

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: Accelerating Discovery into Cures for Unmet Need

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

Current obstacles to AAV-mediated gene therapy in neuromuscular disorders

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

11.30 Keynote Lecture – Catalyzing Translational Innovation

Chris Austin, Director, NIH National Center for Advancing Translational Sciences

12.30 Lunch

13.30 Session 2 Metabolic Diseases

Tay-Sachs: an inborn metabolic disease

Tim Cox, Department of Medicine, University of Cambridge

Developing substrate reduction therapy for rare metabolic disorders

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

Birmingham Rare Disease Network

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

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

Rare Bleeding disorders: the clinical benefits from genomics

Mike Laffan, Professor of Haemostasis and Thrombosis, Imperial College London

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

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 Disease Discovery at Roche

Madhurima Benekareddy, Project Lead and Lab Head, Roche

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

SNP-targeted allele specific therapies for the treatment of rare genetic diseases

Jaya Goyal, Vice President, Bioanalytical, Pharmacology and Biomarker Development, Wave Life Sciences

Rare disease programmes at PTC Therapeutics

Nikolai Naryshkin, Vice President, External Innovation and Alliance Management, PTC Therapeutics

11.20 Coffee Break

11.50 **Session 6 Haematology and Immunology**

Emerging therapies for hemophilia and sickle cell anemia

Seng Cheng, CSO Rare Diseases, Pfizer

Autologous Haematopoietic stem cell gene therapy for severe Immunodeficiencies and neurometabolic disorders

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

13:00 **Closing Remarks**