

AGENDA

Thursday - May 5th 2011

12:00-14:00 – Registration - IGBMC

13:45-14:00 - Welcome Address

14:00-16:00 - Session 1: Clinical Discovery & Impact

Session Co-Chairs: Massimo Pandolfo and Mark Payne

Introduction – 5 min

Miriam Cnop (invited) 20 min; 5-10 Q&A	Pancreatic β cell dysfunction and insulin resistance contribute to diabetes in Friedreich's ataxia
Lauren Seyer 10 min; 3 min Q&A	Optical Coherence Tomography in Friedreich Ataxia
Louise Corben 10 min; 3 min Q&A	Are impairments in planning and online control of movement in Friedreich ataxia an indication of reduced cerebellar-cortico connectivity?
Arnie Koeppen 10 min; 3 min Q&A	Friedreich's ataxia: Iron, copper, and zinc in the dentate nucleus
Eric Deutsch 10 min; 3 min Q&A	Diagnostic utility of a rapid, noninvasive immunoassay for frataxin in atypical patients
Devin Oglesbee 10 min; 3 min Q&A	Development and Validation of a High-Throughput, Quantitative, Luminex Immunoassay for Frataxin in Whole Blood or Dried Blood Spots: An Assay for Newborn Screening, Diagnosis, and Treatment Monitoring
Stéphane Schmucker 10 min; 3 min Q&A	Identification of an atypical Friedreich ataxia patient with no GAA expansion but with a homozygous point mutation in the mitochondrial targeting sequence of frataxin

Wrap-up & discussion – 5 min

16:00-18:00 - Session 2: Pathways Impacted in FA

Session Co-Chairs: Tracey Rouault & Pierre Rustin

Introduction – 5 min

Armando Moreno-Cermeño 10 min; 3 min Q&A	Metabolic rearrangements in conditional yeast frataxin mutant cells
Marcia Haigis 10 min; 3 min Q&A	Role of mitochondrial SIRT3 in Friedreich Ataxia
Robert Schoenfeld 10 min; 3 min Q&A	Behavioral, oxidative and neuritic defects and microarray of the Friedreich's Ataxia Mouse Model YG8
Gregory Wagner 10 min; 3 min Q&A	Cardiac mitochondrial proteins are hyperacetylated in a mouse model of Friedreich's Ataxia
Alain Martelli (invited) 20 min; 5-10 min Q&A	Consequences of Iron Regulatory Protein 1 activation in mouse models of Friedreich's ataxia
Marek Napierala (invited) 20 min; 5-10 min Q&A	Crosstalk between microRNAs and iron metabolism in pathogenesis of Friedreich's ataxia.

Wrap-up & discussion – 5 min

18:00 – 18:45 - Keynote Lecture: Mitochondrial dynamics in neurodegeneration: what we have learned from the ALS models.

Giovanni Manfredi, MD, PhD, Professor of Neurology and Neuroscience, Weill Medical College of Cornell University, New York, NY

Friday – May 6th 2011

8:30-10:30 - Session 3: Genetics, Epigenetics & Regulation (instability)

Session Co-Chairs: Richard Festenstein & Joel Gottesfeld

Introduction – 5 min

Ed Grabczyk (invited) 20 min; 5-10 min Q&A	Transcription-coupled GAA•TTC Expansion Via Mismatch Repair in Human Cells
Jintang Du (presented by J. Gottesfeld) 10 min; 3 min Q&A	Recapitulation of GAA•TTC Triplet Repeat Expansion in Friedreich's Ataxia iPSCs
Cihangir Yandim 10 min; 3 min Q&A	GAA-repeats induce heterochromatinisation, and proteasome-dependent degradation of stalled RNA-PolIII providing a novel way of upregulating <i>FXN</i> in Friedreich's Ataxia
Rodrigo Villaseñor 10 min; 3 min Q&A	Long intronic GAA repeats causing Friedreich ataxia impede transcription elongation
Karen Usdin 10 min; 3 min Q&A	<i>FXN</i> expression from unaffected and <i>FRDA</i> alleles
Dave Lynch 10 min; 3 min Q&A	Association of a Non synonymous SIRT6 polymorphism with improved neurological abilities in <i>FRDA</i>
Yogesh Chutake 10 min; 3 min Q&A	RNA-mediated transcriptional gene silencing of the <i>FXN</i> gene in Friedreich ataxia
Angela Castro 10 min; 3 min Q&A	Deficiency of CTCF-mediated chromatin insulation in Friedreich ataxia results in <i>FXN</i> transcriptional deficiency

Wrap-up & Discussion – 5 min

10:30 – 11:00 - Break

11:00 – 13:00 – Session 4: Frataxin Function

Session Co-Chairs: Grazia Isaya & Annalisa Pastore

Introduction – 5 min

David Barondeau (invited speaker) 20 min; 5-10 min Q&A	Frataxin-based activation of the human Fe-S assembly complex
Timothy Stemmler (invited speaker) 20 min; 5-10 min Q&A	Molecular details regarding Frataxin's roles in the activation of eukaryotic Fe-S cluster biosynthesis
Stéphane Schmucker 10 min; 3 min Q&A	The interaction of mammalian frataxin with a preformed <i>iscu/nfs1/isd11</i> iron-sulfur cluster assembly complex defines the essential function of frataxin in vivo
Clara Iannuzzi 10 min; 3 min Q&A	A Mössbauer and resonance Raman study of the role of CyaY in iron sulfur cluster assembly on the <i>E. coli</i> IscU scaffold protein
Andrew Dancis 10 min; 3 min Q&A	Mutation in ISU bypasses frataxin

Eva-Christina Ahlgren 10 min; 3 min Q&A	The importance of the N-terminus: Structural insights into human frataxin in monomeric and oligomeric forms
Grazia Isaya 10 min; 3 min Q&A	Oligomeric frataxin protects NFS1 from oxidative inactivation during Fe-S cluster assembly

Wrap-up & Discussion – 5 min

13:00 – 14:30 - Lunch and Posters

14:30 – 17:00 Session 5: Models of FA

Session Co-Chairs: Mark Pook & H  l  ne Puccio

Introduction – 5 min

Mirella Dottori (invited) 15 min; 5 min Q&A	Using induced pluripotent stem cells to establish neuronal cellular models of Friedreich Ataxia
Alice P��bay 10 min; 3 min Q&A	FRDA-induced pluripotent stem cell potentials for differentiation
Greg Dusting 10 min; 3 min Q&A	Stem Cell Signaling and Generation of Heart Tissue – Potential for cardiac repair and test tissue for new therapeutics for Friedreich Ataxia
Marie Wattenhofer- Donz�� 10 min; 3 min Q&A	Development of new models for Friedreich ataxia using induced pluripotent stem cells
Sherman Ku 10 min; 3 min Q&A	<i>In vitro</i> differentiation of FRDA induced pluripotent stem cells to neurons provides a cellular model for small molecule therapeutic development
Satyan Chintawar 10 min; 3 min Q&A	Generation of Purkinje cells from human iPS cells
Nadia Soussi-Yanicostas 10 min; 3 min Q&A	Frataxin depletion in zebrafish embryo, toward a novel animal model for Friedreich ataxia
Sirena Soriano 10 min; 3 min Q&A	Study of the metal content in a Drosophila model of Friedreich ataxia
Chiranjeevi Sandi 10 min; 3 min Q&A	Prolonged Treatment with Pimelic <i>o</i> -Aminobenzamide HDAC Inhibitors Ameliorates the Disease Phenotype of a Friedreich Ataxia Mouse Model
Michele Lufino (invited) 20 min; 5-10 min Q&A	Drug screening based on <i>FRDA</i> genomic-reporter fusion vectors identifies two candidate molecules able to up-regulate FXN expression

Wrap-up & Discussion – 5 min

17:00 – 17:45 Keynote Lecture: Human Pluripotent Stem Cells: The State of the Art

Martin Pera, *W.M. Keck Professor of Cell and Neurobiology, Foundation Director, Eli and Edythe Broad Center for Regenerative Medicine and Stem Cell Research, University of Southern California*

*Program Leader, Stem Cells Australia
Chair of Stem Cell Science
University of Melbourne*

17:45 – 19:30 Cocktails and Posters

Saturday – May 7th 2011

8:30 – 10:30 – Session 6: Drug Discovery & Development

Session Co-Chairs: Sidney Hecht & Robert Wilson

Introduction – 5 min

Rob Wilson (invited speaker) 20 min; 5-10 min Q&A	High-throughput and Secondary Drug Screening Assays for Friedreich Ataxia
Joe Sarsero 10 min; 3 min Q&A	Pharmacological screening for the therapy of Friedreich ataxia
Sunil Sahdeo 10 min; 3 min Q&A	A novel cell-based model for high-throughput screening of potential Friedreich's therapeutics
Ping Kei Chan 10 min; 3 min Q&A	GAA-repeat induced heterochromatinisation in intron 1 of the Frataxin gene can be decondensed by HDAC inhibitor (HDACi), Nicotinamide
Vincent Jacques 10 min; 3 min Q&A	Second generation stable brain-penetrant HDAC inhibitors for Friedreich's Ataxia
Omar Khmour 10 min; 3 min Q&A	Multifunctional radical quenchers for the treatment of Friedreich's ataxia
Javier Diaz-Nido 10 min; 3 min Q&A	Brain-derived neurotrophic factor (BDNF) inhibits neurodegeneration triggered by frataxin gene silencing

Wrap-up & Discussion – 5 min

10:30 – 12:00 Break / Posters

12:00 – 13:00: Session 7: Clinical Trials

Session Co-Chairs: Petra Kaufmann & David Lynch

Introduction – 5 min

Francesco Saccà 10 min; 3 min Q&A	Delayed effect of Epoetin alfa on frataxin production in Friedreich's ataxia
Antonio Piga 10 min; 3 min Q&A	Effects of Deferiprone on cardiac disease in patients with Friedreich's ataxia
Fernando Tricta 10 min; 3 min Q&A	A six-month double-blind randomized, placebo-controlled study investigating the safety and tolerability of Deferiprone in subjects with Friedreich's ataxia
Thomas Meier (presented by D. Lynch) 10 min; 3 min Q&A	Efficacy of idebenone (Catena®) in pediatric patients with FRDA: data from a 6 month controlled study (IONIA) followed by a 12-month open label extension study (IONIA-E)

Wrap-up & Discussion – 5 min

13:00 – 14:30 Lunch and Posters

14:30 – 16:00 Session 8: Biomarkers & Functional Measures & Clinical Design

Session Co-Chairs: Martin Delatycki & Alexandra Durr

Introduction – 5 min

Petra Kaufmann (invited) 20 min; 5 min Q&A	Implementation data, endpoints, biomarkers
Martin Delatycki 10 min; 3 min Q&A	Measuring the progression of Friedreich ataxia
Cecilia Marelli 10 min; 3 min Q&A	Annual changes in Friedreich's Ataxia evaluated by SARA (Scale for the Assessment and Rating of Ataxia) are independent of disease severity
Günther Metz 10 min; 3 min Q&A	Rating the disease state of FRDA patients by ICARS: An analysis using a large database of 600 patients
David Lynch 10 min; 3 min Q&A	Measures of Neurologic progress in FRDA: The experience of the CCRNFA
Marguerite Evans-Galea 10 min; 3 min Q&A	FXN methylation in Friedreich ataxia reveals insight into disease mechanism and new tools for the clinic

Wrap-up & Discussion – 5 min

16:00 – 16:45 Keynote: Gene therapy for intractable CNS diseases

Patrick Aubourg, MD, *Director of Inserm 745 Unit "Genetics and biotherapies of degenerative and proliferative diseases of the nervous system", Saint-Vincent de Paul Hospital, Paris*