

SUMMER 2016

the Advocate

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What Does 'Patient Engagement' Mean for the FA Family?

By Ron Bartek

Dear Friends,

Beginning about three years ago, few phrases in the therapy-development arena have been used as frequently and created as much buzz as “patient engagement” and “patient-focused drug development.” What do those phrases really mean and what do they mean to us?

Put simply, these phrases are clear indications that all of our stakeholders—government partners such as the Food and Drug Administration (FDA), National Institutes of Health (NIH), and Congress, academic scientists, pharmaceutical companies and other patient groups—all now recognize that success requires placing the patient at the center of all therapy development efforts. I like to say that we must increase the frequency and volume of the patient's voice early and often, from the beginning to the end of the research and development process.

Many of you are saying, “Well, of course! That's obvious!” But, that is your reaction because the FA community has always put our patients at the center of the research effort. Consequently you might not recognize that in many other communities, scientists and drug developers are still not reaching out to engage patients until they recruit participants for their clinical trials.

Let's take a quick look at some of the ways we all work together to accelerate FA therapy development by getting personally involved early and often. I have often said that the first step for each patient is to get enrolled in the patient registry (see <http://www.curefa.net/registry>). The registry is our primary communication tool for new clinical trials. Furthermore, every additional FA patient that enrolls in FARA's patient registry helps attract additional pharmaceutical companies to our cause. This is the first way we show pharmaceutical companies that we are an active and engaged community.

Each time an FA patient goes to a FARA clinical site (see <http://www.curefa.org/network>) to see an FA clinician, that patient's voice is being registered to record changes since

the last visit, which tests are most sensitive to those changes, and which symptoms are most important to the patient. All that information is referred to collectively as a “natural history database” and is invaluable when drug companies meet with the FA clinicians and FARA to select drugs with potential for addressing those symptoms and to design the optimum clinical trials to test them in FA patients.

Additionally, many FA families have invested extra time in research by participating in clinical studies (see Patient Perspective: Participating in a Biomarker Study, *page 11*). The goal of a clinical study is to learn more about what is happening in FA patients on a metabolic, cardiac, or neurological level. An increasing number of clinical studies have recruited people with FA in an effort to identify biomarkers—ways to measure changes in the disease through blood samples or via imaging studies (see *page 12* for a list of ongoing clinical studies). The hope is to be able to measure change quicker and more efficiently and shorten the length of future trials.

Over the past year, FARA's academic labs and pharmaceutical partners have submitted an increasing number of requests to engage the patient perspective, beyond clinic visits, clinical studies and registry numbers. Whether that means an in-person visit to tell their story, a feedback session on the patient journey, or a focus group to test possible methods of drug administration, the FA community quickly and enthusiastically responds positively to each request. Representatives from pharmaceutical companies remark at how rich these sessions have been in both information and motivation.

Meanwhile, of course, FARA continues to increase the frequency and volume of the FA patient's voice in working with our government partners at the FDA, NIH, and U.S. Congress to help create a financial and policy environment in which FA research and development can thrive. An important aspect of this work with government partners is FARA's intensifying effort to fine tune the FA patient's voice in educating our colleagues at the FDA. For example, FARA leadership regularly participates in meetings scheduled by our drug-company partners with the FDA to seek consent to conduct clinical trials. At the FDA's request, FARA has made a number of presentations at the Agency explaining FA and the ways in which the FA patient community has become the central driving force in FA drug development. FARA is

President's Message

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currently working to schedule additional “patient-focused drug development” meetings with the FDA, including one meeting in which we hope some patients and families will be asked to participate.

FARA's intention in all these “patient engagement” efforts is to accelerate FA therapy development by inserting the patient experience and perspective from the very beginning so that targets important to our patients set the basic science, clinical trials are designed to be beneficial to patients, and the FDA understands FA and FA patients sufficiently to recognize when a therapy's benefits outweigh its risks.

So, again, “patient-focused drug development” is nothing new to you because you and the FA community have long been helping lead the way in this important direction. I would like to ask, then, that you continue to lead the way by enrolling additional patients in the FARA patient registry, scheduling annual examinations at an FA clinical site, participating in FA clinical studies and trials, and participating in visits to pharma partners and academic labs. Together, the FA community continues to build its irresistible force that is closing in on the once-immovable object of FA. We will overcome and, together, we will cure FA.

Warm regards,

Ron

What is Friedrich's Ataxia?

Friedreich's ataxia (FA) is a debilitating, life shortening, degenerative neuro-muscular disorder. Onset of symptoms can vary from childhood to adulthood and can include loss of coordination (ataxia) in the arms and legs; energy deprivation and muscle loss; vision impairment, hearing loss, and slurred speech; aggressive scoliosis (curvature of the spine); diabetes mellitus; and a serious heart condition. While the mental capabilities of people with FA remain completely intact, the progressive loss of coordination and muscle strength in FA leads to motor incapacitation and the full-time use of a wheelchair. There is currently no treatment or cure for FA. FARA is a 501 (c) (3) tax exempt, non-profit organization dedicated to supporting research that will improve the quality and length of life for those diagnosed with Friedrich's ataxia and will lead to treatments that eliminate its symptoms.

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FARA Updates Strategic Initiatives

By Jennifer Farmer

Every few years the FARA board, advisors and staff meet to focus on strategic planning. FARA's strategic initiatives keep us focused on the highest research priorities and organizational needs that will advance our mission, to treat and find a cure for FA. In December 2015, the board met for two days of strategic planning, which resulted in FARA's Strategic Initiatives 2016-2019. Because the FA community is such an integral part of all of FARA's work, it is important that you, too, are aware of these initiatives so that we may continue to move forward efficiently together.

Initiative 1. A deep and diverse FA treatment pipeline is required. FARA will help fill the pipeline with vetted, robust treatment candidates.

- The FA community will require multiple treatments to treat all symptoms and individuals with FA. This necessitates a treatment pipeline that targets the underlying disease and symptoms using a variety of approaches. We currently have several candidates in the pipeline advancing through clinical trials that target various aspects of mitochondrial function and down-stream consequences of frataxin deficiency as well as emerging genetic and protein replacement strategies. FARA identified a need for additional drug discovery of compounds or strategies that directly target increasing frataxin transcription and/or translation (gene and protein) and frataxin mimetics (compounds that could substitute for frataxin). To this end, FARA partners with larger pharma companies with high-throughput screening experience, extensive compound libraries, and resources to support small molecule drug discovery. Also, FARA will continue to support more basic science that enables discovery of new biological or cellular targets to diversify drug discovery approaches.

Initiative 2. FARA will advance FA pipeline candidates from drug discovery through development by addressing gaps in translational research and identifying stalled candidates which we can accelerate.

- FARA will invest time and resources toward de-risking

activities such as funding FARA grants for new cell and animal models to create a comprehensive and validated toolbox for early testing of candidates.

- FARA will work closely with academic and industry partners advancing gene and protein replacement strategies as these therapeutic approaches are novel and have unique challenges while having the potential for significant therapeutic benefit.
- FARA will also identify any priority pipeline programs that stagnate due to lack of funding, scientific challenge, or the need for a partner and develop an action plan that supports the forward movement of the compound.

Initiative 3. FARA will support and optimize the number of industry partners with a strategic interest in FA.

- Over the past three to four years, FARA has made significant strides in outreach and partnership with industry, including small biotech, medium to large pharmaceutical companies and venture capital companies. We believe that we must have industry engagement at all stages of drug development and that we can be valuable partners to actively address their needs and issues challenging forward progress in FA. FARA fosters such engagement by advancing and supporting the development of infrastructure in the Patient Registry, Collaborative Clinical Research Network/Natural History, models and biomarkers. We also continue to develop industry relationships in areas of new technology or innovation, for example, gene editing companies and wearable or remote monitoring devices.

Initiative 4. FARA believes that investment in Clinical Research Infrastructure expedites and promotes clinical development and attracts industry to FA. This infrastructure can improve access the treatments, modify standards of care and monitor long-term clinical outcomes.

- The Collaborative Clinical Research Network, which has been engaged in FA research for 12 years, has developed an experienced investigator network with an understanding of disease progression and natural history data that informs clinical trial design and expedites trial initiation and a biorepository of blood and tissue samples. FARA advisors believe that we need to continue to grow this natural history study by:
 - supporting individuals who participate so that they can continue to return for follow-up visits (longitudinal data on individuals is critical)

Continued from Page 4

- identifying and enrolling more children and individuals at earliest symptom onset
- facilitating and supporting investigator-initiated clinical research through the network and encouraging research to support evidence-based clinical treatment guidelines

Initiative 5. FARA will grow the scientific and medical community.

- From day one, FARA has increased the scientific and medical community focus on FA. It is essential to bring new ideas and expertise to advance and challenge the field and to nurture the development of young scientists and clinicians. We believe in proactively seeking investigators, clinicians and technologies to join the field, investigating barriers to entry to the field and why people leave, as well as providing mechanisms for funding young clinical researchers and clinicians to obtain FA training.

Initiative 6. FA is a global condition that requires international engagement of all stakeholders. The involvement of patients, researchers, medical community, industry, advocacy organizations and government agencies is essential for leveraging knowledge, resources and influence.

- FARA has established relationships with many international advocacy organizations, funds research globally, and has a Global Patient Registry, however there needs to be further coordination and collaboration. International research conferences have been very effective, and FARA continues to work with other advocacy organizations and the international research community to host these conferences every two years. FARA is also working with international partners to rebrand and redesign the patient registry to improve utility, responsiveness and inclusion and to allow the registry to be used as a research tool.
- FARA needs to partner and support international advocacy organizations to reach out to government agencies with regulatory authority (e.g., European Medical Agency) to raise awareness about FA and facilitate clinical trials and drug approvals, internationally.

Initiative 7. Advocacy! FARA will advocate on behalf of government partners (FDA and NIH) and in front of federal/national representatives to raise awareness and resources for rare disease.

- FARA's Advocacy Committee actively engages with efforts to pass government bills that support research, promote industry interest in rare disease drug development and greater

resources for NIH and FDA, and protect and engage patients in the research process.

- FARA regularly seeks NIH and FDA participation in research and clinical meetings. FARA has reached out to the FDA, via a letter of intent, to host an FA Patient-Focused Drug Development in 2017.

Initiative 8. Regulatory science. Identifying, supporting and communicating the regulatory approval path can accelerate movement of drug development and avoid unnecessary delays, ensuring that the patient voice is represented in the process.

- FARA is developing relationships with key committees and review groups within FDA divisions of Center of Drug Evaluation and Research (CDER) and Center for Biologics Evaluation and Research (CBER) to increase knowledge and understanding of FA (e.g., sharing data on natural history of FA) and for us to understand their current standards and operations. For example, as we further develop new outcome measures and biomarkers for clinical trials we are seeking FDA feedback and input and ultimately acceptance as valid endpoints.
- FARA is actively involved in assisting pharma partners in planning and participating in regulatory filings and meetings and navigating the regulatory process.

Initiative 9. Patient engagement goes beyond organizational leadership and representation and requires Research Ambassadors within the FA community who can be called upon to work with FARA leadership, industry partners and government agencies.

- FARA is launching a new training program for Research Ambassadors so that patient representatives are prepared to engage directly with industry in pre-clinical and clinical development.

Initiative 10. Awareness. FARA believes that regular and consistent communications to stakeholders, especially the patient community is of utmost importance. Each actively involved patient adds significant value to research and clinical advancements.

- Whether it is through social media, e-blasts, symposia or this newsletter, FARA's communications focus primarily on the patient and donor community. We must first be informed if we are to function as a cohesive unit. The patient story is also a key tool in raising awareness in traditional media and in person presentation, potentially reaching patients unknown to FARA.

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FARA Announces Recipients of Named Grant Awards

By Bronya Keats, Ph.D.

Research funding provided by FARA is crucial, not only for the ongoing support of outstanding research projects that move us closer to establishing effective treatments for Friedreich's ataxia (FA) but also to encourage expansion of the FA research community. Of note, FARA funding is providing major support for drug discovery, biomarker development, and the Collaborative Clinical Research Network, all of which are critical for FA clinical trials.

In addition to funding general research projects, each year FARA awards three named grants. We congratulate the recent recipients of the FARA named awards: Dr. Andrew Nichols, Dr. M. Dolores Moltó and Dr. Hélène Puccio.

Dr. Nichols (Catabasis Pharmaceuticals, MA) received the Kyle Bryant Translational Research Award for pre-clinical studies of a promising therapeutic candidate, CAT-4001, in mouse and cellular models of FA. CAT-4001 is a novel small molecule designed to activate the transcription factor Nrf2, which is decreased in frataxin deficient cells. Dr. Jordi Magrané (Weill Cornell Medical College, NY) and Dr. Mark Payne (University of Indiana School of Medicine) are collaborating with Dr. Nichols on this project.

Dr. Moltó (Universitat de València, Spain) received the Bronya J Keats International Research Collaboration Award for her project



Dr. M. Dolores Moltó

to identify genetic factors that modify the low frataxin levels caused by the GAA expansion and result in normal frataxin expression. To do this, Dr. Moltó and Dr. Llorens, a postdoctoral researcher in her laboratory, are performing a high-throughput screen using her FA *Drosophila melanogaster* (fruit fly) model, which contains a GAA expansion. The evaluation of positive hits will be done in close collaboration with Dr. Aaron Voigt (Aachen University, Germany), who has extensive experience with genetic screens in *Drosophila*. This study has the potential to discover new therapeutic approaches as well as advance our understanding of the molecular mechanisms underlying the GAA expansion inhibition of frataxin levels.

Dr. Puccio (IGBMC, University of Strasbourg, France) received the Keith Michael Andrus Award for Cardiac Research. She and Dr. Brahim Belbellaa, a postdoctoral fellow in her group, are investigating the potential adverse effects of overexpression of cardiac frataxin, generating a new cardiac-specific conditional knockout mouse



Kyle Bryant with Catabasis CEO Jill C. Milne and Senior VP of Research & Non-Clinical Development, Dr. Andrew Nichols.



Dr. Hélène Puccio with Gavin Lambert at the FARA Energy Ball.

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- FARA believes an investment in educational materials for clinicians can shorten the time to diagnosis and more quickly identify new people living with FA.

Initiative 11. Fundraising. FARA must raise \$6 million annually to ensure adequate funding for effective execution of our strategic plan.

- FARA continues to expand our research initiatives and grant program to accelerate progress, which requires an increasing level of funding. FARA's research funding comes from the families affected by FA and the friends, families, and business associates that know and love them. Whether members of the community make a personal donation, host a fundraiser, organize a team for rideATAXIA, or attend the FARA Energy Ball, they have proved to be engaged and committed to raising the necessary funds for critical research. We are grateful for their support and generosity. ●

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model, and identifying potential biomarkers of cardiac frataxin deficiency. These pre-clinical studies are critical follow-up steps to Dr. Puccio's successful correction of FA cardiomyopathy using gene therapy in a mouse model and they must be done before a FA cardiac gene therapy clinical trial can begin.

In addition to the three named grants, FARA is presently supporting 21 FA research projects. Titles and summaries of most of the projects funded by FARA are available at:

www.curefa.net/RPMP/public/pggrantlist.aspx

We truly appreciate and extend our heartfelt thanks to all our reviewers who devote their time and effort to reviewing grant applications submitted to FARA and are essential for ensuring the high quality of research projects funded by FARA.

Grant application instructions, including deadlines for letters of intent (LOIs) and full applications are available at

www.curefa.org/grant

Additionally, a comprehensive list of research publications resulting from FARA funding, as well as other publications on FA, is provided at: www.curefa.org/scientific-news ●

Join The FARA Patient Registry!

Get notified about new trials and help advance FA research!

www.curefa.net/registry

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Team FARA is made up of people around the globe who participate in endurance events on behalf of the Friedreich's Ataxia Research Alliance (FARA) with the goal of raising awareness and funds for FA research. Keep the momentum going by participating in local events such as rides, 5K runs marathons, ½ marathons, and triathlons.

Email info@curefa.org for more information on how to join Team FARA.

<http://www.curefa.org/team-fara>



THE FARA ENERGY BALL

September 15
Understanding Energy
for a Cure
Scientific Symposium

September 16
Patron Party

September 17
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Sept. 15, 16, and 17, 2016
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Rallying Support Across the Country

By Jamie Young

“CURE...FA! CURE...FA! CURE...FA!—LET’S GO!” These words are cheered by cyclists at the start of every ride to rally everyone to ride with intention.

This year is off to a great start with four successful rideATAXIA events fundraising over \$300,000 in Dallas, Atlanta, NorCal and Chicago. It truly takes an army of community members at each ride location to get the job done. These armies are made up of several FAmilies and their family, friends, and co-workers. They raise funds, recruit riders, fold t-shirts, mark the course, store and transport supplies, set-up tables, take down tables and staff the whole event with volunteers. This is the starting point to generating research funds and progress!

There are so many amazing FAmilies that make rides happen across the country. Each year we highlight a couple of them to get their personal perspective on the ride. The Dagley Family, aka The A-Team, from Frisco, Texas makes up a part of our amazing committee for rideATAXIA Dallas. Inspired by brothers Jason and Alex, both living with FA, the team brought a huge number of riders and were in the top three of fundraising teams this year bringing in \$7,415.

The A-Team helped grow and cultivate a necessary component of the ride- sponsorship. They brought in three new bronze-level sponsors that also attended the event. This has been a priority area for the ride program—to not only to grow funds for research but to establish sustainable partnerships in the local community. The A-Team said they were able to accomplish this by starting with the local businesses they know and trust, leaving no stone



The A Team at rideATAXIA Dallas 2016.

untuned. “Make as many contacts as you can and whatever you do, don’t stop promoting the ride,” says Alex. They also advise to take advantage of social media to post regularly information about the ride.

The Dagley Family got involved because the ride was something they felt they needed to do as a family. They especially enjoy the overall atmosphere that includes the camaraderie of FAmilies getting together with the local community to have fun and fund research. “The ride is a great way to be a part of something bigger than yourself,” Alex states. “I never thought my family and I would ever get this kind of support from friends and FAmily that we have through the ride.”

Interested in getting more involved in rideATAXIA? Email

jamie.young@curefa.org •

rideATAXIA 2016 Schedule

RA Philadelphia	October 16, 2016
RA Orlando	November 13, 2016



Nine Grants Awarded in 2016 Ataxian Athlete Initiative

By Kyle Bryant

The Ataxian Athlete Initiative (AAI), a unique program that provides adaptive cycling equipment to people with ataxia, selected nine recipients in 2016 who have demonstrated a strong desire to stay healthy and fit despite their progressive disabilities.

In 2016, the AAI's grants for adaptive cycling equipment reached seven states and two continents. Alena Wolfson of Glendale, Arizona; Candace Honeycutt of Grinnell, Iowa; Greg Ostrom of Glencoe, Ontario, Canada; Jolin Kowalski of Vicksburg, Michigan; Joshua Lamascus of Marina, California; Leslie Ballard of Yuma, Tennessee; Marinda Cauley of Chatanooga, Tennessee; Jacob Ferguson of Montgomery, Texas; and Endrit Januzaj of Desan, Kosovo received cycling equipment funded by the AAI initiative.

"With no access to adaptive equipment in my country, this grant from the AAI will open up a world of mobility and opportunity for me," said Endrit Januzaj of Desan, Kosovo.

This year, the AAI was sponsored by Catrike, UVA Sun Systems, Front Burner Brands, The Texas Irish Foundation, and a new partnership with Global Genes through the RARE Patient Impact Grant program to fund the U.S. recipients.



2016 AAI recipient Josh Lamascus riding his new Catrike Trail.

"The goal of the RARE Patient Impact Grant program is to fund projects that make tangible differences in patients' lives," said Global Genes' Director of Patient Advocacy, Amanda Knitter. "The Ataxian Athlete Initiative is a program that is the epitome of true impact and we are proud to support the Friedreich's Ataxia Research Alliance's efforts. We hope to expand the grant program each year so we can support more worthy projects in the rare community."

AAI grants are administered through a competitive application process. Applicants research the most appropriate adaptive cycling equipment to suit their abilities and describe how such equipment would help them to reach their fitness goals and improve their quality of life.

"When I'm on my trike, I don't think about atrophy or neurodegeneration, I don't think about heart complications, or life expectancy. I carry these thoughts and feelings with me, but I am mostly just focused on where I'm going, and the pedal strokes it will take me to get there," said Liam Dougherty, a previous AAI grant recipient.

The AAI has provided equipment for 33 individuals since 2009. Visit curefa.org/aai for more information and to view more photos of past recipients. The next application cycle begins in Spring 2017. ●

FARA Store

For yourself, your family and friends, or to have customized FARA items at fundraising events, visit the FARA store!

www.cureFAstore.com.

FARA caps, polo and t-shirts, wristbands, etc.



Patient Perspective: Participating in a Biomarker Study

By Tricia Herman-Maul



I was diagnosed with Friedreich's Ataxia via a DNA test at the age of 28. My diagnosis was no surprise as my sister, Tara had been diagnosed with FA in 1997. When I received my diagnosis, the neurologist also handed me a paper about FARA and information on how to enroll in their patient registry (which I immediately did). In 2009, my

husband, sister, and I attended the symposium in Philadelphia where we met many wonderful people and started connecting with the FA community. Since then we have attended FA Woodstock, RideATAXIA Chicago and held our own FARA Fundraisers. Through FA Woodstock I have been able to participate in blood draws, skin biopsies, hearing studies, and network with FARA representatives.

In November 2015, I received a message from FARA about a research study at the University of Rochester, New York entitled "In-Vivo Confocal Imaging of Meissner's Corpuscles as a Biomarker in Friedreich's Ataxia (FA)." Since being diagnosed with FA in 2008, I had been patiently waiting for my opportunity to be part of a trial that wasn't demanding time away from home. Living in western New York with my husband, CJ and three children: Amelia (11), Calvin (9), and Justin (8), the location and requirements of the trial were very important to me. After speaking with the study coordinator, Janet Snowden, I found this study would only require three day visits to Rochester, which is only 90 minutes from my home. Arrangements were made for CJ and I to spend the night in a hotel just minutes away from the University of Rochester Medical Center for the following week.

The day of the study we arrived before 8 a.m. to the Medical Center, excited and anxious for what the day would entail. I learned that there will only be 16 FA patients whose trial results will be paired

to a healthy person of the same age to measure how FA progresses in the peripheral nervous system and how useful these methods may be to look at changes during future clinical trials. After some paperwork and a routine exam I was eligible to participate. We soon began with the study tests—that's when the real fun began!

After my blood draw, I met with a physician who completed a series of physical movements with my mouth, speech, arm/leg strength, and reflexes for the Friedreich's Ataxia Rating Scale (FARS). I had a few minutes of rest while the team brought in the In-vivo Confocal Reflectance Microscope. We were able to watch on the computer screen as the microscope showed layers of skin on my hands and feet revealing nerve "pits". The purpose of this test was to show how far the nerves travel to the outermost layers of skin.

Next was the nerve conduction study, which was definitely my least favorite. During this test, small electrodes were placed on my skin and shocks of varying intensity and duration were sent through my hands, arms, and feet. Michelle, who administered the test, used a lot of pressure and differing shocks to get good readings, but it was a very uncomfortable test as the muscles contract during each shock. The purpose of this test is to calculate the speed by which nerves carry information.

After lunch, I met with the physical therapist to do the walking and standing portion of the FARS test. This included timed tests like the peg board with both hands, stand with eyes open, stand with eyes closed, one footed stand, and a distance walk.

Next, I completed the Quantitative Sensory Testing. A sensor was strapped to my hand and foot that delivered an instant cold, and I had to press a button when I felt it. The intensity of the cold varied to find the threshold at which I could feel the change in temperature. Likewise, there was a vibration test to report the threshold when I could feel a certain intensity of movement. The picture shows my hand on bean bags to prevent me from feeling any vibration transferred to the table.

My favorite test was the monofilament touch-pressure threshold test. During this test, a nylon monofilament with varying strengths (like fishing line) touched to my fingers as I sat with my eyes closed and it was my job to tell the study coordinator if she was touching my finger or not. This was repeated 10 times per filament until I got at least seven right in the sequence of 10. The threshold at which I could feel the filament was very obvious. What I didn't like was that study guidelines stated we could not move on to the next filament



Gift from the Eloise Sitzer Trust Supports Biomarker Research in FA

By Felicia DeRosa

Eloise Carrie Sitzer of Saint Louis, MO. The name is likely unfamiliar, but she deserves our sincere gratitude. I am sorry I only learned of Ms. Sitzer several months after her passing when a significant financial gift arrived in her name at the FARA office. Ms. Sitzer, predeceased by all her family members including two children who had Friedreich's ataxia, designated half of her trust estate to FARA to advance FA research. Based on her age when she passed (93), Ms. Sitzer's children were likely diagnosed with FA long before the FA gene was discovered and FARA existed.

Her generous gift is a moving and meaningful tribute to her children, Larry William O'Connell and Beverly C. O'Connell and an impactful gift to the entire FA community. FARA will utilize these funds to support an ongoing investment in biomarker research. Biomarkers are biological measures that indicate change in a disease and they have the potential to shorten the length of clinical trials. In the Winter issue of *The Advocate*, FARA's Vice President of Research Development, Jane Larkindale wrote an article detailing some of the clinical studies to find effective biomarkers in FA. Many members of the FA community have blogged about their experience participating in clinical studies (see Patient Perspective: Participating in a Biomarker Study, page 11). It seems fitting that Ms. Sitzer's significant gift designed to speed our path to effective therapies be used to find concrete measures to tell us whether a therapy is effective in someone living with FA.

If you would like more information about how to include FARA in your planned giving, please either contact your lawyer, financial advisor or FARA at info@curefa.org.

Continued from Page 11

if I could not feel it. This test really made me concentrate patiently and took about a half hour to complete. Before I left a skin biopsy was taken from my thigh for researchers to freeze, cut in layers, and study under a microscope.

Overall, the experience was awesome! The 10-hour day was longer than expected since I was the first patient in the study, but definitely worth it. I am hopeful from my experience that one test will show a positive and non-intrusive method to measure FA progression in the peripheral nervous system that can be used to monitor patients in future clinical trials. ●



Tricia and her family at rideATAXIA Chicago in 2013.

Study Recruitment

The clinical trials and studies below are actively seeking patient participation. To access the recruitment notices for any of these trials, visit click on the study title(s).

Phase 2 Trial with Reata Pharmaceutical's RTA 408

<http://www.curefa.org/active-clinical-trials/reata-phase-2-trial-for-rta-408>

Reata and the clinical sites are actively enrolling a seventh cohort of 8 patients to test at a dose of 160mg (the same as cohort six) of study drug. We need to enroll this cohort quickly.

In-Vivo Confocal Imaging of Meissner's Corpuscles as a Biomarker in Friedreich's Ataxia (FA)

<http://www.curefa.org/pdf/UnivRochesterStudy2015.pdf>

Study of Cardiomyopathy in Friedreich's Ataxia Patients

<http://www.curefa.org/active-clinical-trials/weill-cornell-study-of-cardiomyopathy-2>



Meg Larson

Interviewed by Abby Yingling

Name: Meg Larson

Age: 21

Where do you call home? Washington, D.C metro area

Education: Senior at George Mason University, studying communication with a concentration in public relations and a minor in psychology.

How long have you known you are living with FA? (When and how were you diagnosed?) I was misdiagnosed with Charcot-Marie-Tooth (CMT) when I was 14. I never questioned the diagnosis, regardless that many of the supposed symptoms did not apply to me. At 16 I went to John's Hopkins University Hospital to meet with Dr. Thomas Crawford, a specialist in CMT. Within 5 minutes, he correctly diagnosed me with Friedreich's ataxia.

Describe your transition from walking to walker/wheelchair. I won't sugarcoat this at all, I promise you that. I was able to walk fine, a bit unsteady, but fine, until I was 20. At 18, I walked at least 32 New York City blocks a day. My transition from walking to a wheelchair/scooter was something I fought every step of the way (pun intended). At first I was embarrassed to use a mobility aid in public. I didn't want people to think that I was this feeble being. Due to my own stubbornness, I fell in public, grew fatigued to the point of near collapse, and looked like a drunken fool fighting the inevitable necessity of a wheelchair.

When I first started using my trusty TravelScoot, I was a bit embarrassed, but I grew to love it. It's the 'holy grail' of mobility assistive devices for long distance (i.e on campus). I got my manual wheelchair after the scooter, and it's been a massive help in home, at restaurants, and in more confined areas in general. For a long time, I thought that use of a wheelchair/scooter would restrict me. Quite the opposite—these devices are not a restriction, they are an asset to my own personal freedom.

When FA gets you down, what do you think/do to feel better? It's a learning process. Don't expect to be positive all the time, especially during the first few years. FA is not something I'd wish



Meg with her service dog Jude.



for anyone, but if you have it, know that it does get better. You will eventually laugh when you fall down. Personally, I use humor to cope when I'm feeling down. I'd rather laugh at my pain than feel bad for myself, and that's something that took me years to recognize.

What is one way FA has positively affected your life? The most positive thing is my service dog, Jude. He is the greatest asset in my arsenal of medical equipment. This living thing is able to exude emotion and care that a hunk of metal cannot. I urge anyone who has FA—especially those recently diagnosed—to look into getting a service dog.

What is a favorite motivational quote of yours? "Those who mind don't matter, and those who matter don't mind." —Dr. Seuss

What is the best advice you could give to someone newly diagnosed with FA? You can do it. We're all rooting for you.

What is the first thing you want to do when a cure/treatment is found for FA? Buy a pair of Christian Louboutin Pigalles, 100mm (red bottom heels).

"I have FA but FA doesn't have me." What does this statement mean to you? FA is a part of my life, there's no way around that, but it is not the entirety of myself. I'm a writer. I can figure out how to cook just about anything. I can tell you weird facts about dogs for days on end. I love public speaking. I will make you laugh. FA has a hold on me, but I am not just a disease. ●



'Hope for ToMORROW' Fundraiser Bringing It Home for FARA

By Ann Musheno

In June 2015, 9-year-old Anna Morrow was diagnosed with FA. The young girl's community of family and friends immediately rallied together, forming "Hope for toMORROW" in support of Anna and her parents, Kristin and Ben. Just five months after Anna's diagnosis, more than 20 people traveled from Baltimore to participate in rideATAXIA Philly and raised over \$30,000! Propelled by this success, Kristin, Ben and their supporters hosted a dinner fundraiser at their church in Baltimore, MD. The March 2016 "Bringing It Home" event featured live music, raffles, food, beer and wine.

The event attracted 400 supporters – far more than the 250 people they predicted would attend. When their guest list went over their capacity three weeks before the event, friend and event planner Kate Taylor arranged for a large tent to be placed on the patio. Kristin says "We were honored and humbled to have not only friends and family, but friends of friends and community members join in the festivities."

Although a very large donation generously covered the event expenses, the Morrows opted to keep expenses down to give the remainder of the donation directly to FARA. They kept expenses low thanks to the church donating the venue, local businesses providing donations and discounts on beer and wine, individuals



Hope for toMORROW volunteers ready to welcome and assist guests.

donating time and materials for invitations and event signage, and an army of volunteers selling tickets, tending bar, baking cookies and sewing linens.

The most significant cost-saving measure entailed volunteers, led by Anna's grandmothers, preparing, cooking and serving all of the food at the event. Feeding a crowd this size is no small task – they had over 1,400 meatballs, 50 pounds of shrimp salad, 200 baked ham and cheese sandwiches, five gallons of crab soup, and 90 pounds of pork BBQ, among other delicacies.

All of the hard work from Ben, Kristin and their wonderful volunteers made the event a huge success – with over \$38,000 raised for FARA. Kristin says, "The outpouring of love and support provided to our family has been unbelievable. We have actually shed more tears of joy than sadness since Anna's diagnosis due to all of the generosity that has been given to us."

The Morrows are continuing to fundraise for FARA this year at rideATAXIA Philly and plan to make "Bringing it Home" an annual event, saying that "We are honored to be a part of this FAfamily. As a way to say thank you for all that has been done thus far, we vow to do our part to continue in [FARA's] amazing efforts."

"Bringing it Home" is one of over 80 grassroots fundraisers held by families across the country each year. For the past two years, Grassroots fundraisers have exceeded the \$1 million annual fundraising goal set by Mission 1 Million. This program is expected to once again reach \$1 million in 2016. To learn more about grassroots events occurring across the country or how you can host your own grassroots fundraiser, visit curefa.org/grassroots. Together we will cure FA! ●



FARA Spokesperson, Kyle Bryant shares his thanks with the guests at the Bringing It Home event.

Grassroots Fundraising



Professional Tennis Star John Isner and volunteers at Ace for a Cure gather on the Avila Tennis Courts. The December 6 event netted over \$90,000 to help fund research to treat and cure FA. Photo courtesy of Sports Writers of America. (Tampa, FL)



Friends and family celebrate Matt Dilorio's 35th Birthday with a fundraising comedy night to benefit FARA. (Rehoboth, MA)



Riders assembled for a photo at rideATAXIA Dallas on April 9th. (Denton, TX)



Christin Haun poses with a flyer advertising her Charitea Event at McAlister's Deli on January 5. (Broken Arrow, OK)



Family gathered for food and cheer at the Flatbread Pizza Night organized by Erin O'Neil. (Bedford, MA)



Margaret Ferrarone, Marilyn Downing, Jen Farmer, Felicia DeRosa and Mary Caruso joined Team FARA to compete in the Cooper River Bridge Run on April 2. (Charleston, SC)

Grassroots Fundraising



Trudy Cooper (second from left) and guests sample delicacies at "A Glimpse of On Swann." Trudy, along with her husband John, Chris and Michelle Ponte, and Chris Arreola graciously hosted this preview of their new restaurant with all proceeds benefiting FARA. (Tampa, FL)



Anna Gordon and the family of Coach Mike Fallon dressed in their Hollywood finest for the Anna's Army Movie Star Masquerade on March 26. (Vienna, WV)



Top fundraisers hold up their certificates at the first ever rideATAxia Atlanta on May 15. (Cumming, GA)



Family gathered at Rare Disease Day at the movies which featured a preview of Upstate Boys, a film featuring Dylan McDonnell (center) who lives with FA. (Saratoga Springs, NY)



Robert Hopkins, Dillon Head, and Alyx Holliday celebrate Family together at FARA Fest on May 7. This Jimmy-Butt themed event features a timed 5k, fun walk/run and food made by Cheesburger in Paradise. The event grossed over \$56,000! (Virginia Beach, VA)



Bikes and trikes took to the road on June 4 for the 7th Annual rideATAxia NorCal. (Winters, CA)

Grassroots Fundraising



The force is strong with the FA community. Daniel Sims, Kyle Bryant, Lealan Sims, Gavin Lambert, and Tye and Emily Baudin hang out with Star Wars characters on April 2 at FARA Family Fun Day on the Farm at Buti T Ful Farms. (Plant City, FL)



FARA President Ron Bartek, RJ Mercure and Family celebrate Rare Disease Day at a fundraiser at Glory Days Grille on February 29. (Stone Ridge, VA)



Matt Fitzgerald at the Icebreaker Indoor Marathon on January 31 – the first of five marathons he is running this year as part of Team FARA! (Milwaukee, WI)



Family spent time together at the Mother's Day 5K Race for Christina that raised over \$18,000! (Newtown, PA)



Tom Bradley, Kevin Lambert, and Sam Bridgman take a break from a fun afternoon of clay shooting at Pull for a Cure on February 13. (Land O Lakes, FL)



Team FARA was well represented at this year's Tour de Palm Springs bike ride. (Palm Springs, CA)

Grassroots Fundraising



The annual Stoneham Open Golf Tournament was held to benefit FARA for a second year (Stoneham, MA).



A Friedrich's Ataxia Symposium was held on January 21 at UCLA. Over 70 people attended the event to learn more about research progress. (Los Angeles, CA)



Walkers stop for a selfie on the boardwalk at the NJ Seaside Stride on May 21. The annual event features a walk along the boardwalk, lunch catered by Carrabba's, and a huge basket raffle. (Seaside Park, NJ)



Taking a break between rounds, participants at the 26th Annual Double Charity Tennis Tournament show their support for FARA. (Oldsmar, FL)

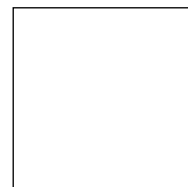


Supporters of Race for Matt & Grace work it out to raise funds at a Zumbathon on March 3. (Johnston, RI)



The players on the #Anna's Army CureFA baseball team dedicated their season to Anna Gordon and raising awareness for FA. They took home the championship for the season! (Parkersburg, WV)

FARA
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SUMMER 2016

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