Hybrid Conference Programme

Start (BST)	Finish (BST)	Presenter details
Monday 10 July 2023		
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: *Chair: Julia Foreman, EMBL-EBI, UK* *Leslie G. Biesecker, NIH-National Human Genome Research Institute, USA* *Aimé Lumaka, University of Kinshasa, Democratic Republic of Congo* *Meredith Weaver, American College of Medical Genetics & Genomics, USA*
13:10	**14:10**	**Keynote**
		Chair: Aimé Lumaka, University of Kinshasa, Democratic Republic of Congo *Moderator: Leslie G. Biesecker, NIH-National Human Genome Research Institute, USA*
		Harnessing our Common African Genomic Variation to Improve Health Globally *Ambroise Wonkam, John Hopkins University, USA*
14:10	**14:55**	**Session 1: Genetic architecture and locus heterogeneity**
		Chair: Leslie G. Biesecker, NIH-National Human Genome Research Institute, USA *Moderator: Julia Foreman, EMBL-EBI, UK*
14:10	14:40	Title TBC *Nadia Carstens, University of the Witwatersrand, South Africa*
14:40	14:55	Selected from abstracts *Short talk speaker*
14:55	15:40	Refreshment break and networking
15:40	**16:25**	**Session 1 cont: Genetic architecture and locus heterogeneity**
		Chair: Leslie G. Biesecker, NIH-National Human Genome Research Institute, USA *Moderator: Julia Foreman, EMBL-EBI, UK*
15:40	16:10	Genetic variation in cardiomyopathy *Elizabeth McNally, Northwestern University, USA*
16:10	16:25	Selected from abstracts *Short talk speaker*
16:25	**17:10**	**Poster pitch talks for poster session 1: odd numbers**
		Chair: Meredith Weaver, American College of Medical Genetics & Genomics, USA
17:10	**18:30**	**Poster session 1: odd numbers, with refreshments**
18:30	20:30	Dinner
18:30	23:00	Bar open (card payments only)

Tuesday 11 July 2023		
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: Genetic pleiotropy and allelic heterogeneity**
		Chair: Julia Foreman, EMBL-EBI, UK *Moderator: Aimé Lumaka, University of Kinshasa, Democratic Republic of Congo*
09:30	10:00	Challenges to gene/disease curation in some dominantly inherited disorders *Andrew Wilkie, University of Oxford, UK*
10:00	10:30	Molecular genetics of inherited retinal diseases: elevated allelic heterogeneity, very low clinical prevalence, and yet an incredibly high number of unaffected carriers *Carlo Rivolta, University of Basel, Switzerland / University of Leicester, UK*
10:30	10:45	Selected from abstracts *Short talk speaker*
10:45	11:00	Selected from abstracts *Short talk speaker*
11:00	11:40	Refreshment break and networking
11:25	*11:40*	*Briefing for Session 3 speakers, microphone runners, chair & moderator - Auditorium*
11:40	**13:10**	**Session 3: Non-coding variation and polygenic risk scores**
		Chair: Aimé Lumaka, University of Kinshasa, Democratic Republic of Congo *Moderator: Meredith Weaver, American College of Medical Genetics & Genomics, USA*
11:40	12:10	Interpreting variation in the non-coding genome *Nicky Whiffin, University of Oxford, UK*
12:10	12:40	Polygenic risk scores in African populations *Segun Fatumo, LSHTM & MRC/UVRI Uganda Research Unit, UK/Uganda*
12:40	12:55	Selected from abstracts *Short talk speaker*
12:55	13:10	Selected from abstracts *Short talk speaker*
13:10	14:20	Lunch and networking
14:05	*14:20*	*Briefing for Session 4 speakers, microphone runners, chair & moderator - Auditorium*
14:20	**15:50**	**Session 4: Variant classification recommendations**
		Chair: Meredith Weaver, American College of Medical Genetics & Genomics, USA *Moderator: Julia Foreman, EMBL-EBI, UK*
14:20	14:50	Title TBC *Leslie G. Biesecker, NIH-National Human Genome Research Institute, USA*
14:50	15:20	Overview of ACMG/ClinGen Technical Standards for Constitutional CNV Classification *Erin R. Riggs, Geisinger Health System, USA*
15:20	15:35	Selected from abstracts *Short talk speaker*
15:35	15:50	Selected from abstracts *Short talk speaker*
15:50	16:20	Refreshment break and networking
14:05	*14:20*	*Briefing for Session 5 speakers, microphone runners, chair & moderator - Auditorium*
16:20	**17:50**	**Session 5: Iterative reporting**
		Chair: Leslie G. Biesecker, NIH-National Human Genome Research Institute, USA *Moderator: Aimé Lumaka, University of Kinshasa, Democratic Republic of Congo*
16:20	16:45	Reanalysing genomic data in rare disease: time to change the paradigm *Zornitza Stark, Victorian Clinical Genetics Services, Australia*
16:45	17:10	Title TBC *Lisenka Vissers, Radboud University, the Netherlands*
17:10	17:50	Panel discussion *Zornitza Stark, Victorian Clinical Genetics Services, Australia* *Lisenka Vissers, Radboud University, the Netherlands*
17:50	**18:30**	**Poster pitch talks for poster session 2: even numbers**
		Chair: Aimé Lumaka, University of Kinshasa, Democratic Republic of Congo
18:30	**19:30**	**Poster session 2: even numbers, with refreshments**
19:30	21:30	Dinner
19:30	23:00	Bar open (card payments only)

Wednesday 12 July 2023		
07:30	09:00	Breakfast
09:15	*09:30*	*Briefing for Session 6 speakers, microphone runners, chair & moderator - Auditorium*
09:30	**11:00**	**Session 6: Computational approaches**
		Chair: Meredith Weaver, American College of Medical Genetics & Genomics, USA *Moderator: Leslie G. Biesecker, NIH-National Human Genome Research Institute, USA*
09:30	10:00	Title TBC *Caroline Wright, University of Exeter, UK*
10:00	10:30	Predictive diagnostic modelling for rare genetic disease using the published literature *Ian Simpson, University of Edinburgh, UK*
10:30	10:45	Selected from abstracts *Short talk speaker*
10:45	11:00	Selected from abstracts *Short talk speaker*
11:00	11:30	Refreshment break and networking
11:15	*11:30*	*Briefing for Session 7 speakers, microphone runners, chair & moderator - Auditorium*
11:30	**13:00**	**Session 7: High input functional assays**
		Chair: Julia Foreman, EMBL-EBI, UK *Moderator: Meredith Weaver, American College of Medical Genetics & Genomics, USA*
11:30	12:00	New evidence-based paradigms for clinical interpretation of splicing variants to augment black-box predictions *Sandra Cooper, University of Sydney, Australia*
12:00	12:15	Selected from abstracts *Short talk speaker*
12:15	12:30	Selected from abstracts *Short talk speaker*
12:30	13:00	High-throughput functional analysis of PALB2 missense variants and their association with breast cancer risk *Haico van Attikum, Leiden University, the Netherlands*
13:00	**13:10**	**Closing remarks and poster prize presentation**
		Scientific Programme Committee: *Chair: Leslie G. Biesecker, NIH-National Human Genome Research Institute, USA* *Julia Foreman, EMBL-EBI, UK* *Aimé Lumaka, University of Kinshasa, Democratic Republic of Congo* *Meredith Weaver, American College of Medical Genetics & Genomics, USA*
13:10	14:00	Lunch and departures
13:45		Coach departures for Stansted and Heathrow airports
14:00		Coach departures for Cambridge train station and city centre