Hybrid Conference Programme

Start (BST)	Finish (BST)	
Thursday 13 July 2023		
09:00	09:50	**Registration, refreshments and networking**
09:35	*09:50*	*Briefing for Keynote & Session 1 speakers, microphone runners, chair, moderator & committee - Auditorium*
09:50	**10:00**	**Welcome** ***Scientific Programme Committee:*** *Matthew Hurles, Wellcome Sanger Institute, UK* *David Adams, Wellcome Sanger Institute, UK* *Clare Turnbull, The Institute of Cancer Research, UK* *Lea Starita, University of Washington, USA* *Lara Muffley, University of Washington, USA*
10:00	11:00	**Keynote speaker** *Chair: Matthew Hurles, Wellcome Sanger Institute, UK and Doug Fowler, University of Washington, USA* *Moderator: Benedetta Bolognesi, Institute for Bioengineering of Catalunya, IBEC, Spain*
10:00	11:00	Towards a contextual atlas of variant effects *Fritz Roth, University of Toronto, Canada*
11:00	11:30	Refreshment break and networking
11:30	13:30	**Session 1: Clinical applications of functional variant maps** *Chair: Clare Turnbull, The Institute of Cancer Research, UK* *Moderator: Elizabeth Radford, Wellcome Sanger Institute, UK*
11:30	12:00	Calibrating Assays for Variant Pathogenicity *Leslie Biesecker, NIH, USA*
12:00	12:30	Unravelling the full spectrum of pathogenic variation using Saturation Genome Editing *Greg Findlay, The Francis Crick Institute, UK*
12:30	13:00	Interpretation of genetic variants in Neurofibromatosis type II by deep mutational scanning *Isaac Jia, Greater Bay Area Institute of Precision Medicine, China*
13:00	13:15	Quantifying Resistance to Targeted Anticancer Therapies at Scale with Duplex Sequencing *Haider Inam, Penn State University, USA*
13:15	13:30	Resolving Uncertainty in Alagille Syndrome Diagnostics *Melissa Gilbert, The Children's Hospital of Philadelphia, USA*
13:30	14:00	**Poster pitches for odd number posters**
14:00	15:30	Lunch with posters
15:15	*15:30*	*Briefing for Session 2 speakers, microphone runners, chair & moderator - Auditorium*
15:30	17:00	**Session 2: Bringing function to the genome** *Chair: Caroline Wright, University of Exeter, UK* *Moderator: Yann Ilboudo, Jewish General Hospital, Canada*
15:30	16:00	Mapping the effects of coding variants in *RBM20* cardiomyopathy *Victoria Parikh, Stanford University, USA*
16:00	16:30	Deep Mutational Scanning for Drug Development *Diane Dickel, Octant Bio, USA*
16:30	16:45	Using Saturation Genome Editing to understand the functional effects of BRCA1 variants *Phoebe Dace, The Francis Crick Institute, UK*
16:45	17:00	A Pilot Study of Large-Scale CRISPR-Based Screening for Variant Effects *Hong Kee Tan, Wellcome Sanger Institute, UK*
17:00	17:30	Refreshment break and networking
17:15	*17:30*	*Briefing for Session 3 speakers, microphone runners, chair & moderator - Auditorium*
17:30	19:00	**Session 3: Computational analysis of genetic screens/saturation mutagenesis** *Chair: James Stephenson, EMBL-EBI, UK* *Moderator: Daniel Tabet, University of Toronto, Canada*
17:30	18:00	Whole proteome modelling of human genetic variation *Mafalda Dias, Centre for Genomic Regulation, Spain*
18:00	18:30	Developing improved, thermostable, viral vaccine formulations through mutational scanning *Raghavan Varadarajan, Indian Institute of Science, India*
18:30	18:45	Cross-protein transfer learning using mutational scanning data substantially improves disease variant prediction across the human proteome *Yun S. Song, University of California Berkeley, USA*
18:45	19:00	Understanding higher-order genetic interactions *David McCandlish, Cold Spring Harbor Laboratory, USA*
19:00	20:30	Dinner
20:30	21:30	Bar on account - Kitchen Garden Bar

Friday 14 July 2023		
07:30	09:00	Breakfast
08:45	*09:00*	*Briefing for Session 6 speakers, microphone runners, chair & moderator - Auditorium*
09:00	10:45	**Session 4: New experimental tools for exploring genes at scale** *Chair: Sounak Sahu, National Cancer Institute, USA* *Moderator: Matt Coelho, Wellcome Sanger Institute, UK*
09:00	09:30	A Proposed Paradigm for Collecting Big Data in Life Science *Erika Debenedicitis, The Francis Crick Institute, UK*
09:30	10:00	High-throughput CRISPR-based genome editing for deep mutational scanning in *Escherichia coli* *Liselot Dewachter, Leuven University, Belgium*
10:00	10:15	Multiplex, prime saturation genome editing identifies drug resistance variants at scale *Lea Starita, University of Washington, USA*
10:15	10:30	CRISPR-CLEAR - In-Situ Investigation of Genotype-to-Phenotype Relationship with Nucleotide Level Resolution CRISPR saturation mutagenesis screens *Basheer Becerra, Harvard Medical School, USA*
10:30	10:45	Single cell sequencing as a universal variant interpretation assay - **virtual** *Francois Aguet, Illumina, USA*
10:45	11:15	Refreshment break and networking
11:00	*11:15*	*Briefing for Session 5 speakers, microphone runners, chair & moderator - Auditorium*
11:15	12:30	**Session 5: Understanding protein function** *Chair: Lea Starita, University of Washington, USA* *Moderator: Mireia Seuma, Institute for Bioengineering of Catalonia, Spain*
11:15	11:45	Mega-scale experimental analysis of protein folding stability in biology and protein design *Gabriel Rocklin, Northwestern University, USA*
11:45	12:00	Understanding kinase-substrate interaction with deep learning and high-throughput scanning *Changhua Yu, California Institute of Technology, USA*
12:00	12:15	Distinct activation mechanisms of the thrombopoietin receptor: A comparison of native ligand, small molecule agonists and patient-derived mutations. *Melissa Call, Walter and Eliza Hall Institute, Australia*
12:15	12:30	Genome-scale quantification and prediction of pathological stop codon readthrough by small molecules - **virtual** *Ignasi Toledano, Institute for Research in Biomedicine, Spain*
12:30	13:00	**Poster pitches for even number posters**
13:00	14:30	Lunch with posters
14:15	*14:30*	*Briefing for Session 6 speakers, microphone runners, chair & moderator - Auditorium*
14:30	16:00	**Session 6: Diverse applications of Mutational Scanning** *Chair: Andrew Waters, Wellcome Sanger Institute, UK* *Moderator: Gabrielle Ferra, University of Washington, USA*
14:30	15:00	Leveraging MAVEs for evolutionary questions *Irene Gallego Romero, University of Melbourne, Australia*
15:00	15:15	iPSC-SGE: assessing variant effects in differentiated cell types at scale *Shawn Fayer, University of Washington, USA*
15:15	15:30	Massively parallel functional scanning of extracellular human insulin receptor variants *Vahid Aslanzadeh, The University of Edinburgh, UK*
15:30	15:45	Rapid Epitope Mapping using Charged Scanning Mutagenesis *Kawkab Kanjo, Indian Institute of Science, India*
15:45	16:00	Saturation mutagenesis screening for FGFR inhibitor sensitivity and resistance point mutations to tailor patient stratification *Sven Diederichs, University Hospital Freiburg, Germany*
16:00	**16:10**	**Closing remarks** ***Scientific Programme Committee:*** *Matthew Hurles, Wellcome Sanger Institute, UK* *David Adams, Wellcome Sanger Institute, UK* *Clare Turnbull, The Institute of Cancer Research, UK* *Lea Starita, University of Washington, USA* *Lara Muffley, University of Washington, USA*
16:20		Coach departures for Stansted and Heathrow airports
16:30		Coach departures for Cambridge train station and city centre