SUMMER 2014

the Advocate

Your connection to the Friedreich's Ataxia Research Alliance

This Issue:

FARA Friedreich's Ataxia Research Alliance

www.cureFA.org

A Dream Come True: Penn Medicine/CHOP Friedreich's Ataxia Center of Excellence

By Ron Bartek

Dear Friends,

Earlier this year, a longstanding FA Family dream came true. In March, the tremendous generosity of the Hamilton and Finneran families and the continued commitment and collaboration of the University of Pennsylvania and the Children's Hospital of Philadelphia (CHOP) enabled FARA to establish the Penn Medicine/CHOP Friedreich's Ataxia Center of Excellence. The Center was launched with a gift of $3.25 million presented by FARA in partnership with the Hamilton and Finneran families.

As many of you know, it was not by accident that Penn Medicine and CHOP were selected as the site for FARA's Center of Excellence. The wonderful scientists there have led the way in FA basic and clinical research since FARA's inception. FARA has always dreamed of being able to provide them additional support and collaborators so they could accomplish even more for FA patients. The Center of Excellence will do just that.

The Center's co-directors are David Lynch, MD, PhD, and Robert Wilson, MD, PhD. Both are FARA scientific advisors and are well known and deeply appreciated throughout the FA community. Dr. Lynch sees more FA patients each year than any other clinician in the world. He led the multi-site program that developed the FA rating scales in use in FA clinical trials -- an effort that has grown into the Collaborative Clinical Research Network (CCRN) in FA with clinics in the United States, Canada, Australia and, soon, in Brazil. Dr. Lynch serves as the principal investigator of the CCRN and leads its vital effort to build the FA natural history database so essential to understanding FA and so important to our pharmaceutical partners designing FA clinical trials, many of which Dr. Lynch continues to conduct at CHOP.

Dr. Wilson was one of FARA's founding board members and, as Chair of the Scientific Review Committee, provided direction for FARA's research grant program. He also served as scientific chair of FARA's first three International FA Scientific Conferences and prepared the NIH grant applications for each of them. Dr. Wilson is an extraordinary FA scientist. He worked with the NIH to conduct a high-throughput screen of 340,000 compounds seeking candidates for FA therapeutics and is working with medicinal chemists to exploit the results. He is also exploring other avenues of approach in his drug discovery work to develop potential FA treatments that could then be tested in the CCRN.

Important additional collaborators the Center brings to the Penn/CHOP FA team include Kimberly Lin, MD, and Ian Blair, PhD. Dr. Lin provides the Center with her much needed cardiology expertise. She is board certified in pediatric cardiology, pediatrics and internal medicine. Dr. Blair is renowned in the field of mass spectrometry and is working closely with Dr. Lynch to identify, in the cells of FA patients, biochemical changes that can be used to measure the impact of potential therapies. The Center is also looking to provide this fine FA team with an additional lab scientist and a clinical neurologist so more potential therapies can be developed and more clinical trials to test them can be conducted. Finally, no explanation of the great value of this new Center would be complete without acknowledging the remarkable contributions made to it by our dear colleague, Jen Farmer. In addition to being FARA's Executive Director, Jen serves as the overall coordinator of the CCRN, helped shape the vision and organization of the Center and plays a major role in its operation.

You can understand why your FARA family is so appreciative of the generosity of the Finneran and Hamilton families and so excited about the establishment of the Penn Medicine/CHOP Friedreich's Ataxia Center of Excellence. The Center is poised to increase and accelerate the FA research this fantastic team will be conducting itself and to help FARA encourage, shape and support promising FA research around the world.

Warm regards to you and your families,

Ron

Research of Treatment Strategies Continues to Expand

By Jennifer Farmer

At FARA's June Board of Directors and Scientific Advisory Board meeting, I opened with the following comments:

Many of you have been at this since the very beginning (since the establishment of FARA), and no matter when or why you have committed your support, I know that it has not gotten easier with time. It is sometimes hard to see progress and the small victories when we are still working toward a first approved treatment, but I see it, hear it and witness it every day in how the work of our organization has changed and evolved. I believe that those first treatment milestones are in sight and the momentum and interest is reaching a new place which also allows us to really envision a path toward stopping the devastation of FA.

Since writing my last Advocate article and providing an update on the treatment pipeline there have been some significant changes – many due to growing interest from new industry partners. We could spend a whole article discussing why there is new and growing interest from all types (large and small pharma, biotech and venture capital) of industry, but one key reason is the work that the FA patient, medical and scientific communities continue to do in building a solid infrastructure to support advancement of drug development through clinical trials. When we share with companies the data and performance of our patient registry, natural history and outcome measure studies, or experience recruiting subjects for clinical trials – they listen and engage. This is not possible without your support and action. Your willingness to register, show up for natural history visits, roll-up your sleeve for another blood draw, volunteer for a clinical trial, and raise funds and awareness has created the foundation for companies to enter into FA clinical research and see the opportunity for success.

Gene Therapy Update

In the past five months there have been many exciting developments in approaches and interest in advancing gene therapy strategies for treatment of FA. Three new companies have been established and made public announcements of financing and critical licensing agreements. In addition, Dr. Hélène Puccio and her colleagues presented significant results in the April issue of *Nature Medicine* demonstrating that gene-replacement therapy prevented and corrected cardiac damage in a FA mouse model using an adeno-associated virus to deliver the frataxin gene.

• AAVLife has been founded with a clear commitment to the rapid development of a gene-therapy program focused on treating the life-threatening cardiac condition suffered by FA patients. FARA has been collaborating closely with the founders of AAVLife since the middle of 2013 and appreciate their dedication to treating the cardiac disease which causes premature mortality in FA. AAVLife has announced successful series A financing, licensing of rAAVrh10 vector from RegenX, and is presently working on the large animal studies to translate Dr. Puccio's mouse studies to clinical studies in individuals with FA. ***www.aavlife.com***

• Agilis Biotherapeutics made public announcements at the end of 2013 with a closing of series A financing, collaboration with Intrexon Corporation, a leader in synthetic biology, and a focus on DNA-based therapeutics for Friedreich's ataxia. FARA has been collaborating with Agilis as they developed and launched their drug discovery efforts in FA. ***www.agilisbio.com***

• Voyager Therapeutics, a new gene therapy company focused on FA and a few other neurodegenerative diseases, was launched earlier this year. Voyager is backed by Third Rock Ventures and just recently announced licensing terms in which REGENX has granted Voyager a non-exclusive worldwide license, as well as sublicensing rights, to REGENX's NAV vectors for the treatment of ALS, FA and HD. FARA leadership have had the opportunity to participate in discussions with many of Voyager's founders since early 2013 and appreciate their interest and commitment to developing a gene therapy approach for the neurological symptoms of FA. ***www.voyagertherapeutics.com***

HDAC Inhibitor Update

Repligen announced earlier this year that BioMarin has acquired the HDAC inhibitor program. This is very encouraging since BioMarin is a leader in developing therapies for rare, inherited conditions and Repligen was no longer in a position to advance the program. BioMarin is continuing the work that Repligen started to identify a new lead candidate with improved characteristics over RG2833 (the previous candidate that went through Phase 1 studies).

Nicotinamide (vitamin B3) is also known to function as an HDAC inhibitor. Dr. Richard Festenstein and colleagues reported results recently of an open-label exploratory clinical trial that evaluated dose, safety, and biochemical and clinical outcomes. At higher doses (>3.5 grams per day of nicotinamide) there were issues with tolerability (e.g.,nausea) and safety (elevated liver enzymes), but at lower doses the drug was well tolerated. An increase in frataxin protein was observed with single oral dosing in a Phase 1 portion of the study, and a sustained increase in frataxin gene expression and protein with daily dosing over eight weeks was observed in the Phase 2 portion of the study. No clinical benefit or improvement was observed. The full results have been published and can be accessed at:
http://www.thelancet.com/journals/lancet/article/PIIS0140-6736(14)60382-2/abstract

The study authors emphasize that these results are preliminary and not adequate to support the use of nicotinamide as a treatment for FA, but do suggest that further studies should be considered.

Pipeline Additions

New lead candidates have been added to the pipeline, with clinical trials either ongoing or anticipated for later this year.

Early this year Reata Pharmaceuticals contacted FARA about RTA 408, a drug they believe could have benefit in FA. RTA 408 is one of a class of drugs that Reata has developed to target the activation of a transcriptional factor Nrf2, a therapeutic target in FA. Several researchers (e.g., Dr. Gino Cortopassi of UC Davis and Dr. Pierre Rustin's group in France) have shown that Nrf2 is paradoxically decreased in cells from FA patients. Nrf2 should be activated when there is mitochondrial dysfunction and oxidative stress, but data has shown that it is not active in FA. It could be that there is a "vicious cycle" causing Nrf2 to be lower in FA; if one could find a way to increase Nrf2 , that cycle may be broken. The hypothesis being tested is that increasing Nrf2 could improve mitochondrial function by reducing oxidative stress; these compounds have been shown to have protection against diseases involving inflammation (there is some limited data and evidence that FA has some inflammatory aspects). Also, Reata has shown in numerous preclinical studies that their compounds increase the number of mitochondria, improve mitochondrial efficiency, and boost energy production by mitochondria, all of which would potentially benefit FA patients. Working with FARA and several FA Investigators in our Clinical Research Network, Reata is moving very quickly and planning for a Phase 2 clinical trial of RTA 408 to begin during the second half of 2014. They are in the process of completing study design and regulatory filings, and we look forward to sharing more details with the FA community later this year.

Acetyl-L-Carnitine or ALCAR is a naturally occurring compound made in the body and available as a supplement. L-carnitine transports fatty acids to the mitochondria for breakdown and is also known to have an important role in glucose metabolism. ALCAR has been studied as a supplement in many diseases, including cardiovascular disease, diabetes, neurodegenerative disorder and Friedreich's ataxia. Early studies showed that the drug was well tolerated and that there were some modest objective benefits.
http://www.ncbi.nlm.nih.gov/pubmed/?term=15480852
http://www.ncbi.nlm.nih.gov/pubmed/?term=10803803

No further studies have been conducted until now. Dr. Theresa Zesiewicz at the University of South Florida has initiated a long term open label study of ALCAR examining cardiac and neurological measures in adults with FA.
http://www.clinicaltrials.gov/ct2/show/NCT01921868

This study is open for enrollment. For more information and coordinator contact info, see the study ad on page 10. Proceeds from the FARA Energy Ball are funding this study.

Ongoing Trials

As new pipeline candidates enter the pipeline, we also anxiously await results of several trials ongoing or recently completed:

- ***Phase 2, Safety and Efficacy of EPI-743 in Patients with FA*** – Double-blind placebo control portion of the study completed, open label extension is ongoing, data being analyzed, results anticipated by early fall. Also, there is a small open label study for FA patients with point mutations ongoing at the University of South Florida.

- ***Open-label pilot study - Interferon gamma-1b in children with FA*** – Study completed late spring, data being analyzed, results anticipated by early fall.

- ***Phase 1 Safety and Pharmacology Study of VP20629/OX1 in Adults with FA*** – Enrollment ongoing, anticipated completion later in 2014.

FARA's website has a fully updated pipeline image with detailed updates on each program at *www.curefa.org/pipeline.html.*

In closing, what has not changed and will not change is our focus, urgency, effort and commitment. ●

Focus On A Cure

By Ed Ramsey, Chairman,
FARA Board of Directors

I am forever grateful for all of the wonderful people in this world who support those who live with disability. The emotional and physical support makes life so much easier and pleasant. You provide us with strength, courage, resolve, and hope. The FARA family is fantastic! What more could a person ask for???

Simply put – not to have this disability. I have had countless hours to dream this dream and wish this wish, not only for myself but for all people who live with disability – including parents, friends and caregivers.

How wonderful to be able to stand toe to toe and share a handshake or a hug …To be able to run with a kite tethered to a string as it soars above, ride a bike, or go for a hike. We would love to play soccer, or frolic with a dog, or just dance. We would be proud to stand to say the pledge of allegiance …To walk unencumbered down the aisle or guide a baby with a smile.

We have these dreams and hopes for ourselves and for all of our loved ones with a disability. Comfort and care are essential but will not eradicate this disease. Only cutting-edge research, clinical trials and investment in a cure will take away this disability.

What is Friedreich'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 Friedreich's ataxia and will lead to treatments that eliminate its symptoms.

FARA's Research Grant Program

By Bronya Keats, MD

New Research

Our congratulations to University of Minnesota researchers, Drs. Pierre-Gilles Henry and Christophe Lenglet, and to Dr. Mark Baker at Newcastle University in the United Kingdom. All three were recently awarded grants for projects submitted in response to a request for proposals (RFP) issued jointly by FARA, Ataxia UK and GoFAR. These projects focus on the development of biomarkers by non-invasive approaches for evaluating the molecular and pathological features of affected neurons in FRDA patients. Once again, we see the importance of the close working relationship we have with our FRDA advocacy group partners in accelerating research to accomplish our mission.

Early and longitudinal assessment of neurodegeneration in the brain and spinal cord in Friedreich's ataxia

Co-Principal Investigators: Pierre-Gilles Henry and Christophe Lenglet (University of Minnesota)

The overall goal of this project is to demonstrate the feasibility of identifying and tracking structural and neurochemical changes in the spinal cord and brain of patients with FRDA, especially at an early stage of the disease. Drs. Henry and Lenglet will use both magnetic resonance spectroscopy and diffusion magnetic resonance imaging, which provide complementary information on neurochemical and microstructural changes. They will continue their follow-up of FRDA patients who have been scanned at the University of Minnesota over the past year in order to assess the year-to-year evolution of the observed microstructural and neurochemical alterations. They will also determine if these alterations can be detected earlier in the course of the disease. These studies may result in novel and unique tools for the quantitative assessment of disease progression and treatment efficacy in prospective therapeutic trials of FRDA. *(see study ad on page 7).*

Beta-band EMG-EMG coherence: a novel, painless and simple screening test for the onset of corticospinal tract disease/dorsal root ganglionopathy in Friedreich's ataxia

Principal Investigator: Mark Baker (Newcastle University, United Kingdom)

This project aims to develop a painless, inexpensive and easy-to-use portable electro-diagnostic screening test of subclinical dorsal root ganglionopathy, dorsal column degeneration and corticospinal tract degeneration in FRDA. The subjects will undergo a number of clinical, laboratory, and neurophysiological evaluations, including analysis of beta band intermuscular coherence (BIMC), which has been extensively studied by Dr. Baker for other neurological diseases. BIMC is calculated based on data acquired from surface electromyogram (EMG) recordings on two target muscles when the subject is asked to intermittently perform a single act (e.g. dorsiflex the ankle). It is a sensitive measure that distinguishes between patients and controls, and has been shown to disappear in diseases affecting the corticospinal tracts and in dorsal root ganglionopathy. The measurement of BIMC may be a simple means by which FRDA patients can be screened early, painlessly and at frequent intervals; thus it has the potential to be an informative biomarker for direct measurement of neuronal degeneration.

We also congratulate the recipient of the 2014 FARA Keith Michael Andrus Memorial Award for Cardiac Research, Dr. Veronique Monnier from the Université Paris Diderot in France.

Keith Michael Andrus Memorial Award for Cardiac Research: Identification of therapeutic compounds on a cardiac Drosophila model of Friedreich's ataxia

Principal Investigator: Veronique Monnier (Université Paris Diderot, France)

Dr. Monnier and her team have developed a Drosophila cardiac model of Friedreich's ataxia that recapitulates the defects of cardiac function observed in patients and FRDA mouse models, in particular heart dilatation and impaired systolic function. In a recent publication describing this FRDA fly model, they showed that methylene blue, a clinically approved drug, was able to prevent the heart dysfunction (Tricoire H, Palandri A, Bourdais A, Camadro JM, Monnier V. Human Molecular Genetics 23:968-979, 2014). The major goal of this project is to use the Drosophila cardiac model to identify new therapeutic compounds through a medium-scale unbiased drug screening of 1280 compounds. Cardiac imaging will be performed in vivo to select compounds that improve the heart function of the fly. This drug screen has great potential for identifying novel compounds that are likely to lead to therapeutic interventions for FRDA.

Ongoing Research

FARA is presently supporting 24 research projects all of which underwent rigorous peer-review and were rated as outstanding, both scientifically and with regard to directly addressing FARA's strategic research initiatives. Ongoing funded research projects include: *(1) Dr. Ed Grabczyk's work (using autopsy neuron and heart samples provided by Dr. Arnulf Koeppen) to determine if progressive GAA repeat expansion caused by mismatch repair proteins is a major factor in FRDA pathology; (2) The development by Professor Javier Diaz-Nido and his collaborator, Professor Ernest Giralt, of novel DNA nanocarriers that cross the blood-brain barrier as a gene delivery system to treat FRDA; (3) Dr. Mark Pook's experiments to determine if a group of compounds known as HMTase inhibitors can increase frataxin; and (4) A study by Dr. Paola Giunti and her co-investigator, Professor Andrey Abramov, using FRDA mouse models to characterize mitochondrial pathophysiology and to explore the pharmacological effects of two types of compounds that are already in the FRDA treatment pipeline.*

Grant application instructions, including deadlines for LOIs and full applications are available at ***http://www.curefa.org/grant.html***

Titles and summaries of most of the projects presently funded by FARA are available at: ***www.curefa.org/RPMP/public/pggrantlist.aspx*** and the grants awarded by year can be accessed at: ***www.curefa.org/grants-awarded.html***

New Publications

FARA-funded research continues to result in valuable contributions to the peer-reviewed literature. Examples of recent publications on FRDA are:

- ***The exciting research demonstrating that AAV gene therapy prevents and reverses cardiomyopathy in the FRDA MCK mouse. (Perdomini M, Belbellaa B, Monassier L, Reutenauer L, Messaddeq N, Cartier N, Crystal RG, Aubourg P, Puccio H. Nature Medicine 20:542-547, 2014)***

- ***Results of an open label pilot study of nicotinamide, an HDAC inhibitor showing that frataxin levels increased in blood samples from participants but the neurological measures did not show significant improvement. While these results are of interest, further studies of longer duration are needed to determine if nicotinamide is safe and effective as a long-term treatment for FRDA. (Libri et al. Lancet, published online 1 May 2014)***

- ***The generation and characterization of novel FRDA fibroblast and neural stem cell models from YG8R mice, which are valuable tools for preclinical testing of potentially therapeutic compounds. (Sandi C, Sandi M, Jassal H, Ezzatizadeh V, Anjomani-Virmouni S, Al-Mahdawi S, Pook MA. PLoS ONE 9:e89488, Feb 2014)***

- ***A clinical study of glucose metabolism in FRDA patients suggesting that increasing age, longer GAA repeat length and higher body mass index (BMI) may be some of the factors that elevate risk of developing insulin resistance, glucose intolerance, and eventually diabetes. (Greeley NR, Regner S, Willi S, Lynch DR. Journal of the Neurological Sciences, published online 24 April 2014)***

A comprehensive list of FARA-funded and other research publications on FRDA is provided at: ***http://www.curefa.org/scientific-news.html***

Recruting Now!

PATIENTS with EARLY SYMPTOMS of FRIEDREICH'S ATAXIA

Age 10 and above needed for an MRI study to evaluate the chemistry and connectivity of the brain and spinal cord in Friedreich's Ataxia at the Center for Magnetic Resonance Research at University of Minnesota

You will lie in the scanner for ~1.5 hour while listening to the music of your choice. Reimbursement for travel expenses is available and you will be compensated for your time.

Please note that we cannot scan you if you have Harrington rods, and we cannot scan people with diabetes at this time.

If you are interested or have questions,
please call Diane Hutter at (612) 625-2350 or
email ***hutte019@umn.edu.***

The FARA Advocate is brought to you by:
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THE FARA ENERGY BALL

Sept. 4, 5, and 6, 2014 in Tampa, Florida.

To benefit FARA and
USF Ataxia Research Center

Book your tickets online now.
www.curefa.org/EnergyBall
Donations in lieu of attendance are gratefully accepted.

September 4
Understanding Energy for a Cure Scientific Symposium

September 5
Patron Party

September 6
The FARA Energy Ball with live performance by the Spazmatics

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Roy's

Team Kendall

By Kyle Bryant

Just because the journey is a challenging one doesn't mean there aren't things to be celebrated along the way.

Team Kendall rolled up to the starting line at Ride Ataxia Dallas 25 people strong, looking determined to ride and have a great time. While they came to get a job done—ride six or 25 or 50 miles in support of Kendall and all people living with FA—they did it with fuzzy whirly-doo's on their helmets and noisemakers in the crowd. Just because the journey is a challenging one doesn't mean there aren't things to be celebrated along the way.

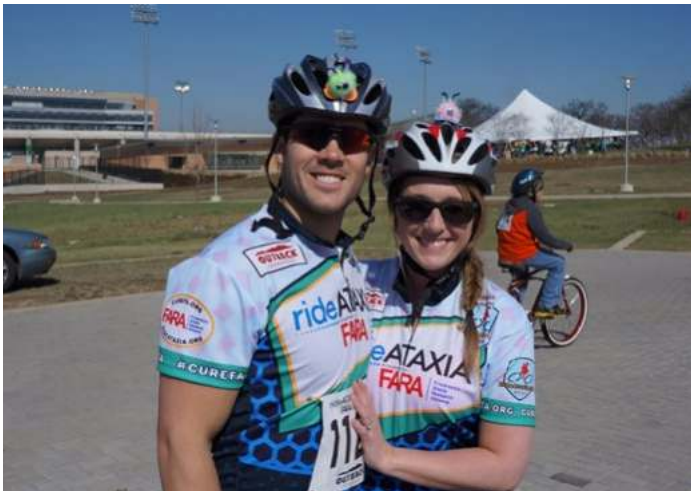
Kendall Harvey's story is similar to the story of others in the FA community; she was an active kid, and FA appeared out of the blue with no warning. Kendall has a very strong circle of support from family and friends, so her diagnosis not only affected her but her entire network. She knew she needed to find a way to tell her loved ones about FA and her diagnosis without them going to Wikipedia and reading all the terrible facts.

The ride was a way to introduce her friends and family to FA in a positive way. Kendall created ***teamkendall.org*** as a way to tell her story and share it on Facebook so that people could understand the situation from her perspective.

"I wanted to say 'The bad news is I have this disease, the good news is there is something we can do about it and this is what we are going to do,'" Kendall says. Kendall and her immediate family – including her husband Kyle and her parents – began by keeping their closest friends and family in constant communication during the initial diagnosis and learning stages. "It is surprising how many people had never heard of it," she says, at the same time recognizing that she herself had never heard of FA before her diagnosis. After they got their feet under them and made the decision to take action with Ride Ataxia as their platform, Kendall began to start spreading the word more widely on Facebook to many friends that she had not seen for years.

"My team and I focused on sharing information about FA," she says."Once we educated our friends and family about the disease and all of the amazing progress that is happening to cure FA, donations began pouring in." Donations totaled $38,000 and Team Kendall rallied around their fearlessly fun leader in great numbers. On the day of the ride, Team Kendall rode strong and celebrated their efforts, knowing that they are able to triumph over FA by coming together and taking action. ●

Team Kendall



Kendall with Husband Kyle Harvey

Join us at one or more of these locations this year:

RA Chicago	July 20
RA Seattle	August 3
RA Philadelphia	October 12
RA Orlando	November 2

Get all the details at www.rideataxia.org.

Ataxian Athlete Initiative Announces 2014 Recipients

By Kyle Bryant

FARA, in partnership with The FA Project, Catrike, The Melting Pot, and The Texas Irish Foundation, is pleased to announce the 2014 Ataxian Athlete Initiative (AAI) grant recipients: Liam Dougherty of Philadelphia, PA, Carl Estabrook of Rockport, MA, Abby Yingling and Chase Yingling of Lemoyne, PA, Amanda Hernandez of Graham, TX, and Mary Fuchs of Sun Lakes, AZ. The AAI provides adaptive cycling equipment to people with all types of ataxia who have demonstrated the desire to stay active and healthy despite their disabilities.

AAI grants are administered through a competitive application process. Applicants submit a short summary of their experience with ataxia and their efforts to stay active. Individuals then select 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.

"I have discovered how much control I have over my own life. My future is not out of my hands. I am able to control myself in ways that are more important than coordination and fine motor movements. I am in control of what I learn, what I do, and the kind of person I will continue to grow into. Before anything else, FA has taught me to focus on the essential parts of who I am, and look at my future through that lens. Reevaluating personal priorities has shown me that it is crucial that I persevere despite the friction I sometimes feel from my disorder." –Liam Dougherty, 2014 AAI Grant Recipient.

Including the 2014 grant awards, the AAI has provided equipment for 27 individuals since 2009. Additionally, Ride Ataxia works with the Texas Irish Foundation to provide equipment specifically in the North Texas area.

Please see ***rideataxia.org/aai.php*** for more information about the AAI program. The next application cycle begins in Spring 2015.

Recruting Now!

USF Ataxia Research Center

The USF Ataxia Research Center is pleased to announce a new research study for Friedreich's ataxia patients ("An open-label study of the effects of Acetyl-L-Carnitine on cardiovascular outcomes in Friedreich's Ataxia."). This study will be a 26-month trial where all subjects will receive Acetyl-L-Carnitine (ALCAR). There are 11 visits to the research clinic at the University of South Florida Morsani Center for Advanced Health Care. Study related procedures and medication will be provided by the sponsor.

The sponsor of the trial is USF's Department of Neurology and the study's lead investigator is Theresa Zesiewicz, MD, of the University of South Florida - Tampa, FL.
The overall goal of the trial is to determine changes in heart function. We will also look at changes in neurological and neuromuscular function.

Patients with Friedreich's ataxia who are interested in participating in the study at the Florida site should contact the USF Ataxia Research Center at 813-974-5909. To be eligible for participation, genetic confirmation of Friedreich's Ataxia is required. For more information on this research study, please click on the following link: ***www.clinicaltrials.gov/ct2/show/NCT01921868***

Acetyl-L-Carnitine (ALCAR) is a derivative of carnitine, a water soluble molecule of importance in lipid metabolism. Studies have shown carnitine derivatives to have some effect on cardiovascular disease as well as possibly improving coordination. This trial will measure changes in cardiac function through a series of electrocardiograms and echocardiography.

IRB Protocol #: Pro00008499
PI: Theresa Zesiewicz, MD

Dr. Christophe Lenglet

By David Woods, PhD

"What's the point of doing research if you can't share or communicate what you've learned?"

Armed with master's degrees in applied mathematics, computer science and engineering from colleges in his native France, Christophe Lenglet, 34, went on to secure a doctorate in biomedical imaging and neuroscience at Sophia Antipolis, located in a technology park situated between Antibes and Nice.

The park is home to companies in computing, electronics, pharmacology and biotechnology as well as being a locus for higher education. But its thrust is not only in science but also in bringing together people from different intellectual horizons and fostering interaction, networking and cross-fertilization of ideas.

Clearly this has influenced Christophe, who has authored or co-authored some 25 articles, more than 30 conference papers, and some 50 abstracts. "What's the point in doing research" he says, "if you can't share or communicate what you've learned?"

From the sunny Côte d'Azur, he made his way to New Jersey for a two-year stint as a research scientist with Siemens Corporation in Princeton, NJ. From there, Christophe moved to the University of Minnesota initially as a research associate in the Department of Electrical and Computer Engineering, and later joined the medical school's faculty as an assistant professor in the Department of Radiology. He jokes that the trajectory of his places of work seems to be one of ever lower temperatures and that perhaps his next location might be Alaska.

On the other hand, he believes that the trail towards a cure for FA is actually getting warmer. In fact, along with FARA, Ataxia UK and its counterpart in Italy, GoFAR, the goal of the three organizations is achieving effective treatments and a cure in the near term.

Christophe and his colleagues at the University of Minnesota have received numerous grants including FARA's Kyle Bryant Translational Research award, and his current one from FARA on early and longitudinal assessment of neurodegeneration in the brain and spinal cord in FA, which is also the subject of a paper that Christophe's team is preparing for publication.

Kyle Bryant is the founder and director of Ride Ataxia, a biking initiative to generate awareness and funds for Friedreich's ataxia research. Christophe will join Kyle for the first time at Ride Ataxia Philly in October. Working with Kyle and other FA patients, he says, provides him and other researchers with a personal connection that adds a human aspect to research.

Christophe Lenglet

An avid triathlon enthusiast, Christophe has completed several Ironman races and will compete in another one scheduled for September in Oklahoma as part of Team FARA. He's also an enthusiast of TaeKwonDo, the Korean martial art that combines combat and self defense techniques with sport and exercise. In fact, he believes that sport is a good way to make friends and settle in to a new job—sports training is how he met his American wife Katherine, who is studying to be a physical therapist.

What's next for Dr. Lenglet? He intends to pursue his research interests in mathematical and computational strategies in neuroimaging; translational neuroimaging for neurodegenerative disorders; and human brain anatomical and functional conductivity.

Oh, and he'd like to travel more … perhaps not to Alaska, but somewhere warmer, like the next Ironman competition in Hawaii.

Bon Voyage, Christophe.

A Day In The Life: Living With FA

By Christina Logan

"Giving up is simply not an option" is a quote I now live by. Even though I am an individual living with Friedreich's ataxia, I do not let that get in the way of my life. My name is Christina Logan, and I am an independent, 23-year-old woman working full-time and living on my own in the city of Philadelphia. I was diagnosed with FA on April 20, 2012. Each day, I have to embrace living in a major city despite facing my challenges living with FA. Yes, it can be difficult getting around and doing things on my own, but I couldn't do it without the strength and willpower of my family, friends, and most importantly, myself.

I moved to the city over a year ago from Bucks County in the suburbs of Philadelphia because I wanted to prove to myself that I could live on my own and survive the real world! I graduated Johnson & Wales University with a Bachelor's Degree in Hotel and Lodging Management. I am proud to say that I am working at my dream job: Front Office Supervisor and Housekeeping Supervisor at The Westin Philadelphia. These positions require me to be on my feet, which sometimes can be challenging, but I've grown to learn how to pace myself and know my limits. Work is aware of my Friedreich's ataxia and they are very accommodating. So, don't be discouraged about living and chasing your dreams!

Living in a major city can be very accessible and accommodating to your needs. Since I don't drive, I either walk or rely on public transportation. Philadelphia is a very walkable city. But if I am feeling fatigued, I have access to public transportation, such as the bus or subway which are readily available and easy to hop on and off. Sometimes I have trouble getting on and off the bus, but the handicap seats are right in front. A few times, generous people have assisted me on or off the bus, and it truly made my day! It's the simple acts in life that truly means the most!

If there is anything tough about living in the city, it has to be facing the public. It's hard to face the fact that there are always going to be judgmental people who make rude remarks. I've had a few comments made to me about my walking or "funny feet." At first, it took me a while to not let these people get to me. But then, I overcame this feeling by reminding myself that I am a better, hardworking, strong-minded person who may have a debilitating disease, but I don't let that affect my life. I still get out of bed every morning and embrace life to its fullest!

Christina Logan

I couldn't be living on my own without the encouragement of my family and friends! I know if I ever need help or support, they are there for me every step of the way. For example, it's physically hard for me to shop for food or run errands on my own. I am fortunate enough to have my family come into the city and help me. It may not mean a lot to some people, but to me, it means the world.

So, I am writing to you all to hopefully inspire and encourage you that you can live on your own even though you have Friedreich's ataxia. It does have its hardships, but don't let that disappoint you. I have the love and comfort of my family and friends who know I can do it. I am working full time at the prestigious Westin Philadelphia, and I face the public living with a rare disease. I am also a FARA Ambassador, and I recently participated in a clinical trial at Children's Hospital of Philadelphia (CHOP). I love to explore the city of brotherly love by revisiting the historic sites and trying new restaurants and writing a food blog ***(espressochristina.blogspot.com)*** for fun! If anything, living in a major city has made me a stronger person with the knowledge that anything is possible!

Upcoming Events

July-August 2014
Summer of Eating
Various Restaurants - Novato, CA

July 17 - 19, 2014
FA Woodstock
Flying H Ranch - LaPorte, IN

July 20, 2014
Ride Ataxia Chicago
Channahon, IL

July 27, 2014
Team FARA - Team Patrick Morrissey
Ironman Lake Placid
Lake Placid, NY

August 3, 2014
Ride Ataxia Seattle
Redmond, WA

August 8, 2014
Welsh Bash in the Backyard
Harrisburg, PA

August 17, 2014
Team FARA - Racing Falmouth for FARA
Falmouth and Woods Hole, MA

August 23, 2014
Team FARA - Team Adirondack
Adirondack Hike
Louisville, KY

August 24, 2014
Team FARA - Dave Norman
Louisville Ironman
Louisville, KY

September 4, 2014
USF Health Friedreich's Ataxia
Scientific Symposium
Tampa, FL

September 6, 2014
The FARA Energy Ball
Tampa, FL

September 7, 2014
The 11th Annual Fuzzy Buzzy Golf Tournament
Windham, NH

September 8, 2014
Hole Out for A Cure
Peoria Heights, IL

September 15, 2014
2nd Annual FARA Invitational Golf Classic
Calgary, AB, Canada

September 20, 2014
5th Annual Race for Matt and Grace
Smithfield, RI

September 20, 2014
McDonnell Musical Festival
Queensbury, NY

September 21, 2014
10th Annual Team Donovan Triathlon
Rye, NY

September 21, 2014
Team FARA - Christophe Lenglet
Redman Triathlon
Oklahoma City, OK

September 25, 2014
Century 21 Golf Tournament
Fontana, CA

September 26, 2014
Rocky Mountain Bird & Birdie
Brighton & Commerce City, CO

September 27, 2014
Slim's Journey: FARA 5K Run/2.5k Walk
Warrenton, MO

October 12, 2014
Ride Ataxia Philly
Blue Bell, PA

October 13, 2014
CHOP Friedreich's Ataxia Symposium
King of Prussia, PA

October 18, 2014
5th Annual Swing Away at FA
Dawsonville, GA

October 26, 2014
Spark Hope
Beverly, MA

November 2, 2014
Ride Ataxia Orlando
Clermont, FL

November 2, 2014
Team FARA -
TCS New York City Marathon
New York, NY

November 8, 2014
Team FARA - Team Mustangs for Alison
Savannah Rock N' Roll Marathon
Savannah, GA

November 15, 2014
Stephanie's Hope Holiday Boutique
Santa Clarita, CA

Please check the FARA website (www.curefa.org/events.html) for contact information and a full list of events.

Team FARA at Tour de Palm Springs

By Jamie Young

Beth Bax led her Biking Beauties at Team FARA Tour de Palm Springs for the 3rd year (Palm Springs, CA)

Every February, thousands of cyclists come together in California to ride the Tour de Palm Springs. Team FARA, which began riding in the Tour three years ago to spread awareness and raise funds for FA research, had its best year yet in 2014. Over 70 cyclists, joined by many others in the FA community cheering them on, raised over $40,000 for research. This event is unique among other Team FARA events because it has formed a strong partnership with the local Outback Steakhouse community that rides and fundraises for FARA.

In addition to its fundraising success, Tour de Palm Springs also engages the FA community in Southern California by providing a platform to raise awareness. Southern California resident Beth Bax has been fundraising since participating in Ride Ataxia II in 2008. She now recruits the largest team, Beth's Biking Beauties, to the Team FARA effort in Palm Springs.

"I enjoy this event; because it is close enough to where I live, I can encourage people to drive out and join me," says Beth. "Since my recumbent is hard to ship and dismantle, this is a local event I can get to easily and still raise money for FARA," she says.

The Team FARA program has allowed FARA to reach and engage new regions because it is accessible to everyone. Participants can scale their event to the difficulty and size they desire: from a team of two that participates in a local 5K or a team of 50 that participates in a local bike ride. It also alleviates a lot of work that comes with planning an event since participants take part in one that is already established. Ways to expand existing Team FARA events include becoming an official event sponsor, hosting a FARA booth or including additional team activities with the event, such as a team dinner at a local restaurant.

"My favorite part of Tour de Palm Springs by far is the Team FARA dinner afterwards," says Beth. "You can actually see and talk with everyone. FARA has really become my extended family – I get to catch up with everyone I know and meet people I don't know but love instantaneously because of what they are doing for FARA," says Beth.

FARA is always looking to expand into new areas. This year alone we will have more than 100 Team FARA members signed up to compete in events. We are looking forward to recruiting more participants and spreading FA awareness to new places. To plan a new Team FARA event, please email ***Jamie.young@curefa.org.***

With support from Outback, Team FARA had its biggest year yet at the Tour de Palm Springs (Palm Springs, CA)

Team Bradley enjoys a tasting from World of Beer at the Pull For a Cure fundraiser (Tampa Bay Sporting Clays - Land O Lakes, FL)

Friends of the DeWitt family organized a Euchre Tournament to raise funds for research in honor of Jack (Howell, MI)

Gavin Lambert rocks purple hair as he runs across the finish at the Run or Dye 5K for Team FARA (Tampa, FL)

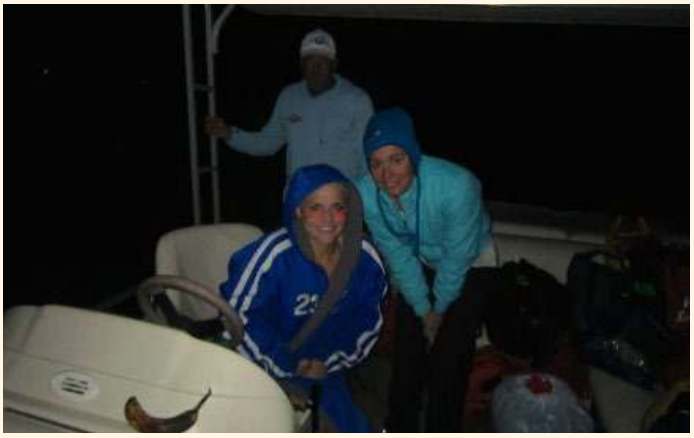
#AnnasArmy attacks ataxia at the Gordon family's Masquerade Ball to Cure FA (Parkersburg, WV)

Amanda Hernandez and Ryan Hernandez get ready to ride at Ride Ataxia Dallas (Denton, TX)

Outback Steakhouse refuels our hungry riders with a great meal at Ride Ataxia Dallas

Grace Haupt and Liam Dougherty helped us present a $3.25M check to establish the Penn Medicine & CHOP Friedreich's Ataxia Center of Excellence (Philadelphia, PA)

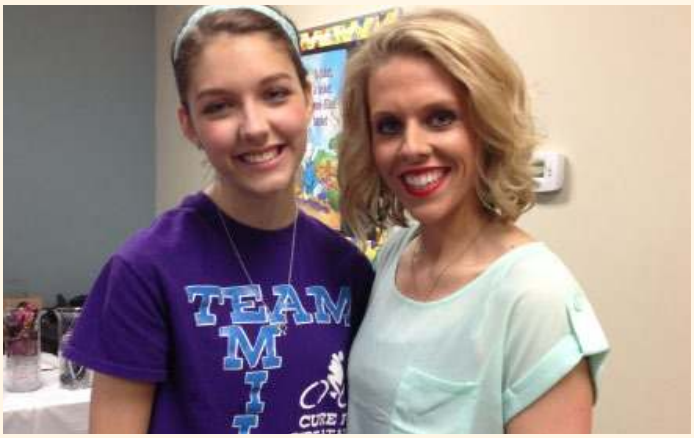
FARA staff members Jen Farmer, Jamie Young, and Felicia DeRosa ran the NYC Half Marathon for Team FARA (New York, NY)



Kristin Jones swam 40+ miles over 4 days in 4 lakes at the AZ SCAR Challenge for Team FARA in honor of her brothers Ryan & Owen (Tempe, AZ)

Dr. Dave Lynch and his team from the Children's Hospital of Philadelphia join Team FARA to raise awareness for rare disease research at the Million Dollar Bike Ride (Philadelphia, PA)

Joined by good friends, Erin O'Neil organized another fun Flatbread Fundraiser (Bedford, MA)



Emily Young picks up new styling tips from her friend Sarah Bielfeldt at the Expressions Hair Extravaganza for Team Emily (Gibson City, IL)

Lauren Williams and John Cernosek helped Robin Nistle's Jazzercise group raise awareness for FA at their halftime performance at a Washington Wizards game (Washington, DC)

The University of Portland's men's basketball team put on another great SamJam wheelchair game in honor of Sam Bridgman (Portland, OR)

Jeff Morgan finished strong at the Boston Marathon for Team FARA in honor of Erin O'Neil (Boston, MA)

Friends from the tri-state area returned to the Jersey shore for the Seaside Stride Luncheon hosted by our NJ FAmilies (Seaside Park, NJ)

It was a beautiful day for golf at the Anytime Fitness Golf Tournament for Team Gavin (Palm Harbor, FL)

Christin Haun and friends sample Broken Arrow's finest at the BA Cure FA Fun Walk and Self Guided Historical Tour for FARA (Broken Arrow, OK)

Aloha from the FARA Fest Lei Day 5K hosted by the Virginia Beach FA Families (Virginia Beach, VA).

The FARA staff joins the fun at the Logan family's Race for Christina Mother's Day 5K (Richboro, PA)

Paul and Brianne Konanz enjoy a beautiful ride at Ride Ataxia NorCal (Davis, CA)

Emily Penn and Natalie Newman out on the course at the LoneStar Benefit Bash golf tournament (Flower Mound, TX)

Join The FARA Patient Registry!

Get notified about new trials and help advance FA research!

www.curefa.org/registry

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SUMMER 2014

the Advocate

Your connection to the Friedreich's Ataxia Research Alliance

www.cureFA.org

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