# Genomics of Rare Disease
25-27 March 2020

Wellcome Genome Campus,
Hinxton, Cambridge, UK

## Conference Programme

### Wednesday 25 March 2020

<table>
<thead>
<tr>
<th>Time</th>
<th>Event</th>
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<tbody>
<tr>
<td>12:00-13:30</td>
<td>Registration with lunch</td>
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<tr>
<td>13:30-13:40</td>
<td><strong>Welcome and introduction</strong>&lt;br&gt;Programme Committee: Kaitlin Samocha, Wellcome Sanger Institute, UK</td>
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<td>13:40-14:40</td>
<td><strong>Lupski lecture</strong>&lt;br&gt;Chair: Kaitlin Samocha, Wellcome Sanger Institute, UK</td>
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<td></td>
<td>Rare variation, rare subtypes of common disease: neuropsychiatry and neurodevelopment</td>
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<td>Mark Daly&lt;br&gt;FIMM, University of Helsinki, Finland</td>
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<td>14:40-15:55</td>
<td><strong>Session 1: What's new in rare disease?</strong>&lt;br&gt;Chair: Lisenka Vissers, Radboudumc University, The Netherlands</td>
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<tr>
<td>14:40</td>
<td>Delineating the structure of chromosome rearrangements using multiple WGS technologies&lt;br&gt;Anna Lindstrand&lt;br&gt;Karolinska Institutet, Sweden</td>
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<td>15:10</td>
<td>Functional genetics in inborn errors of immunity&lt;br&gt;Cecilia Poli Harlowe&lt;br&gt;Universidad del Desarrollo, Chile</td>
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<td>15:40</td>
<td>Patterns of mitochondrial DNA variation across 15,000 individuals in the gnomAD database&lt;br&gt;Nicole Lake&lt;br&gt;Yale University, USA</td>
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<td>15:55-16:30</td>
<td><strong>Afternoon tea</strong></td>
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16:30-17:00  
**Session 1 continued: What’s new in rare disease?**  
*Chair: Lisenka Vissers, Radboudumc University, The Netherlands*

16:30  
**Integrating healthcare and research genetic data empowers the discovery of 48 novel developmental disorders**  
*Kaitlin Samocha*  
*Wellcome Sanger Institute, UK*

16:45  
**The Androgen Receptor CAG repeat expansion is unexpectedly common in the general population**  
*Arianna Tucci*  
*Queen Mary University, UK*

17:00-17:30  
**Lightning talks**  
*Chair: Kaitlin Samocha, Wellcome Sanger Institute, UK*

17:30-19:00  
**Poster session 1 (odd numbers) with drinks reception**

19:00  
**Dinner**

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**Thursday 26 March 2020**

09:00-10:45  
**Session 2: Beyond the coding genome**  
*Chair: James Ware, Imperial College London, UK*

09:00  
**Polygenic contribution to rare inherited cardiac disorders**  
*Connie Bezzina*  
*AMC, University of Amsterdam, The Netherlands*

09:30  
**Coding plus non-coding, common plus rare: lessons learned from TBX6-associated congenital scoliosis (TACS)**  
*Nan Wu*  
*Peking Union Medical College Hospital, China*

10.00  
**Short talk TBC**

10.15  
**Pervasive and CpG-dependent promoter-like characteristics of transcribed enhancers**  
*Robin Steinhaus*  
*Charité – Universitätsmedizin Berlin, Germany*

10:30  
**Identification of repeat expansions with whole exome and whole genome sequencing in epilepsy patients**  
*Melanie Bahlo*  
*Walter and Eliza Hall Institute, Australia*

10:45-11:20  
**Morning coffee**
11:20-12:50  
**Session 3: Informatics**  
Chair: Kaitlin Samocha, Wellcome Sanger Institute, UK

11:20  
Phenotype aware prioritisation of rare disease variants  
Damian Smedley  
Queen Mary University of London and Genomics England, UK

11:50  
CADD-SV – a framework to score the effect of structural variants in health and disease  
Philip Kleinert  
Berlin Institute of Health, Germany

12:05  
InDelible: detection and evaluation of clinically-relevant structural variation from whole exome sequencing  
Eugene Gardner  
Wellcome Sanger Institute, UK

12:20  
Genetic intolerance across homologous protein domains identifies TTN missense variants with a role in dilated cardiomyopathy  
Xiaolei Zhang  
Imperial College London, UK

12:35  
Gene-specific modelling of mutational hotspots and in-silico evidence in a large hypertrophic cardiomyopathy cohort  
Adam Waring  
University of Oxford, UK

12:50-14:15  
**Lunch**

14:15-15:45  
**Session 4: Functional genomics**  
Chair: Mark Daly, FIMM, University of Helsinki, Finland

14:15  
Understanding the functional effects of coding variation at scale  
Lea Starita  
University of Washington, USA

14:45  
Phenotypic impacts of mono- and biallelic variation at a locus  
Tamar Harel  
Hadassah-Hebrew University Medical Center, Israel

15:15  
Saturation genome editing of developmental disorder genes  
Hong Kee Tan  
Wellcome Sanger Institute, UK

15:30  
The role of genetic modifiers in a mild LAMA2-RD case associated with a LAMA2 loss-of-function mutation  
Veronica Pini  
UCL, UK

15:45-16:20  
**Afternoon tea**
Session 5: New therapeutic approaches
Chair: Clare Turnbull, Institute of Cancer Research, UK

16:20 Targeted therapy in patients with PIK3CA-related overgrowth syndrome
Guillaume Canaud
Hôpital Necker Enfants Malades, France

16:50 Splice modulation therapy in inherited retinal diseases
Rob Collin
Radboudumc University Medical Center, The Netherlands

17:20 Fast and scalable RNA-seq splicing analysis for the clinical setting
Joseph Aicher
University of Pennsylvania, USA

Lightning talks
Chair: James Ware, Imperial College London, UK

Poster session 2 (even numbers) with drinks reception

Conference dinner

Friday 27 March 2020

Session 6: Clinical genetics
Chair: Jennifer Posey, Baylor College of Medicine, USA

09:00 Identifying diagnoses beyond the exome: lessons from genetically undiagnosed cases with compelling clinical phenotypes
Anne O'Donnell-Luria
The Broad Institute and Boston Children's Hospital, USA

09:30 Cancer susceptibility genomics: harnessing the full spectrum of variant frequency-penetrance
Clare Turnbull
Institute of Cancer Research, UK

10:00 Neuromuscular channelopathy gene, SCN4A, and the risk of sudden infant death syndrome
Emma Matthews
Institute of Neurology, UCL, UK

10:15 The added value of RNA sequencing on top of WES for variant interpretation and diagnosis of patients with Mendelian disorders
Vicente Yepez
Technical University of Munich, Germany

Morning coffee
**Session 7: Developmental genetics**
*Chair: Tamar Harel, Hadassah-Hebrew University Medical Center, Israel*

11:10  Neurodevelopmental proteasomopathies  
Sébastien Kury  
CHU de Nantes, France

11:40  Mutation-specific pathophysiological mechanisms in a new SATB1-associated neurodevelopmental disorder  
Elke de Boer  
Radboud University Medical Center, The Netherlands

11:55  Detection of mosaic chromosomal alterations in children with developmental disorders  
Ruth Eberhardt  
Wellcome Sanger Institute, UK

12:10  Heterozygous variants disturbing the transcriptional repressor activity of FOXP4 cause a disorder with congenital abnormalities, expressive language deficits and divergent growth parameters  
Lot Snijders Blok  
Radboud University Medical Center, The Netherlands

12:25  GREB1L variants contribute to the development of Mayer Rokitansky Kuster Hauser syndrome  
Angad Jolly  
Baylor College of Medicine, USA

12:40-12:50  **Closing remarks**  
*Chair: Lisenka Vissers, Radboudumc, The Netherlands*

12:50-14:00  **Lunch**

13:45  **Coach to Heathrow airport via Stansted airport departs**

14:00  **Coach to Cambridge departs**