Conference Agenda

Tuesday 10th September

Pembroke College, Oxford

16.00 **Registration**

17.00 **Opening Remarks**
Professor Matthew Wood, Director, Oxford-Harrington Rare Disease Centre

17.15 **Keynote Lectures**

The Harrington Discovery Institute
Jonathan Stamler, President, Harrington Discovery Institute

Gene Therapy for Retinal Diseases
Robert MacLaren, Professor of Ophthalmology, Nuffield Department of Clinical Neurosciences, University of Oxford

Gene therapy for Cystic Fibrosis
Eric Alton, Professor of Gene Therapy and Respiratory Medicine, National Heart & Lung Institute, Imperial College London

19.00 **Drinks and canapés reception**

Wednesday 11th September

9.00 **Session 1 Neurological Diseases**

Gene therapy in Duchenne Muscular Dystrophy and Spinal Muscular Atrophy
Laurent Servais, Professor of Paediatric Neuromuscular Diseases, Department of Paediatrics, University of Oxford

Title tbc
Thomas Voit, Director of the NIHR Great Ormond Street Hospital (GOSH) BRC, Professor and Honorary Consultant of Paediatrics at GOSH

Novel target and drug discovery in Friedreich's ataxia
Richard Wade-Martins, Professor of Molecular Neuroscience, Department of Physiology, Anatomy and Genetics, University of Oxford

Modelling early phase pathology in amyotrophic lateral sclerosis
Kevin Talbot, Professor of Motor Neuron Biology, Nuffield Department of Clinical Neurosciences, University of Oxford

Antisense oligonucleotides in neuromuscular disorders
Francesco Muntoni, Chair of Paediatric Neurology, UCL

Targeting a common pathway in spinocerebellar ataxia
Esther Becker, Associate Professor of Neurobiology, Department of Physiology, Anatomy & Genetics, University of Oxford

11.00 **Coffee**

Keynote Lecture – Catalyzing Translational Innovation
Chris Austin, Director, NIH National Center for Advancing Translational Sciences

12.30 **Lunch**

13.15 **Session 2 Metabolic Diseases**

Tay-Sachs: an inborn metabolic disease
Tim Cox, Department of Medicine, University of Cambridge

Title tbc
Wyatt Yue, Structural Genomics Consortium, University of Oxford

Human Genetics Aided Discovery of a New Metabolic Hormone
Atul Chopra, Investigator, Harrington Discovery Institute, University Hospitals, Cleveland

Searching for mitochondrial therapeutics
Bob Lightowlers, Professor of Molecular Neuroscience, University of Newcastle

14.35 **Session 3**

Developing new treatments for patients with inherited skin diseases
John McGrath, Professor of Molecular Dermatology, King's College London

Rare Bleeding disorders: the clinical benefits from genomics
Mike Laffan, Professor of Haemostasis and Thrombosis, Imperial College London

How to secure funding for development of a rare disease medicine?
Catriona Crombie, Philanthropic Fund Manager, LifeArc

15.35 **Coffee break**

15.55 **Session 4 Genomics and Genetic Approaches**

The 100,000 Genomes Project
Richard Scott, Clinical Lead, Rare Diseases, Genomics England

The Next Generation Childrens Project: rapid whole genome analysis of severely ill children
Lucy Raymond, Professor of Medical Genetics and Neurodevelopment, Department of Medical Genetics, University of Cambridge

Title tbc
Jenny Taylor, Wellcome Trust Centre for Human Genetics, University of Oxford

Neurogenetics and repeat expansion disorders
Henry Houlden, Consultant and Professor of Clinical Neurology and Neurogenetics, UCL

Inherited cardiomyopathies: ready for nucleic acid therapies?
Hugh Watkins, Radcliffe Professor of Medicine, University of Oxford

Title tbc
Tim Barrett, Leonard Parsons Professor of Paediatrics and Child Health, University of Birmingham

Monogenic disorders and inflammatory bowel disease
Holm Uhlig, Professor of Paediatric Gastroenterology, University of Oxford

Conference residential dinner

Thursday 12th September

Pembroke College, Oxford

8.30 **Registration and coffee**
9.00 **Session 5 Industry**

Rare is the new common: From Gaucher to Parkinson's disease
Pablo Sardi, Head of Lysosomal Storage and Metabolic Diseases Research, Sanofi

Pharmaceutical Development for Rare and Orphan Diseases
Steve Murray, VP Strategy and Innovation, Mallinckrodt Pharmaceuticals

Translational Research for Repositioning TAK063 in Fragile X Syndrome – A Potential New Treatment
Christine Charman, Senior Director, External Asset Lead, Centre for External Innovation, Takeda Pharmaceuticals

Exosome-based therapies for the treatment of rare genetic diseases
Per Lundin, Chief Operating Officer, Evox Therapeutics

Title tbc
Michael Panzara, Chief Medical Officer, Wave Life Sciences

Title tbc
Madhurima Benekareddy, Project Lead and Lab Head, Roche

Title tbc
Stuart Hughes, Vertex Pharmaceuticals

11.00 Coffee Break

11.20 Session 6 Haematology and Immunology

Emerging therapies for hemophilia and sickle cell anemia
Seng Cheng, CSO Rare Diseases, Pfizer

Title tbc
Bobby Gaspar, CSO, Orchard Therapeutics

The NIHR BioResource - Rare Diseases pilot study for the 100,000 Genomes Project
Willem Ouwehand, Professor of Experimental Haematology, University of Cambridge

Birmingham Rare Disease Network
David Adams, Director, NIHR Birmingham Biomedical Research Centre, University of Birmingham

13:20 Closing Remarks