NIHR | BioResource

CRDN Cambridge Rare Disease Network

Cambridge Rare Disease Day Evening Lecture

Thursday 28 February 2019 - 6:00PM - 7:30PM

William Harvey Lecture Theatre, School of Clinical Medicine, Addenbrooke's Hospital, Cambridge, CB2 0SP

5.30pm	Registration, tea and coffee
6.00pm	**Welcome and Introduction** Professor Patrick Chinnery, NIHR BioResource Co- Chair
6.10pm	**Improving patient health care in CNO and SAPHO** Dr Jagtar Singh Nijjar, NIHR Clinical Lecturer in Rheumatology, University of Cambridge
6.20pm	**Next Generation Children Project:** Rapid genome sequencing for severely ill children. A model for translational genomics research with direct integration into healthcare. Dr Isabelle Delon, Clinical Scientist in Molecular Genetics
6.30pm	**Imaging in the diagnosis of Rare Diseases** Dr Tomasz Matys, University Lecturer and Honorary Consultant in Radiology, University of Cambridge
6.40pm	**Whole Genome Sequencing for susceptibility to infectious disease, potential for patient benefits** Dr James Thaventhiran, Consultant Clinical Immunologist, Department of Medicine, University of Cambridge
6.55pm	**Q&A Discussion with all speakers** Facilitator: Dr Gemma Chandratillake, Course Director, ICE Genomic Medicine Programme
7.25pm	**Closing remarks** Dr Nathalie Kingston, NIHR BioResource Director
7.30pm	Drinks, nibbles and networking
8.00pm	Event ends

RAREDISEASEDAY.ORG

We'd like to thank you for coming and hope you enjoy this evening.

For tweets on the day, the hashtag is #rarediseaseday
- please include @NIHRBioResource and @camraredisease

Speakers' Biographies

Chair: Professor Patrick Chinnery, NIHR BioResource Co-Chair,
Head of the Department of Clinical Neurosciences, University of Cambridge

Patrick Chinnery qualified in medicine in Newcastle in 1992, trained in clinical neurology and developed a research programme studying the genetic basis of rare mitochondrial disorders, leading to new treatment trials for this group of diseases. A Wellcome Trust Principal Research Fellow, his laboratory is based in the MRC Mitochondrial Biology Unit on the Cambridge Biomedical Campus where he continues to study the inheritence of rare mitochondrial diseases and their treatment.

Dr Jagtar Singh Nijjar, NIHR Clinical Lecturer in Rheumatology, University of Cambridge

Jagtar Singh Nijjar is a Rheumatology registrar at the University of Cambridge and Addenbrooke's Hospital with an active research interest in rare inflammatory bone disease. He is the recruitment lead for a new NIHR BioResource cohort called ImPaHCS (improving Patient Health in CNO and SAPHO) By investigating rare inflammatory diseases like SAPHO (Synovititis, Acne, Pustulosis, Hyperostosis and Osteitis) and CNO (chronic non-bacterial osteomyelitis) we'll understand more about these conditions.

Dr Isabelle Delon, PhD, Clinical Scientist, EMEE Genomic Laboratory

Isabelle Delon is a trainee consultant clinical scientist (Higher Specialist Scientist) with 10 years' experience in molecular genetics at Cambridge University Hospitals Regional Genetics Laboratory, now East Midlands and East of England Genomic Laboratory Hub. She trained in Developmental Biology and Genetics at the Centre for Developmental Biology in Toulouse, France where she gained her MSc and PhD and worked as a research associate at the Gurdon Institute, Cambridge. Isabelle's expertise lies in rare disease diagnostics and she leads the NHS Genetics Laboratory team delivering rapid whole genome sequencing for children in intensive care – the Next Generation Children project, in partnership with the NIHR BioResource for Translational Research and the University of Cambridge

Dr Tomasz Matys, University Lecturer and Honorary Consultant in Radiology,
Department of Radiology, University of Cambridge

Tomasz Matys is a consultant Neuroradiologist for Addenbrooke's Hospital and a University Lecturer in Radiology. He undertook a specialist Neuroradiology fellowship in Cambridge and his main research interest is Neuro-oncology with a particular interest in using MRI for predicting treatment response and prognostication.

Dr James Thaventhiran, Consultant Clinical Immunologist at the University of Cambridge

James Thaventhiran is a Consultant Clinical Immunologist, who looks after patients with inherited susceptibility to infectious disease. Over the last 4 year he has run a large-scale whole genome project important for driving research into primary immune conditions. He will present a talk regarding how his motivation comes from directly experiencing the impact this technology has on patients.

Q&A Facilitator: Dr Gemma Chandratillake, Course Director, ICE Genomic Medicine Programme

Gemma Chandratillake is trained as a molecular geneticist and genetic counsellor. She is the Education and Training Lead for the East of England Genomic Medicine Centre (NHS) and coordinates the Masters course in Genomic Medicine at the University of Cambridge. Gemma is a Trustee of the Cambridge Rare Disease Network and works with stakeholders to find creative ways to mainstream genomics within the NHS for the benefit of patients with rare diseases.

Closing Remarks: Dr Nathalie Kingston, Director for NIHR BioResource for Translational Research in Common and Rare Diseases

Nathalie Kingston is the Director of the NIHR BioResource for Translational Research. She trained in Immunology at the University of Strathclyde and moved to the University of Cambridge to carry out research work in autoimmune and infectious diseases. She then joined the Wellcome Trust Sanger Institute for a position in Scientific Administration and subsequently became the Director's Executive Assistant. Nathalie joined the NIHR BioResource in 2013.