Rare diseases affect fewer than 5 in 10,000 of the general population in the UK (EU definition). There are over 6000 recognised rare diseases which, are often chronic and life-threatening and have an enormous impact on the lives of patients and their families. At some point in their lives, it is estimated that 7% (4.2million) of the UK population will be affected by a rare disease. This meeting will examine the genetic information that predisposes an individual to a rare disease; explore the mechanisms of rare disease and introduce examples of current personalised medicine and therapeutic approaches.

PROGRAMME

14:00   Welcome and opening remarks
         Philip Beales, UCL

14:10   Tess Harris, President, PKD International - ‘Personal Experiences of Living with Rare Diseases’

Introduction to genomics and personalised medicine
14:40   Tim Aitman, Imperial College London - ‘The new genomics and precision medicine’

Genetics/Genomics
15:00   Nick Wood, UCL - ‘Genetics of rare neurodegenerative diseases’
15:20   Michael Simpson, King’s College London - ‘Genomics and Rare Diseases’
15:40   Break

Functional Biology
16:10   Veronica van Heyningen, University of Edinburgh Western General Hospital - ‘Genes and Mechanisms in Developmental Eye Disease’
16:30   Matt Hurles, Wellcome Trust Sanger Institute - ‘Deciphering Developmental Disorders’

Therapeutics and Personalised Medicine
16:50   Fran Platt, University of Oxford - ‘Development of a small molecule therapy for lysosomal disorders’
17:10   Francesco Muntoni, UCL ‘RNA therapies of neuromuscular diseases’
17:30   Concluding Remarks
         Philip Beales, UCL
17:45   Drinks reception

The event is free to attend but you must register at cricksymp6.eventbrite.co.uk