Advancing Research for Friedreich's Ataxia

Each Breakthrough Brings us closer to a cure.
Life-altering breakthroughs in Friedreich’s ataxia research are made possible by the passionate will of parents and patients, scientific collaboration, and the generosity of supporting corporations and individuals.

At age nine, Keith started showing painful and frustrating symptoms. For two years, his parents, Raychel and Ron Bartek, struggled to find the cause. Doctors hypothesized but lacked definitive answers. Then one day, finally, a diagnosis: Friedreich’s ataxia (FA), a rare, debilitating, life-shortening neuromuscular disorder. The diagnosis was initially a relief – with a named disorder the Barteks could move forward with treatment. Quickly, relief gave way to deep despair: no treatment or cure existed for FA.

In search of hopeful news, the Barteks met with the lead rare disease specialist at the National Institutes of Health (NIH), setting into motion a powerful movement that would forever change the outlook for FA families and patients.

Encouraged by the NIH, the Barteks engaged a team of passionate families and three of the world’s leading FA research scientists to launch a dedicated and urgent grassroots effort to advance FA research. They established the Friedreich’s Ataxia Research Alliance (FARA) in 1998. Its goal: to slow, stop, and reverse the course of FA, ultimately curing the disease. Its work: funding FA research and driving scientific collaboration among scientists, the pharmaceutical industry, families, physicians, and other rare disease organizations.

Since then, FARA’s efforts have led to major scientific breakthroughs in the fight against FA, bringing tremendous hope to people who just ten years ago had none. Today, FARA’s leading scientific advisors and respected researchers have strong conviction that advanced treatments and ultimately a cure are on the horizon.
Roadmap to a Cure

From day one, the broad vision, collaborative nature, and tenacious dedication of its founders have driven FARA’s work. They developed a unique roadmap for moving swiftly and directly toward treatments and a cure for FA. To avoid the common barriers that deter rapid progress in rare disease research, FARA designed an infrastructure that efficiently stewards important laboratory breakthroughs through pharmaceutical development to patients’ bedside.

Key elements of this infrastructure include:

A World-Class Scientific Advisory Board that sets FARA’s research agenda and is comprised of leading scientists from diverse disciplines. This global team directs a comprehensive strategy for FA research that focuses on multiple approaches and maximizes the pace and efficiency of FARA’s research program.

International Scientific Conferences that bring together researchers from around the world to share their FA research progress, propose new hypotheses, and discuss challenges/hurdles to their progress.

An Expedited Grant Program that operates 24/7/365, thereby assuring that all promising research applications receive funding within 60 days of submission.

A Global Patient Registry that retains detailed patient records and facilitates recruitment and patient access for clinical trials – frequently a prohibitively costly and tedious process for pharmaceutical companies. FARA’s is the only worldwide registry of FA patients.

A Collaborative Clinical Research Network that monitors the natural history and progression of the disease while providing patients with access to the best clinical care. FARA’s drug development partners consider this network a significant attribute in advancing their promising drugs through clinical trials.

FA Mouse and Cell Models, continuously being developed and improved with FARA funding, that are critical to discovering and evaluating new treatments.

FARA’s Ripple Effect

Since FA shares similar traits with other diseases, both rare and common, FARA’s work has powerful implications for a broad disease community. Research discoveries for FA may provide significant insights into:

- Alzheimer’s Disease
- Ataxia-telangiectasia
- Cerebellar ataxias
- Diabetes
- Fragile X Syndrome
- Huntington’s Disease
- Parkinson’s Disease
- Mitochondrial diseases (MELAS, MERRF, Leber’s)
- Muscular Dystrophies (Duchenne, ALS, SMA)

FARA collaborates extensively with related disease advocacy groups – including the Muscular Dystrophy Association, American Heart Association, and a growing number of partner organizations around the world – to co-fund research and conferences that benefit all collaborating organizations and the patients they represent.

“Few advocacy groups can do it right and FARA is one of the groups that can. FARA funds basic research to understand FA and builds partnerships with others who can help move discoveries into cures, faster and more effectively than if working in isolation.”

STEPHEN GROFT, Pharm. D, Director, NIH Office of Rare Disease Research

“The signs and symptoms of Friedreich’s ataxia come from mitochondrial dysfunction. Just the research on mitochondrial dysfunction and how to reverse that dysfunction for energy production can have implications and a huge ripple effect on many other diseases.”

ROB WILSON, MD, PHD, Scientist, Hospital of the University of Pennsylvania
Emphasis has shifted to Drug Development and Clinical Research

Basic Research | Drug Discovery and Development | Clinical Trials
---|---|---
2006 | 10 | 4 | 1
2007 | 6 | 8 | 3
2008 | 5 | 9 | 6
2009 | 7 | 10 | 6

Accelerated Research Progress

FARA's 2010 Research Pipeline Demonstrates Significant Advancements Since 2004

2004: 1 Clinical Trial - 3 Potential Treatments/Approaches
2010: 8 Clinical Trials - 9 Potential Treatments/Approaches

Unraveling the Mysteries of FA

In its first decade, FARA unraveled the mysteries of Friedrich's ataxia. An intense research focus, funded by FARA supporters, led to critical understandings about the disease’s genesis and progression.

Today, the FARA scientific community is translating these groundbreaking laboratory discoveries into the beginning stages of treatment, bringing real hope to FA sufferers. As FARA works to expand its network of collaborators, it continues to facilitate additional discoveries and expedite the movement of potential treatments along the critical path to clinical testing in FA patients.

BRIDGET DOWNING, 29, diagnosed at age nine, currently enrolled in a FARA-funded clinical trial for a neurotransmitter. The Downing family has raised more than $435,000 for FA research.

*Once you're diagnosed, it is very scary to think of what the future holds. Thanks to FARA, patients and families find that they have become part of a loving and supportive community. They can also see how much research is being done and how close science is to finding something to help treat FA. Now that I can participate in clinical trials, I not only hope, but believe, that treatments will make a difference in my life.*

The FARA research pipeline is an illustration of the multiple therapies in development. On the horizontal axis, each leading drug candidate is represented by a bar and is grouped by its mechanism of action. Each mechanism of action targets one of the known causes of Friedrich’s ataxia. The milestones for drug development are listed on the vertical axis with treatment available to patients at the top.

Advancing Research for Friedrich's Ataxia
Help us Reach the Horizon

Today, FA patients are hopeful due to the FARA-funded breakthroughs over the past decade; tomorrow, they will benefit from treatments with your help. Freedom from the debilitating, devastating effects of FA is on the horizon. Each day, the FARA scientific community moves closer to realizing our goal of slowing, stopping, reversing, and ultimately curing FA.

As we get closer and closer to treatments and a cure, we need your help more than ever. Please join FA families and friends, along with the many dedicated researchers who have brought us to this pivotal point. Your support will help accelerate research and save lives.

Together, we WILL make the ultimate breakthrough to cure FA.

Turn Opportunities into Treatments

We are now in the "treatment era" and are supporting vital and costly clinical trials, without which we cannot achieve our goal. At the same time, we cannot stop funding basic, discovery research and filling the research pipeline until we have the cure. As a consequence, FARAs funding needs have grown exponentially along with our progress.

FARA FUNDING COMPARISON

<table>
<thead>
<tr>
<th>Year</th>
<th>Grants</th>
<th>Total Grant Funding</th>
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In 2009, FARA funded one clinical trial at a cost of $500,000. Funding needs of this magnitude will increase as our research moves closer and closer to approved treatments.

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"Disease treatment and research is a complex problem and takes the efforts of everybody to solve… It’s not about competition but about sharing the science that we all learn to help people who suffer…I am certain that we are six to 12 months ahead of where we would have been without FARA — and every day counts in the lives of these patients.”

Jennifer Good, CEO, PENWEST PHARMACEUTICALS

"FARA provided the finances to get things going with FA research. Now, more importantly, they provide infrastructure, collaboration, and the drive to turn small advances into large advances.”

David Lynch, MD, PhD, Neurologist, Principal Investigator, FA clinic, Children’s Hospital of Philadelphia

"I pulled myself out of deep despair in the early days of my diagnosis and committed myself to doing everything I could to change the course of this disease. Like FARA does with disease research and advocacy, I pushed my personal limits to get things done. It’s why FARA as an organization appeals to me: the people behind it are willing to go that extra mile to figure out and do the things that need to be done. FARA is a wonderful model for the opportunities that exist at even the bleakest moments.”

Kyle Bryant, 27, diagnosed at 16, founder of Ride Ataxia, a national cycling fundraiser for FA research. Kyle has raised over $800,000 for FA research.

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FARA’s Guiding Principles

**Wisdom.** Being inquisitive, innovative, thorough, and collaborative; mining knowledge and experience for breakthrough insights; and sharing information and other resources in a timely manner.

**Caring.** Behaving with concern for the dignity, comfort, and well-being of people; respecting their individuality and perspective; and giving them our undivided attention.

**Urgency.** Being passionate and focused on FARA’s mission and goal and moving quickly on opportunities to advance them.

**Quality.** Setting extraordinarily high standards for the fight against FA, constantly improving, and attending to the details of excellence.

**Spirit.** Lifting the mood of others, and being a source of hope and confidence and the kind of organization of which people are proud to be a part.

**Integrity.** Being honest, transparent, forthright, and ethical – challenging anything that might compromise FARA’s mission and goal.

“Acting alone, there is very little we can accomplish, but acting together, there is very little we will NOT accomplish.”

RON BARTEK, President and Founder, FARA