# 2017 FARA Awarded Grants

## Principal Investigator/Organization

- **Robert B. Wilson** - Children's Hospital of Philadelphia
- **David Corey** - University of Texas Southwestern
- **Richard Wade-Martins** - University of Oxford
- **Gino Cortopassi** - University of California Davis; Paola Giunti - University College London; Mark Pook - Brunel University
- **Hugo Bellen** - Baylor College of Medicine
- **Javier Santos** - University of California Los Angeles; Katia Aquilano - University of Rome
- **David Lynch** - Children's Hospital of Philadelphia; University of California Davis; Chad Hoyle - Ohio State University
- **Joel Gottesfeld** - The Scripps Research Institute
- **Pierre Gilles Henry & Christophe Lenglet** - University of Minnesota
- **Cat Lutz** - The Jackson Laboratories
- **David Corey** - Children's Hospital of Philadelphia; University of California Los Angeles; Chuanli Li - Cincinnati Children's Hospital Medicine
- **Kathy Mathews** - University of Iowa; Sub Subramony - University of Florida; Chad Hoyle – Ohio State University; Grace Yoon - Sick Kids; Martin Delatycki - Murdoch Children's Research Institute
- **Theresa Zesiewicz** - University of South Florida
- **Christian Rummey**, **Biostatistician working on Collaborative Clinical Research Network in FA data set
- **David Lynch** - Children's Hospital of Philadelphia; University of California Davis; Southwester

## Grant Description

- **Center of Excellence: Drug Discovery**
- **Development of Oligonucleotide Activators of FXN Expression**
- **Identification of FXN-increasing drug targets by CRISPR-mediated mutagenesis**
- **2016-2017 Bronya J. Keats International Research Collaboration Award:** Drug rescue of frataxin-dependent neural and cardiac pathophysiology in FA models.
- **Suppressing the iron/sphingolipid/PDK/Mef2 pathway implicated in FA for therapeutic evaluation**
- **Evaluating Novel Capsid Engineered for Efficient CNS Transduction, as Frataxin Gene Deliver Vehicles**
- **Identification of FXN-increasing drug targets by CRISPR-mediated mutagenesis**
- **2016-2017 Bronya J. Keats International Research Collaboration Award:** Is FXN DNA Methylation a determinant of response to HDAC inhibitor treatment in Friedreich ataxia?
- **Identification of genetic factors involved on FXN transcriptional silencing mediated by the GAA repeat expansion**
- **Structural dynamics and the consolidation of protein function in protein complexes involved in the biosynthesis of iron-sulfur clusters: Quaternary addition of small Trojan tutor proteins**
- **Studying the role of brown fat in Friedreich's ataxia**
- **Functional analysis of primary sensory neurons and ( proprio) sensory pathology in Friedreich's ataxia**
- **Endurance and resistance exercise mitigate Friedreich's ataxia**
- **Development of a novel iPSC-derived neuronal cell model for Friedreich's ataxia: generation of stable doxycycline-inducible expression of progerin in FRDA neuronal cells (Fellowship: Baohu Ji, PhD)**
- **Longitudinal MR Imaging and Spectroscopy at 9.4T in a Conditional Mouse Model of FA**
- **The pathogenesis of Friedreich ataxia**
- **Collaborative Clinical Research Network in FA - Clinical Site Activity for Natural History and Biorepository**
- **Collaborative Clinical Research Network in FA - University of South Florida site activity, Symposium, Biomarker and Clinical Research**
- **Center of Excellence - Translational & Clinical Research**

## Type of Research

- Basic/Translational
- Translational
- Clinical
- Translational/Clinical
- Clinical
- Clinical
- Clinical
- Clinical
- Clinical
- Clinical
- Translational/Clinical
- Translational/Clinical
- Clinical
- Clinical
- Clinical
- Clinical

## Co-funding

- Cure FA Foundation & Hamilton & Finneran Family
- FARA Australia
- FARA Ireland
- Cure FA Foundation
- Cure FA Foundation & Hamilton & Finneran Family
- Cure FA Foundation
| Outcome Measures & Biomarkers                                                                 | Clinical                                                                 | Translational                                                                 | Cure FA Foundation & Hamilton & Finneran Family |
|------------------------------------------------------------------------------------------------|---------------------------------------------------------------------------|------------------------------------------------------------------------------|-------------------------------------------------
| Pierre Gilles Henry & Christophe Lenglet - University of Minnesota                             | Early and Longitudinal Assessment of Neurodegeneration in the Brain and Spinal Cord in Friedreich's Ataxia |                               |                                                 |
| Matthew Hirschey - Duke University                                                             | Protein biomarkers in FRDA cardiomyopathy to monitor disease progression and therapeutic efficacy |                               |                                                 |
| Manuela Corti - University of Florida                                                          | Clinical outcome measures of efficacy in the treatment of Friedreich's ataxia |                               |                                                 |
| Ian Blair - University of Pennsylvania                                                        | Center of Excellence - Metabolic Biomarkers                                 |                               |                                                 |
| Pierre Gilles Henry - University of Minnesota                                                 | Measurement of TCA Cycle Rate in the Dentate Nucleus in Friedreich's ataxia |                               | Cure FA Foundation                             |
| David Lynch - Children's Hospital of Philadelphia; Massimo Pandolfo - Erasme University Hospital| Biomarker Consortium: Neurophysiologic Biomarkers in FA                    |                               | Voyager Therapeutics                           |
| David Herrmann - University of Rochester                                                       | Biomarker Consortium: In vivo confocal imaging of Meissner's Corpuscles as a biomarker in FA |                               | Voyager Therapeutics                           |
| Martin Delatycki - Murdoch Children's Research Institute, Melbourne, Australia; Theresa Zesiewicz - University of South Florida | Biomarker Consortium: Longitudinal gait and balance measurement |                               | Voyager Therapeutics & Agilis                  |

| Cardiac Research                                                                               | Clinical                                                                 | Translational                                                                 | Cure FA Foundation & Hamilton & Finneran Family |
|------------------------------------------------------------------------------------------------|---------------------------------------------------------------------------|------------------------------------------------------------------------------|-------------------------------------------------
| Martin Delatycki - Murdoch Children's Research Institute, Melbourne Australia; Kim Lin - Children's Hospital of Philadelphia | Interstitial fibrosis, the renin-angiotensin-aldosterone system and biomarkers in the cardiac disease of Friedreich ataxia |                               | FARA Australia                                  |
| Kim Lin - Children's Hospital of Philadelphia                                                  | Center of Excellence - cardiac research and educational                   |                               | Cure FA Foundation & Hamilton & Finneran Family |