Hybrid Conference Programme

Start (GMT)	Finish (GMT)	Presenter details
Monday 25 March 2024		
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
		Hilary Martin,Wellcome Sanger Institute, UK
		Jennifer Posey, Baylor College of Medicine, USA
13:10	**14:10**	**Keynote lecture**
		Chair: Jennifer Posey, Baylor College of Medicine
		Moderator: Anna Lindstrand, Karolinska Institute, Sweden
		Towards interventional genetics
		Timothy Yu, Boston Children's Hospital, USA
14:10	**14:55**	**Session 1: What's new in rare disease?**
		Chair: Anna Lindstrand, Karolinska Institute, Sweden
		Moderator:Jennifer Posey, Baylor College of Medicine
14:10	14:40	Undiagnosed disease programme in South Africa
		Shahida Moosa, Stellenbosch University, South Africa
14:40	14:55	Rare disease gene association discovery from burden analysis of the 100,000 Genomes Project data
		Valentina Cipriani, Queen Mary University of London, UK
14:55	15:40	Refreshment break and networking
15:25	*15:40*	*Briefing for Session 1 speakers, microphone runners, chair, moderator & committee - Auditorium*
15:40	**17:10**	**Session 1: What's new in rare disease continued**
		Chair: Evan Eichler, University of Washington, USA
		Moderator: Lisenka Vissers, Radboud University, Netherlands
15:40	16:10	Positive selection in male germ cells and its impact on rare disorders
		Raheleh Rahbari, Wellcome Sanger Institute, UK
16:10	16:25	Harmonized framework for RNA-seq-based rare disease diagnostics in a pan-continental consortium - Solve-RD
		Vicente Yepez, Technical University of Munich, Germany
16:25	16:40	Structural variant allelic heterogeneity in MECP2 Duplication Syndrome provides insight into clinical severity and variability of disease expression
		Claudia Carvalho, Pacific Northwest Research Institute, USA
16:40	16:55	Using regional nonsense constraint for clinical and biological insights into rare genetic conditions
		Alexander Blakes, University of Manchester, UK
16:55	17:10	Identification of mitochondrial genome constraint in gnomAD provides new tools for variant classification
		Nicole Lake, Yale, USA
17:10	**17:35**	**Poster pitch talks for odd number posters**
		Chair: Hilary Martin, Wellcome Sanger Institute, UK
17:35	**19:00**	**Poster session 1 - odd number posters**
19:00	21:00	Dinner
		Bar open (card payments only)

Tuesday 26 March 2024

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: Pangenome** *Chair: Lisenka Vissers, Radbound University, Netherlands* *Moderator:Jennifer Posey, Baylor College of Medicine*
09:30	10:00	Complex recurrent structural polymorphisms and susceptibility to genomic disorders *Evan Eichler, University of Washington, USA*
10:00	10:15	Mind the Reference Gap *Kristine Bilgrav Saether, Karolinska Institutet, Sweden*
10:15	10:30	Unveiling the Genetic Tapestry: Rare Diseases Among Georgia's Ethnic Azerbaijani Population *Tinatin Tkemaladze, Tbilisi State Medical University, Georgia*
10:30	11:00	Rare disease multi-omics and the Qatar Mendelian Program *Khalid Fakhro, Hamad Bin Khalifa University, Qatar*
11:00	11:45	Refreshment break and networking
11:30	*11:45*	*Briefing for Session 3 speakers, microphone runners, chair & moderator - Auditorium*
11:45	**13:15**	**Session 3: Genomic Screening** *Chair: Hilary Martin, Wellcome Sanger Institute, UK* *Moderator: Lisenka Vissers, Radbound University, Netherlands*
11:45	12:15	Outcome of publicly funded nationwide first-tier noninvasive prenatal screening *Joris Vermeesch, KU Leuven, Belgium*
12:15	12:30	Analytical validity of variant prioritisation algorithm in whole genome sequencing data for newborn screening in the Generation Study *Joanna Kaplanis, Genomics Engalnd Ltd, UK*
12:30	12:45	Predicting the severity of recessive ADSL deficiency with deep mutational scanning and biallelic pathogenicity scores *Hasan Çubuk, The University of Edinburgh, UK*
12:45	13:15	Genomic Sequencing as a First-Tier Screening Test and Outcomes of Newborn Screening *Ting Chen, Xinhua Hospital, Shanghai Jiaotong University School of Medicine, China*
13:15	14:30	Lunch and meet the speaker networking
14:15	*14:30*	*Briefing for Session 4 speakers, microphone runners, chair & moderator - Auditorium*
14:30	**16:00**	**Session 4: Blurring boundaries between common and rare disease** *Chair: Hilary Martin, Wellcome Sanger Institute, UK* *Moderator: Evan Eichler, University of Washington, USA*
14:30	15:00	Monogenic and polygenic stroke- not as district as we thought *Hugh Markus, Cambridge University, UK*
15:00	15:15	A genotype-first approach to interrogate the allelic series of variants in the calcium sensing receptor associated with autosomal dominant hypocalcemia type 1 *Sun-Gou Ji, BridgeBio Pharma Inc., USA*
15:15	15:30	Dissecting the contribution of common variants to risk of rare neurodevelopmental conditions *Qinqin Huang, Wellcome Sanger Institute, UK*
15:30	16:00	The shifting landscape of penetrance and expressivity in a world of biobanks, genomes and precision meds *Valerie Arboleda, UCLA, USA*
16:00	**16:30**	**Poster pitch talks for even number posters** *Chair: Jennifer Posey, Baylor College of Medicine*
16:30	**16:45**	**Sponsored talk**
16:30	16:45	Addressing barriers to inclusion in rare disease research *Hannah Stark, Operations Lead at the NIHR BioResource, UK*
17:00	**18:30**	**Poster session 2 - even number posters**
18:30	20:30	Dinner
		Bar open (card payments only)

Wednesday 27 March 2024		
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: Therapeutics** *Chair: Anna Lindstrand, Karolinska Institute, Sweden* *Moderator: Hilary Martin, Wellcome Sanger Institute, UK*
09:30	10:00	Advances in haemophilia A gene therapy *Johnny Mahlangu, University of the Witwatersrand, Johannesburg, South Africa*
10:00	10:15	First In Class ASO Targeting IGHMBP2 Cryptic Splice Variant: Efficacy and Safety *Sandra Smieszek, Vanda Pharmaceuticals, USA*
10:15	10:30	Genome-scale quantification and prediction of pathological stop codon readthrough by small molecules *Ignasi Toledano, IRB, Spain*
10:30	11:00	Transforming drug discovery using large-scale genomics *Keren Carss, Astra Zeneca, UK*
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: Beyond the genome** *Chair: Anna Lindstrand, Karolinska Institute, Sweden* *Moderator: Hilary Martin, Wellcome Sanger Institute, UK*
11:30	12:00	RNA in Genomic Medicine *Diana Baralle, Southampton University, UK*
12:00	12:15	Full-length transcript atlas of the developing human cortex uncovers novel candidate diagnoses in developmental disorders *Kartik Chundru, University of Exeter, UK*
12:15	12:30	Using long read genomics to identify methylation outliers in rare disease *Tanner Jensen, Stanford University, USA*
12:30	13:00	From phenotype to AI-based phenomics *Lisenka Vissers, Radbound University, Netherlands*
13:00	**13:15**	**Closing remarks and prize presentation** ***Scientific Programme Committee:*** *Fowzan Alkuraya, King Faisal Specialist Hospital & Research Centre, Saudi Arabia* *Anna Lindstrand, Karolinska Institute, Sweden* *Hilary Martin,Wellcome Sanger Institute, UK* *Jennifer Posey, Baylor College of Medicine, USA*
13:15	14:15	Lunch and departures
14:10		Coach departures for Stansted and Heathrow airports
14:20		Coach departures for Cambridge train station and city centre