

Hybrid Conference Programme

Start (BST)	Finish (BST)	Time allocated	Format	Presenter details
Monday 24 April 2023				
12:00	13:00	60 mins		Registration, lunch and networking
12:45	13:00			Briefing for Keynote & Session 1 speakers, microphone runners, chair, moderator & committee - Auditorium
13:00	13:10	10 mins		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	60 mins		Keynote 1 - Lupski lecture
				Chair: Fowzan Alkuraya, King Faisal Specialist Hospital & Research Centre, Saudi Arabia Moderator: Jennifer Posey, Baylor College of Medicine, USA 50 + 10 Q&A mins In-person Title TBC Cynthia Morton, Brigham and Women's Hospital, USA
14:10	14:55	45 mins		Session 1: Gene regulation in rare disease
				Chair: Anna Lindstrand, Karolinska Institute, Sweden Moderator: James Ware, Imperial College London, UK 14:10 14:40 25 + 5 Q&A mins In-person Title TBC Peter Robinson, The Jackson Laboratory, USA 14:40 14:55 10 + 5 Q&A mins Selected from abstracts Short talk speaker
14:55	15:40	45 mins		Refreshment break and networking
15:40	16:25	45 mins		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 25 + 5 Q&A mins In-person Title TBC Alexandre Reymond, University of Lausanne, Switzerland 16:10 16:25 10 + 5 Q&A mins Selected from abstracts Short talk speaker
16:25	17:10	45 mins	Virtual & in-person	Lightning talks for poster session 1: odd numbers
				Chair: James Ware, Imperial College London, UK
17:10	18:30	80 mins	Virtual & in-person	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	90 mins		Session 2: Clinical applications
				<i>Chair: Jennifer Posey, Baylor College of Medicine, USA</i> <i>Moderator: Fowzan Alkuraya, King Faisal Specialist Hospital & Research Centre, Saudi Arabia</i>
09:30	10:00	25 + 5 Q&A mins	In-person	Genomics of rare diseases in Latin America <i>Claudia Gonzaga-Jauregui, International Laboratory for Human Genome Research, UNAM, Mexico</i>
10:00	10:30	25 + 5 Q&A mins	In-person	Founder mutations in rare disease: a Middle Eastern experience <i>Dana Marafi, Kuwait University, Kuwait</i>
10:30	10:45	10 + 5 Q&A mins		Selected from abstracts <i>Short talk speaker</i>
10:45	11:00	10 + 5 Q&A mins		Selected from abstracts <i>Short talk speaker</i>
11:00	11:40	40 mins		Refreshment break and networking
11:25	11:40			Briefing for Session 3 speakers, microphone runners, chair & moderator - Auditorium
11:40	13:10	90 mins		Session 3: Translational genomics
				<i>Chair: James Ware, Imperial College London, UK</i> <i>Moderator: Anna Lindstrand, Karolinska Institute, Sweden</i>
11:40	12:10	25 + 5 Q&A mins	In-person	Making use of population datasets in variant interpretation <i>Kaitlin Samocha, Massachusetts General Hospital, USA</i>
12:10	12:40	25 + 5 Q&A mins	In-person	Brain organoid modeling of neurogenetic disorders uncovers defective cilia dynamics as a significant underlying mechanism <i>Jay Gopalakrishnan, Heinrich Heine University Düsseldorf, Germany</i>
12:40	12:55	10 + 5 Q&A mins		Selected from abstracts <i>Short talk speaker</i>
12:55	13:10	10 + 5 Q&A mins		Selected from abstracts <i>Short talk speaker</i>
13:10	14:20	70 mins		Lunch and networking
14:05	14:20			Briefing for Session 4 speakers, microphone runners, chair & moderator - Auditorium
14:20	15:50	90 mins		Session 4: Public health, prevention and policy
				<i>Chair: Fowzan Alkuraya, King Faisal Specialist Hospital & Research Centre, Saudi Arabia</i> <i>Moderator: Jennifer Posey, Baylor College of Medicine, USA</i>
14:20	14:50	25 + 5 Q&A mins	In-person	Preventing rare disease - Mackenzie's Mission, the Australian Reproductive carrier screening project <i>Nigel Laing, Harry Perkins Institute of Medical Research, Australia</i>

14:50	15:20	25 + 5 Q&A mins	In-person	Preconception carrier screening for rare diseases <i>Lidewij Henneman, Amsterdam UMC, the Netherlands</i>
15:20	15:35	10 + 5 Q&A mins		Selected from abstracts <i>Short talk speaker</i>
15:35	15:50	10 + 5 Q&A mins		Selected from abstracts <i>Short talk speaker</i>
15:50	16:20	30 mins		Refreshment break and networking
16:20	17:20	60 mins		Keynote 2 <i>Chair: Anna Lindstrand, Karolinska Institute, Sweden</i> <i>Moderator: Jennifer Posey, Baylor College of Medicine, USA</i> 50 + 10 Q&A mins Virtual Title TBC David Liu, The Broad Institute, Harvard University, USA
17:20	18:00	40 mins	Virtual & in-person	Lightning talks for poster session 2: even numbers <i>Chair: Jennifer Posey, Baylor College of Medicine, USA</i>
18:00	19:00	60 mins	Virtual & in-person	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	90 mins		Session 5: New technologies <i>Chair: James Ware, Imperial College London, UK</i> <i>Moderator: Jennifer Posey, Baylor College of Medicine, USA</i>
09:30	10:00	25 + 5 Q&A mins	In-person	Title TBC <i>Danny Miller, University of Washington, USA</i>
10:00	10:30	25 + 5 Q&A mins	In-person	Title TBC <i>Gosia Borowiak, Institute of Molecular Biology & Biotechnology, Faculty of Biology, Poland</i>
10:30	10:45	10 + 5 Q&A mins		Selected from abstracts <i>Short talk speaker</i>
10:45	11:00	10 + 5 Q&A mins		Short talk selected from abstracts <i>Short talk speaker</i>
11:00	11:30	30 mins		Refreshment break and networking
11:15	11:30			Briefing for Session 6 speakers, microphone runners, chair & moderator - Auditorium

11:30	13:00	90 mins		Session 6: Genomics and multi-omics
				<i>Chair: Jennifer Posey, Baylor College of Medicine, USA</i>
				<i>Moderator: Anna Lindstrand, Karolinska Institute, Sweden</i>
11:30	12:00	25 + 5 Q&A mins	In-person	Multi-omic approaches to dissect mitochondrial pathology <i>Robert Taylor, Newcastle University, UK</i>
12:00	12:15	10 + 5 Q&A mins		Selected from abstracts <i>Short talk speaker</i>
12:15	12:30	10 + 5 Q&A mins		Selected from abstracts <i>Short talk speaker</i>
12:30	13:00	25 + 5 Q&A mins	In-person	Genetic effects on gene dosage affect cellular phenotypes and disease risk <i>Tuuli Lappalainen, KTH Royal Institute of Technology, Sweden</i>
13:00	13:10			Closing remarks and poster prize presentation
		10-15 mins		Scientific Programme Committee: <i>Fowzan Alkuraya, King Faisal Specialist Hospital & Research Centre, Saudi Arabia</i> <i>Anna Lindstrand, Karolinska Institute, Sweden</i> <i>Jennifer Posey, Baylor College of Medicine, USA</i> <i>James Ware, Imperial College London, UK</i>
13:10	14:00	50 mins		Lunch and departures
13:40				Coach departures for Stansted and Heathrow airports
14:00				Coach departures for Cambridge train station and city centre