

Workshop programme

Start (GMT)	Finish (GMT)	Duration (mins)	Format	Presenter details
Wednesday 25 January 2023				
12:00	13:00	60 mins		Registration, lunch and networking
13:00	13:10	10 mins		Welcome
			In-person	Scientific Programme Committee: Helen Firth, Cambridge University Hospitals Trust, UK Julia Foreman, EMBL-EBI, UK Alexander Ross, University of Cambridge, UK
13:10	14:00	50 mins		Keynote lecture
				Chair: <i>Caroline Wright, University of Exeter, UK</i>
		45 + 5 Q&A	In-person	Mechanisms of dominance Andrew Wilkie, University of Oxford, UK
14:00	15:00	60 mins		Session 1: Tools for variant interpretation
				Learning objective: Familiarity with core tools available for genomic variant interpretation Chair: <i>Caroline Wright, University of Exeter, UK</i> Moderator: <i>Julia Foreman, EMBL-EBI, UK</i>
14:00	14:30	25 + 5 Q&A	In-person	Resources for genomic medicine Eamonn Sheridan, University of Leeds, UK
14:30	15:00	25 + 5 Q&A	In-person	gnomAD and missense constraint Kaitlin Samochoa, Center for Genomic Medicine, Massachusetts General Hospital, USA
15:00	15:45	45 mins		Refreshment break and networking
15:45	16:35	50 mins		Session 2: ACMG framework
				Learning objective: Awareness of the structure and application of the ACMG framework for variant classification Chair: <i>Julia Foreman, EMBL-EBI, UK</i>
		45 + 5 Q&A	In-person	The 2015 and 2023 ACMG/AMP frameworks for variant pathogenicity classification Leslie Biesecker, NIH/NHGRI, USA
16:35	16:40	5 mins		Comfort break
16:40	18:10	90 mins		Session 3: Workshop I - Introduction to DECIPHER
				Learning objective: Ability to independently utilise DECIPHER to assess a genomic variant Chair: <i>Julia Foreman, EMBL-EBI, UK</i>
			In-person	Julia Foreman and DECIPHER team, EMBL-EBI, UK
18:10	19:00	50 mins		Drinks reception
19:00	21:00			Dinner
19:00	23:00			Bar open (card payments only)

Thursday 26 January 2023

07:30	09:00			Breakfast
09:00	11:00	120 mins		Session 4: Workshop II - Cardiac variant interpretation
				Learning objective: Understanding of some of the specific aspects of variant interpretation as applied to cardiac genetics <i>Chair: Caroline Wright, University of Exeter, UK</i>
09:00	09:30	30 mins	In-person	Introduction James Ware, Imperial College London, UK
09:30	11:00	90 mins	In-person	Workshop James Ware, Imperial College London, UK Julia Foreman, EMBL-EBI, UK
11:00	11:30	30 mins		Refreshment break and networking
11:30	13:00	90 mins		Session 5: Workshop III - Cancer variant interpretation
				Learning objective: Understanding of some of the specific aspects of variant interpretation as applied to cancer genetics <i>Chair: Alexander Ross, University of Cambridge, UK</i>
11:30	12:00	25 + 5 Q&A	In-person	Cancer whole genome sequencing analysis and interpretation Serena Nik-Zainal, University of Cambridge, UK
12:00	13:00	60 mins	In-person	Workshop Marc Tischkowitz, University of Cambridge, UK Alexander Ross, University of Cambridge, UK
13:00	14:00	60 mins		Lunch and networking
14:00	16:00	120 mins		Session 6: Workshop IV: CNVs and SNVs
				Learning objective: Ability to utilise online tools for CNV interpretation <i>Chair: Leslie Biesecker, NIH/NHGRI, USA</i> <i>Moderator: Julia Foreman, EMBL-EBI, UK</i>
14:00	14:30	25 + 5 Q&A	Virtual	Introduction Erin Riggs, Geisinger, USA - virtual presentation
14:30	16:00	90 mins	In-person	Workshop Julia Foreman, EMBL-EBI, UK
16:00	16:30	30 mins		Refreshment break and networking
16:30	17:30	60 mins		Session 7: Mutation and mosaicism
				Learning objective: Understanding of the nature of the mutational process and the aetiology of mosaicism <i>Chair: Nicky Whiffin, University of Oxford, UK</i>
16:30	17:00	25 + 5 Q&A	Virtual	De novo mutation Matt Hurler, Wellcome Sanger Institute, UK - virtual presentation
17:00	17:30	25 + 5 Q&A	In-person	Mosaicism Andrew Wilkie, University of Oxford, UK
17:30	18:30	60 mins		Free time
18:30	20:30	120 mins		Dinner
18:30	23:00			Bar open (card payments only)

Friday 27 January 2023

07:30	09:00			Breakfast
09:00	10:50	110 mins		Session 8: Workshop V - A protein view of variants
				Learning objective: Understanding of the effects of protein coding variants and assessing them from a perspective of protein structure <i>Chair: Kaitlin Samocha, Massachusetts General Hospital, USA</i>
09:00	09:20	15 + 5 Q&A	Virtual	Using CRISPR-based saturation genome editing to improve the diagnosis of neuro-developmental disorders Elizabeth Radford, University of Cambridge/WSI, UK - virtual presentation
09:20	09:50	25 + 5 Q&A	In-person	<i>In silico</i> approaches to modelling the structural effect of protein-altering variation Caroline Wright, University of Exeter, UK
09:50	10:50	60 mins	In-person	Workshop Caroline Wright, University of Exeter, UK
10:50	11:30	40 mins		Refreshment break and networking
11:30	12:30	60 mins		Session 9: Non-coding variation
				Learning objective: An appreciation of the nature and assessment of variants in non-coding elements of the genome <i>Chair: Andrew Wilkie, University of Oxford, UK</i>
11:30	12:00	25 + 5 Q&A	Virtual	Structural variations in the 3D genome Stefan Mundlos, Max Planck Institute, Germany - virtual presentation
12:00	12:30	25 + 5 Q&A	In-person	Title TBC Nicky Whiffin, University of Oxford, UK
12:30	12:45	15 mins		Closing remarks
			In-person	Scientific Programme Committee: <i>Helen Firth, Cambridge University Hospitals Trust, UK</i> <i>Julia Foreman, EMBL-EBI, UK</i> <i>Alexander Ross, University of Cambridge, UK</i>
12:45	13:45	60 mins		Lunch and departures
13:45				Coach departure for Cambridge train station and city centre