Hybrid conference programme

Start (BST)	Finish (BST)	Presenter details
Monday 24 April 2023		
12:00	**13:00**	**Registration, lunch and networking**
12:45	*13:00*	*Briefing for Keynote & Session 1 speakers, microphone runners, chair, moderator & committee - Auditorium*
13:00	**13:10**	**Welcome** ***Scientific Programme Committee:*** *Fowzan Alkuraya, King Faisal Specialist Hospital & Research Centre, Saudi Arabia* *Anna Lindstrand, Karolinska Institute, Sweden* *Jennifer Posey, Baylor College of Medicine, USA* *James Ware, Imperial College London, UK*
13:10	**14:10**	**Keynote 1 - Lupski lecture** *Chair: Fowzan Alkuraya, King Faisal Specialist Hospital & Research Centre, Saudi Arabia* *Moderator: Jennifer Posey, Baylor College of Medicine, USA* Exploring the noncoding genome with chromosomal structural rearrangements *Cynthia Morton, Brigham and Women's Hospital, USA*
14:10	**14:55**	**Session 1: Gene regulation in rare disease** *Chair: Anna Lindstrand, Karolinska Institute, Sweden* *Moderator: James Ware, Imperial College London, UK*
14:10	14:40	Shedding light on the dark matter of the genome to improve diagnostic yield in rare disease diagnostics *Peter Robinson, The Jackson Laboratory, USA*
14:40	14:55	The role of start-stops elements in rare disease *Nechama Wieder, University of Oxford, UK*
14:55	15:40	Refreshment break and networking
15:40	**16:25**	**Session 1 cont: Gene regulation in rare disease** *Chair: Anna Lindstrand, Karolinska Institute, Sweden* *Moderator: James Ware, Imperial College London, UK*
15:40	16:10	Blurring the boundaries between rare and common diseases *Alexandre Reymond, University of Lausanne, Switzerland*
16:10	16:25	Recalibrating differential gene expression analysis by variance in gene dosage: A novel method for prioritizing functionally relevant genes in rare diseases *Philipp Rentzsch, SciLifeLab, Sweden*
16:25	**17:10**	**Poster pitch talks for poster session 1: odd numbers** *Chair: James Ware, Imperial College London, UK*
17:10	**18:30**	**Poster session 1: odd numbers, with refreshments**
18:30	20:30	Dinner
18:30	23:00	Bar open (card payments only)

Tuesday 25 April 2023

07:30	09:00	Breakfast
09:15	*09:30*	*Briefing for Session 2 speakers, microphone runners, chair & moderator - Auditorium*
09:30	**11:00**	**Session 2: Clinical applications**
		Chair: Jennifer Posey, Baylor College of Medicine, USA *Moderator: Fowzan Alkuraya, King Faisal Specialist Hospital & Research Centre, Saudi Arabia*
09:30	10:00	Genomics of rare diseases in Latin America *Claudia Gonzaga-Jauregui, International Laboratory for Human Genome Research, UNAM, Mexico*
10:00	10:30	Founder mutations in rare disease: a Middle Eastern experience *Dana Marafi, Kuwait University, Kuwait*
10:30	10:45	More than a fancy exome: unique capabilities of genome sequencing for rare disease diagnosis *Gabrielle Lemire,Broad Institute of MIT and Harvard, USA*
10:45	11:00	Mendelian hunt gone awry: the diagnostic implications of pitfalls in causal variant identification based on 4,510 molecularly characterized families *Lama AlAbdi, King Faisal Specialist Hospital & Research Centre, Saudi Arabia*
11:00	11:40	Refreshment break and networking
11:25	*11:40*	*Briefing for Session 3 speakers, microphone runners, chair & moderator - Auditorium*
11:40	**13:10**	**Session 3: Translational genomics**
		Chair: James Ware, Imperial College London, UK *Moderator: Anna Lindstrand, Karolinska Institute, Sweden*
11:40	12:10	Making use of population datasets in variant interpretation *Kaitlin Samocha, Massachusetts General Hospital, USA*
12:10	12:40	Brain organoid modeling of neurogenetic disorders uncovers defective cilia dynamics as a significant underlying mechanism *Jay Gopalakrishnan, Heinrich Heine University Düsseldorf, Germany*
12:40	12:55	Refining the pathogenicity of rare variants in monogenic obesity genes using large-scale population data: Implications for genomic medicine *Nathalie Charmi, Mount Sinai Hospital, USA*
12:55	13:10	Strategic identification of pathogenic variant carriers from tumour-only sequencing: analysis of 49,264 cancer patients *Zeid Kuzbari, Institute of Cancer Research, UK*
13:10	14:20	Lunch and networking
14:05	*14:20*	*Briefing for Session 4 speakers, microphone runners, chair & moderator - Auditorium*
14:20	**15:50**	**Session 4: Public health, prevention and policy**
		Chair: Fowzan Alkuraya, King Faisal Specialist Hospital & Research Centre, Saudi Arabia *Moderator: Jennifer Posey, Baylor College of Medicine, USA*
14:20	14:50	Preventing rare disease - Mackenzie's Mission, the Australian reproductive carrier screening project *Nigel Laing, Harry Perkins Institute of Medical Research, Australia*
14:50	15:20	Preconception carrier screening for rare diseases *Lidewij Henneman, Amsterdam UMC, the Netherlands*
15:20	15:35	Three years' experience of BeGECS at the Center for Medical Genetics Ghent: results from 400 couples *Hannah Verdin, Ghent University, Belgium*
15:35	15:50	Genetic variations in cardiac channelopathy disorders in the Indian population: implications for public health and screening strategies *Anjali Bajaj, CSIR-Institute of Genomics and Integrative Biology, India*

15:50	16:20	Refreshment break and networking
16:20	**17:20**	**Keynote 2**
		Chair: Anna Lindstrand, Karolinska Institute, Sweden *Moderator: Jennifer Posey, Baylor College of Medicine, USA* Base editing and prime editing: correcting mutations that cause genetic disease in cells, animals, and patients *David Liu, The Broad Institute, Harvard University, USA*
17:20	**18:00**	**Poster pitch talks for poster session 2: even numbers**
		Chair: Jennifer Posey, Baylor College of Medicine, USA
18:00	**19:00**	**Poster session 2: even numbers, with refreshments**
19:00	21:00	Dinner
19:00	23:00	Bar open (card payments only)

Wednesday 26 April 2023

07:30	09:00	Breakfast
09:15	*09:30*	*Briefing for Session 5 speakers, microphone runners, chair & moderator - Auditorium*
09:30	**11:00**	**Session 5: New technologies**
		Chair: James Ware, Imperial College London, UK *Moderator: Jennifer Posey, Baylor College of Medicine, USA*
09:30	10:00	The potential of long-read sequencing in rare disease *Danny Miller, University of Washington, USA*
10:00	10:30	Towards better understanding of atypical diabetes using human pluripotent stem cell technology *Gosia Borowiak, Institute of Molecular Biology & Biotechnology, Faculty of Biology, Poland*
10:30	10:45	Deep mutational scanning improves functional variant interpretation in enzymes associated with rare diseases *Kaiyue Ma, Yale University, USA*
10:45	11:00	Long-read sequencing and optical genome mapping enable full characterization of previously unresolved structural variation *Griet De Clercq Ghent University, Belgium*
11:00	11:30	Refreshment break and networking
11:15	*11:30*	*Briefing for Session 6 speakers, microphone runners, chair & moderator - Auditorium*
11:30	**13:00**	**Session 6: Genomics and multi-omics**
		Chair: Jennifer Posey, Baylor College of Medicine, USA *Moderator: Anna Lindstrand, Karolinska Institute, Sweden*
11:30	12:00	Multi-omic approaches to dissect mitochondrial pathology *Robert Taylor, Newcastle University, UK*
12:00	12:15	Deciphering transposable elements by short- and long read sequencing *Kristine Bilgrav Saether, Karolinska Institutet, Sweden*
12:15	12:30	Systematic assessment of polygenic score associations in a large-scale, phenotypically diverse pediatric rare disease cohort *Craig Smail, Children's Mercy Hospital, USA*
12:30	13:00	Genetic effects on gene dosage affect cellular phenotypes and disease risk *Tuuli Lappalainen, KTH Royal Institute of Technology, Sweden*
13:00	**13:10**	**Closing remarks and poster prize presentation**
		Scientific Programme Committee: *Fowzan Alkuraya, King Faisal Specialist Hospital & Research Centre, Saudi Arabia* *Anna Lindstrand, Karolinska Institute, Sweden* *Jennifer Posey, Baylor College of Medicine, USA* *James Ware, Imperial College London, UK*
13:10	14:00	Lunch and departures
14:00		Coach departures for Stansted and Heathrow airports
14:10		Coach departures for Cambridge train station and city centre