

Workshop programme

Start (GMT)	Finish (GMT)	Time allocated (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: <i>Helen Firth, Cambridge University Hospitals Trust, UK</i> <i>Julia Foreman, EMBL-EBI, UK</i> <i>Alexander Ross, Cambridge University Hospitals Trust, UK</i>
13:10	14:00	50 mins		Keynote lecture
				<i>Chair: Helen Firth, Cambridge University Hospitals Trust, UK</i>
		45 + 5 Q&A	In person	Mechanisms of dominance <i>Andrew Wilkie, University of Oxford, UK</i>
14:00	15:00	60 mins		Session 1: Tools for variant interpretation
				Learning objective: Familiarity with core tools available for genomic variant interpretation <i>Chair: Caroline Wright, University of Exeter, UK</i>
14:00	14:30	25 + 5 Q&A	In person	Resources for genomic medicine <i>Helen Firth, Cambridge University Hospitals Trust, UK</i>
14:30	15:00	25 + 5 Q&A	In-person	gnomAD and missense constraint <i>Kaitlin Samocha, Center for Genomic Medicine, Massachusetts General Hospital, USA</i>
15:00	15:30	30 mins		Refreshment break and networking
15:30	17:00	90 mins		Session 2: Workshop I - Introduction to DECIPHER
				Learning objective: Ability to independently utilise DECIPHER to assess a genomic variant <i>Chair: Julia Foreman, EMBL-EBI, UK</i>
			In person	<i>Julia Foreman and DECIPHER team, EMBL-EBI, UK</i>
17:00	17:30	30 mins		Refreshment break and networking
17:30	18:20	50 mins		Session 3: ACMG framework
				Learning objective: Awareness of the structure and application of the ACMG framework for variant classification <i>Chair: Helen Firth, Cambridge University Hospitals Trust, UK</i>
		45 + 5 Q&A	In-person	Title TBC <i>Leslie Biesecker, NIH/NHGRI, USA</i>
18:20	19:10	50 mins		Drinks reception
19:10	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 <i>James Ware, Imperial College London, UK</i>
09:30	11:00	90 mins	In person	Workshop <i>James Ware, Imperial College London, UK</i> <i>Julia Foreman, EMBL-EBI, UK</i>
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: Helen Firth, Cambridge University Hospitals Trust, UK</i>
11:30	12:00	25 + 5 Q&A	In person	Title TBC <i>Serena Nik-Zainal, University of Cambridge, UK</i>
12:00	13:00	60 mins	In person	Workshop <i>Mark Tischkowitz, University of Cambridge, UK</i> <i>Alexander Ross, Cambridge University Hospitals Trust, UK</i>
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>
14:00	14:30	25 + 5 Q&A	Virtual	Title TBC <i>Erin Riggs, Geisinger, USA - Virtual</i>
14:30	16:00	90 mins	In person	Workshop <i>Helen Firth, Cambridge University Hospitals Trust, UK</i> <i>Julia Foreman, EMBL-EBI, UK</i>
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 <i>Matt Hurles, Wellcome Sanger Institute, UK - Virtual</i>
17:00	17:30	25 + 5 Q&A	In person	Mosaicism <i>Andrew Wilkie, University of Oxford, UK</i>

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 Saturation mutagenesis for variant effect prediction <i>Lizzie Radford, University of Cambridge/WSI, UK - Virtual</i>
09:20	09:50	25 + 5 Q&A	In person Title TBC <i>Caroline Wright, University of Exeter, UK</i>
09:50	10:50	60 mins	In person Workshop <i>Caroline Wright, University of Exeter, UK</i>
10:50	11:20	30 mins	Refreshment break and networking
11:20	12:20	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>
11:20	11:50	25 + 5 Q&A	In-person Title TBC <i>Stefan Mundlos, Max Planck Institute, Germany</i>
11:50	12:20	25 + 5 Q&A	In person Title TBC <i>Nicky Whiffin, University of Oxford, UK</i>
12:20	12:40	20 mins	Refreshment break and networking
12:40	12:55	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, Cambridge University Hospitals Trust, UK</i>
12:55	14:00		Lunch and departures
14:15			Coach departure for Cambridge train station and city centre