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**
		Wellcome Connecting Science: Michelle Bishop, Wellcome Connecting Science, United Kingdom ***Scientific programme committee:*** *Hilary Martin, Wellcome Sanger Institute, United Kingdom* *Malte 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*	
13:10	**14:00**	**Opening keynote**	**Auditorium**
		Chair: Hilary Martin *Moderator: Claudia Gonzaga-Jauregui*	
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**
		Chair: Claudia Gonzaga-Jauregui *Moderator: Zané Lombard*	
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 *Pamela Bowman, University of Exeter / Royal Devon University Healthcare NHS Foundation Trust, United Kingdom*	
15:20	15:35	Whole Genome Sequencing of Euploid Products of Conception Identifies Diverse Genetic Causes of Recurrent Pregnancy Loss *Andrew Jia, Yale University, United States*	
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**
		Chair: Hilary Martin *Moderator: Malte Spielmann*	
16:15	16:45	Title TBC *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 *Anthony McGuigan, University of Oxford, United Kingdom*	
17:00	17:15	Saturation Genome Editing to Clarify Variant Effect within 5'UTRs of Neurodevelopmental Disorder Genes *Vanessa Burns, Wellcome Sanger Institute, United Kingdom*	
17:15	17:30	MethaDory: training open and local DNA methylation signature classifiers for rare diseases using synthetic cases for arrays and long-read sequencing data *Federico Ferraro, Erasmus MC, The Netherlands*	
17:30	17:45	Optical Genome Mapping and Long-Read Nanopore Sequencing for CSTB repeat expansion molecular diagnosis in Unverricht–Lundborg Disease *Flavia Fineschi, Children's Hospital A. Meyer, Italy*	
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**
		Chair: Caroline Wright *Moderator: Stephen Montgomery*	
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 in over 800,000 individuals *Sanna Gudmundsson, Karolinska Institutet, Sweden*	
10:30	10:45	Beyond Background Genetics: Stochastic Drivers of Phenotypic Diversity in NDDs –Evidence from Mice *Gabriela Gurria, Wellcome Sanger Institute, United Kingdom*	
10:45	11:00	Partial NMD targeting contributes to incomplete penetrance of inherited protein-truncating variants in severe developmental disorders *Juliet Hampstead, Radbound University Medical Center, The Netherlands*	
11:00	11:45	Refreshment break and networking	Event space
11:20	*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**
		Chair: Hilary Martin *Moderator: Malte Spielmann*	
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 *Steven Brenner, University of California, Berkeley, United States*	
12:30	12:45	ProtVar2 Launch - The Next Dimension of Missense Variant Interpretation *James Stephenson, EMBL-EBI, United Kingdom*	
12:45	13:00	DECIPHER: Enabling accurate clinico-molecular diagnosis and sharing of approved and emerging therapies for rare genetic disease *Julia Foreman, EMBL-EBI, United Kingdom*	
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 *Kartik Chundru, University of Exeter, United Kingdom*	
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**
		Chair: Caroline Wright *Moderator: Stephen Montgomery*	
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 *Qiaowei Liang, Yokohama City University, Japan*	
15:20	15:35	DECIPHERD and CHANGER: Addressing Gaps in Rare and Undiagnosed Disease Genomics in Chile *Gabriela Repetto, Facultad De Medicina, Clinica Alemana Universidad Del Desarrollo, Chile*	
15:35	15:50	Genetic constraint in small nuclear RNA genes *Elston Dsouza, University of Oxford, United Kingdom*	
15:50	16:05	Accelerating genetic diagnosis in the NICU via machine-learning assisted selection of infants for genome sequencing *Stephen Meyn, School of Medicine and Public Health, University of Wisconsin - Madison, United States*	
16:05	16:45	Refreshment break and networking	Event space
16:45	**17:25**	**Poster pitch talks for even number posters**	**Auditorium**
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**	**Final day keynote**	**Auditorium**
		Chair: Claudia Gonzaga-Jauregui *Moderator: Hilary Martin*	
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**
		Chair: Zané Lombard *Moderator: Caroline Wright*	
11:00	11:30	Title TBC *Robert Wynn, Manchester University NHS, 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 *Uri Sprecher, Tel Aviv University, Isreal*	
11:45	12:00	N-of-1 personalized therapy for CMT2S: From patient-specific preclinical models to first-in-human dosing *Sandra Smieszek, Vanda Pharmaceuticals Inc., United States*	
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, University of Lübeck, Germany* *Stephen Montgomery, Stanford Medicine, United States* *Claudia Gonzaga-Jauregui, International Laboratory for Human Genome Research, 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