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Genomics of Rare Disease

28-29 March 2022

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 clinial 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

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17:20	17:40	Networking in Spatial Chat