

From the Executive Director



FARA Expands Research Programs

By Jennifer Farmer

FARA continues to strengthen its focus on advancing research with multiple programs and strategic initiatives that include a robust grant program, the FA patient registry, research conferences and workshops, outreach and awareness to the medical and scientific communities, and partnerships with government, academic and industry organizations to advance leading drug candidates and direct support and involvement in the Collaborative Clinical Research Network in FA.

Grant Program The economic struggle facing many of us and our friends and families at home is also affecting our research. The National Institutes of Health (NIH) is a governmental medical research agency that is the largest source of funding for medical research in the world. The NIH has been a significant partner and contributor to FA research, however over the past two years there has been a significant decrease in NIH resources. Specifically, several years ago NIH funded the top 20-25% of proposals received, but now it funds only the top 10-15%. The net result is that the NIH has fewer resources to devote to FA research and the demand on FARA and other organizations to find other ways to support such scientific research is even more important.

So far in 2010 FARA has received more proposals than ever before and has committed almost \$2 million in grant funding to more than 20 research projects as well as the Collaborative Clinical Research Network in Friedreich's Ataxia. A full listing of grants funded for January - July 2010 is available at www.curefa.org/grants-awarded.html.

Continued on p. 3

In this issue:

- Ride Ataxia
- Grants Update
- FARA Grassroots Fundraising
- President's Message
- Race Across America Article
- Featured Scientist

Accelerating Progress: Meeting on FA Cellular Models and Cell Therapy

By Bronya Keats

Tremendous excitement is spreading throughout the scientific community as a result of the development of what are known as induced pluripotent stem (iPS) cells. These cells have many of the properties of embryonic stem cells, but a huge advantage of iPS cells is that they can be obtained from sources such as skin. Basically, skin cells are reprogrammed into iPS cells by over-expressing certain genes, and these cells are then capable of becoming virtually any cell type in the body. Researchers are already generating iPS cell lines using skin biopsies from people with FA, and rapid progress is being made with differentiating these cells into neuronal and cardiac cells. To facilitate communication, cooperation, and collaboration among researchers working on these major advances, the Cellular Models and Cell Therapy meeting was held in Chicago in March, 2010 as a satellite to the NAF Ataxia Investigators Meeting.

Funding from international patient advocacy groups

Underscoring the need for standardized FA cellular models was the fact that the meeting was co-organized and supported by FARA-Australasia (FARA-A), FARA, Ataxia UK, FASI, & NAF, with representatives from each of these patient advocacy groups participating in the meeting. In addition to the 16 invited FA researchers, we were fortunate to have three internationally renowned stem cell experts:

Continued on p. 8

What is Friedreich's ataxia?

Friedreich's ataxia 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.



Dear friends,

As 2010 shows many signs of being an extremely successful year for FA research (with several promising clinical trials under way and more to come before year's end), I wanted to share with you my view of why FARA is making such progress. The clear message from the many non-profit organizations who come to FARA for advice, from NIH which often refers such organizations to FARA as a model, and from FARA's many partners is this: the characteristic that most differentiates FARA from other organizations and contributes to its success to date is one of its guiding principles—collaboration.

Knowing from the beginning that, Acting alone there is little we can accomplish, we set out to collaborate with every individual and every organization with any interest in supporting FA research. Of course, we began by collaborating with FA patients and families who remain the heart, soul and lifeblood of FARA. FARA set its initial sights on "growing the field"—increasing the number of scientists doing FA research and the resources being devoted to supporting their work, instilling in them the same collaborative spirit, and beginning to draw some interest from the pharmaceutical industry.

Testimony to our success in that arena comes in the fact that, whereas FARA's first international scientific conference in 1999 drew 80 scientists, one other advocacy organization and no drug companies, the conference we are planning for next year should gather about three times as many scientists, six to eight other advocacy organizations with whom FARA is co-funding FA research, and six to eight drug companies with active FA programs. FARA's collaborations have become so fruitful and we are so relentless in pursuing them that we like to say, If you are interested in supporting FA research and FARA is not collaborating with you, it is because we have not yet found you—but we will!

This has been a stellar year for FARA's scientific collaboration. Working with several other advocacy partners around the world, we co-hosted an exciting workshop in March that brought several of the world's leaders in cell models together with FA researchers working hard to develop FA nerve- and heart-cell models. These cell models—one of FARA's strategic initiatives—are being derived from FA patients' skin cells and will be extremely helpful in drug discovery and screening. For an excellent summary of that meeting, please see this issue's article by Dr. Bronya Keats. In June, FARA held its second FA Cardiac Summit, bringing a gifted group of basic and clinical FA researchers together with leading cardiologists to share insights and chart the course for intensified collaborations aimed at improved heart health for FA patients. Jen Farmer's article in this issue has a detailed report on this meeting.

FARA has collaborative relationships with a growing number of pharmaceutical companies involved in advancing promising FA drugs and we are working constantly to find additional

pharmaceutical partners for each new discovery in the FA research pipeline. Some additional such opportunities are developing from Raychel and my involvement this year with the Pharmaceutical Research and Manufacturers of America (PhRMA) and with PhRMA's Committee on Rare and Specialty Diseases. It is clear that pharmaceutical companies are showing more and more interest in rare diseases and this forum is introducing us to a number of companies interested in FARA and FA.

As most of you know, FARA's collaborative nature is not limited to research and drug development. While Jen and I focus largely on collaborations with research and development partners, Felicia, Kyle and all the fantastic FARA fundraising volunteers are working around the clock, with many of you, to build and expand the collaborations essential to increase awareness of FA and the resources needed to support the research. I am thrilled to say, too, that Miss Jamie Young recently joined this very important FARA staff effort as Office Manager of the Exton, PA office. Jamie is a recent graduate of Illinois State University in Public Relations. Jamie's younger sister, Emily, has FA and she shares the passion we all have in our quest for treatments and a cure.

The recent experience with the Race Across America (RAAM), brought to the fore so much of the collaborative spirit of the ever-growing Team FARA. Felicia and Kyle use the RAAM experience to illustrate how the FA community mobilizes around a cause or issue and, with a whatever-it-takes attitude, gets it done. They describe, for example, how the initial call for RAAM volunteers was answered by individuals with diverse backgrounds, talents and expectations who united in their commitment to make a difference for FA families, formed a team far stronger than the sum of its individual parts, and struck out together across the country. That team was joined and cheered all along the way by others in the FA community, right down to the finish line in Annapolis.

That RAAM Team FARA embodied the FARA spirit of collaboration and the spirit of the entire FA Family that rallied around it. The courage, commitment and teamwork of the RAAM Team FARA helps fuel the collaboration, urgency and momentum building in other Team FARAs among FA families, scientists and pharmaceutical partners. That taste of victory at the Annapolis finish line was sweet, and was a foretaste of the victory at the finish line of treatments and a cure.

Warmest regards,
Ron

Acting alone, there is very little any of us can accomplish. Acting together, there is very little we will NOT accomplish.

A few selected grants include:

Project Title: *Multifunctional Radical Quenchers for the Treatment of Friedreich s Ataxia*

Principal Investigator: Dr. Sidney Hecht, Arizona State University

Summary: This project is focused on the development of drugs to treat Friedreich s ataxia. The strategy is based on the belief that the absence of frataxin in Friedreich s ataxia patients leads to progressive mitochondrial dysfunction and associated oxidative stress, resulting in increasing degradation of the mitochondrion (especially the mitochondrial membranes), and in diminished adenosine triphosphate (ATP) production. The objectives of this project involve the synthesis and evaluation of potential drugs that have shown promise by implementation of a strategy for which proof-of-principle has been obtained. The drugs are designed to blunt mitochondrial degradation resulting from oxidative stress and to augment ATP production in partially dysfunctional mitochondria.

Project Title: *Translational Measures of Auditory Function in Individuals with Friedreich Ataxia*

Principal Investigator: Dr. Gary Rance, University of Melbourne

Summary: Individuals with Friedreich s ataxia despite having normal hearing (sound detection thresholds) can have difficulty understanding speech in everyday listening conditions. These hearing difficulties are due to disruption of auditory nerve activity that typically cannot be corrected by increasing the loudness of the signal. The primary purpose of this study is to measure changes in the function of the auditory nerve in individuals with Friedreich s ataxia, particularly those occurring before the hearing impairment is evident. These changes will allow a better understanding of the cause of the auditory nerve damage which will in turn, guide the development of intervention strategies. We will be able to quantify the degree of auditory nerve involvement and define the natural course of change in FRDA as well as monitor the effects of new therapies in this disorder. FRDA changes in the auditory pathway will be further investigated in a secondary study involving mice with the FRDA genetic profile.

Drug Development and Clinical Trials

To succeed in our mission, FARA needs a broad and deep pipeline of drugs/potential treatments. Getting discoveries from the laboratory to approved treatments in people is fraught with pitfalls, wrong-turns and failure thus our analogy of needing lots of shots on goal. Also, we know that a single victory of a single drug may not lead to an all-encompassing therapy and we need

to consider treatment coming more in the combined drug or cocktail form with iterative improvements over time. As drugs like Idebenone, Deferiprone, Varenicline and others are completing phase II and III clinical trials, we are experiencing some frustrations but also valuable lessons. When a drug does not meet its primary endpoint in a study, or negative findings from a trial are presented, it does not mean failure. There is a wealth of data collected during a clinical trial and often these data bring new insights or direction. We are gaining valuable experience that will only improve our planning and implementation of future studies. We are learning what resources are needed or gaps that exist, such as validated functional outcome measures.

FARA is excited to report that some of our lead drug candidates have made substantial progress this year and we are beginning to see new candidates emerge and join the pipeline. The FARA team continues to support our existing pharmaceutical partners in their efforts to advance drugs through development. The team is also focused on outreach to nurture new partnerships and opportunities; since the beginning of the year we ve gained several new pharmaceutical partners with drug candidates under evaluation.

- *RepliGen Pharmaceuticals has announced several milestones of progress in moving forward the lead HDAC inhibitor candidate, RG2833. They announced recently that they have received orphan drug status for RG2833 and they have submitted an application for an Investigational New Drug (IND) to the FDA. All efforts are being made to begin first-in-human studies later in 2010.*
- *Penwest Pharmaceuticals launched a phase II study of A0001, at Children s Hospital of Philadelphia, earlier this year. A0001 is an alpha-tocopherolquinone, a compound similar to Coenzyme Q10 that functions in the mitochondria. FARA has supported the development of this compound from its early discovery at Edison Pharmaceuticals to present by awarding research grants to Edison, partnering with Edison and a leading FA researcher to secure NIH support for its development, and putting the full force of our Patient Registry to use in recruiting patients for the Penwest study.*

Collaborative Clinical Research Network in Friedreich s Ataxia (CCRN in FA)

The CCRN in FA is an international network of clinical research centers that work together to advance treatments and clinical care for individuals with Friedreich s ataxia. The network collaborates with pharmaceutical companies, government agencies and other research centers and the patient community to facilitate clinical research and trials needed to identify new therapies.

We currently have more than 600 individuals with FA participating in clinical research through the CCRN in FA, and the network is growing a biorepository of DNA, RNA and plasma samples available for FA research.

New sites have been launched at:

- *The Hospital for Sick Kids in Toronto, Canada with Dr. Grace Yoon*
- *The University of South Florida with Dr. Theresa Zesiewicz*

Studies ongoing through the CCRN in FA:

- *Natural history, performance measures and quality of life studies*
- *Frataxin and mitochondrial protein biomarker studies*
- *Speech and hearing studies*
- *Genetic modifier studies*

More participation is needed at all sites to learn more about the CCRN in FA and how to get involved, visit www.curefa.org/network.html, or call or email Jen Farmer at (484) 875 3015 or jen.farmer@curefa.org. ■



A Request From FARA's Grant Writing Team

Needed: Continual Funding for FARA's Programs

How you can help:

Do you or someone you know have ties to corporate or philanthropic foundations that grant funds for programs aligned with FARA's mission to treat and cure FA?

Call FARA at 484-875-3015 or email at info@cureFA.org.

FARA 2010-2011 New Investigator Award

FARA believes in welcoming, mentoring and supporting new scientists in the FA research community. One way we do this is through offering a special grant award for young or new investigators who have demonstrated an interest in FA research and want to further that commitment. We are committed to creating opportunities for new investigators who bring fresh ideas and energy to our research community and providing the necessary resources and motivation to help them launch successful independent careers or translate their experience from a related field to FA.

We are proud to announce that our 2010 New Investigator Award is going to Dr. Lata Mahishi, Weill Medical College of Cornell University, New York.

Dr. Mahishi is an Instructor at the Weill Medical College of Cornell University and a Goldsmith Fellow at Burke Cornell Research Institute. She received her Ph.D. in biotechnology from the University of Pune, India and then was visiting fellow at the National Institutes of Health where she developed an interest in repeat expansion disease – specifically FA.

Project Title: MicroRNAs in Friedreich ataxia

Lay Summary: Today different drugs are at various stages of clinical/preclinical studies for FRDA treatment. We use frataxin levels as one of the few biochemical/ molecular biology markers to identify new drugs. However, we need additional biomarkers to better evaluate every new treatment option. MicroRNAs are a type of small molecules that have been shown to be disrupted in certain other neurodegenerative diseases. Dr Mahishi's proposal outlines an approach to identify additional biomarkers in the form of microRNAs and explore use of Anti- and Pre-microRNA molecules as potential therapeutic agents.

Sign Up in the Patient Registry Database for Future Clinical Trials:

www.cureFA.org/registry

New Locations for Ride Ataxia

By Kyle Bryant



Earlier this year FARA organized two very successful Ride Ataxia events.

Tampa

The inaugural Ride Ataxia Tampa Bay took place on April 13, 2010. It was a great day to ride with 75 new teammates and some old friends. The planning and in-kind support from the Outback Steakhouse Pro-Am and the generous contributions from cyclists, friends and family helped us surpass our fundraising goal to raise over \$30,000 in support of Friedreich's Ataxia (FA) research!



After overnight camping in Grass Valley, CA, Ride Ataxia Nor Cal cyclists prepare for day 2 of riding.

People traveled from near and far to be a part of the first Ride Ataxia event of 2010, including employees from one of our New York-based pharmaceutical partners, Penwest Pharmaceuticals. Penwest is sponsoring a clinical trial of A0001 in FA patients at Children's Hospital of Philadelphia, and we are thankful for their tireless efforts to find a treatment for FA. Special thanks also to the local team for Kraft Foods for coming out in numbers to ride with us. We sincerely appreciate this great local support. We are also grateful for the strong showing from our FA community with representatives from 15 different FA families who either cycled or volunteered for the event.

Thank you again to the Outback Steakhouse Pro-Am for inviting us to be a part of an amazing week. Special thanks also to the Hillsborough County Sheriff's Office and the Hillsborough and Pasco County Parks Department for their incredible support during the ride.

NorCal

Ride Ataxia NorCal took place on May 15 and 16, 2010 in the California Foothills outside of Sacramento. It was a very challenging ride. The event consisted of 10-, 25-, and 2-day 100-mile routes designed to allow participation from as many participants as possible. The 10- and 25-mile

options took off at around 9 a.m. on May 15, starting and ending at Negro Bar State Park in Folsom, California. The 100-mile route started at the same time and travelled 50 miles up to Grass Valley, climbing a total of 5,000 feet. That night the riders enjoyed a great feast, compliments of Outback Steakhouse before tent camping at the Nevada County Fairgrounds. The cyclists travelled back to Folsom on May 16 on a route with another 3,000 feet of climbing, resulting in 8,000 vertical feet of climbing during the 2-day 100-mile trek. This ride was very challenging some might even say too challenging. Therefore I know that all the participants proved something to themselves when they crossed the finish line!

Ride Ataxia NorCal participants came from all over and included leaders from one of our San Jose-based pharmaceutical partners, Edison Pharmaceuticals, who generously sponsored our event jersey. We also had a participant from one of our Canadian Pharmaceutical Partners, ApoPharma. Special thanks also to the team from Brown and Caldwell for their jersey sponsorship and for coming out in numbers to ride with us. We sincerely appreciate this great local support. We are also grateful for the strong showing from our FA community with representatives from 10 different FA families who either cycled or volunteered for the event.

The Ride Ataxia Nor Cal Team consisted of 200 riders and 75 volunteers who helped raise \$130,000 for research.

More Opportunities To Get Involved

FARA has more Ride Ataxia events planned, so join us as we turn the crank toward treatments and a cure!

Philly October 10, 2010

SoCal December 4-5, 2010

Dallas March 26, 2011

For additional details, visit www.rideataxia.org.



Matt Rupel enthusiastically cheers on the riders at the start of Ride Ataxia Nor Cal.

Ride Ataxia Tampa Bay cyclists, Blair DeSaw, Marilyn Downing, Kyle Bryant and Mary Caruso prepare for a day of cycling in the sun.



Joel Gottesfeld

Interviewed by Paul Marcotte

Joel Gottesfeld, Ph.D., is one of the world's leading researchers seeking to find a cure for Friedreich's ataxia. Dr. Gottesfeld is a professor in the Department of Molecular Biology at The Scripps Research Institute, in La Jolla, California. His lab has focused on a very promising area of research: the identification of histone deacetylase inhibitors that reverse frataxin gene silencing. His lab's research is the foundation for compounds now being developed by Repligen Corporation as a possible future treatment for Friedreich's ataxia.



How did you get involved in FA research?

The short answer: by serendipity! I am one of the Associate Editors of the Journal of Biological Chemistry, and I had a paper back in 2003 submitted to the journal by Bob Wells and colleagues describing the unusual DNA conformation that the GAA repeats within the frataxin gene can adopt. Bob and his colleagues had shown that the GAA repeats cause a block in RNA synthesis on pathogenic frataxin genes, and proposed that it was this unusual DNA structure, called "sticky DNA," that was responsible for gene silencing in FRDA. The last paragraph of the paper proposed that small molecules that could reverse this unusual DNA structure might activate the frataxin gene in patient cells and prove to be therapeutic. At that time my lab was working on a class of small molecules called polyamides that could be designed to bind any predetermined DNA sequence. So we were in a position to test Bob's hypothesis. After the paper was accepted for publication, I phoned Bob and told him my idea and asked if he wished to collaborate, receiving a positive answer, of course. While we were successful in showing that our polyamides reversed the block in transcription in patient cells, subsequent experiments showed that these molecules did not penetrate the brain (at least in mice) and so we needed to look for another approach to reactivate the silent frataxin gene in FRDA. Hence, the origin of our interest in histone deacetylase inhibitors.

Tell us about HDAC inhibitors and why they are a promising area for possible treatment of Friedreich's ataxia?

Unlike Friedreich's ataxia, most other triplet repeat disorders (such as Huntington's disease, many of the spinocerebellar ataxias, and myotonic dystrophy) are caused by mutations that either affect the sequence of the protein itself or cause cells to produce a toxic RNA sequence. Neither

of these scenarios is true in FRDA. The GAA repeats in the frataxin gene are in a region of the gene that does not code for the frataxin protein sequence; the repeats are in a part of the gene called an intron. Also, the GAA repeats have no effect on the sequence of the mature RNA that encodes frataxin, as the repeats are rapidly removed from the RNA copy of the frataxin gene. Experiments in our lab show that the repeats do not affect RNA processing or synthesis of the protein per se, they only affect the output of the gene; that is, transcription of the DNA into messenger RNA. Therefore, molecules that reverse gene silencing and cause patient cells to produce more frataxin protein may indeed prove to be therapeutic.

Is it your expectation that HDAC inhibitors will raise the level of frataxin in the body?

In collaboration with Myriam Rai and Massimo Pandolfo in Brussels, we have shown that our HDAC inhibitors will indeed increase frataxin protein in the affected tissues (brain and heart) in their mouse model for the disease. While there is no guarantee that what is true of a mouse can be repeated in humans, our expectation (or rather hope) is that the same will be true in humans and our molecules will indeed increase frataxin protein levels in humans suffering from FRDA.

Why is that important for FA patients?

Since FRDA is a disease due to a mutation in a single gene, causing gene silencing, we believe that molecules that increase frataxin protein should be therapeutic. A worry, however, is whether increases in frataxin will indeed have the expected therapeutic benefit to patients, that is, slowing or reversing the course of the disease. While we hope that this will prove to be true, only clinical trials will tell us whether our molecules are of benefit to patients.

What is the status of your HDAC research today?

While our current molecules do indeed increase frataxin mRNA and protein in patient cells and in the mouse model, these molecules suffer from two limitations; namely, less than optimal brain penetration and stability in animals. Thus, a postdoctoral research associate in my lab, Chunping Xu, is currently synthesizing new derivatives that may overcome these limitations. This work is a collaboration with our pharmaceutical industry partner,

Continued on p. 7

Thinking about fundraising?
Contact marilyn.downing@cureFA.org

Repligen Corporation. We are testing these molecules in patient cells and in new cellular models for FRDA based on induced pluripotent stem (iPS) cells, which are derived from patient skin cells. These iPS cells can be turned into neurons or heart cells in the laboratory, and may represent more relevant cell types for testing our compounds. We are also interested in the exact biochemical mechanism of how the HDAC inhibitors increase frataxin mRNA. Answers to questions such as this may give us clues as to how to improve on our molecules.

You are currently working with Repligen Corporation in developing RG2833, an inhibitor of histone deacetylase 3 (HDAC-3) for FA. How far along is the development of that compound?

Repligen synthesized a large number of derivatives of our lead HDAC inhibitors and identified RG2833 as the best candidate to take forward into human clinical trials. This decision was based on the results of experiments at Repligen and in our lab at Scripps. Repligen has done all of the pre-clinical work necessary to file an Investigational New Drug (IND) application with the US Food and Drug Administration (FDA). Once all of the work required by the FDA is completed, Repligen will be in a position to commence testing in humans. Our role in this process is limited to laboratory experiments with RG2833, showing its efficacy in patient cells and in the iPS cell models that I described.

What are the next steps in development?

Clinical trials are a long process: first, the drug candidate molecule must be shown to be safe in both unaffected human subjects and then in patients. This is called a phase I study. Then a larger phase II study is initiated to show efficacy, both in terms of increases in frataxin, which can be shown in white blood cells taken from patients, and in terms of neurological performance on the various rating scales that neurologists use to measure disease severity. Lastly, a much larger phase III trial or trials are initiated. Unfortunately, all of this takes years. But hopefully, we will know some results from the phase I study in patients to show whether the molecule is effective in increasing frataxin in blood cells. This would be a good indication that Repligen should proceed with larger and more costly patient studies.

What other promising research do you see underway as possible treatments for FA?

Currently, there are several areas of exciting research going on. First, in terms of increasing frataxin protein, scientists in Vienna have shown that the blood cell-enhancing protein, erythropoietin (EPO), causes increases in frataxin protein both in patient cells and in blood cells taken from patients treated with EPO. These are exciting findings, but more

work is necessary to find safer versions of EPO for FRDA patients. Additionally, Mark Payne and colleagues are pursuing frataxin protein replacement, using a version of the protein coupled to a small signal sequence that causes the protein to be taken up by cells. Whether this approach will work in animals and ultimately in humans, of course, will require further work. Other labs, including Richard Festenstein in London, are investigating molecules to increase frataxin synthesis by targeting other classes of protein deacetylases, such as a family of enzymes called sirtuins. Lastly, efforts at Edison Pharmaceuticals are aimed at circumventing the cellular energy defect caused by the reduction of normal levels of frataxin in patients. Each of these approaches, including our own with HDAC inhibitors, offers promise for patients, but each of these approaches will require validation that will only come through human clinical trials.

You've spoken about how working with FA families has affected your career and your life. Can you describe what that impact has been?

I spent the first two decades or more of my scientific career working on basic research in molecular biology. Although this work was exciting and fulfilling, I had no idea of just how much more meaningful life would be working on a problem of real significance to patients. Meeting the young people affected with FRDA has been a moving experience, giving me, and the members of my lab, the inspiration to work hard toward finding a treatment for the disease. In meeting both patients and their families, time and again I am inspired by their grace and selflessness. The question that family members often pose to me is what can I do for you? But really, it is my hope that I can do something for you and your affected family members.

How soon do you think there will be effective treatments for FA?

I wish I had a crystal ball to foresee just when there will be an effective treatment. However, I am hopeful that with all of the effort being expended worldwide toward development of various therapies for FRDA, something good will come of these studies within this decade. ■

**Sign Up in the Patient Registry
Database for Future Clinical Trials:**

www.cureFA.org/registry

Dr. Ole Isacson (Professor of Neurology and Director of the Center for Neuroregeneration Research at Harvard Medical School), Dr. Jack Kessler (Professor and Chair of Neurology and Director of the Stem Cell Institute at Northwestern University), and Professor Bob Williamson (Honorary Senior Principal Fellow at the University of Melbourne and the Australian Academy of Science Secretary of Science Policy). They were extremely positive and complimentary about the ongoing collaborative research in FA, and stated that the science presented at this meeting was on a par with any they have seen. This was especially meaningful given the global leadership roles of these three scientists in this arena. They also emphasized the value of standardized characterization and sharing of FA iPS cell lines for therapeutic advances.

A key funding initiative

The availability of well-characterized cellular models is highly beneficial for drug discovery and testing, as well as for developing gene and stem cell therapies. All of the international FA advocacy partners recognize the critical need for these models and are supporting their development. In particular, FARA is providing funding to Dr. Helene Puccio, and FARA and FARA-A are co-funding grants to Drs. Mirella Dottori and Alice Pebay for the development and differentiation of FA iPS cells. Dr. Joel Gottesfeld, whose groundbreaking work on HDACi therapeutic candidates has been supported by FARA, is also working on iPS cells because he sees their potential for testing and identifying the most effective HDACi for FA therapy.

While numerous FA cell models have been used in research laboratories, none have been truly representative of FA. The discovery that skin cells, which are easy to obtain, can be reprogrammed into iPS cells is a monumental breakthrough; it makes research on neuronal and cardiac cell lines from FA patients possible.

Latest developments

The Meeting on Cellular Models and Cell Therapy provided an outstanding opportunity for the establishment of communication, cooperation, and collaboration, and there is no doubt that this highly productive meeting was successful in achieving this goal. Those involved in advancing stem cell and gene therapy approaches for FA are working together on the studies that need to be done to make such therapies a reality.

All participants agreed that we need reproducible, stable, well-characterized FA iPS cell lines that are appropriate for research (including drug screening), maintained in a reputable facility, and distributed among researchers. To this end, discussions are underway with Dr. Jeremy Crook,

Director of Stem Cell Medicine and the Australian Stem Cell Bank (Cytentia) at the O'Brien Institute in Melbourne. Cytentia will derive, store, and distribute high-quality iPS cell lines for pre-clinical and clinical research. This work will be done in accordance with strict standardized procedures. Particularly exciting is the fact that FA will be a major research focus at the O'Brien Institute; thus, they will not only be providing standardized FA iPS cell lines to researchers around the world, they will also be accelerating progress in the development of cell therapies for FA. ■



Barnfly Racing Team

Scotty Haggard, his wife, Jana, his dad, Tim and family make up the Barnfly Racing Team. They do all of the mechanics and work themselves. Scotty's dad, Tim, drives the #17 car, which features the FARA logo on the hood. The #17 car is a circle track race car that is having great success at the Fairgrounds Speedway Nashville. On July 17, the team won its first race of the season.

Scotty has been into racing his whole life and saw an opportunity to help increase awareness for FARA and FA while he and his team are out winning races and doing what they all love.



Second FA Cardiac Summit identifies needs and goals

By Jennifer Farmer

We are all well aware that cardiac disease in FA is under appreciated in the medical and research communities. It is not well described or studied even though it can be the most dangerous and life shortening aspect of FA. In a continued effort to bring more research and attention to cardiac involvement in FA, Dr. Mark Payne, FARA Scientific Advisor, cardiologist and FA researcher, organized and led the 2nd FA Cardiac Summit on June 11, 2010. Twenty participants were invited, representing diverse areas of basic science, clinical research, and cardiology.

The objectives and goals of the meeting included:

- *Identify clinical and basic science questions that are important to solve to better understand cardiac disease in FA, such as establishing hypotheses and explanations for how mitochondrial dysfunction in FA might lead to cardiac disease in FA.*
- *Review and document the cardiac phenotype from clinical and pathology data currently available.*
- *Discuss approaches to advanced therapy (LVAD, transplant, etc.) to treat patients with rapidly progressive and/or severe cardiomyopathy*
- *Plan prospective treatment trials that will allow for the establishment of informed treatment recommendations.*
- *Bring new clinical and basic researchers into this field to advance discovery.*

The summit was filled with targeted presentations and robust discussion that engaged the entire group of participants, bringing new ideas forward and solidifying several next steps. There was overwhelming consensus and support for achieving improvements in cardiac care for individuals with FA. This would be done through the development of standards of care and further development of the cardiac expertise at our Collaborative Clinical Research Network in FA sites and beyond. One of the goals is to establish cardiac referral centers for disseminating information, and providing treatment and management for cardiac patients. Another high priority for the Cardiac Summit participants and FARA is to support necessary research into reasonable and FDA-acceptable outcomes of cardiac function for clinical trials. On the basic research side, the development of an FA cardiac specific cell culture model is needed to test various hypotheses of underlying mechanisms of the disease and for testing and development of new therapeutic approaches.

FARA gratefully thanks Dr. Payne for organizing the 2nd Cardiac Summit, and all of the participants for sharing their knowledge, contributing new ideas and facilitating good discussion that has led us forward. We look forward to continued work with this group as we begin to tackle the questions, needs, and desired outcomes identified in the summit. ■

Keith Michael Andrus Memorial Award Announced

The Friedreich s Ataxia Research Alliance (FARA) invites proposals, under a competitive Request for Applications (RFA) process, for a grant focusing on the cardiac disease of Friedreich s ataxia. We anticipate funding one, one-year award. The total award is limited to \$75,000 (direct costs only).

Keith Michael Andrus was the inspiration for the formation of FARA. Keith s diagnosis of Friedreich s ataxia at the age of 11 led his stepfather and mother, Ron and Raychel Bartek, to establish FARA in 1998. Keith ended his battle with FA on January 22, 2010 at the age of 24 due to congestive heart failure. Even during the final and very challenging months of Keith s life, he courageously continued to advocate not only for himself but for others with FA, leaving no stone unturned as he explored advanced and experimental treatment options.

The Keith Michael Andrus Memorial Award has been established as a tribute to Keith s legacy and to help improve the lives of others diagnosed with FA. This award is specifically designated for cardiac research.

RFA Applications due: September 1, 2010

Award announcement: November 1, 2010

Grant start date: November 15, 2010

For more information please contact Jen Farmer:
Jen.Farmer@cureFA.org

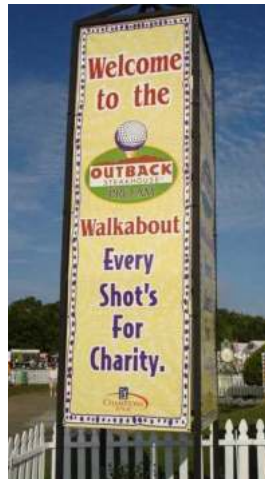
**Sign Up in the Patient Registry
Database for Future Clinical Trials:**

www.cureFA.org/registry

Outback Steakhouse Huge Supporter of FA Research

By Felicia DeRosa

At both the corporate and local level, Outback Steakhouse continues to be a strong partner of the Friedrich's Ataxia Research Alliance (FARA), both in the person-to-person connection between Outbackers and FA Families and through OSI corporate leadership support of FA research.



Act Locally. . .

Partnering with the Zies, Golanec, Simpson, Richard, Sweeny, Dalton, Bartek, Laird, and Konanz families, Outback Steakhouse restaurants have continued to be successful in raising funds and awareness through the luncheon fundraising program in the following locations: Maumee, OH; White Plains, NY; Rohnert Park, CA; Springfield, NJ; Springfield, VA; and Bakersfield, CA.

The Heineken with a Heart program also continues to bring FA families and communities together for a fun, festive and fundraising happy hour in Atlanta, GA, Tempe, AZ, Chicago, IL, Baltimore, MD and Royersford, PA. Special thanks to Julie Robertson at Outback and Hillary Johnson at Heineken for the success of this program.

Several Outback teams have also organized and participated in sporting events, such as golf, sporting clays and running, to benefit FARA. This year FARA is one of the beneficiaries of the DC Golf Classic, Yorktown Lions Club Down Under Scramble, and the Chesapeake Bay Area Charity Clay Shoot. Additionally, the San Francisco local Outbackers put on their running shoes again this year as part of Team FARA in the San Francisco half marathon.



Connor Sweeny poses for a picture with Kyle Bryant while people fill their plates with some delicious Outback food at the luncheon in Springfield, NJ, hosted by the Sweeny and Dalton families.

Outback Pro-Am

FARA is incredibly grateful to have been selected as one of the beneficiaries of the 2010 Outback Steakhouse Pro-Am. The Outback Steakhouse Pro-Am is a weeklong official tournament on the PGA Champions Tour. Celebrities such as Michael J Fox, Vince Gill, and George Lopez were paired with professional golfers for the crowd drawing tournament this year. FARA was fortunate to work with Amy Hawk- Tournament Director and Mary Margaret Schexnayder- Volunteer Coordinator, who gave FARA the opportunity to raise awareness at the event through program and billboard ads, a FARA information booth at the golf course, and the Ride Ataxia Tampa bike ride prior to the tournament.

Ride Ataxia & The FARA Energy Ball

Outback has also been a key supporter of both Ride Ataxia and the upcoming FARA Energy Ball. Pat Kruk and the Outback Team in Northern California not only provided in-kind catering and food support to 200 cyclists and 75 volunteers during the actual Ride, but supported a number of fundraising parties in the area leading up to the event. Ride Ataxia is also grateful to be working with John Jackson and his Philadelphia team, Joe Gatto and his Southern California team, and Blaise Hadley and his Dallas team, to plan upcoming rides in those areas. Outback is a presenting sponsor of the FARA Energy Ball in Tampa at the end of August. In addition to this top level sponsorship, the OSI Restaurant Partners have been incredibly generous with donated auction items for the event.

Thank you to the individuals organizing and acting locally at Outback on FARA's behalf as well as the OSI leadership for your commitment to supporting FARA events such as Ride Ataxia and the FARA Energy Ball. You have made our rare disease feel a lot less lonely and truly become part of our Family. ■



The Outback Steakhouse in White Plains, NY, in holding a very successful fundraising luncheon, also brought together members of the Caruso/Bode, Trovinger, Richard, Golanec and Simpson families.



Stephanie's Hope Holiday Boutique, Valencia, California

By Marilyn Downing

In 2004 Stephanie Magness found out her true diagnosis after years of being misdiagnosed. It was not easy to hear that she had Friedreich's ataxia but her mother, Sharon, was prepared to jump in and raise money for research. Stephanie was not ready to be so "visible"—to have people "see what her life was like." Sharon decided to hold off until her 23-year old daughter was ready.

While she watched the 2005 MDA telethon, Stephanie was inspired by a young girl with muscular dystrophy talking about raising awareness and funds. This was the catalyst, explains Stephanie, because she knew she could do the same. She was finally ready and Stephanie's Hope Holiday Boutique was created.

The Magness family started with backyard dinners and silent auctions but moved on to a new idea for them and for FARA—a benefit boutique! This year will be the fifth boutique for the Magness family and Stephanie is the heart and soul of this event. She speaks to the attendees about FA, helping them to understand the disease and the urgency for research funds. She wants her community to know what FA is and this is becoming a reality as the boutique becomes an annual must-attend event, with media attention and as many as 26 vendors, each donating 20% of their proceeds. Stephanie's four boutiques have brought in about \$23,000—enough to completely fund a research grant.

Stephanie feels that this event has not only helped her to accept her diagnosis but it also gives her a voice in the FA community. It has given me the opportunity to grow into it, accept it and be OK with it, said Stephanie. Meeting so many new people who can see past it and just see *me*, empowers her.

Being involved with fundraising has led many of us to develop new relationships and to grow in confidence to take our story beyond—to spread awareness about FA and to help move the science along that much quicker. With family events such as Stephanie's Hope Holiday Boutique, along with Ride Ataxia events, the Energy Ball and the FARA presence on Facebook, many more young adults with FA are becoming involved with the greater FA community.

For more information go to www.StephaniesHope.com. This year's boutique will take place November 13. ■



Board of Directors

Ronald J. Bartek, President/Director/
Co-Founder
Retired U.S. Government Official;
Business Consultant

Thomas A. DeCotiis, Chairman of the Board
Founder and Chief Executive Officer,
CorVirtus

Rob Dhole, Director
President, Diversified Agency Services Healthcare
Omnicom Group

Marilyn E. Downing, Secretary/Director
Teacher/Diagnostician, Special Education

Steve Klasko, Director
CEO, USF Health
Dean, College of Medicine
University of South Florida

J. Ed Ramsey, Treasurer/ Director
VP, Secretary/ Treasurer, & Co-Owner,
Taylor- Ramsey Corp.
President, Summit Hardwood
VP, BEPCP

Paul Avery, Director
Former Chief Operating Officer,
OSI Restaurant Partners, LLC

Dr. Holly Hedrick, Director
Surgeon, Children's Hospital of Philadelphia

Nicholas A. Johnson, Director
Associate & Senior Mechanical Engineer, Bard,
Rao + Athanas Consulting Engineers, LLC

Laura Kalick, Director
Nonprofit Tax Director, BDO Seidman, LLP

Paul Marcotte, Director
Attorney & Communications Consultant

Peter Pitts, Director
Partner & Director, Global Health, Porter Novelli
Senior Fellow, Center for Medicine in the
Public Interest
Former Associate Commissioner, US Food and
Drug Administration

Dr. Earl Giller, Scientific Director
Consultant, Global CNS Pharmacology
Consulting, LLC

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



www.cureFAstore.com

**Very reasonably priced FARA caps,
polo and t-shirts, wristbands, etc.**



**Thank you to Performance Award Center (PAC)
for their support.**

The FARA Advocate is brought to you by:

Contributors: Ron Bartek, Kyle Bryant, Felicia DeRosa, Marilyn Downing,
Jennifer Farmer, Dr. Bronya Keats, Paul Marcotte

Editor: Karen Smaalders ***Design/ Layout:*** Anne Myers



Team FARA cyclists - John Lockwood, Sean Baumstark, Mike Mellott and Kyle Bryant receive their official Race Across America finisher medals in Annapolis, MD.

Team FARA Races Across America

by Felicia DeRosa

Team FARA completed the 2010 Race Across America (RAAM) in 8 days, 8 hours, and 14 minutes, making us believe that all things are possible with a clear goal, a team with a whatever it takes attitude, and the support of a united community. The remarkable team traveled 3,005 miles across the country with 100,000 feet of climbing, showing us how powerful a voice can truly be when committed to a cause and an act of endurance.

The Decision

Prior to cycling in RAAM, Ride Ataxia founder Kyle Bryant told an interviewer, I think that dealing with FA I constantly feel an urgency to do things now while I still can, before they pass me by. . . He was quickly joined by fellow cyclists and friends Sean Baumstark, John Lockwood, and Mike Mellott to form Team FARA's 4-man relay team in RAAM. A strong voice for the cause took root in the decision to do something now.

The Preparation

The decision to act was followed by preparation. Team FARA in RAAM cyclists spent countless hours on the bike in preparation for race riding around the clock and covering nearly 375 miles as a team each day. In the early mornings they turned the crank on the trainer as they rolled out of bed before work, and on the repeated long weekend rides they built the strength to climb the mountain passes of Colorado and fight the crosswinds in Kansas. The crew also spent countless hours on Sunday night teleconferences methodically researching and planning the trip to ensure efficient and safe movement across the country.

The Commitment to Action

Though the journey was long and included sleepless nights, rainstorms, seemingly never ending uphill climbs, heat,

humidity, and mosquitoes, the commitment to the goal and the support of the community kept the team going. If the geography required all four cyclists to ride in the vans rather than resting in the RV between shifts, the cyclists rose to that challenge. If a minor injury took one cyclist off the course, another filled in with back-to-back shifts. If a bike light was lost as night approached, the crew got a closed bike shop to open their doors and replace the light within minutes. If the RV generator blew, the crew persisted until they found a repair shop close to the route to fix it. Though weary, the team and crew was committed to each other and to constantly moving forward. As a result, the team successfully crossed the finish at 1:14 a.m., welcomed by a huge crowd of FA families and friends.

Join Team FARA

You can be a part of Team FARA, and the event need not be an extreme endurance event. It doesn't matter if the event is a 5K run or a marathon, a 10 mile ride or century, some laps in the pool or a several mile swim in the sound; It just needs to be something that physically challenges you and gives you a platform to raise your voice, awareness, and funds in support of Friedreich's ataxia research. To join Team FARA, contact **Felicia DeRosa** at info@curefa.org. We will help you set up your fundraising webpage and send you a Team FARA running jersey to wear during your event.

Team FARA in RAAM

Kyle Bryant
Sean Baumstark
John Lockwood
Mike Mellott

Team FARA in RAAM Crew

Mike Bryant- Crew Chief
Tracy Allegeier
Mike Andresen
Blake Andrews
Diane Bryant
Mary Caruso
Felicia DeRosa
Aaron Farley

Team FARA in RAAM Crew

(Continued)
Mike Gore
Paul Konanz
Stephen Parsons
Bob O Neil
Phil Vickers



Team FARA crew member Mary Caruso and cyclist John Lockwood share a moment of triumph at the finish line.

Visit theataxian.com for information about the documentary that captured Team FARA as they pedaled across the country.

UPCOMING EVENTS for 2010

AUGUST

August 7, 2010

Outback Steakhouse Luncheon
Tacoma, WA

August 13, 2010

Welsh Family Backyard Bash
Harrisburg, PA

August 15, 2010

Team FARA, Moms Triathlon
Seattle, WA

August 21, 2010

Beat Ataxia Blues at
Buddy Guy Legends
Chicago, IL

August 21, 2010

2nd Annual Harry Hunt Rusty Nail
Golf Tournament
Howey-in-the-Hills, FL

August 25-29, 2010

Team FARA, Fool Killer Climb
White Mountains, NH

August 26-28, 2010

FARA Energy Ball
Tampa, FL

SEPTEMBER

September 11, 2010

Team FARA,
Adirondack Great Range Hike
Adirondack Mountains, NY

September 12, 2010

Fuzzy Buzzy Golf Tournament
Windham, NH

September 13, 2010

Peters Hole Out for a Cure
Peoria, IL

September 20, 2010

Hughes Golf Tournament
Herndon, VA

September 23, 2010

Century 21 King Charity
Golf Tournament
Fontana, CA

September 2010

Olsen Golf Tournament
New Jersey

September 26, 2010

Team Donovan,
Jarden Westchester Triathlon
Rye, NY

OCTOBER

October 9, 2010

Hopkins Annual 5K Race for FARA
Bristol, Rhode Island

October 9, 2010

Team FARA, Hawaii Ironman
World Championships
Kona, Hawaii

October 10, 2010

Team FARAMones,
Chicago Marathon
Chicago, IL

October 10, 2010

Ride Ataxia Philadelphia
Limerick, PA

October 11, 2010

Outback Steakhouse
Golf Tournament for FARA
Jacksonville, FL

October 16, 2010

Martinez Ride Ataxia Party
Martinez, CA

NOVEMBER

November 7, 2010

Team FARA, NYC Marathon
New York, NY

November 13, 2010

Stephanie's Hope Holiday Boutique
Valencia, CA

DECEMBER

December 4-5, 2010

Ride Ataxia SoCal
Los Angeles, CA

**Please check the FARA website for contact
information and a full list of events.**



Anxiously watching the bidding at the pre-Northern California Ride Ataxia party planned by Becky Prater and family.



After walking in the O'Brien's Seaside Stride, these kids just have one message...Cure FA!



Kyle Bryant and Tom Trovinger rolling in for a cold one at the Heineken with a Heart fundraiser in Baltimore.



Walking along the beautiful New Jersey shore in the Seaside Stride, organized by Karen O'Brien.



Tiffany Gambill is surrounded by her sorority sisters who held an event in her honor at the University of Massachusetts, Dartmouth.



Marilena Wilkinson participated in the Atacama Desert Run as a member of Team FARA.



THE FARA ENERGY BALL

August 26th
Cultivating a Cure
Scientific Symposium

August 27th
"Martinis & Poker"
Patron Party

August 28th
The FARA Energy Ball
with live performance
by Sister Hazel

AUGUST 26-28, 2010
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.

Thanks to Our Generous Sponsors*

PLATINUM SPONSORS

A La Carte Event Pavilion
Paul and Mary Jacobs
Standard Meat Company

GOLD SPONSORS

Bill and Janine Allen
Anheuser Busch
Bob and Debbie Basham
Beam Global Spirits and Wine, Inc.
Kraft Food Foodservice
Pernod Ricard
Southern Wine and Spirits of America
Chris and Sandi Sullivan
Trinchero Family Estates
USF Health
Dr. Paul and Avery Lowry Zaritsky
Zeno Office Solutions

SILVER SPONSORS

Bacardi
Bar One
Mr. Rick Berman
Mr. & Mrs. Franklin & Barbara Carson
Chateau Ste. Michelle Wine

SILVER SPONSORS (Continued)

Constellation Wines
Mr. & Mrs. Herman & Tanya Fernandez
Finest Call
Foster's Wine Estates
Mr. & Mrs. Keith & Gaylaine Harvey
Heaven Hill
Hot Schedules
Jacobs Air Water Systems (JAWS)
Monin Gourmet Flavorings
Proximo
Thomas Financial Group
William Grant & Sons, Inc.

MEDIA SPONSORS

Broad Reach Television, Inc.
Tampa Bay Metro Magazine
Tampa Bay Online
Centro Tampa
The Tampa Tribune
News Channel 8
Design donated by:
Brand Architecture, Inc.
Printing donated by:
Central Florida Press

*As of July 20, 2010

Presenting Sponsors:

clw real estate services group

 **MillerCoors™**

**The Avery Family
Foundation**


RESTAURANT
PARTNERS, LLC®

OUTBACK
STEAKHOUSE®

CARRABBA'S
ITALIAN GRILL


Fleming's

BONEFISH
GRILL


Roy's



Walkers enjoy a nice stroll in the sun during FA Awareness Weekend in Jonestown, PA.



The Helms Family gears up for the 10 mile route at Ride Ataxia Nor Cal. Team Helms was led by Dylan riding the course on his trike. Go Dylan!



Zac Zies and Zane with Patrick and Susan Short at the Maumee, OH Outback luncheon.



FARA Executive Director Jen Farmer sports her Team FARA jersey prior to the Broad Street 10 miler.



Breanne Moen runs the American River Parkway 1/2 Marathon as part of Team FARA and in honor of her friend Rachel Gill.



Tom Trovinger and Dennis Wood with their families at Dennis's Ham Supper Fundraiser.



Guests show their appreciation at the Flatbread's Charity Night for FA. Erin O'Neil organized this special event which took place in Bedford, MA.



Sisters Tammy Luebbe and Pamela Rasey invited Nelda Van Schoick (center) to be their featured speaker at their dinner dance held in honor of Evan Luebbe, in Liberty Township, OH.



Attending the Van Schoick family's annual Bash in Athens, GA, included members of the Bryant, Zilles, Bartek, Jacquin, LaRoche and Stack families. The event is given in honor of Robbi and Becca Van Schoick (front row, right).



The Helms First Annual FARA fundraiser was a party for the whole family as the kids show off their moves on the dance floor.



The guy with the great smile is Steven Cheatwood, who attended the Heineken with a Heart event in Atlanta.



Laura Beth Jacquin and Jon Zilles enjoy some time together in Atlanta at the Heineken with a Heart event.

Contact Us



Friedreich's Ataxia Research Alliance
P.O. Box 1537
Springfield, VA 22151

Phone: **(484) 875-3015**

E-mail:
info@cureFA.org

Web Site:
www.cureFA.org