life expectancy of 5 years now live longer and healthier lives, yet they still only have a 50 percent chance of reaching age 40. Our work is unfinished, but with your help we will continue to pursue our ultimate goals: a cure for FD and prevention of new FD births.

Dysautonomia Foundation

Board of Directors

President
Faye Ginsburg

Vice Presidents
Eddie Baranoff
Jeffrey Goldberger
Laurent Landau
Steven Kietz
Lisa Newman
Paul Wexler

Secretary
Steven Fass

Treasurer
Allan Cohen

Directors
Josh Goldberg
Adam Posnack
Rabbi Larry Sernovitz
Cindy Singer
Jennifer Sonenshein
Barbra Waldfogel
Howard Weiser

Executive Director
David Brenner

315 W. 39th St., Suite 701
New York, NY 10018
www.famdys.org

© 2012 Dysautonomia Foundation, Inc.



famdys.org



facebook.com/famdys



twitter.com/famdys



youtube.com/famdys



Foundation President Faye Ginsburg Shares Her Vision for the Future

Faye Ginsburg began her term as president of the Dysautonomia Foundation board of directors in January 2012. At the Foundation's annual FD Day Conference in June, she delivered an inspirational speech with some recollections of her first visit to FD Day twenty years ago.

"Maybe it's because I am an anthropologist, but I remember walking into the auditorium filled with FD families and feeling like I was being initiated into a new tribe, a world I never knew existed," she said. "I am stepping into the job of president of the Dysautonomia Foundation with humility and commitment, knowing how well this job has been filled by many extraordinary and dedicated people who came before me."



Faye Ginsburg with her husband, Fred Myers, and her daughter, Samantha Myers.

Founder of the Center for Media, Culture and History at New York University, she is the author of several books, a professor of anthropology, and a 1994 MacArthur Fellow. She is the mother of Samantha Myers, who was born with FD in 1989 when the life expectancy for FD was thought to be only ten years.

"In the last dozen years, we have made progress in so many directions – more than doubling the size and capacity of

the NYU Dysautonomia Center, finding the FD gene, developing carrier testing, introducing new medical options, extending research along new scientific frontiers, and enhancing the quality of life for those with FD," she said.

Ginsburg remembers first meeting Dr. Axelrod, director of the Dysautonomia Center, when the facility was nothing more than a tiny one-room office where the doctor not only saw patients, but also vacuumed and dusted. Over the years, the Center has grown, with many dramatic improvements in the health of people with FD, but with these successes, new struggles have emerged.

Knowing firsthand by watching her daughter become a young adult, Ginsburg described the FD population as increasingly challenged as they grow older and enter uncharted territory for the disease. Problems with gait, vision, kidney disease and respiration plague the FD adult population. "The fact that we have an adult population is amazing. And the range of research out there is staggering, but if you ask the kids what's important, it's the quality of life improvements that really make the difference to them," she said.

Looking ahead, Ginsburg has set her goals as president on sustaining and expanding the clinical care and research for those living with FD through the Dysautonomia Treatment center at NYU School of Medicine. "Samantha is my lodestone, inspiring me to stay creative and committed in equal measure, always a tremendous reminder as to what really counts in life, that every day is a gift," she said.

FD Research Publications

These publications from the last six months were co-authored by researchers who have been funded by the Foundation.

Familial dysautonomia (Riley-Day syndrome): When baroreceptor feedback fails. Norcliffe-Kaufmann L and Kaufmann H; *Autonomic Neuroscience*. 2012 Dec 24

IKAP expression levels modulate disease severity in a mouse model of familial dysautonomia. Dietrich P, Alli S, Shanmugasundaram R, Dragatsis I. *Human Molecular Genetics*. 2012 Dec 1

A rating scale for the functional assessment of patients with familial dysautonomia (riley day syndrome). Axelrod FB, Rolnitzky L, Gold von Simson G, Berlin D, Kaufmann H; *Journal of Pediatrics*. 2012 Dec

Cardiac-locked bursts of muscle sympathetic nerve activity are absent in familial dysautonomia. Macefield VG, Norcliffe Kaufmann L, Axelrod FB and Kaufmann H; *Journal of Physiology*. 2012 Nov 19 (Epub).

Lack of migraine in headaches of familial dysautonomia patients. Shihman B, Steiner I, Yovchev I, Maayan C. *Journal of Neurology Transmission*. 2012 Oct 9 (Epub).

Researcher Leads The Way *(Continued from Page 1)*

Q. What are you working on right now?

A. I am working on finding the best way to stabilize the cardiovascular system in FD. With the rest of the team, I'm trying to find alternative ways that we can suppress the sudden surges in autonomic activity without causing sedating or depressing the drive to breathe like we've done in the past with drugs like Clonidine and Valium.

Q. This FD Day will be the 5th year for the FD photography exhibition, why did you start this event?

A. People with FD lead fascinating lives. Only they know how it feels to grow up with next to no feedback from the body. "The world through my eyes" is a way for people with FD to tell their own stories in a single snap-shot. Blood pressure numbers never tell you how a person is feeling on the inside. The brave people who put forward their images have taught me so much.

Over the past two years, Norcliffe-Kaufmann has co-authored half a dozen papers on FD-related findings published in prestigious journals including Neurology and Brain. She is collaborating with several visiting experts, including Vaughan Macefield, PhD, whose nerve activity and gait studies were featured in our Spring 2012 DYS/course. The Foundation is fortunate to have such a dedicated and enthusiastic member of our team.

Recent FD Events

FD Benefit Dinner and Concert - November 27, 2012

With over 600 attendees and nearly \$1 million raised, this event, chaired by Laurie and Jeffrey Goldberger, was a fabulous success. The Counting Crows delivered a memorable performance, and Laurie's heartfelt speech moved the entire audience and spread the word about FD to the greater community. Visit our website for photos and a look at one of our new videos, which premiered at the event.



*Event organizer,
Laurie Goldberger*

FD Cycle Tour – September 23, 2012



The sun was shining and spirits were soaring for our 9th Annual Tour de Foliage. With more than 100 riders on the 50 and 15-mile courses, the event raised \$200,000 for FD treatment and research. A special thanks goes to Lisa Rudley, Lisa and Jeff Newman and Melissa Slive for chairing the event in Pleasantville, NY.

FD Golf Classics – July 30, 2012

Hundreds of people golfed in support of FD research and treatment for FD on July 30th in New York and Chicago. Our two golf events, the 16th annual in Long Island, NY and 9th annual in Deerfield, IL, were both great successes. Friends of the Foundation enjoyed 18-holes of golf and an evening of dinner, cocktails and auction.



With prizes including a tropical getaway to the Ritz Carlton in Jamaica, a Caribbean cruise and autographed sports memorabilia, the auctions were a fantastic finale to these great events. Our 2013

golf events will be in May (NY) and July (Chicago). Check our website for exact dates. Thanks go to Paul Wexler, Steven Fass, Adam Posnack and Paul Sunderland for their efforts on the Long Island event and to Gregg Meyers for the Chicago event.

Upcoming FD Events

Walk for FD, NYC – March 10, 2013

FD Bowlathon, Woodmere, NY – April 14, 2013

Long Island Golf Outing, Old Westbury, NY – May 13, 2013

FD Day, NYC – June 3, 2013

Chicago Golf Outing – July 2013

FD Cycle Tour – Fall 2013

FD Journal

Submit a page for the 2012 Journal, our annual publication honoring and remembering loved ones from the FD community. There's no better way to show you care or to celebrate accomplishments and milestones. Don't be left out, submit a page now! Details at www.famdys.org/journal.



FD Community News

Congratulations

Gabi Jassie graduated valedictorian of her class at The Henry Viscardi School in Albertson, Long Island June 21, 2012. She is now attending Hunter College in New York City.



Judy Fettman married Marc Dreyfuss October 14, 2012. The ceremony was attended by Drs. Axelrod and Kaufmann.

Congratulations to Judy and Marc; we hope to see many more FD weddings!

In Memorium

Fay Lager, founder of our South Florida Chapter, passed away on her 96th birthday September 20, 2012. Upon learning her granddaughter had FD, Lager started the South Florida Chapter in 1979. Since then, the chapter has contributed more than half a million dollars to the treatment and research of FD. Our hearts go out to her family and loved ones and to all those she touched with her generosity and kind spirit.

Robert Porges, MD, beloved husband of Dr. Felicia B. Axelrod, passed away on November 1, 2012. Dr. Porges delivered the first baby born to a woman with FD; he was the OBGYN for many women with FD and delivered a total of five of their babies. He published two research papers on FD babies: Neonatal Recognition of Familial Dysautonomia, and Breech Presentation among Infants with Familial Dysautonomia. He was a wonderful and charming man, as well as a generous supporter of the Dysautonomia Foundation. He will be missed.

