Programme overview

Wednesday 25 March

11:00	–	13:00	Registration and light lunch
13:00	–	13:15	Welcome/ housekeeping
13:15	–	17:45	**Session 1: New genes and developments in diagnosis of the ataxias**
18:30	–	20:15	**Poster sessions 1, 2 and 3** and welcome drinks
20:15	–		Dinner and Prof Sophie Scott (UCL) on the neurology of laughter

Thursday 26 March

8:30	–	12:45	**Session 2: Genetic and molecular mechanisms of the ataxias**
12:45	–	14:00	Lunch
14:00	–	16:00	**Session 3a: Cellular and animal models of Friedreich's ataxia**
14:00	–	15:45	**Session 3b: Cellular and animal models of other ataxias**
16:15	–		Coaches leave for Oxford
17:15	–	19:15	Free time
19:15	–	19:45	Drinks reception at Oxford Town Hall
19:45	–		Dinner in Oxford Town Hall
22:45	–		Coaches leave for Beaumont Estate

Friday 27 March

8:30	–	12:00	**Session 4: Cellular and systemic pathways**
12:00	–	14:00	Lunch and **poster sessions 4 and 5**
14:00	–	18:00	**Session 5: Drug discovery and emerging therapeutic strategies**
18:00	–	19:30	**Poster sessions 6 and 7**
19:40	–		Conference Dinner and pre-dinner talk from Eurordis

Saturday 28 March

8:30	–	12:45	**Session 6: Biomarkers and functional measures**
12:45	–	14:00	Lunch
14:00	–	17:30	**Session 7: Clinical trials and trial design**
17:30	–	17.40	Close of conference

Detailed programme

Wednesday 25 March

13:00 Welcome/ housekeeping

13:15 Helen Kearney: *My life with ataxia*

Session 1 - New genes and developments in diagnosis of the ataxias

Chairs: Andrea Nemeth (University of Oxford and Oxford NHS Trust, UK) and Michel Koenig (Institut Universitaire de Recherche Clinique, Montpellier, France)

Invited speakers

13:25 Michel Koenig (Institut Universitaire de Recherche Clinique, Montpellier, France):
Recessive ataxia by partial loss of function: a common theme

13:55 Andrea Nemeth (University of Oxford and Oxford NHS Trust, UK):
The future of diagnostics in ataxias and implications for therapeutics

Selected presentations

14:25 Marie Coutelier (INSERM, Paris, France):
GRID2 mutations span from congenital to mild adult onset cerebellar ataxia

14:45 Marios Hadjivassiliou (Royal Hallamshire Hospital, Sheffield, UK):
The aetiology of progressive cerebellar ataxia: Prospective evaluation of 1234 patients

15:05 Rebekah Jobling (The Hospital For Sick Children, Toronto, Canada):
PMPCA mutations cause abnormal mitochondrial protein processing in patients with non-progressive cerebellar ataxia

15:25 Break

15:55 Stefania Magri (Fondazione IRCCS, Istituto Neurologico Carlo Besta, Italy):
A comprehensive NGS gene panel for the genetic diagnosis of hereditary cerebellar ataxias

16:15 Rebecca Schule (Hussman Institute for Human Genomics, University of Miami,USA):
Collaborative gene identification strategies in hereditary ataxias

16:35 Matthis Synofzik (Hertie-Institute for Clinical Brain Research, Germany):
Exome and panel sequencing in a large cohort of early-onset ataxias: novel genes and genetic mechanisms

16:55 Dineke Verbeek (University of Groningen, Netherlands):
The identification of novel spinocerebellar ataxia disease genes using next generation sequencing approaches

17:15 Margit Burmeister (University of Michigan, USA):
Surprising results from next generation sequencing: Summary of >50 ataxia exomes sequenced

17:35-17:45 Close of session

Thursday 26 March

Session 2 – Genetic and molecular mechanisms of the ataxias

Chairs: Sanjay Bidichandani (University of Oklahoma, US) and Olaf Riess (University of Tubingen, Germany)

Invited speakers

8:30 Sanjay Bidichandani (University of Oklahoma, USA):
Epigenetic promoter silencing in Friedreich ataxia

9:00 Steve Jackson (University of Cambridge, UK):
Assembly and disassembly of protein complexes at sites of DNA damage: mechanistic insights and therapeutic applications

Selected presentations

9:30 Eleonora Di Gregorio (University Hospital, Torino, Italy):
Unraveling molecular pathogenesis of SCA38, a novel autosomal dominant ataxia

9:45 Conceição Bettencourt (UCL Institute of Neurology, UK):
Transcriptome-wide analysis of the human brain as a route to SCA pathways and biomarkers

10:00 Marguerite Evans-Galea (Murdoch Childrens Research Institute, Melbourne, Australia):
The impact of compound heterozygous mutations in FXN on clinical outcome in Friedreich ataxia: insights from frataxin structure and function

10:15 Natalia Gromak (Sir William Dunn School of Pathology, University of Oxford, UK):
R-loop function in pathology of Friedreich ataxia and implications for other expansion disorders

10:30 Break

11:00 Arnulf Koeppen (VA Medical Center Albany, NY, USA):
Cardiac remodeling in Friedreich ataxia

11:15 Marek Napierala (University of Alabama at Birmingham, US):
Expanded GAA repeats induce transcriptional silencing restricted to the FXN locus and decrease the elongation rate through the FXN gene

11:30 Thorsten Schmidt (University of Tuebingen, Netherlands):
Targeting the intracellular localization of ataxin-3 as a road to therapy of Spinocerebellar Ataxia Type 3 (SCA3)

11:45 Ana M Silva (University of Oxford, UK):
Expanded GAA repeats impair frataxin gene expression and promote repositioning to the nuclear periphery at single-cell level

12:00 Ana Teresa Simões (University of Coimbra, Portugal):
Identification of the calpain cleavage sites in ataxin-3 protein

12:15 Michael Themis (Brunel University, UK):
Lentivirus mediated FXN gene delivery restores genome stability and DNA damage repair potential in human and mouse FRDA fibroblasts

12:30-12:45 Close of session

12:45-14:00 Lunch

Session 3a – Cellular and animal models of Friedreich's ataxia

Chairs: Helene Puccio (Inserm, Illkirch, France) and Michele Lufino (University of Oxford, UK)

Invited speakers

14:00 Helene Puccio (Inserm, Illkirch, France):
Progress in the development of new mouse and cell models for deciphering the neurological defects in Friedreich Ataxia and testing therapies

14:25 Michele Lufino (University of Oxford, UK):
Cell and animal FXN genomic reporter models of Friedreich's ataxia

Selected presentations

14:45 Vijayendran Chandran (University of California, Los Angeles, US):
Inducible and reversible frataxin knock-down mouse model for Friedreich's ataxia

15:00 Simona Donatello (University libre de Bruxelles, Belgium):
Induced pluripotent stem cell-derived neurons from Friedreich's ataxia patients have a cellular phenotype that can be reversed by frataxin inducers

15:15 Jordi Magrane (Brain and Mind Research Institute, New York, US):
Analysis of mitochondrial dynamics in cultured sensory neurons and in in vivo mouse models of Friedreich's Ataxia

15:30 Juan Antonio Navarro Langa (University of Regensburg, Germany):
Impact of frataxin-deficiency on mitochondrial dynamics

15:45 Jose V. Llorens (University of Uppsala, Sweden):
A new Drosophila melanogaster model to identify genetic modifiers of transcriptional repression caused by GAA expansion in FXN

16:00 Close of session

16:15 Coaches leave for Oxford

17:15 – 19:15 Free time

19:15 – 19:45 Drinks reception at Oxford Town Hall

19:45 Dinner in Oxford Town Hall
There will be presentations by Jeffrey Sherman (Horizon Pharma) who are sponsoring this event and Kyle Bryant (FARA) who has Friedreich's ataxia and is the Founder and director of Ride ataxia.

Session 3b – Cellular and animal models of other ataxias

Chair: Michel Koenig (Institut Universitaire de Recherche Clinique, Montpellier, France)

Invited speaker

14:00 Olaf Riess (University of Tubingen, Germany):
Deciphering the pathogenesis of SCA3 using animal models

Selected presentations

14:30 Olga Baron (King's College London, UK):
Relevance of autophagy in neurodegeneration in DRPLA

14:45 Cecilia Mancini (University of Torino, Italy):
SCA28-Knockin mouse model: severe impairment of mitochondrial fission/fusion network in MEF cells

15:00 Isabel Onofre (Center for Neuroscience and Cell Biology, Coimbra, Portugal):
Characterization of a human Machado-Joseph disease neuronal cell model derived from Induced Pluripotent Stem Cells

15:15 Liliana Santos (University of Minho, Braga, Portugal):
Determinants of neuron-specific pathogenesis in Machado-Joseph Disease: study in a C. elegans model

15:30 Natascia Ventura (Leibniz Institute for Environmental Medicine and the Heinrich Heine University of Duesseldorf, Germany):
In vivo phenotypic-based screening to identify suppressors of mitochondrial-associated ataxias

15:45 Close of session

16:15 Coaches leave for Oxford

Friday 27 March

Session 4 – Cellular and systemic pathways

Chairs: Giovanni Manfredi (Cornell University, USA) and Henry Paulson (University of Michigan, USA)

Invited speakers

8:30 Kamran Khodakhah (Albert Einstein College of Medicine, USA):
Aberrant cerebellar output in ataxia – a common theme

9:00 Henry Paulson (University of Michigan, USA):
Why does polyglutamine expansion cause dominantly inherited ataxia? Lessons from SCA3

Selected presentations

9:30 David Alsina (Universitat de Lleida, Spain):

Yeast Flavohemoglobin (YHB1) and Nitric Oxide, new players in frataxin deficient yeast

9:50 Javier Diaz-Nido (Universidad Autonoma de Madrid, Spain):
DNA repair deficit and neuroinflammation as potential contributors to the physiopathology of Friedreich's ataxia

10:10 Break

10:40 Sofia Esteves (Life and Health Sciences Research Institute (ICVS), Portugal)*:*
Citalopram treatment ameliorates motor impairment and suppresses ataxin-3 aggregation in a Machado-Joseph disease mouse model

11:00 Angelical Martin (Duke University, USA):
The role of acetylation in the pathogenesis of Friedreich's ataxia

11:20 Joaquim Ros (Universitat de Lleida, Spain):
Viability of frataxin-deficient dorsal root ganglia neurons is recovered by calcium chelators and mitochondrial pore inhibitors

11:40 Amanda Stram (Indiana University School of Medicine, USA):
Mitochondrial protein hyperacetylation is associated with early diastolic dysfunction in a model of Friedreich's ataxia hypertrophic cardiomyopathy

12:00 Close of session

12:00-14:00 Lunch and poster sessions 4 and 5

Session 5 – Drug discovery and emerging therapeutic strategies

Chairs: Roberto Testi (University of Rome Tor Vergata, Italy) and Rob Wilson (University of Pennsylvania, USA)

14:00 Memorial to the late Earl Giller to whom this session is dedicated – Ron Bartek (FARA)

Invited speaker

14:05 Joel Gottesfeld (Scripps research Institute, USA):
Mechanism of action of 2-aminobenzamide HDAC inhibitors in reversing gene silencing in Friedreich's ataxia

Selected presentations

14:35 Hagar Greif (Bioblast Pharma Ltd., Tel Aviv, Israel):
BB-FA (TAT-MTS(cs)-Frataxin) exhibits promising potential as a protein replacement drug candidate for Friedreich's Ataxia

14:55 Kevin Kemp (University of Bristol, UK):
The neuroprotective and neuroregenerative properties of bone marrow stem cell mobilising drugs in Friedreich's ataxia

15:15 Fatih Ozsolak (RaNA Therapeutics, Cambridge, US):
Stabilization of FXN mRNA using oligonucleotides for the treatment of Friedreich's ataxia

15:35 Break

15:55 Giorgio Casari (San Raffaele Scientific Institute, Milan, Italy):
Genetic and pharmacological rescues of spinocerebellar ataxia in the SCA28 model open to human therapy

16:15 Luis Pereira de Almeida (University of Coimbra, Coimbra, Portugal):
Transplantation of cerebellar neural stem cells alleviates motor coordination and neuropathological deficits of a transgenic mouse model of Machado-Joseph disease

16:35 Alessandra Rufini (University of Rome "Tor Vergata, Italy):
Therapeutic strategies to prevent the ubiquitin/proteasome-dependent degradation of frataxin

16:55 Jacques P. Tremblay (Laval University, Quebec, Canada):
An AAV9 coding for frataxin clearly improved the symptoms and prolonged the life of Friedreich ataxia mouse models

17:15 Joana Duarte-Nueves (Center for Neuroscience and Cell Biology, University of Coimbra, Portugal):
Therapeutic role of Neuropeptide Y in mouse models of Machado-Joseph Disease

17:35 Florence Malisan (Department of Biomedicine and Prevention, University of Rome "Tor Vergata", Italy):
Src inhibitors modulate frataxin protein levels

17:55-18:00 Close of session

18:00-19:30 Poster sessions 6 and 7

19:40 Conference dinner and pre-dinner talk by Avril Daly (Eurordis)

Saturday 28 March

8:30 Harriet Bonney: *The patient in the chair*

Session 6 – Biomarkers and functional measures

Chairs: David Lynch (University of Pennsylvania, US) and Bernard Ravina (Voyager Therapeutics, USA)

Invited speaker

8:40 David Lynch (University of Pennsylvania, USA):
Biomarkers in Friedreich ataxia

Selected presentations

9:10 Ian Blair (University of Pennsylvania, Philadelphia, US):
Platelet biomarkers of metabolic disturbances in Friedreich's ataxia

9:25 Martin Delatycki (Murdoch Childrens Research Institute, Parkville, Australia):
A longitudinal study of the Friedreich Ataxia Impact Scale

9:40 Zofia Fleszar (Hertie-Institute for Clinical Brain Research, Tübingen, Germany):
PreAtaxia: characterizing ataxia-specific movement changes at a preclinical stage

9:55 Louise Corben (Monash University, Melbourne, Australia):
Abnormal brain function and connectivity in cerebello-cerebral circuits underlying cognitive function in Friedreich ataxia: The IMAGE-FRDA study

10:10 Pierre-Gilles Henry (University of Minnesota, US):
MRS and diffusion MRI of the spinal cord in Friedreich's Ataxia

10:25 Break

10:55 Marinela Vavla (E. Medea Scientific Research Institute, Conegliano and Bosisio Parini, Italy):
Potential neuroimaging biomarkers validated in Friedreich's ataxia: DTI and functional magnetic resonance findings

11:10 Mafalda Raposo (Centre of Research in Natural Resources, University of the Azores, Portugal):
Novel candidate blood-based transcription biomarkers of spinocerebellar ataxia type 3 (SCA3)

11:25 Louise Corben (Physiotherapy Department, Monash Health, Cheltenham, Australia):
Sensitivity of spatiotemporal gait parameters in Friedreich ataxia

11:40 Michael Parkinson (UCL Institute of Neurology, UK):
OCT in Diagnosing ARSACS

11:55 R. Mark Payne (Indiana University School of Medicine, Indianapolis, US):
Fatty acid oxidation is disrupted in the FRDA heart

12:15 Jorg B. Schulz (Department of Neurology, RWTH Aachen University, Germany):
Biological and clinical characteristics of the European Friedreich Ataxia Consortium for Translational Studies (EFACTS): cross-sectional analysis of baseline data

12:30 Martin Delatycki (Murdoch Childrens Research Institute, Melbourne, Australia):
The views of individuals with, and parents of individuals with Friedreich ataxia regarding pre-symptomatic testing of minors

17:45 Close of session

12:45-14:00 Lunch

Session 7 – Clinical trials and trial design

Chairs: Paola Giunti (University College London/UCL Hospital, UK) and Massimo Pandolfo (Université Libre de Bruxelles)

Selected presentation

14:00 Richard Festenstein (Imperial College London, UK):
Reversing FXN gene silencing in vivo in humans -towards a disease-modifying therapy?

Invited speakers

14:20 Massimo Pandolfo (Université Libre de Bruxelles)
Clinical trials in Friedreich's ataxia

14:35 Interview with Pavel Balabanov (European Medicine Agency, London, UK)

Selected presentations

15:10 Paola Giunti (University College London, Institute of Neurology, UK):
Can sensorimotor processing abnormalities explain or contribute to balance impairment in cerebellar disease?

15:30 Break

15:55 Zohar Argov (Bioblast Pharma, Chief Medical Officer, Israel):
Double blind, randomized, controlled phase 3 trial of high dose IV trehalose (Cabaletta) for treatment of spinocerebellar ataxia 3 (SCA3)

16:15 Robert Molinari (Retrotope Inc., US):
An upcoming clinical trial testing the safety and efficacy of a stabilized polyunsaturated fatty acid in Friedreich's ataxia

16:35 Colin Meyer (Reata Pharmaceuticals, Chief Medical Officer, US):
Rationale and design of a clinical study of RTA 408 in patients with Friedreich's ataxia

16:55 Gino Cortopassi (UC Davis School of Veterinary Medicine, US):
Repurposed dyclonine for Friedreich's ataxia therapy

17:15 General discussion

17:30-17:40 Close of conference