

Virtual Conference Agenda

Start (BST)	Finish (BST)	Presenter details
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Wednesday 12 May 2021

13:00 13:10 Welcome

		<p>Scientific Programme Committee: Helen Firth, Cambridge University Hospitals, UK Steven Harrison, Broad Institute, USA Subha Madhavan, Georgetown University Medical Center, USA Gert Matthijs, University of Leuven, Belgium Sharon Plon, Baylor College of Medicine, USA Caroline Wright, University of Exeter, UK</p>
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13:10 13:55 Keynote

		<p>Introduction to the session Chair: Helen Firth, Cambridge University Hospitals, UK</p>
13:10	13:40	<p>Curating the variome John Burn, Newcastle University, UK</p>
13:40	13:55	<p>Q&A Chair: Helen Firth, Cambridge University Hospitals, UK Moderator: Caroline Wright, University of Exeter, UK</p>
13:55	14:15	Break

14:15 15:45 Session 1: Population and Penetrance I

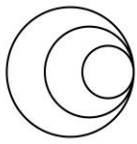
		<p>Introduction to the session Chair: Steven Harrison, Broad Institute, USA</p>
14:15	14:35	<p>Reference population databases improve variant curation Anne O'Donnell Luria, Broad Institute, USA</p>
14:35	14:55	<p>Challenging the traditional definition of penetrance using monogenic diabetes as an example Kashyap Patel, University of Exeter, UK</p>
14:55	15:05	<p>Launch of the Gene Curation Coalition Database Marina DiStefano, Geisinger, USA</p>
15:05	15:15	<p>Towards updated recommendations and protocols for the use of computational tools in missense variant pathogenicity assessment Marc Greenblatt, University of Vermont, USA</p>
15:15	15:45	<p>Q&A Chair: Steven Harrison, Broad Institute, USA Moderator: Helen Firth, Cambridge University Hospitals, UK</p>
15:45	16:00	Break

16:00 16:50 Poster Session 1 - for poster numbers P1-P26

16:00	16:20	Poster session 1 lightning talks - P1-P26
16:20	16:50	Poster session 1 - P1-P26

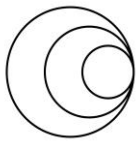
16:50 18:20 Session 2: Population and Penetrance II

		<p>Introduction to the session Chair: Caroline Wright, University of Exeter, UK</p>
16:50	17:10	<p>Polygenic risk scores: moving from research to implementation Cathryn Lewis, King's College London, UK</p>
17:10	17:30	<p>Tantalizing dilemma in genetics risk prediction: case of African genomics Emile Chimusa, University of Cape Town, South Africa</p>
17:30	17:40	<p>Investigating the penetrance of rare genetic variants in developmental disorder genes in UK Biobank Rebecca Kingdom, University of Exeter, UK</p>
17:40	17:50	<p>Determining if polygenic scores provide clinical utility Sowmiya Moorthie, PHG Foundation, UK</p>
17:50	18:20	<p>Q&A Chair: Caroline Wright, University of Exeter, UK Moderator: Steven Harrison, Broad Institute, USA</p>



Thursday 13 May 2021

13:00		14:30		Session 3: Testing and Screening I
		Introduction to the session <i>Chair: Gert Matthijs, University of Leuven, Belgium</i>		
13:00	13:20	Outcome of 153,575 non-invasive prenatal tests within a publicly-funded nationwide first-tier screening scheme <i>Kris Van Den Bogaert, Center of Human Genetics, University of Leuven, Belgium</i>		
13:20	13:40	Direct to consumer genetic testing: issues to consider <i>Anneke Lucassen, University of Southampton, UK</i>		
13:40	13:50	Exome CNV calling and analysis in a large cohort of families with undiagnosed rare genetic disease <i>Gabrielle Lemire, Broad Institute, USA</i>		
13:50	14:00	The role of exome sequencing in newborn screening <i>Steven Brenner, UC Berkeley, USA</i>		
14:00	14:30	Q&A <i>Chair: Gert Matthijs, University of Leuven, Belgium</i> <i>Moderator: Caroline Wright, University of Exeter, UK</i>		
14:30	14:40	Break		
14:40		15:30		Poster Session 2 - for poster numbers P27-P51
14:40	15:00	Poster session 2 lightning talks - P27-P51		
15:00	15:30	Poster session 2 - P27-P51		
15:30		17:10		Session 4: Testing and Screening II
		Introduction to the session <i>Chair: Gert Matthijs, University of Leuven, Belgium</i>		
15:30	15:50	Full genome analysis for rare disease diagnosis <i>Pui-Yan Kwok, University of California, San Francisco, USA</i>		
15:50	16:10	Identification of neuropsychiatric CNVs in a health system population: high prevalence, penetrance, and personal utility <i>Christa Martin, Geisinger, USA</i>		
16:10	16:30	How about RNA in genomic testing? <i>Diana Baralle, University of Southampton, UK</i>		
16:30	16:40	Integration of proteomics with genomics and transcriptomics increases the diagnostic rate of Mendelian disorders <i>Robert Kopajtich, Technical University Munich, Germany</i>		
16:40	17:10	Q&A <i>Chair: Gert Matthijs, University of Leuven, Belgium</i> <i>Moderator: Steven Harrison, Broad Institute, USA</i>		
17:10	17:20	Break		
17:20		18:00		DECIPHER Demonstration
		Introduction to the session <i>Chair: Helen Firth, Cambridge University Hospitals, UK</i>		
17:20	17:40	DECIPHER – Facilitating the sharing and interpretation of rare disease variant data and associated clinical phenotypes in the genomic era <i>Julia Foreman, Wellcome Sanger Institute, UK</i>		
17:40	18:00	Discussion and Q&A <i>Chair: Helen Firth, Cambridge University Hospitals, UK</i> <i>Moderator: Caroline Wright, University of Exeter, UK</i>		



Friday 14 May 2021

13:00		14:50		Session 5: Precision Oncology
				Introduction to the session <i>Chair: Sharon Plon, Baylor College of Medicine, USA</i>
13:00	13:20			Master protocol clinical trial for cancer precision medicine <i>Yeul Hong Kim, Korea University College of Medicine, Republic of Korea</i>
13:20	13:40			Genomics in metastatic breast cancer: the AURORA molecular screening programme <i>Philippe Aftimos, Jules Bordet Cancer Institute, Belgium</i>
13:40	14:00			Precision medicine in cancer: incorporating molecular testing in the treatment decision making process <i>Michael Pishvaian, John Hopkins University School of Medicine, USA</i>
14:00	14:10			The singleton rare missense variant: Pheochromocytoma and SDHB/SDHD as an exemplar for quantifying phenotypic specificity <i>Clare Turnbull, Institute of Cancer Research, UK</i>
14:10	14:20			Exploring rare germline variants in childhood cancer patients with features suggestive of an underlying genetic predisposition to cancer <i>Dianne Sylvester, Kids Research, Australia</i>
14:20	14:50			Q&A <i>Chair: Sharon Plon, Baylor College of Medicine, USA</i> <i>Moderator: Steven Harrison, Broad Institute, USA</i>
14:50	15:10			Break
15:10		16:40		Session 6: Patients and Treatments
				Introduction to the session <i>Chair: Sharon Plon, Baylor College of Medicine, USA</i>
15:10	15:20			The shared responsibility of recontact: patients obtaining updated genetic test results through patient registries <i>Juliann Savatt, Geisinger, USA</i>
15:20	15:40			Cardiovascular precision medicine: diagnostics and therapeutics <i>Euan Ashley, Stanford University Medical Center, USA</i>
15:40	16:00			Genetic-guided therapies in the epilepsies <i>Heather Mefford, St Jude Children's Research Hospital, USA</i>
16:00	16:30			Q&A <i>Chair: Sharon Plon, Baylor College of Medicine, USA</i> <i>Moderator: Helen Firth, Cambridge University Hospitals, UK</i>
16:30	16:50			Closing remarks
				Scientific Programme Committee: <i>Helen Firth, Cambridge University Hospitals, UK</i> <i>Steven Harrison, Broad Institute, USA</i> <i>Subha Madhavan, Georgetown University Medical Center, USA</i> <i>Gert Matthijs, University of Leuven, Belgium</i> <i>Sharon Plon, Baylor College of Medicine, USA</i> <i>Caroline Wright, University of Exeter, UK</i>