

FALL 2018

DYS/COURSE

NEWS OF FAMILIAL DYSAUTONOMIA (FD)

RESEARCH UPDATE:
NEW FOCUS ON
SPLICING THERAPIES

NEW EQUIPMENT
FOR THE NYU
DYSAUTONOMIA CENTER

WORKING OUT WITH FD

FD DAY 2018 BRINGS
MANY SMILES

THE TEL AVIV RAPPER

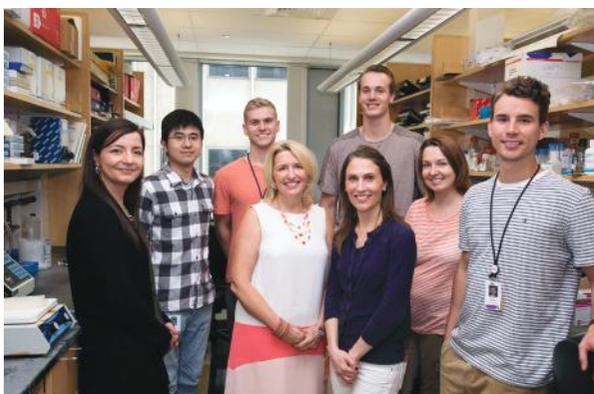


The FD Newsletter was endowed with a gift in memory of Elaine Jamie Lipson, whose love of life will remain forever in our hearts

RESEARCH UPDATE: NEW FOCUS ON SPLICING THERAPIES

Meet **Monica Salani**, a scientist working in the lab of, **Dr. Susan Slaughaupt**, a member of the Foundation's Scientific Advisory Board. Sue's lab has been researching FD for more than 20 years, and Monica has been at the **Massachusetts General Hospital Research Institute (MGH)** for six years working on the FD program. The work in Dr. Slaughaupt's lab is now focused on splicing therapies. According to Monica, "We are working together with PTC Therapeutics to find a better, newer drug than kinetin that will correct splicing and increase IKAP protein."

Recently, the Slaughaupt lab has received two grants from the prestigious National Institute of Health. "We are thrilled by the support of NIH," notes Monica. "One of the grants is being used to test new molecules in our animal model of FD to better understand how they are working and how they might benefit FD patients. Our goal is to bring the best possible drug to patients." The other grant is being used to study the eyes in the FD mouse. Explains Monica, "The loss of vision is one of the most debilitating aspects of FD. This grant is a collaboration with **Dr. Franco Pagani** and **Dr. Luk Vandenberghe**, and our goal is to optimize an RNA-based therapy and see if we can deliver it directly to the eye to increase IKAP protein in the retina. Our lab is dedicated to exploring all possible routes towards a therapy for FD."



The Slaughaupt lab group (L to R): Monica Salani, Dadi Gao, Connor Montgomery, Susan Slaughaupt, Elisabetta Morini, Shawn Herron, Yulia Grishchuk and Anthony Brenner

THE TEL AVIV RAPPER

Meet **Shir Goldman**, a 33-year-old rapper with FD, who lives in Ramat Gan, Israel. Shir, whose stage name is 'The Happy Patient,' writes all of his own songs, mostly about empowering people to fulfill their dreams. Adds Shir, "I think it's important to use your time to get closer to what you want to achieve. I want society to look at handicapped people equally and give us the same chances on the stage as well as on the street." Shir performs every



Shir Goldman rapping it up in Tel Aviv

night at various bars in the Tel Aviv area, sometimes hitting a few bars each night. Once a year, Shir, who also works at a hospital doing tech work in between creating new songs, organizes a fundraiser to coincide with his birthday week. "I've been doing this for the past five years. I invite famous artists to sing with me and this year, about 170 people attended the event and we raised a record \$5,000 for FD," notes Shir."

HOLIDAY SHOPPING EVENT



Help support the Familial Dysautonomia (FD) Foundation by doing your holiday shopping at participating **Americana Manhasset** and select **Wheatley Plaza** stores on **Thursday, November 29** to **Sunday, December 2**.

25% of your designated purchase will be donated to the Participating Organizations of your Choice!

HERE'S HOW TO PARTICIPATE

- Visit championsforcharity.org
- Register and select Dysautonomia Foundation as your preferred charity.
- Bring your registration number with you when you shop at Americana Manhasset or Wheatley Plaza from November 29-December 2.
- If you can't get there in person or on those dates, you can pre-shop using Americana Manhasset's personal shopping service at 516-627-2277 or dmerollo@americanamanhasset.com
- Shop till you Drop!

#LET'S MATCH FD ANNUAL FALL MATCHING GRANT CHALLENGE

Our second annual crowdfunding campaign was a huge success! We raised more than \$247,000 to support the critical work of the Dysautonomia Treatment Center at NYU. Thanks to the 630 donors and 25 fundraisers who participated in this effort! This is a more than thirty percent increase over the \$180,000 raised during our first crowdfunding campaign in 2017.

WORKING OUT WITH FD

Judy Fettman, a 48-year-old woman from New York with FD, loves to get a good sweat going. That's why she makes it her business to show up at the health club at North Riverdale's Skyview-on-the-Hudson as often as four times each week. "I do leg raises,



Judy Fettman working out at the gym

lat pull-downs and a brisk trot on the treadmill for a full-body workout and it makes me feel really good," says Judy. "I basically live a good life. I'm married. I have a dog that I love. I try to keep busy and do as much as I can for myself." Judy loves doing artwork such as collage and mixed media. Although she lives in the same building as her parents, she tries not to ask for help too often. "It may take me longer to do things than others but I get it all done," adds Judy. "I hope others with FD will follow my lead."

HAVE YOU CONSIDERED A PLANNED GIFT?

Planned giving offers options to make a charitable contribution that may cost you little or nothing today, but will make a meaningful difference to FD in the future. In addition to supporting a cause that is important to you, there may be tax benefits for you or your estate. Anyone can make a planned gift. Options include: bequests, trusts, gifts of life insurance and retirement funds. To learn more, please contact **Lanie Etkind** at 212-279-1066 or letkind@famdys.org.

ER RESIDENT AND MOTHER OF FD PATIENT HAVE TRUE "SIMPATICO"

It started out as a total coincidence. "I was trying to hail a doctor for my daughter," explains **Norma Trager**, the mother of FD patient **Leigh Berkowitz**. "I heard a woman in the ER looking for someone to see her daughter by 5pm before the FD Center closes. I responded immediately since I had two sisters who passed away from FD, and they were the reason I went into medicine," explains third year NYU emergency medicine resident **Dr. Michael Coplin**, who grew up going to FD Day every year. When Dr. Coplin was introduced to the patient, Leigh Berkowitz, he noticed that she was about the same age as his sister Diane. "Before Diane passed, she used to talk about her friend Leigh. Both Leigh and my sister were married and in their forties and they used to all go out together," explains Coplin. "They had similar attitudes. They had overcome so much and never complained. Norma seemed very happy I was there since I knew the special needs of an FD patient. It was an honor for me to care for her," says Coplin. "He was a remarkable young man," adds Trager. "He stayed well beyond his shift and his level of empathy was very rare. He even came all the way to Riverdale to pay a Shiva call after Leigh passed. We had a real simpatico thing going on."

GETTING A GOOD NIGHT'S SLEEP WITH FD

One of our youngest FD patients recently started sleeping with a BIPAP machine and it is working exceptionally well. According to **Christy Spalink**, an acute care nurse practitioner at the Dysautonomia Center, "The BIPAP machine protects from serious breathing issues and really helps improve the quality of life for those with FD. A lot of times our FD patients don't even realize that they are suffering from apnea, and the ongoing lack of sleep even affects their aptitude in school. I'm so glad that some of the patients are open to using the machine and that it is working so well for them."

ARE WE CONNECTED?

If you are not receiving email communications from the Foundation, we may not have your current email address. If you'd like to add or update your email address, please send an email to info@famdys.org. In the subject line, include your full name and the words "add my email." We look forward to connecting!

MANY THANKS TO:

• **Buzz Neimark** who celebrated his Bar Mitzvah on May 20th and dedicated his Mitzvah project to FD in honor of his good friend **Jack “Jack Attack” Posnack**. Buzz raised \$1700 from



L to R: **Buzz Neimark** and **Jack Posnack** share a joke

donations from friends and family and then matched the amount from his own Bar Mitzvah gifts. According to Buzz, “My wish for my bar mitzvah was to make the world a better place, one mitzvah at a time. I chose the Dysautonomia Foundation as the recipient of my mitzvah project because I want to make Jack’s life and all the people afflicted with FD’s life better.

• **Joshua Goldberg** from Newtown, Pennsylvania, who celebrated his October 27 Bar Mitzvah with a mitzvah project to benefit FD. As a tribute to his cousin Rebekah Lieberman, who has FD, Joshua hosted a fundraiser to benefit FD at a local restaurant on September 8th.

• **Erin Boas** and the MEGA (Making Everyone Greatly Aware) Club at the **West Chester Rustin High School** for donating to FD in June. A recent graduate, Erin started the club in her junior year. MEGA raises money and awareness for nonprofit organizations and charities, and we thank Erin and the Club for selecting FD as a recipient of their fundraising!



Evan Kaplan with his new WWE belt

• **Mitchell Stuart, Paul Heyman** and **Brock Lesnar** for making **Evan Kaplan’s** dream come true! Evan, who is 21-years-old and lives in Rhinebeck, NY, loves WrestleMania and the WWE. Last fall Evan brought with him to the production of an FD video the WWE belt that his grandpa gave him for his birthday. Videographer Mitchell Stuart told Evan that he knew Paul Heyman, a famous pro wrestling manager. Not long after, Evan was thrilled to

receive a package containing a Universal Championship belt signed by Brock Lesnar, Evan’s hero. “I couldn’t believe it,” said Evan. “It started out as a huge coincidence but they all just made something great happen and I can’t thank them enough.”

FD DAY 2018 BRINGS MANY SMILES

On June 3rd, more than 150 FD family members and friends gathered from around the world to learn about major breakthroughs in the FD world at the 33rd annual FD Day in New York City. The day was filled with long-awaited reunions, informative presentations by renowned physicians and exciting activities for the kids. View videos from the morning presentations here: <https://vimeo.com/album/5243303>



FD family and friends reunite

IN LOVING MEMORY



Leigh Berkowitz
3/26/71-3/30/18



Martin Shandler
7/24/62-8/23/18

**Their Memories Inspire Us In
the Ongoing Battle For A Cure For FD**

FD CHAPTER NEWS:

CHICAGO:

Congratulations to the Chicago Chapter of the Dysautonomia Foundation and its golf event chairs **Gregg** and **Laura Meyers**,



Sophie Meyers rides the golf cart at Ravinia Green Country Club

parents of **Sophie Meyers**.

Many thanks also to Ladies Lunch chairs **Corrie Delevitt** and **Joey Rosen** and their hard working committee. The 15th Annual Golf Classic and 10th Annual Ladies Lunch & Play, held on July 23, brought together more than 400 people at **Ravinia Green Country Club** in Riverwoods, and raised more than \$95,000 for FD.

MONTREAL:

We are thrilled that the Montreal chapter funded a new, state-of-the-art blood pressure machine, the Finapres Nova, to monitor hemodynamic stability for patients in the clinic at the Dysautonomia Center at NYU. This donation was made in memory of **Ben Bigman** who passed away in January. **Norma Bigman**, the founding President of the Montreal Chapter along with her husband Ben, started this chapter after the loss of their son, Neil, who was only 7 years old and was born with FD.

Many thanks to the Montreal chapter for their contribution to NYU Langone representing funds raised by the **Schwartzberg family** in memory of their beloved son and brother **Steven Schwartzberg**. We are extremely honored that the Schwartzberg family chose to pay tribute to Stevie's memory by funding a mental health initiative at the Dysautonomia Center that will benefit FD patients by offering much-needed counseling and emotional support services.

NEW EQUIPMENT FOR THE NYU DYSAUTONOMIA CENTER

In October, the Dysautonomia Center ordered a highly anticipated optical scanner for the retina. The Zeiss Optical Coherence Tomography® (OCT) has been at the top of the Center's equipment "wish list" for a long time. After the Center secured grants from the **Michael J. Fox Foundation** and **PTC Therapeutics, Inc.**, friends of the Foundation came together to match funds needed to secure the machine. The Zeiss OCT® will be used to obtain high definition scans of the retina and will monitor degenerative changes. Now, the Center will be able to track the progression of the disease and identify compounds that help save vision. The much sought after machine will help the Center determine response to treatments aimed at slowing the loss of nerves in the eye.



A MESSAGE FROM **LANIE ETKIND,** EXECUTIVE DIRECTOR

It's hard to believe that on September 18 I completed my first year as Executive Director of the Familial Dysautonomia Foundation. Having known Jack Posnack since his birth, I was certainly aware of the physical challenges faced by someone with FD. But over the past twelve months, I have learned so much more about the impact of the disease as well as this foundation's longtime efforts to both find a cure and to enhance quality of life for those affected. I am deeply impressed by all that this "small-but-mighty" foundation has accomplished over the years, particularly considering that FD is an "ultra-rare" disease, and the community affected so limited in size. That considerable funds have been raised, and so much learned about this disease, is a testament to the fierce love and devotion of families, the dedicated and hard-working board of directors, a deeply caring medical staff, brilliant researchers worldwide who continue to push the boundaries of science, a wise and knowledgeable Scientific Advisory Board, and an extremely generous circle of supporters. There is still so much more to accomplish, and I look forward to all of us working together towards reaching our shared goals.

DR. LEFCORT RECEIVES \$2.9 MILLION NIH GRANT FOR FD RESEARCH

This September, neuroscientist **Dr. Frances Lefcort** and her team at **Montana State University** in Bozeman, MT, announced a five-year grant for \$2.9 million from the **National Institute of Health** for "Metabolic control of gut-brain axis in Familial Dysautonomia." **Dr. Frances Lefcort** is a professor at Montana State and is a member of the FD Foundation's Scientific Advisory Board. According to Dr. Lefcort, who wrote the grant in collaboration with the Dysautonomia Treatment Center, "The grant will allow us to look at FD patients and examine their source of gastrointestinal issues that affect their metabolism." Adds Lefcort, "We are going to study the gut microbiome and metabolism and how that affects neuronal health in both FD patients and FD mouse models. If we can find a difference in bacteria living in the gut, we could develop a special cocktail to treat the gut function. It's an exciting time for us."



President's Message

Dear Friends,
As always, I write with gratitude for your support and interest in the lives of people with Familial Dysautonomia and their allies. We are eager to present this latest edition of DYScourse, update you on the work of the Familial Dysautonomia Foundation, news regarding the constant progress of the Treatment Center, the tireless efforts of those who support our fundraising, and of course, on the lives of the people with FD who are at the heart of our mission. Under Executive Director Lanie Etkind's leadership we are happy to report progress on a number of fronts in this newsletter, and encourage you to check us out online for the release of our updated website before the end of the year. Best wishes for a happy holiday season with family and friends and a very happy Chanukah..

Faye Ginsburg
President of the Board of Directors

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THE MAYOR OF LONDON WELCOMES THE FD COMMUNITY WITH OPEN ARMS

This summer, **Faye Ginsburg** and her husband **Fred Myers**, took their daughter **Samantha** to London for a visit. They were extremely impressed with how accessible the city is for people with disabilities. According to Faye, "The UK disability care was amazing! After



Sam and her Father, **Fred** in London

facing long lines at Buckingham Palace, a warden noticed Sam in her chair and whisked us off to the front gate (where royals enter!), had us picked up on a golf cart adapted to hold a wheelchair to get us across a very large, gravelly courtyard, where another set of wardens set up our accessible tour, took us on a wheelchair lift, got us to the elevator in the back area when we were ready to see the State rooms, and sprayed Sam with a little Evian bottle when she nearly fainted from the heat. They even had an accessible gift shop!" Faye shared these comments with her dear friend who lives in London, and her friend forwarded these sentiments to the Mayor! Almost immediately, the Mayor's office responded with the following: "I am so glad to hear that your friend and her daughter had such a wonderful and enjoyable trip to London. The Mayor has made it clear that London is open to anyone who wants to work, live or indeed visit, no matter their age, gender, race or ability. It is wonderful to hear that this is a reality for those visiting. The Mayor hopes that your friend and her daughter will come back and visit again soon."

2018 GOLF OUTING A SWINGING SUCCESS



Golfers await the tee off at the 22nd annual FD Golf Outing

We could not have asked for a more beautiful day for the 22nd Annual FD Golf outing on Long Island on May 22. Thanks to all those who came together to support and raise funds for those with familial dysautonomia!

With 124 golfers attending the event, it was a swinging success. Many thanks to Golf Chairs **Paul Wexler, Steve Fass, Steve Kietz** and **Rachael Schlau** for their endless support.

KIDS OF COURAGE

In August, **Kids of Courage**, an organization dedicated to improving the lives of children and young adults with serious medical diagnoses, hosted a trip to Las Vegas for more than 100 campers, including 13 with FD. According to **Sam Landau**, who has participated in this camp experience for the last nine years, “I look forward to these trips all year long. I see tons of my friends and we go to different places each time. This year we went to see David Copperfield and a Cirque du Soleil show. It was awesome!”



2018 Kids of Courage Trip (L to R): Melanie Cohen, Sara Zucker, Frannie Cohen, Sam Landau, Evan Kaplan, Rebecca Newman, Rachael Eisenson and Gabi Jassie.
Not pictured: Steven Wexler, Danny Goldstein, Michael Brenner, Dovi Porisch and Esra Kress

FOR FURTHER READING, CHECK OUT THESE ARTICLES OF INTEREST

- “Respiratory care in familial dysautonomia: Systematic review and expert consensus recommendations,” authored by **Dr. Mikhail Kazachkov**, members of the **Dysautonomia Center** team and others, was published in the August issue of *Respiratory Medicine*. Read the full article here: <https://files.acrobat.com/a/preview/8ee2638d-9dea-4e5b-b030-f79e50077e06>
- “Retina-specific loss of Ikbkap/Elp1 causes mitochondrial dysfunction that leads to selective retinal ganglion cell degeneration in a mouse model of familial dysautonomia,” authored by FD family member and Scientific Advisory Board member **Dr. Frances Lefcort** and her colleagues, and published in the June 2018 issue of *The Company of Biologists*. The Dysautonomia Foundation was credited as a funder of the study. Read the full article here: <https://files.acrobat.com/a/preview/1532080c-4488-4e7d-aff7-123e9a616f6a>

SPOTLIGHT ON CHRISTY SPALINK

This past Spring, acute care nurse practitioner **Christy Spalink** gave several live interviews on Doctor Radio at NYU on SiriusXM about her work with FD patients. The groundbreaking 24/7 national radio channel



features live, call-in shows hosted by leading NYU Langone Health professionals from a vast array of fields. Christy spoke about raising awareness of FD and the importance of partnering with patients and families in order to improve the patient’s quality of life. Christy is currently working on a clinical trial offering cognitive behavioral therapies to address different mental health issues such as anxiety and depression that are common in FD. “The great thing about this trial is that we are doing it by telemedicine so patients can participate from anywhere,” notes Christy. To learn more, contact the Dysautonomia Center at 212-263-7225.

BEFORE YEAR-END, MAKE A TAX-SAVVY GIFT TO SUPPORT FD

If you are 70½ years of age or older, you can make a tax-free charitable gift from your IRA directly to FD (up to \$100,000 a year). This provision offers a unique opportunity to individuals who wish to support their favorite charities – we hope we are one of them! To learn more, contact your financial advisor or **Lanie Etkind**, the Dysautonomia Foundation’s Executive Director at 212-279-1066 or email letkind@famdys.org.

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-  <https://www.instagram.com/foundationdysautonomia/>

**Our new website is in the works.
Check back soon for a whole new look!**



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**YOUR SUPPORT MEANS A LOT
TO US. PLEASE DON'T FORGET TO
USE THE ENCLOSED ENVELOPE!**

DYS/COURSE 2018-2

SAVE THE DATE

Sunday, April 7: FD Art Expo, Allouche Gallery, New York. We are looking for artists who have FD to participate. Please contact Lanie Etkind at letkind@famdys.org if you are interested.

Monday, May 20: 23rd Annual FD Golf Outing, Glen Oaks Club, Old Westbury, NY

Sunday, June 2: 34th Annual FD Day, NYU Langone

DYSAUTONOMIA FOUNDATION

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info@familialdysautonomia.org
Lanie Etkind, Executive Director

A copy of our latest annual report may be obtained from the FD Foundation (315 W. 39th St., Ste. 701, New York, NY 10018) or from the New York State Attorney General's Charities Bureau (28 Liberty Street, 15th Floor, New York, New York 10005).



FD NEWSLETTER FALL 2018

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The Dysautonomia Foundation is a 501 (c) (3) non-profit organization dedicated to funding treatment of, research on and awareness about familial dysautonomia, a rare, life-threatening, Jewish genetic disease that affects the autonomic nervous system.