WELLCOME GENOME CAMPUS | ADVANCED COURSES AND SCIENTIFIC CONFERENCES

Genomics of Rare Disease 2018
26-28 March 2018

Wellcome Genome Campus,
Hinxton, Cambridge, UK

Conference Programme

Monday 26 March 2018

12:00-13:30 **Registration with lunch**

13:30-13:40 **Welcome and introduction**
Programme Committee: Helen Firth, Cambridge University Hospitals, UK

13:40-14:40 **Lupski lecture:**
The generation of new sequence diversity
Kári Stefánsson
deCODE genetics, Iceland

14:40-15:50 **Session 1: What's new in rare disease?**
Chair: Kym Boycott, Research Institute - Children's Hospital of Eastern Ontario, Canada

14:40 NAD deficiency, congenital malformations and niacin supplementation
Sally Dunwoodie
Victor Chang Cardiac Research Institute, Australia

15:20 Contribution of retrotransposition to developmental disorders
Eugene Gardner
Wellcome Sanger Institute, UK

15:50-16:30 **Afternoon tea**

16:30-17:00 **Session 1 continued:**
Chair: Kym Boycott, Research Institute - Children's Hospital of Eastern Ontario, Canada

16:30 Identifying genetic interactions with therapeutic implications in rare disease
Ramsay Bowden
University of Cambridge, UK

16:45 Significant burden of inherited nonsynonymous variation in children with developmental disorders
Kaitlin Samocha
Wellcome Sanger Institute, UK

17:00-17:40 **Lightning talks for poster session 1**

17:40-19:15 **Poster session 1 (odd numbers) with drinks reception**

19:15 **Dinner**

Tuesday 27 March 2018

09:00-10:50 **Session 2: Lessons from large-scale WGS studies**
Chair: Matt Hurles, Wellcome Sanger Institute, UK

09:00 Lessons from the 100,000 Genomes Project Rare Disease Programme
Richard Scott
Genomics England, UK

09:40 Insights from population-scale genome sequencing
Ira Hall
Washington University, USA

10:20 Complex structural variants resolved by short-read and long-read whole genome sequencing are an under-recognised cause of Mendelian disease
Alba Sanchis Juan
University of Cambridge, UK

10.35 Next Generation Children Project: Whole genome sequencing for rapid diagnosis of severely ill children in intensive care
Courtney French
University of Cambridge, UK

10:50-11.30 **Morning coffee**

11:30-13:00 **Session 3**: **Data sharing – legal and ethical issues**
Chair: Helen Firth, Cambridge University Hospitals, UK

11:30 Genomic futures: who is liable for what and when?
Bartha Maria Knoppers
McGill University, Canada

12:00 How is my genomic data mine? Private interests and proprietary rights
Jonathan Montgomery
University College London, UK

12:30 Panel discussion
Bartha Maria Knoppers
McGill University, Canada

Jonathan Montgomery
University College London, UK

Sobia Raza
PHG Foundation, UK

13:00-14:30 **Lunch**

14.30-16:05 **Session 4: Multiomic and single cell technologies**
Chair: Jay Schendure, University of Washington, USA

14:30 The power of ONE: Immunology in the age of single cell genomics
Ido Amit
Weizmann Institute of Science, Israel

15:10 Oligogenic inheritance and effects of modifier genes in cardiovascular disease
Casey Gifford
Gladstone Institutes, UCSF, USA

15:50 Integrating genomic and metabolomic data for variant prioritization in patients with unexplained neurometabolic phenotypes
Emma Graham
University of British Columbia, Canada

16:05-16:35 **Afternoon tea**

16:35-18:00 **Session 5: Informatics**
Chair: Kaitlin Samocha, Wellcome Sanger Institute, UK

16:35 Detecting and discovering pathogenic short tandem repeat expansions from short-read data
Harriet Dashnow
Royal Children's Hospital, Australia

17:15 A Bayesian adaptation of the ACMG/AMP pathogenicity recommendations
Leslie Biesecker
NIH, USA

17:30 Quantifying the proportion of patients in a rare disease cohort attributable to different classes of genotype
Hilary Martin
Wellcome Sanger Institute, UK

17:45 Predicting genomic instability and hotspots for Alu/Alu-mediated rearrangements in the human genome
Xiaofei Song
Baylor College of Medicine, USA

18:00-18:40 **Lightning talks for poster session 2**

18:40-20:10 **Poster session 2 (even numbers) and drinks reception**

20:10 **Conference dinner**

Wednesday 28 March 2018

08:30-10:20 **Session 6: Developmental genetics**
Chair: Han Brunner, Radboud University Medical Centre, The Netherlands

08:30 Enhancers and human disease: insights from mouse knockout studies
Axel Visel
Lawrence Berkeley National Laboratory, USA

09:10 Tridimensional analysis of human embryo development
Alain Chédotal
Institut De La Vision, France

09:50 Novel syndrome due to de novo missense mutations of CHD3 in 35 patients
Lot Snijders Blok
Radboud University Medical Center, The Netherlands

10:05 Biallelic mutations in human DCC cause developmental split brain syndrome
Saumya Jamuar
KK Women's and Children's Hospital, Singapore

10:20-11:00 **Morning coffee**

11:00-12:50 **Session 7: Model organisms and systems**
Chair: Matt Hurles, Wellcome Sanger Institute, UK

11:00 Defects in *Drosophila* high-throughput habituation as an endophenotype of autism/intellectual disability disorders – of mechanisms and clinical applications
Annette Schenck
Radboud University Medical Center, The Netherlands

11:40 From bed to benchside: developmental and translational research for skeletal dysplasia
Valérie Cormier-Daire
Imagine Institute, Necker Hospital, France

12:20 Systematic functional phenotyping of disease variants
Mikko Taipale
University of Toronto, Canada

12:35 Cyclin D2 stabilisation: a shared end-point for mTOR and p38 MAPK activation disorders?
James Poulter
University of Leeds, UK

12:50-14:10 **Lunch**

14:10-16:00 **Session 8: Therapy**
Chair: Kym Boycott, Research Institute - Children's Hospital of Eastern Ontario, Canada

14:10 Novel genetic therapies for neuromuscular disorders
Francesco Muntoni
University College London, UK

14:50 CAD mutations and uridine-responsive epileptic encephalopathy
Ingrid Bader
Paracelsus Medical University, Austria

15.05 Systemic upregulation of compensatory disease modifier ameliorates muscular dystrophy phenotypes in vivo
Dwi Kemaladewi
SickKids Hospital/University of Toronto, Canada

15:20 Achondroplasia: The journey from diagnosis to precision therapies: a template for rare genetic diseases
Ravi Savarirayan
Murdoch Children's Research Institute, Australia

16:00-16:10 **Closing remarks**
Programme Committee: Han Brunner, Radboud University Medical Centre, The Netherlands

16:20 **Coaches depart to Cambridge City Centre via Train Station, and Heathrow Airport via Stansted Airport**