Conference Agenda

Thursday 11th May

Richard Doll Lecture Theatre

8:30 Registration

9:00 Opening Remarks

Dame Kay Davies, Chair of Oxford Rare Disease Initiative, Dr Lee's Professor of Anatomy

9:20 Plenary Lecture – Antisense oligonucleotides to modify splicing in Duchenne muscular dystrophy and Spinal muscular atrophy

Francesco Muntoni, Chair of Paediatric Neurology, UCL

9:50 Session 1 – Neurological Diseases

Genetic and pathological investigations into human neurodegeneration
Henry Houlden, Consultant Neurologist, UCL and Head of Neurological GeCIP Domain, Genome England

Allosteric, non-inhibitory pharmacological chaperones for the treatment of lysosomal storage diseases
Richard Roberts, Director of Drug Discovery, Minoryx

SMN2 splicing modifiers in the treatment of Spinal Muscular Atrophy
Friedrich Metzger, Head of Discovery Rare Diseases, F. Hoffmann-La Roche

Novel genetic and mechanistic insights into rare cerebellar disorders
Esther Becker, Department of Physiology, Anatomy and Genetics, University of Oxford

11:10 Coffee Break

11:45 Plenary Lecture – Inherited Retinal Degeneration

Julian Howell, CMO, ReNeuron

12:15 Session 2 – Genomics and Genetic Approaches

Progress in understanding the genomic aetiology of osteoarthritis
Eleftheria Zeggini, Wellcome Trust Sanger Institute, Cambridge

DECIPHER- Mapping the Clinical Genome
Helen Firth, Consultant Clinical Geneticist, Cambridge University Hospitals Trust

Oxford Consortium for Single-Cell Biology (Capturing Biology, One Cell at a Time)
Rory Bowden, Oxford Genomics Centre, Wellcome Trust Centre for Human Genetics

13:15 Lunch

14:15 Plenary Lecture – Birmingham Rare Disease Network (Title TBC)

Timothy Barrett, Leonard Parsons Professor of Paediatrics and Child Health, University of Birmingham

14:45 Session 3 – Haematology and Immunology

Genetic haematopoitic failure syndromes
Inderjeet Dokal, Chair of Child Health, Queen Mary University London

Rare congenital pathologies of the thymus stroma
Georg Holländer, Department of Paediatrics, University of Oxford

Statistical approaches for identifying the genetic causes of rare diseases applied to bleeding and platelet disorders
Ernest Turro, University of Cambridge

15:45 Plenary Lecture – UK Biobank: a resource for genetic-epidemiological research

Rory Collins, BHF Professor of Medicine and Epidemiology, University of Oxford

16:15 Coffee Break

16:40 Plenary Lecture – Genetic studies in rare diseases can provide new drug targets to help meet severe clinical need – the otulin story

Duncan McHale, Vice President, Global Exploratory Development, UCB Pharma

17:10 Session 4 – Metabolic Diseases

Enzyme Replacement Therapy in AKU
Farid Khan, Chairman, Protein Technologies

Rationalizing substrate reduction therapy for pyridoxine-dependent epilepsy
Wyatt Yue, Associate Professor, Structural Genomics Consortium, University of Oxford

Conference Agenda
Friday 12th May

Richard Doll Lecture Theatre

8:30 Registration

9:00 Plenary Lecture – Title TBC
Christelle Perros-Huguet, Alexion Pharmaceuticals

9:30 Session 5 – Drug Repurposing
Using social finance to fund generic drug repurposing
Rick Thompson, Findacure

A new model of pursuing rare disease development: repurposing in a rare metabolic autoinflammtory disease - MKD.
Tauhid Ali, VP & Head of TAK-celerator, Takeda Pharmaceuticals

Repurposing saracatinib for FOP treatment
Alex Bullock, Structural Genomics Consortium, Oxford

10:30 Coffee Break

11:00 Plenary Lecture – Emerging Molecular Therapies for Inherited Metabolic and Related Diseases
Seng Cheng, Global Head of Research, Rare Diseases, Sanofi-Genzyme

11:30 Session 6 – Cardiovascular Medicine
Rare disease in Cardiology: Changing the paradigm
Perry Elliott, Professor in Inherited Cardiovascular Diseases, UCL

Metabolic substrate modification as a therapy for heart muscle diseases
Michael Frenneaux, Clinical Professor, University of East Anglia

12:15 Plenary Lecture – Becoming I'mpatient – Open Science platform enabling early-stage drug discovery by rare disease foundations
Wenhwa Lee, Programme Director, Disease Foundations Network Strategic Alliances

12:45 Closing Remarks and Poster Prize