

2018 FARA Meeting on Biomarkers and Outcome Measures

Tampa, Florida

Friday, September 28th 8:30 am – 5:30 pm

Introduction and Welcome

8:30- 8:45	FARA Charles Lockwood, MD, MHCM (Dean, <i>USF Health Morsani College of Medicine</i>)
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Session 1

Biomarkers for monitoring frataxin and downstream metabolic changes

Moderator: David Lynch, MD, PhD (*Children's Hospital of Philadelphia*)

8:45-9:05	Ian Blair, PhD (<i>University of Pennsylvania Perelman School of Medicine</i>) Erythrocyte frataxin: a new N-terminally acetylated extra-mitochondrial proteoform of frataxin
9:05-9:25	Sanjay Bidichandani, MBBS, PhD (<i>University of Oklahoma College of Medicine</i>) Hypermethylation of the FXN gene as a predictive biomarker of drug response for HDAC inhibitor treatment in Friedreich ataxia
9:25-9:35	Pierre-Gilles Henry, PhD (<i>University of Minnesota</i>) Measurement of the TCA cycle rate in the dentate nucleus in Friedreich's Ataxia
9:35-9:55	Marek Napierala, PhD (<i>University of Alabama at Birmingham</i>) Targeted Proteomics Screen to Identify FRDA Biomarkers
9:55-10:15	Shana McCormack, MD, MTR (<i>Children's Hospital of Philadelphia</i>) Muscle mitochondrial oxidative phosphorylation capacity and whole body glucose and lactate metabolism in Friedreich's Ataxia.
10:15-10:35	Hélène Puccio, PhD (<i>Institut de Génétique et de Biologie Moléculaire et Cellulaire</i>) One-carbon metabolism and amino acid dysregulation as potential biomarkers for FA
10:35- 10:50	Session 1 discussion and summary
10:50-11:10	Coffee break

Session 2

Monitoring and pharmacodynamics biomarkers in the heart

Moderator: R. Mark Payne, MD (*Indiana University School of Medicine*)

11:10-12:00	Barry Byrne, MD, PhD (<i>University of Florida</i>), Marcondes França, MD, PhD (<i>University of Campinas</i>), Kim Lin, MD (<i>Children's Hospital of Philadelphia</i>) Panel discussion of use of cardiac MRI as a monitoring or pharmacodynamic biomarker for FA cardiomyopathy
12:00-12:10	Kim Lin, MD (<i>Children's Hospital of Philadelphia</i>) A Serum Biomarker Panel Reveals Evidence of Fibrosis and Correlation with MRI-based Cardiac Phenotype in a Cross-Sectional Analysis of Asymptomatic Friedreich Ataxia Patients

Late Breaking Abstract

- 12:10-12:20 **Kristin Obrochta, PhD** (*Biomarin Pharmaceutical Inc*) - Neurofilament-Light chain and cardiac Troponin are circulating biomarkers of neurological and cardiac disease pathology in Friedreich's Ataxia for FA cardiomyopathy
- 12:20-12:30 Session 2 discussion and summary
- 12:30-1:45 Lunch Break and poster session (posters will be up throughout the day)

Session 3

Monitoring and pharmacodynamics biomarkers in the nervous system

Moderator: **Steve Hersch, MD, PhD** (*Mass General Hospital*)

- 1:45- 2:45 **Marcondes França, MD, PhD** (*University of Campinas*), **Ian Harding, MD** (*Monash University*), **Pierre-Gilles Henry, PhD** (*University of Minnesota*), **Christophe Lenglet, PhD** (*University of Minnesota*) Panel discussion of use of MRI as a monitoring or pharmacodynamic biomarker for FA neurological progression.
- 2:45-3:05 **Peter Creigh, MD** (*University of Rochester Medical Center*) Longitudinal evaluation of in-vivo reflectance confocal microscopy of Meissner's corpuscles and other conventional peripheral nerve measures as biomarkers in Friedreich's Ataxia
- 3:05-3:25 **Massimo Pandolfo, MD** (*Université Libre de Bruxelles*) – Exploring the proprioceptive system in FA by MEG
- 3:25-3:40 Session 3 discussion and summary
- 3:40-4:00 Coffee break

Session 4

Functional outcome measures

Moderator: **Sub Subramony, MD** (*University of Florida*)

- 4:00-4:20 **Sarah Milne, PhD** (*Murdoch Childrens Research Institute*) Longitudinal gait and balance outcome measurement in individuals with Friedreich ataxia
- 4:20-4:40 **Louise Corben, PhD** (*Murdoch Children's Research Institute*) Validation of an instrumented measure of upper limb function in Friedreich ataxia.
- 4:40-5:00 **Adam Vogel, PhD** (*The University of Melbourne*) Quantifying objective analysis of speech in a supervised statistical machine learning framework: data across 10 years of recordings in Friedreich ataxia
- 5:00-5:15 Session 4 discussion and summary
- 5:15– 5:30 **Steve Hersch, MD, PhD** (*Mass General Hospital*) Summary and state of the FA biomarker field