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 Retinitis Pigmentosa
Robert MacLaren, Professor of Ophthalmology, Nuffield Department of Clinical Neurosciences, University of Oxford

Gene therapy for Cystic Fibrosis
Eric Alton, Chair in Gene Therapy, Faculty of 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

Title tbc- new drug targets 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

Title tbc
Atul Chopra, Investigator, Harrington Discovery Institute, University Hospitals, Cleveland

Title tbc
Bob Lightowlers, Professor of Molecular Neuroscience, University of Newcastle

14.35 **Session 3**

Title tbc Gene therapy in Dermatology
John McGrath, King's College London

Rare blood disorders
Mike Laffan, Professor of Haemostasis and Thrombosis, Department of Medicine, Imperial College London

LifeArc Philanthropic Fund for Rare Disease Translational Research
Catriona Crombie, Philanthropic Fund Manager, LifeArc

15.35 **Coffee break**

15.55 **Session 4 Genomics and Genetic Approaches**

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

Title tbc
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

Title tbc
Henry Houlden, Consultant Neurologist, UCL and Head of Neurological GeCIP Domain, Genome England

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

Title tbc
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**

Title tbc
Pablo Sardi, Rare and Neurological Diseases Therapeutic Area, Sanofi

Title tbc
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

Title tbc
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

Title tbc
Willem Owehand, 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**