

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, 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 Lightowers, 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 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

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

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

Rare is the new common: From Gaucher to Parkinson's disease

Pablo Sardi, Head of Lysosomal Storage and Metabolic Diseases Research, 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

The rare diseases pilot for the 100K genomics 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