

Hybrid Conference Programme

Start (BST)	Finish (BST)	Presenter details	
Monday 27 April 2026			
12:00	13:00	Registration, lunch and networking	Event space
12:45		Briefing for opening keynote speaker, session 1 speakers, all microphone runners, chair, moderator and committee	Auditorium
13:00	13:10	Welcome and introductions	Auditorium
		<p>Wellcome Connecting Science: Michelle Bishop, Wellcome Connecting Science, United Kingdom</p> <p>Scientific programme committee: Hilary Martin, Wellcome Sanger Institute, United Kingdom Maite Spielmann, Charité, Germany Stephen Montgomery, Stanford University, USA Claudia Gonzaga-Jauregui, National Autonomous University Of Mexico, Mexico Caroline Wright, University of Exeter, United Kingdom Zané Lombard, University of Witwatersrand, South Africa</p>	
13:10	14:00	Opening keynote	Auditorium
		<p>Chair: Hilary Martin Moderator: Claudia Gonzaga-Jauregui</p>	
13:10	14:00	Somatic instability of repetitive DNA: Lessons from Huntington's Disease Bob Handsaker, Broad Institute, United States	
14:00	14:05	Comfort break	
14:05	15:35	Session 1: NIPT/ Prenatal/ Perinatal	Auditorium
		<p>Chair: Claudia Gonzaga-Jauregui Moderator: Zané Lombard</p>	
14:05	14:35	The Future of Prenatal Diagnostics Mike Talkowski, Broad Institute, United States	
14:35	15:05	Reducing transmission of pathogenic mtDNA variants Mary Herbert, Monash, Australia	
15:05	15:20	In utero treatment of rare disease: using non-invasive prenatal testing (NIPT) to inform targeted sulphonylurea treatment in pregnancies at risk of KATP channel-related neonatal diabetes and neurodevelopmental disorders <i>Pamela Bowman, University of Exeter / Royal Devon University Healthcare NHS Foundation Trust, United Kingdom</i>	
15:20	15:35	Whole Genome Sequencing of Euploid Products of Conception Identifies Diverse Genetic Causes of Recurrent Pregnancy Loss <i>Andrew Jia, Yale University, United States</i>	
15:35	16:15	Refreshment break and networking	Event space

16:00	16:15	Briefing for session 2 speakers, chair and moderator	Auditorium
16:15	17:45	Session 2: Multiomics and Rare Disease	Auditorium
		<i>Chair: Hilary Martin</i>	
		<i>Moderator: Malte Spielmann</i>	
16:15	16:45	Multi-omics for Gene Discovery and Precision Medicine in the Pediatric Epilepsies Heather Mefford, St. Jude Children's Research Hospital, USA	
16:45	17:00	Noncoding deletions across 125,730 individuals highlight promoter disruption as a recurrent cause of rare disease <i>Anthony McGuigan, University of Oxford, United Kingdom</i>	
17:00	17:15	Saturation Genome Editing to Clarify Variant Effect within 5'UTRs of Neurodevelopmental Disorder Genes <i>Vanessa Burns, Wellcome Sanger Institute, United Kingdom</i>	
17:15	17:30	Lessons for genomic newborn screening from carefully curated gene-disease dyad studies in population cohorts <i>Leigh Jackson, University of Exeter, United Kingdom</i>	
17:30	17:45	Optical Genome Mapping and Long-Read Nanopore Sequencing for CSTB repeat expansion molecular diagnosis in Unverricht-Lundborg Disease <i>Flavia Fineschi, Children's Hospital A. Meyer, Italy</i>	
17:45	18:25	Poster pitch talks for odd number posters	Auditorium
18:25	19:30	Poster session 1 - odd number posters	Event space
19:30	21:00	Dinner	Hinxton Hall Restaurant
19:25		Bar open (card payments only)	Graham Cameron Bar

Tuesday 28 April 2026

09:15		Briefing for Session 3 speakers, chair and moderator	Auditorium
09:30	11:00	Session 3: Factors affecting penetrance	Auditorium
		<i>Chair: Caroline Wright</i> <i>Moderator: Stephen Montgomery</i>	
09:30	10:00	Somatic mutations in normal tissues in Neurofibromatosis Type 1 Henry Lee-Six, Wellcome Sanger Institute, United Kingdom	
10:00	10:30	Exploring rare disease penetrance across cohorts Sanna Gudmundsson, Karolinska Institutet, Sweden	
10:30	10:45	Beyond Background Genetics: Stochastic Drivers of Phenotypic Diversity in NDDs –Evidence from Mice <i>Gabriela Gurria, Wellcome Sanger Institute, United Kingdom</i>	
10:45	11:00	Partial NMD targeting contributes to incomplete penetrance of inherited protein-truncating variants in severe developmental disorders <i>Juliet Hampstead, Radboud University Medical Center, The Netherlands</i>	
11:00	11:45	Refreshment break and networking	Event space
11:30	11:45	Briefing for session 4 speakers, chair and moderator	Auditorium
11:45	13:15	Session 4: Whats new in rare disease? Part 1	Auditorium
		<i>Chair: Hilary Martin</i> <i>Moderator: Malte Spielmann</i>	
11:45	12:15	Integrating AI tools into rare disease diagnosis and reanalysis Daniel Macarthur, Centre for Population Genomics, Garvin Institute of Medical Research, Australia	
12:15	12:30	Findings from the Critical Assessment of Genome Interpretation, a community experiment to evaluate phenotype prediction <i>Steven Brenner, University of California, Berkeley, United States</i>	
12:30	12:45	ProtVar2 Launch - The Next Dimension of Missense Variant Interpretation <i>James Stephenson, EMBL-EBI, United Kingdom</i>	
12:45	13:00	DECIPHER: Enabling accurate clinico-molecular diagnosis and sharing of approved and emerging therapies for rare genetic disease <i>Julia Foreman, EMBL-EBI, United Kingdom</i>	
13:00	13:15	Comprehensive characterisation of non-coding and coding effects of de novo mutations in a large-scale rare disease case-control cohort <i>Kartik Chundru, University of Exeter, United Kingdom</i>	
13:15	14:35	Lunch and networking	Event space

14:20	14:35	Briefing for Session 5 speakers, chair and moderator	Auditorium
14:35	16:05	Session 5: Whats new in rare disease? Part 2	Auditorium
		<i>Chair: Caroline Wright</i>	
		<i>Moderator: Stephen Montgomery</i>	
14:35	15:05	The value of investing in rare disease research in Africa: Diagnostic yield and implementation insights from the DDD-Africa study Zané Lombard, University of Witwatersrand, South Africa	
15:05	15:20	Integrative multi-platform genomic analysis for resolving hidden structural variations: insights from MECP2 duplication syndrome <i>Qiaowei Liang, Yokohama City University, Japan</i>	
15:20	15:35	DECIPHERD and CHANGER: Addressing Gaps in Rare and Undiagnosed Disease Genomics in Chile <i>Gabriela Repetto, Facultad De Medicina, Clinica Alemana Universidad Del Desarrollo, Chile</i>	
15:35	15:50	Genetic constraint in small nuclear RNA genes <i>Elston Dsouza, University of Oxford, United Kingdom</i>	
15:50	16:05	Accelerating genetic diagnosis in the NICU via machine-learning assisted selection of infants for genome sequencing <i>Stephen Meyn, School of Medicine and Public Health, University of Wisconsin - Madison, United States</i>	
16:05	16:45	Refreshment break and networking	Event space
16:45	17:25	Poster pitch talks for even number posters	Auditorium
		<i>Chair: Stephen Montgomery</i>	
17:25	18:30	Poster session 2 - even number posters	Event Space
18:30	20:30	Dinner	Hinxton Hall Restaurant
18:30		Bar open (card payments only)	Graham Cameron Bar

Wednesday 29 April 2026			
09:15		Briefing for Session 6 speakers, chair & moderator	Auditorium
09:30	10:20	Lupski keynote lecture	Auditorium
		<i>Chair: Claudia Gonzaga-Jauregui</i> <i>Moderator: Hilary Martin</i>	
09:30	10:20	GUARDIAN and opportunities to improve population genomic health Wendy Chung, Harvard Medical School and Boston Children's Hospital, USA	
10:20	11:00	Refreshment break and networking	Event space
10:45	11:00	Briefing for closing keynote speaker, chair, moderator and committee	Auditorium
11:00	12:30	Session 6: Therapeutics	Auditorium
		<i>Chair: Zané Lombard</i> <i>Moderator: Caroline Wright</i>	
11:00	11:30	Nucleic Acid Therapies for Rare Diseases: Progress and Prospects in the UK Haiyan Zhou, University College London, United Kingdom	
11:30	11:45	The autophagic/lysosomal enhancer GHF-201 can ameliorate pathological features in patient-derived fibroblasts and a mouse model of Pompe disease <i>Uri Sprecher, Tel Aviv University, Israel</i>	
11:45	12:00	N-of-1 personalized therapy for CMT2S: From patient-specific preclinical models to first-in-human dosing <i>Sandra Smieszek, Vanda Pharmaceuticals Inc., United States</i>	
12:00	12:30	Huntington's disease – mechanisms to therapeutics Sarah Tabrizi, UCL, United Kingdom	
12:30	12:45	Closing Remarks	Auditorium
		Scientific Programme Committee: Hilary Martin, Wellcome Sanger Institute, United Kingdom Malte Spielmann, Charité, Germany Stephen Montgomery, Stanford University, United States Claudia Gonzaga-Jauregui, National Autonomous University Of Mexico, Mexico Caroline Wright, University of Exeter, United Kingdom Zané Lombard, University of Witwatersrand, South Africa	
12:45	13:45	Lunch and departures	Hinxton Hall Restaurant
13:45		Coach departures for Stansted and Heathrow airports	Main entrance
13:55		Coach departures for Cambridge train station and city centre	Main entrance