

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 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</p>	
13:10	14:00	Opening keynote	Auditorium
		<p>Chair: TBC Moderator: TBC</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: TBC Moderator: TBC</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	Short talk selected from abstracts <i>Short talk speaker</i>	
15:20	15:35	Short talk selected from abstracts <i>Short talk speaker</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: TBC</i>	
		<i>Moderator: TBC</i>	
16:15	16:45	Title TBC Vicente Yepez, OmicsDiscoveries, Germany	
16:45	17:15	Title TBC Heather Mefford, St.Jude Children's Research Hospital, USA	
17:15	17:30	Short talk selected from abstracts <i>Short talk speaker</i>	
17:30	17:45	Short talk selected from abstracts <i>Short talk speaker</i>	
17:45	18:25	Poster pitch talks for odd number posters	Auditorium
18:25	19:25	Poster session 1 - odd number posters	Event space
19:25	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: TBC</i>	
		<i>Moderator: TBC</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 in over 800,000 individuals Sanna Gudmundsson, KTH, Sweden	
10:30	10:45	Short talk selected from abstracts <i>Short talk speaker</i>	
10:45	11:00	Short talk selected from abstracts <i>Short talk speaker</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: TBC</i>	
		<i>Moderator: TBC</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	12:45	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	
12:45	13:00	Short talk selected from abstracts <i>Short talk speaker</i>	
13:00	13:15	Short talk selected from abstracts <i>Short talk speaker</i>	
13:15	14:30	Lunch and networking	Event space
14:15	14:30	Briefing for Session 5 speakers, chair and moderator	Auditorium
14:30	16:00	Session 5: Whats new in rare disease? Part 2 - Panel discussion	Auditorium
		<i>Chair: TBC</i>	
		<i>Moderator: TBC</i>	
14:30	16:00	Panel session <i>Panel members to be confirmed</i>	
16:00	16:40	Refreshment break and networking	Event space
16:40	17:20	Poster pitch talks for even number posters	Auditorium
17:20	18:20	Poster session 2 - even number posters	
18:20	20:30	Dinner	Hinxton Hall Restaurant
18:20		Bar open (card payments only)	Graham Cameron Bar

Wednesday 29 April 2026			
09:15		Briefing for Session 6 speakers, chair & moderator	Auditorium
09:30	11:00	Session 6: Therapeutics	Auditorium
		<i>Chair: TBC</i>	
		<i>Moderator: TBC</i>	
09:30	10:00	Title TBC Sarah Tabrizi, UCL, United Kingdom	
10:00	10:30	Title TBC Robert Wynn, Manchester University NHS, United Kingdom	
10:30	10:45	Short talk selected from abstracts <i>Short talk speaker</i>	
10:45	11:00	Short talk selected from abstracts <i>Short talk speaker</i>	
11:00	11:40	Refreshment break and networking	Event space
11:25	11:40	Briefing for closing keynote speaker, chair, moderator and committee	Auditorium
11:40	12:30	Closing keynote	Auditorium
		<i>Chair: Khadijah Bakur, LiferaOmics, Saudi Arabia</i>	
		<i>Moderator: TBC</i>	
		GUARDIAN and opportunities to improve population genomic health Wendy Chung, Harvard Medical School and Boston Children's Hospital, USA	
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