Virtual Conference Agenda

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

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

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

Break and networking in Spatial Chat

Poster Session 1: Odd Numbers

Lightning talks for poster session 1: odd numbers
Poster session 1: odd numbers

Session 2: Non-mendelian forms of rare disease

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

Genome versus locus mutation: genetics & genomics of somatic & organismal disease
James R Lupski, Baylor College of Medicine, USA

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

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

The female protective effect against autism spectrum disorder
Emilie Wigdor, Wellcome Sanger Institute, UK

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

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

<table>
<thead>
<tr>
<th>Time</th>
<th>Speaker</th>
<th>Title</th>
</tr>
</thead>
</table>
| 12:00  |         | **Advances in the treatment of genetic diseases: from gene search to drug discovery and clinical trials**  
Arnold Munnich, INSERM, France |
| 12:20  |         | **Prospects for genetic cures in inherited cardiomyopathy**  
Hugh Watkins, University of Oxford, UK |
| 12:40  |         | **PhenoApt leverages clinical expertise to prioritize candidate genes via machine learning**  
Zefu Chen, Peking Union Medical College, China |
| 12:50  |         | **Identifying Orphan drugs as potential therapies in rare diseases that they were not designated for**  
Hassan Shakeel, Sanger / CUH / NIHR, UK |
| 13:00  |         | **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  |         | **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

<table>
<thead>
<tr>
<th>Time</th>
<th>Activity</th>
</tr>
</thead>
<tbody>
<tr>
<td>14:00</td>
<td><strong>Lightning talks for poster session 2: odd numbers</strong></td>
</tr>
<tr>
<td>15:10</td>
<td><strong>Break and networking in Spatial Chat</strong></td>
</tr>
</tbody>
</table>

#### 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

<table>
<thead>
<tr>
<th>Time</th>
<th>Speaker</th>
<th>Title</th>
</tr>
</thead>
</table>
| 15:30  |         | **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