Draft Conference Programme

Wednesday 27 March 2019

12:00-13:30  Registration with lunch

13:30-13:40  Welcome and introduction
             Programme Committee

13:40-14:40  Lupski lecture

             Nancy Cox
             Vanderbilt University Medical Center, USA

14:40-16:00  Session 1: Solving the unsolved

14:40        Harnessing transmission genetics to build models of complex disease
             from the ground up
             Jennifer Posey
             Baylor College of Medicine, USA

15:20        The NIH undiagnosed diseases program: expansion to a national and
             international networks
             William Gahl
             NIH, USA

16:00-16:40  Afternoon tea

16:40-17:10  Session 1 continued: Solving the unsolved

16:40        Short talk selected from submitted abstract

16:55        Short talk selected from submitted abstract

17:10-17:45  Lightning talks

17:45-19:15  Poster session 1 (odd numbers) with drinks reception

19:15        Dinner
Thursday 28 March 2019

09:00-10:30  **Session 2: Informatics**

09:00  Regulatory variant in rare disease: causal and modifier effects  
*Tuuli Lappalainen*  
*Genome Center, USA*

09:40  Short talk selected from submitted abstract

09:55  Short talk selected from submitted abstract

10:10  Short talk selected from submitted abstract

10:25  Short talk selected from submitted abstract

10:25-11:00  **Morning coffee**

11:00-12:25  **Session 3: Developmental genetics**

11:00  Rare disorders caused by mutations in genes encoding epigenetic regulators: novel diagnostic opportunities  
*Rosanna Weksberg*  
*The Hospital for Sick Children, Canada*

11:40  Short talk selected from submitted abstract

11:55  Short talk selected from submitted abstract

12:10  Short talk selected from submitted abstract

12:25-14:00  **Lunch**

14:00-15:30  **Session 4: New therapeutic approaches**

14:00  P4 medicine for intellectual disabilities: integrated -omics the way forward?  
*Clara van Karnebeek*  
*Amsterdam University Medical Centres, The Netherlands*

14:40  Hope for escape from a prison of bone  
*Frederick Kaplan*  
*University of Pennsylvania, USA*

15:20  Short talk selected from submitted abstract

15:35  Short talk selected from submitted abstract

15:50-16:30  **Afternoon tea**
16:30-17:30  
**Session 5: Electronic health records**

16:00  Assessing the pathogenicity, penetrance and expressivity of apparently monogenic disease variants in UK Biobank  
*Caroline Wright*  
*University of Exeter, UK*

16:40  Genomic disorders in the general population. What can we learn and how can we help?  
*Katrin Mannik*  
*University of Lausanne, Switzerland*

17:20  Short talk selected from submitted abstract

17:35  Short talk selected from submitted abstract

17:50-18:20  
**Lightning talks**

18:20-19:50  
**Poster session 2 (even numbers) with drinks reception**

19:50  
**Conference dinner**

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**Friday 29 March 2019**

08:30-10:20  
**Session 6: Data sharing – legal and ethical issues**

08:30  Advancing medicine through data sharing and collaboration on a global scale  
*Heidi Rehm*  
*Broad Institute, USA*

09:10  Talk title TBC  
*Ewan Birney*  
*EMBL-EBI, UK*

09:50  Short talk selected from submitted abstract

10:05  Short talk selected from submitted abstract

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

11:00-12:50  
**Session 7: Functional genomics**

11:00  Development and disease at single cell resolution  
*Malte Spielmann*  
*Max Planck Institute for Molecular Genetics, Germany*

11:40  Functional characterization and therapeutic targeting of gene regulatory elements  
*Nadav Ahituv*  
*UCSF, USA*
12:20  Short talk selected from submitted abstract
12:35  Short talk selected from submitted abstract

12:50-14:15  Lunch

14:15-16:05  Session 8: Intersection of polygenic and monogenic
14:15  The Finnish population isolate: examples of polygenic and rare variants in brain diseases
   Aarno Palotie
   FIMM, University of Helsinki, Finland
14:55  Short talk selected from submitted abstract
15:10  Short talk selected from submitted abstract
15:25  Using polygenic scores to define lifetime trajectories of disease
   Michael Inouye
   University of Cambridge, UK

16:05-16:15  Closing remarks
   Programme Committee

16:25  Coaches depart to Cambridge city centre via train station, and Heathrow airport via Stansted airport