

# 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*

**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*

**Human Genetics Aided Discovery of a New Metabolic Hormone**

*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*

**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*

**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**