

Virtual Conference Agenda

Start (BST)	Finish (BST)	Presenter details
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Monday 28 March 2022

13:00	13:10	Welcome
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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](#)

13:10	13:55	Keynote - Lupski Lecture
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Introduction to the session

Chair: James Ware, Imperial College London, UK

13:10	13:40	Deciphering Developmental Disorders – How the DDD study advanced diagnosis and discovery
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[Helen Firth, Cambridge University Hospitals, UK](#)

13:40	13:55	Q&A
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Chair: James Ware, Imperial College London, UK

Moderator: Lisenka Vissers, Radboudumc University, Netherlands

13:55	14:20	Networking using Spatial Chat
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14:20	14:30	Break
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14:30	16:00	Session 1: Diverse genomes as drivers of precision medicine
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Introduction to the session

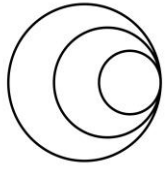
Chair: Lisenka Vissers, Radboudumc University, Netherlands

14:30	14:50	Implementation of precision medicine in an Asian population
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[Saumya Januar, KK Women's and Children's Hospital, Singapore](#)

14:50	15:10	Rare diseases in diverse populations: clinical WGS experience from Istanbul, an EMEA hub
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[Yasemin Alanay, Acibadem University School of Medicine, Turkey](#)



- 15:10 15:20 Short talk selected from abstracts
- 15:20 15:30 Short talk selected from abstracts
- 15:30 16:00 Q&A
Chair: Lisenka Vissers, Radboudumc University, Netherlands
Moderator: Jennifer Posey, Baylor College of Medicine, USA

16:00 16:10 Break

16:10 16:40 Networking using Spatial Chat

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 Short talk selected from abstracts

17:10 17:20 Short talk selected from abstracts

17:20 17:30 Short talk selected from abstracts

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

Tuesday 29 March 2022

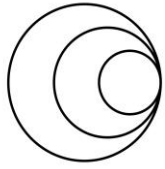
13:00 14:30 Session 3: Treatment of rare diseases

Introduction to the session

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

13:00 13:20 Talk title TBC
[Arnold Munnich, INSERM, France](#)

13:20 13:40 Prospects for genetic cures in inherited cardiomyopathy
[Hugh Watkins, University of Oxford, UK](#)



13:40 13:50 Short talk selected from abstracts

13:50 14:00 Short talk selected from abstracts

14:00 14:30 Q&A

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

Moderator: Lisenka Vissers, Radboudumc University, Netherlands

14:30 14:50 Break

14:50 16:00 Poster Session

14:50 15:20 Poster lightning talks

15:20 16:00 Poster session and networking using Spatial Chat

16:00 16:10 Break

16:10 17:40 Session 4: High throughput functional assays to improve variant interpretation

Introduction to the session

Chair: Jennifer Posey, Baylor College of Medicine, USA

16:10 16:30 Progress and promise of MAVE for clinical variant interpretation

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

16:30 16:50 Using saturation genome editing to improve the diagnosis of developmental disorders

[*Elizabeth Redford, Wellcome Sanger Institute, UK*](#)

16:50 17:00 Short talk selected from abstracts

17:00 17:10 Short talk selected from abstracts

17:10 17:40 Q&A

Chair: Jennifer Posey, Baylor College of Medicine, USA

Moderator: James Ware, Imperial College London, UK

17:40 17:50 Closing remarks

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