***Hybrid Conference Programme***

| Start (BST) | Finish (BST) | Presenter details |
|---|---|---|
| **Monday 10 July 2023** | | |
| **12:00** | **12:50** | **Registration, lunch and networking** |
| *12:45* | *13:00* | *Briefing for Keynote & Session 1 speakers, microphone runners, chair, moderator & committee - Auditorium* |
| **12:50** | **13:00** | **Welcome**<br>***Scientific Programme Committee:***<br>*Chair: Julia Foreman, EMBL-EBI, UK*<br>*Leslie G. Biesecker, NIH-National Human Genome Research Institute, USA*<br>*Aimé Lumaka, University of Kinshasa, Democratic Republic of Congo*<br>*Meredith Weaver, American College of Medical Genetics & Genomics, USA* |
| **13:00** | **14:00** | **Keynote**<br>*Chair: Aimé Lumaka, University of Kinshasa, Democratic Republic of Congo*<br>*Moderator: Leslie G. Biesecker, NIH-National Human Genome Research Institute, USA*<br>Harnessing our Common African Genomic Variation to Improve Health Globally<br>*Ambroise Wonkam, John Hopkins University, USA* |
| 14:00 | 14:05 | Comfort break |
| **14:05** | **14:55** | **Session 1: Genetic architecture and locus heterogeneity**<br>*Chair: Leslie G. Biesecker, NIH-National Human Genome Research Institute, USA*<br>*Moderator: Julia Foreman, EMBL-EBI, UK* |
| 14:05 | 14:35 | Deciphering Developmental Disorders in Africa (DDD-Africa) Study – insights and opportunities from the first 120 cases<br>*Nadia Carstens, University of the Witwatersrand, South Africa* |
| 14:35 | 15:05 | Genetic variation in cardiomyopathy<br>*Elizabeth McNally, Northwestern University, USA* |
| 15:05 | 15:20 | Identification of novel and reported pathogenic variants in ten FANC genes in Mexican patients with Fanconi anemia<br>*Leda Torres, Instituto Nacional de Pediatria, México* |
| 15:20 | 15:35 | Challenges in evaluating clinical actionability of genomic findings over nine years of ClinGen evidence-based assessments<br>*Heidi Cope, RTI International, USA* |
| 15:35 | 16:15 | Refreshment break and networking |
| *16:00* | *16:15* | *Briefing for Session 2 speakers, microphone runners, chair & moderator - Auditorium* |
| **16:15** | **17:45** | **Session 2: Genetic pleiotropy and allelic heterogeneity**<br>*Chair: Julia Foreman, EMBL-EBI, UK*<br>*Moderator: Aimé Lumaka, University of Kinshasa, Democratic Republic of Congo* |
| 16:15 | 16:45 | Challenges to gene/disease curation in some dominantly inherited disorders<br>*Andrew Wilkie, University of Oxford, UK* |
| 16:45 | 17:15 | Molecular genetics of inherited retinal diseases: elevated allelic heterogeneity, very low clinical prevalence, and yet an incredibly high number of unaffected carriers<br>*Carlo Rivolta, University of Basel, Switzerland / University of Leicester, UK* |
| 17:15 | 17:30 | Polycystic kidney disease: Biallelic PKD1 and monoallelic PKHD1 ClinGen gene curations<br>*Tam Sneddon, University of North Carolina USA* |
| 17:30 | 17:45 | Check the test: assessment of genotype interpretation in Direct-to-Consumer genetic tests<br>*Peter Taschner, UAS Leiden Netherlands* |
| **17:45** | **18:10** | **Poster pitch talks for poster session 1: odd numbers**<br>*Chair: Meredith Weaver, American College of Medical Genetics & Genomics, USA* |
| **18:10** | **19:15** | **Poster session 1: odd numbers, with refreshments** |
| 19:15 | 20:30 | Dinner |
| 19:15 | 23:00 | Bar open (card payments only) |

| Tuesday 11 July 2023 | | |
|---|---|---|
| 07:30 | 09:00 | Breakfast |
| **09:00** | **09:30** | **Resources, tools and database networking - event space** |
| *09:15* | *09:30* | *Briefing for Session 3 speakers, microphone runners, chair & moderator - Auditorium* |
| **09:30** | **11:00** | **Session 3: Non-coding variation and polygenic risk scores**<br>*Chair: Aimé Lumaka, University of Kinshasa, Democratic Republic of Congo*<br>*Moderator: Meredith Weaver, American College of Medical Genetics & Genomics, USA* |
| 09:30 | 10:00 | Polygenic risk scores in African populations - VIRTUAL<br>*Segun Fatumo, LSHTM & MRC/UVRI Uganda Research Unit, UK/Uganda* |
| 10:00 | 10:30 | Interpreting variation in the non-coding genome<br>*Nicky Whiffin, University of Oxford, UK* |
| 10:30 | 10:45 | Characterizing clinical actionability in the context of polygenic risk assessment<br>*Jessica Hunter, RTI International, USA* |
| 10:45 | 11:00 | BRCA1 secondary mutations at splice-sites drive exon-skipping and PARP inhibitor resistance<br>*Matthew Wakefield, The Walter and Eliza Hall Institute, Australia* |
| 11:00 | 11:30 | Refreshment break and networking |
| *11:20* | *11:30* | *Briefing for Session 4 speakers, microphone runners, chair & moderator - Auditorium* |
| **11:30** | **13:15** | **Session 4: Variant classification recommendations**<br>*Chair: Meredith Weaver, American College of Medical Genetics & Genomics, USA*<br>*Moderator: Julia Foreman, EMBL-EBI, UK* |
| 11:30 | 12:00 | Variant classification recommendations<br>*Leslie G. Biesecker, NIH-National Human Genome Research Institute, USA* |
| 12:00 | 12:30 | Overview of ACMG/ClinGen technical standards for constitutional CNV classification<br>*Erin R. Riggs, Geisinger Health System, USA* |
| 12:30 | 12:45 | Utilization of REVEL at increased strength affects inborn errors of metabolism genes differently<br>*Alexa Dickson, Washington University in St Louis, USA* |
| 12:45 | 13:00 | *CanVIG-UK (Cancer Variant Interpretation Group-UK) national survey of variant workflows across molecular diagnostics laboratories reveals commonality of problematic steps*<br>*Sophie Allen, The Institute of Cancer Research, UK* |
| 13:00 | 13:15 | CNV classification on exomes from a large rare disease cohort - VIRTUAL<br>*Gabrielle Lemire, Broad Institute, USA* |
| 13:15 | 14:45 | Lunch and networking |
| *14:30* | *14:45* | *Briefing for Session 5 speakers, microphone runners, chair & moderator - Auditorium* |
| **14:45** | **15:45** | **Session 5: Iterative reporting**<br>*Chair: Leslie G. Biesecker, NIH-National Human Genome Research Institute, USA*<br>*Moderator: Aimé Lumaka, University of Kinshasa, Democratic Republic of Congo* |
| 14:45 | 15:15 | Reanalysing genomic data in rare disease: time to change the paradigm<br>*Zornitza Stark, Victorian Clinical Genetics Services, Australia* |
| 15:15 | 15:45 | It has got to be genetic: the power of longitudinal cohorts to diagnose the undiagnosed<br>*Lisenka Vissers, Radboud University, the Netherlands* |
| 15:45 | 16:15 | Refreshment break and networking |
| **16:15** | **17:15** | **Session 5: Iterative reporting continued**<br>Panel discussion<br>*Steven Harrison, Ambry Genetics, USA*<br>*Zornitza Stark, Victorian Clinical Genetics Services, Australia*<br>*Lisenka Vissers, Radboud University, the Netherlands*<br>*Caroline Wright, University of Exeter, UK*<br>*Sarah Wynn, Unique, UK* |
| **17:15** | **17:40** | **Poster pitch talks for poster session 2: even numbers**<br>*Chair: Aimé Lumaka, University of Kinshasa, Democratic Republic of Congo* |
| **17:40** | **18:45** | **Poster session 2: even numbers, with refreshments** |
| 18:45 | 21:30 | Dinner |
| 18:45 | 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**<br>*Chair: Meredith Weaver, American College of Medical Genetics & Genomics, USA*<br>*Moderator: Leslie G. Biesecker, NIH-National Human Genome Research Institute, USA* |
| 09:30 | 10:00 | Evaluating and improving computational variant classification<br>*Caroline Wright, University of Exeter, UK* |
| 10:00 | 10:30 | Modelling gene-to-phenotype relationships in monogenic neurological diseases<br>*Ian Simpson, University of Edinburgh, UK* |
| 10:30 | 10:45 | C2S2: Phenotype-driven clustering for discovery of disease subgroups<br>*Daniel Danis, The Jackson Laboratory for Genomic Medicine, USA* |
| 10:45 | 11:00 | The likelihood ratio calculator: bridging the Bayesian-Frequentist divide to enable flexible allocation of evidence weighting for case-control data (PS4)<br>*Chey Loveday, Institute of Cancer Research, UK* |
| 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: Functional assays**<br>*Chair: Julia Foreman, EMBL-EBI, UK*<br>*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<br>*Sandra Cooper, University of Sydney, Australia* |
| 12:00 | 12:15 | Resolving the full functional spectrum of POT1 alleles via saturation genome editing<br>*Sofia Obolenski, Wellcome Sanger Institute, UK* |
| 12:15 | 12:30 | Saturation genome editing to classify all possible mis sense variants of BRCA2-C-terminal DNA binding domain<br>*Sounak Sahu, National Cancer Institute, USA* |
| 12:30 | 13:00 | High-throughput functional analysis of PALB2 missense variants and their association with breast cancer risk<br>*Haico van Attikum, Leiden University, the Netherlands* |
| **13:00** | **13:10** | **Closing remarks and poster prize presentation**<br>***Scientific Programme Committee:***<br>*Chair: Leslie G. Biesecker, NIH-National Human Genome Research Institute, USA*<br>*Julia Foreman, EMBL-EBI, UK*<br>*Aimé Lumaka, University of Kinshasa, Democratic Republic of Congo*<br>*Meredith Weaver, American College of Medical Genetics & Genomics, USA* |
| 13:10 | 14:00 | Lunch and departures |
| 14:00 | | Coach departures for Stansted and Heathrow airports |
| 14:10 | | Coach departures for Cambridge train station and city centre |