

Virtual Conference Agenda

Start (BST)	Finish (BST)	Presenter details
-------------	--------------	-------------------

Monday 28 March 2022

12:00 12:10 Welcome

Scientific Programme Committee:

[Fowzan Sami Alkuraya, King Faisal Specialist Hospital & Research Centre, Saudi Arabia](#)

[Jennifer Posey, Baylor College of Medicine, USA](#)

[Lisenka Vissers, Radboudumc University, Netherlands](#)

[James Ware, Imperial College London, UK](#)

12:10 12:55 Keynote - Lupski lecture

Introduction to the session

Chair: James Ware, Imperial College London, UK

12:10 12:40 Deciphering Developmental Disorders – How the DDD study advanced diagnosis and discovery

[Helen Firth, Cambridge University Hospitals, UK](#)

12:40 12:55 Q&A

Chair: James Ware, Imperial College London, UK

Moderator: Lisenka Vissers, Radboudumc University, Netherlands

12:55 13:20 Break and networking in Spatial Chat

13:20 15:00 Session 1: Diverse genomes as drivers of precision medicine

Introduction to the session

Chair: Lisenka Vissers, Radboudumc University, Netherlands

13:20 13:40 Implementation of precision medicine in an Asian population

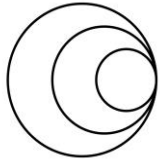
[Saumya Jamuar, KK Women's and Children's Hospital, Singapore](#)

13:40 14:00 Rare diseases in diverse populations: clinical whole genome sequencing experience from Istanbul, an EMEA hub

[Yasemin Alanay, Acibadem University School of Medicine, Turkey](#)

14:00 14:10 Deciphering Developmental Disorders in Africa (DDD-Africa) study – expanding phenotypes using exome sequencing in an African setting

Nadia Carstens, University of the Witwatersrand, South Africa



14:10 14:20 Recessive burden analysis and gene discovery in ~40,000 developmental disorder trios with diverse ancestries
Kartik Chundru, Wellcome Sanger Institute, UK

14:20 14:30 Tissue-specific exon skipping driving genetic pleiotropy in CEP290-related ciliopathies presents opportunity for splice-switching oligonucleotide therapies
Rowan Taylor, University of Leeds, UK

14:30 15:00 Q&A
Chair: Lisenka Vissers, Radboudumc University, Netherlands
Moderator: Jennifer Posey, Baylor College of Medicine, USA

15:00 15:20 Break and networking in Spatial Chat

15:20 16:40 Poster Session 1: Odd Numbers

15:20 15:50 Lightning talks for poster session 1: odd numbers

15:50 16:40 Poster session 1: odd numbers

16:40 18:00 Session 2: Non-mendelian forms of rare disease

Introduction to the session
Chair: Jennifer Posey, Baylor College of Medicine, USA

16:40 17:00 Genome versus locus mutation: genetics & genomics of somatic & organismal disease
[*James R Lupski, Baylor College of Medicine, USA*](#)

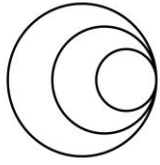
17:00 17:10 Compound inheritance gene dosage model for lethal lung developmental disorders
Justyna Karolak, Poznan University of Medical Sciences, Poland

17:10 17:20 Haplotype-based analysis highlights the contribution of common regulatory and protein-coding TYR variants in the genetic architecture of albinism
Panos Sergouniotis, University of Manchester, UK

17:20 17:30 The female protective effect against autism spectrum disorder
Emilie Wigdor, Wellcome Sanger Institute, UK

17:30 18:00 Q&A
Chair: Jennifer Posey, Baylor College of Medicine, USA
Moderator: James Ware, Imperial College London, UK

18:00 18:20 Networking in Spatial Chat



Tuesday 29 March 2022

12:00 13:35 Session 3: Treatment of rare diseases

Introduction to the session

Chair: Fowzan Sami Alkuraya, King Faisal Specialist Hospital & Research Centre, Saudi Arabia

12:00 12:20 Advances in the treatment of genetic diseases: from gene search to drug discovery and clinical trials

[Arnold Munnich, INSERM, France](#)

12:20 12:40 Prospects for genetic cures in inherited cardiomyopathy

[Hugh Watkins, University of Oxford, UK](#)

12:40 12:50 PhenoApt leverages clinical expertise to prioritize candidate genes via machine learning

Zefu Chen, Peking Union Medical College, China

12:50 13:00 Identifying Orphan drugs as potential therapies in rare diseases that they were not designated for

Hassan Shakeel, Sanger / CUH / NIHR, UK

13:00 13:05 Highly commended abstract lightning talk: Experience of genomic testing and personalised management of genetic epilepsy syndromes from India

Vivekananda Bhat, Kasturba Medical College, India

13:05 13:35 Q&A

Chair: Fowzan Sami Alkuraya, King Faisal Specialist Hospital & Research Centre, Saudi Arabia

Moderator: Lisenka Vissers, Radboudumc University, Netherlands

13:35 14:00 Break and networking in Spatial Chat

14:00 15:10 Poster Session 2: Even Numbers

14:00 14:30 Lightning talks for poster session 2: odd numbers

14:30 15:10 Poster session 2: even numbers

15:10 15:30 Break and networking in Spatial Chat

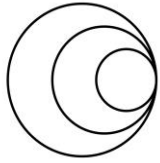
15:30 17:10 Session 4: High throughput functional assays to improve variant interpretation

Introduction to the session

Chair: Jennifer Posey, Baylor College of Medicine, USA

15:30 15:50 Progress and promise of MAVE for clinical variant interpretation

[Lea Starita, University of Washington, USA](#)



- 15:50 16:10 Using CRISPR-based saturation genome editing to improve the diagnosis of neurodevelopmental disorders
[Elizabeth Radford, Wellcome Sanger Institute, UK](#)
- 16:10 16:20 Loss of function variants in MYCBP2 cause neurodevelopmental phenotypes and corpus callosum dysgenesis
[Lama AlAbdi, King Faisal Specialist Hospital & Research Centre, Saudi Arabia](#)
- 16:20 16:30 Properties of altered function versus loss of function mutations in developmental disorders: bioinformatic insights to aid functional assays, gene discovery and precision medicine
[Katrina Andrews, Wellcome Sanger Institute, UK](#)
- 16:30 16:40 Quantifying the contribution of near-coding variation to rare disease
[Alexandra Geary, University of Oxford, UK](#)
- 16:40 17:10 Q&A
Chair: [Jennifer Posey, Baylor College of Medicine, USA](#)
Moderator: [James Ware, Imperial College London, UK](#)

17:10 17:20 Closing remarks

Scientific Programme Committee:

[Fowzan Sami Alkuraya, King Faisal Specialist Hospital & Research Centre, Saudi Arabia](#)

[Jennifer Posey, Baylor College of Medicine, USA](#)

[Lisenka Vissers, Radboudumc University, Netherlands](#)

[James Ware, Imperial College London, UK](#)

17:20 17:40 Networking in Spatial Chat