wellcome connecting science

Curating the Clinical Genome

12-14 May 2021

Virtual Conference Agenda

Start (BST)	Finish (BST)	Presenter details
Wednesday 12 May 2021		
13:00	13:10	**Welcome**
		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*
13:10	13:55	**Keynote**
		Introduction to the session *Chair: Helen Firth, Cambridge University Hospitals, UK*
13:10	13:40	Curating the variome *John Burn, Newcastle University, UK*
13:40	13:55	Q&A *Chair: Helen Firth, Cambridge University Hospitals, UK* *Moderator: Caroline Wright, University of Exeter, UK*
13:55	14:15	Break
14:15	15:45	**Session 1: Population and Penetrance I**
		Introduction to the session *Chair: Steven Harrison, Broad Institute, USA*
14:15	14:35	Reference population databases improve variant curation *Anne O'Donnell Luria, Broad Institute, USA*
14:35	14:55	Challenging the traditional definition of penetrance using monogenic diabetes as an example *Kashyap Patel, University of Exeter, UK*
14:55	15:05	Launch of the Gene Curation Coalition Database *Marina DiStefano, Geisinger, USA*
15:05	15:15	Towards updated recommendations and protocols for the use of computational tools in missense variant pathogenicity assessment *Marc Greenblatt, University of Vermont, USA*
15:15	15:45	Q&A *Chair: Steven Harrison, Broad Institute, USA* *Moderator: Helen Firth, Cambridge University Hospitals, UK*
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**
		Introduction to the session *Chair: Caroline Wright, University of Exeter, UK*
16:50	17:10	Polygenic risk scores: moving from research to implementation *Cathryn Lewis, King's College London, UK*
17:10	17:30	Tantalizing dilemma in genetics risk prediction: case of African genomics *Emile Chimusa, University of Cape Town, South Africa*
17:30	17:40	Investigating the penetrance of rare genetic variants in developmental disorder genes in UK Biobank *Rebecca Kingdom, University of Exeter, UK*
17:40	17:50	Determining if polygenic scores provide clinical utility *Sowmiya Moorthie, PHG Foundation, UK*
17:50	18:20	Q&A *Chair: Caroline Wright, University of Exeter, UK* *Moderator: Steven Harrison, Broad Institute, USA*

Thursday 13 May 2021

13:00	14:30	**Session 3: Testing and Screening I**
		Introduction to the session *Chair: Gert Matthijs, University of Leuven, Belgium*
13:00	13:20	Outcome of 153,575 non-invasive prenatal tests within a publicly-funded nationwide first-tier screening scheme *Kris Van Den Bogaert, Center of Human Genetics, University of Leuven, Belgium*
13:20	13:40	Direct to consumer genetic testing: issues to consider *Anneke Lucassen, University of Southampton, UK*
13:40	13:50	Exome CNV calling and analysis in a large cohort of families with undiagnosed rare genetic disease *Gabrielle Lemire, Broad Institute, USA*
13:50	14:00	The role of exome sequencing in newborn screening *Steven Brenner, UC Berkeley, USA*
14:00	14:30	Q&A *Chair: Gert Matthijs, University of Leuven, Belgium* *Moderator: Caroline Wright, University of Exeter, UK*
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 *Chair: Gert Matthijs, University of Leuven, Belgium*
15:30	15:50	Full genome analysis for rare disease diagnosis *Pui-Yan Kwok, University of California, San Fransisco, USA*
15:50	16:10	Identification of neuropsychiatric CNVs in a health system population: high prevalence, penetrance, and personal utility *Christa Martin, Geisinger, USA*
16:10	16:30	How about RNA in genomic testing? *Diana Baralle, University of Southampton, UK*
16:30	16:40	Integration of proteomics with genomics and transcriptomics increases the diagnostic rate of Mendelian disorders *Robert Kopajtich, Technical University Munich, Germany*
16:40	17:10	Q&A *Chair: Gert Matthijs, University of Leuven, Belgium* *Moderator: Steven Harrison, Broad Institute, USA*
17:10	17:20	Break

17:20	18:00	**DECIPHER Demonstration**
		Introduction to the session *Chair: Helen Firth, Cambridge University Hospitals, UK*
17:20	17:40	DECIPHER – Facilitating the sharing and interpretation of rare disease variant data and associated clinical phenotypes in the genomic era *Julia Foreman, Wellcome Sanger Institute, UK*
17:40	18:00	Discussion and Q&A *Chair: Helen Firth, Cambridge University Hospitals, UK* *Moderator: Caroline Wright, University of Exeter, UK*

Friday 14 May 2021

13:00	14:50	**Session 5: Precision Oncology**
		Introduction to the session *Chair: Sharon Plon, Baylor College of Medicine, USA*
13:00	13:20	Master protocol clinical trial for cancer precision medicine *Yeul Hong Kim, Korea University College of Medicine, Republic of Korea*
13:20	13:40	Genomics in metastatic breast cancer: the AURORA molecular screening programme *Philippe Aftimos, Jules Bordet Cancer Institute, Belgium*
13:40	14:00	Precision medicine in cancer: incorporating molecular testing in the treatment decision making process *Michael Pishvaian, John Hopkins University School of Medicine, USA*
14:00	14:10	The singleton rare missense variant: Phaeochromocytoma and SDHB/SDHD as an exemplar for quantifying phenotypic specificity *Clare Turnbull, Institute of Cancer Research, UK*
14:10	14:20	Exploring rare germline variants in childhood cancer patients with features suggestive of an underlying genetic predisposition to cancer *Dianne Sylvester, Kids Research, Australia*
14:20	14:50	Q&A *Chair: Sharon Plon, Baylor College of Medicine, USA* *Moderator: Steven Harrison, Broad Institute, USA*
14:50	15:10	Break

15:10	16:40	**Session 6: Patients and Treatments**
		Introduction to the session *Chair: Sharon Plon, Baylor College of Medicine, USA*
15:10	15:20	The shared responsibility of recontact: patients obtaining updated genetic test results through patient registries *Juliann Savatt, Geisinger, USA*
15:20	15:40	Cardiovascular precision medicine: diagnostics and therapeutics *Euan Ashley, Stanford University Medical Center, USA*
15:40	16:00	Genetic-guided therapies in the epilepsies *Heather Mefford, St Jude Children's Research Hospital, USA*
16:00	16:30	Q&A *Chair: Sharon Plon, Baylor College of Medicine, USA* *Moderator: Helen Firth, Cambridge University Hospitals, UK*
16:30	16:50	**Closing remarks**

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