WELLCOME GENOME CAMPUS | ADVANCED COURSES AND SCIENTIFIC CONFERENCES

Genomics of Rare Disease
5-7 April 2017

Wellcome Genome Campus,
Hinxton, Cambridge, UK

Conference Programme

Wednesday, 5 April

12:30-13:50	**Registration with Lunch**
13:50-14:00	**Welcome and Introductions** *Programme Committee*
14:00-15:00	**Lupski Lecture:** **Lgr5 Stem Cell-based organoids in human disease** *Hans Clevers - confirmed* *Hubrecht Institute, The Netherlands*
15:00-16:00	**Session 1: The emerging role of non-coding variants in disease** *Chair: Matt Hurles*

15:00 The contribution of de novo mutations in regulatory elements to neurodevelopmental disorders
Matthew Hurles
Wellcome Trust Sanger Institute, UK

15:30 Improving genetic diagnosis in Mendelian disease with transcriptome sequencing
Beryl Cummings
Broad Institute, USA

16:00-16:30 **Afternoon Tea**

16:30-17:30 **Session 1 continued: The emerging role of non-coding variants in disease**
Chair: Matt Hurles

16:30 Dissecting the Causal Mechanism of X-Linked Dystonia-Parkinsonism by Integrating Genome and Transcriptome Assembly
Tatsiana Aneichyk
Harvard Medical School, USA

17:00 Genetic diagnosis of Mendelian disorders via RNA sequencing
Laura Kremer
Helmholtz Zentrum München, Germany

17:15 Exome-wide evaluation of splice-disrupting mutations in 7833 families with severe developmental disorders
Jenny Lord
Wellcome Trust Sanger Institute, UK

17:30-18:00 Lightning Talks

18:00-19:30 **Poster Session 1 (odd numbers) with drinks reception**

19:30 Dinner

Thursday, 6 April

09:00-10:30 **Session 2: The intersection of monogenic and polygenic disease**
Chair: Han Brunner

09:00 On the spectrum from rare, mendelian to common, complex disease.
Ben Neale
Massachusetts General Hospital, USA

09.30 MTOR-related genes contribute to brain growth in patients with intellectual disability and in the population
Han Brunner
Radboud University Medical Center, The Netherlands

10.00 Common variation contributes to risk of severe developmental disorders
Mari Niemi
Wellcome Trust Sanger Institute, UK

10.15 Rare and common epilepsies converge on a shared gene regulatory network: opportunities for novel antiepileptic drug discovery
Michael Johnson
Imperial College London, UK

10:30-11.00 **Morning Coffee**

11:00-12:30 **Session 3: Overgrowth Mosaicism and imprinting in disease and health**
Chair: Helen Firth

11:00 *Phenotypic and genotypic aspects of cutaneous mosaicism*
Veronica Kinsler
UCL Institute Child Health, UK

11:30 *Multilocus imprinting disorders and new imprinting phenotypes*
Karen Temple
University of Southampton, UK

12:00 Revertant mosaicism repairs skin lesions in a patient with keratitis-ichthyosis-deafness (kid) syndrome by second-site mutations in Connexin 26
Marie Louise Bondeson
Uppsala University Hospital, Sweden

12:15 HIST1H1E and a growing family of overgrowth genes: clarifying the genetic architecture of overgrowth syndromes
Kate Tatton Brown
Institute of Cancer Research, UK

12:30-14:00 **Lunch**

14.00-15:30 **Session 4: New technologies to understand rare disease mechanisms**
Chair: Jay Shendure

14:00 Genome-first detection of genetic variation
Mark Chaisson
University of Washington, USA

14:30 Prenatal detection of fetal single gene diseases and de novo mutations non-invasively
Rossa Chiu
The Chinese University of Hong Kong

15:00 New approaches to interpreting variation in human genomes
Jay Shendure
University of Washington

15:30-16:00 **Afternoon Tea**

16:00-16:45 **Session 5: Informatic Tools**
Chair: Han Brunner

16:00 Bioinformatics for prioritization of functional cis-regulatory variants in patient genomes
Wyeth Wasserman
University of British Columbia, Canada

16:15 A fast association test for identifying pathogenic variants involved in rare diseases
Daniel Greene
MRC Biostatistics Unit, UK

16:30 RareConnect + PhenoTips: integrating a patient-facing portal into the daily practice of genomic medicine
Michael Brudno
Hospital for Sick Children, Canada

16:45-17:15 Lightning Talks

17:15-18:45 **Poster Session 2 (even numbers) with drinks reception**

19:00 **Dinner**

Friday, 7 April

09:00-10:30 **Session 6: Developmental genetics**
Chair: Kym Boycott

09:00 The mutation origin and functional consequences of multi-nucleotide variants in the Deciphering Developmental Disorders Study
Joanna Kaplanis
Wellcome Trust Sanger Institute, UK

09:15 Overgrowth and undergrowth/ID: Two faces of a unique CDKN1C-mutation
Siren Berland
Haukeland University Hospital, Norway

09:30 Gain of function variants in SAMD9 cause a complex multisystem condition with dynamic genomic rescue
Federica Buonocore
UCL GOS ICH, UK

09:45 The landscape of recessive mutations in 7,833 trios with developmental disorders
Hilary Martin
Wellcome Trust Sanger Institute, UK

10:00 Mutations in the cadherin-catenin complex in Blepharo-Cheilo-Dontic Syndrome
Annelies de Klein
ERASMUS MC, The Netherlands

10.15 Multiple molecular diagnoses uncovered by re-analysis of cases with phenotypic expansion
Jennifer Posey
Baylor College of Medicine, USA

10:30-11:00 **Morning Coffee**

11:00-12:30 **Session 7: Cellular and animal models**
Chair: Matt Hurles

11:00 Understanding cellular heterogeneity
Sarah Teichmann
Wellcome Trust Sanger Institute, UK

11:30 Regulatory principles governing enhancer function
Emma Farley
University of California, USA

12:00 Cellular proliferation and apoptosis underlie the primary mechanism of rare CNVs associated with neurodevelopmental disorders
Santhosh Girirajan
Pennsylvania State University, USA

12:15 Models of human disease from embryonic lethal lines
Cecilia Icoresi Mazzeo
Wellcome Trust Sanger Institute, UK

12:30-14:00 **Lunch**

14:00-15:30 **Session 8: Trials and large scale health-care implementation**
Chair: Kym Boycott

14:00 A tale of two trials – drug treatments of PIK3CA-related overgrowth spectrum and proteus syndrome
Kim Keppler-Noreuil
NIH, USA

14:30 Rare disease at the interface: The accessibility of skin for genetic discovery and therapeutic delivery as exemplified by CYLD cutaneous syndrome
Neil Rajan
Newcastle University, UK

14:45 The GABAergic system as a therapeutic target for the fragile X syndrome and related neurodevelopmental disorders
Frank Kooy
Universitiy of Antwerp, Belgium

15:00 Harnessing social media to transform clinical trials in rare disease; the North American airway collaborative experience in idiopathic subglottic stenosis
Alexander Gelbard
Vanderbilt University, USA

15:15 A clinical utility study on exome sequencing versus conventional genetic testing in pediatric neurology
Lisenka Vissers
Radboudumc, The Netherlands

15:30-15:45 Closing remarks

15:45	Coaches depart to Cambridge City Centre and Train Station, Heathrow Airport via Stansted Airport