Genomics of Rare Disease
27-29 March 2019

Wellcome Genome Campus,
Hinxton, Cambridge, UK

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**

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**	