WELLCOME GENOME CAMPUS | ADVANCED COURSES AND SCIENTIFIC CONFERENCES

Genomics of Rare Disease 2018
26-28 March 2018

Wellcome Genome Campus,
Hinxton, Cambridge, UK

Draft Programme

Monday 26 March 2018

12:00-13:30	**Registration with lunch**
13:30-13:40	**Welcome and introductions** *Programme Committee: Helen Firth, Cambridge University Hospitals, UK*
13:40-14:40	**Lupski lecture:** **Generation of sequence diversity** *Kari Stefansson* *deCODE genetics, Iceland*
14:40-16:00	**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 Speaker (from submitted abstracts)
16:00-16:40	**Afternoon tea**
16:40-17:10	**Session 1 continued:** *Chair: Kym Boycott, Research Institute - Children's Hospital of Eastern Ontario, Canada* 16:40 Short talk (from submitted abstracts) 16:55 Short talk (from submitted abstracts)
17:10-17:40	**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 Short talk (from submitted abstracts)

10.35 Short talk (from submitted abstracts)

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

Panel will include:
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 Polygenetics of congenital heart disease
Casey Gifford
Gladstone Institutes, UCSF, USA

15:50 Short talk (from submitted abstracts)

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

16:35-17:15 **Session 5: Informatics session**
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 Short talk (from submitted abstracts)

17:30 Short talk (from submitted abstracts)

17:45-18:15 **Lightning talks for poster session 2**

18:15-19:45 **Poster session 2 (even numbers) and drinks reception**

19:45 **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 Distant-acting enhancers in human disease: insights from mouse models
Axel Visel
Lawrence Berkeley National Laboratory, USA

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

09:50 Short talk (from submitted abstracts)

10:05 Short talk (from submitted abstracts)

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 Short talk (from submitted abstracts)

12:35 Short talk (from submitted abstracts)

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

15:30 Short talk (from submitted abstracts)

15.45 Short talk (from submitted abstracts)

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**