

**Course Programme**

Start (GMT)	Finish (GMT)	Presenter details
<b>Monday 3 February 2025</b>		
12:00	13:00	Registration, lunch and networking
13:00	13:10	<b>Welcome</b>
13:00	13:05	<b>Wellcome Connecting Science:</b> <i>Nagehan Ramazanoglu Bahadir, Wellcome Connecting Science, UK</i>
13:05	13:10	<b>Scientific Programme Committee:</b> <a href="#">Helen Firth, Cambridge University Hospitals Trust, UK</a> <a href="#">Julia Foreman, EMBL-EBI, UK</a> <a href="#">James Fasham, University of Exeter, UK</a>
13:10	14:00	<b>Keynote lecture</b> <i>Chair: Helen Firth, Cambridge University Hospitals Trust, UK</i>
13:10	14:00	Incomplete penetrance and variable expressivity: lessons from population cohorts <a href="#">Caroline Wright, University of Exeter, UK</a>
14:00	15:00	<b>Session 1: Tools for variant interpretation</b> Learning objective: Familiarity with core tools available for genomic variant interpretation <i>Chair: James Fasham, University of Exeter, UK</i>
14:00	14:30	Resources for genomic medicine <a href="#">Eamonn Sheridan, University of Leeds, UK</a>
14:30	15:00	gnomAD and missense constraint [virtual] <a href="#">Kaitlin Samocha, Center for Genomic Medicine, Massachusetts General Hospital, USA</a>
15:00	15:30	Refreshment break and networking
15:30	15:45	Gene disease curation (G2P) <a href="#">Michael Yates, University of Edinburgh, UK</a>
15:45	16:35	<b>Session 2: ACMG framework</b> Learning objective: Awareness of the structure and application of the ACMG framework for variant classification <i>Chair: Isabelle Delon, Cambridge University Hospitals, UK</i>
15:45	16:35	ACMG/AMP v4 frameworks for variant pathogenicity classification [virtual] <a href="#">Leslie Biesecker, NIH/NHGRI, USA</a>
16:35	16:40	Comfort break
16:40	18:10	<b>Session 3: Introduction to DECIPHER</b> Learning objective: Ability to independently utilise DECIPHER to assess a genomic variant <i>Chair: Julia Foreman, EMBL-EBI, UK</i>
16:40	18:10	<a href="#">Julia Foreman, EMBL-EBI, UK</a> <a href="#">Julie Lecarpentier-Guillou-Keredan, EMBL-EBI, UK</a> <a href="#">Isabelle Delon, Cambridge University Hospitals, UK</a>  <i>Instructors from DECIPHER team:</i> <i>Blessing Ashimi, EMBL-EBI</i> <i>Maged Eladawy, EMBL-EBI</i> <i>Yusra Haider, EMBL-EBI</i> <i>Sarah Hunt, EMBL-EBI</i>
18:10	18:30	Free time
18:30	20:30	Dinner
18:30		Bar open (card payments only)

<b>Tuesday 4 February 2025</b>	
07:30	09:00 Breakfast
<b>09:00</b>	<b>11:00 Session 4: Cardiac variant interpretation</b>
	Learning objective: Understanding of some of the specific aspects of variant interpretation as applied to cardiac genetics Chair: Nicola Whiffin, University of Oxford, UK
09:00	09:30 Cardiovascular Genetics <a href="#">James Ware, Imperial College London, UK</a>
09:30	11:00 Workshop <a href="#">James Ware, Imperial College London, UK</a> <a href="#">Julia Foreman, EMBL-EBI, UK</a>
11:00	11:30 Refreshment break and networking
<b>11:30</b>	<b>13:00 Session 5: Cancer variant interpretation</b>
	Learning objective: Understanding of some of the specific aspects of variant interpretation as applied to cancer genetics Chair: Helen Firth, Cambridge University Hospitals Trust, UK
11:30	12:00 Interpretation of variants in cancer susceptibility genes <a href="#">Clare Turnbull, Institute of Cancer Research, UK</a>
12:00	13:00 Workshop Clare Turnbull, Institute of Cancer Research, UK Alice Garrett, Institute of Cancer Research, UK Julia Foreman, EMBL-EBI, UK Helen Firth, Cambridge University Hospitals
13:00	14:00 Lunch and networking
<b>14:00</b>	<b>16:00 Session 6: CNVs</b>
	Learning objective: Ability to utilise online tools for CNV interpretation and correlate genotype with phenotype Chair: Diana Baralle, University of Southampton, UK
14:00	14:30 Complex de novo structural variants are an underestimated cause of rare disorders <a href="#">Raheleh Rahbari, Wellcome Sanger Institute, UK</a>
14:30	16:00 Workshop <a href="#">Julia Foreman, EMBL-EBI, UK</a> <a href="#">Helen Firth, Cambridge University Hospitals Trust, UK</a> <a href="#">Isabelle Delon, Cambridge University Hospitals, UK</a> Instructors from DECIPHER team: Blessing Ashimi, EMBL-EBI Maged Eladawy, EMBL-EBI Yusra Haider, EMBL-EBI Sarah Hunt, EMBL-EBI
16:00	16:30 Refreshment break and networking
<b>16:30</b>	<b>17:30 Session 7: Non-coding variation</b>
	Learning objective: An appreciation of the nature and assessment of variants in non-coding elements of the genome Chair: Caroline Wright, University of Exeter, UK
16:30	17:00 RNA and splicing analyses in Genomics <a href="#">Diana Baralle, University of Southampton, UK</a>
17:00	17:30 The role of non-coding exons in rare disease <a href="#">Nicola Whiffin, University of Oxford, UK</a>
17:30	18:30 Drinks reception
18:30	20:30 Dinner
18:30	Bar open (card payments only)

**Wednesday 5 February 2025**

07:30	09:00	Breakfast
<b>09:00</b>	<b>11:00</b>	<b>Session 8: A protein view of variants</b>
		Learning objective: Understanding of the effects of protein coding variants and assessing them from a perspective of protein structure <i>Chair: Julia Foreman, EMBL-EBI, UK</i>
09:00	09:30	MAVEs <a href="#">Elizabeth Radford, University of Cambridge/WSI, UK</a>
09:30	10:00	Decoding protein variant effects and molecular mechanisms through the lens of protein structure <a href="#">Joe Marsh, University of Edinburgh</a>
10:00	11:00	Workshop <a href="#">Caroline Wright, University of Exeter, UK</a>
11:00	11:30	Refreshment break and networking
<b>11:30</b>	<b>13:05</b>	<b>Session 9: Therapies for genomic disorders</b>
		Learning objective: gaining understanding of the potential of nucleic acid therapies in genomic medicine <i>Chair: James Fasham, University of Exeter, UK</i>
11:30	11:55	Therapeutic Genomics, who can and could be treated? <a href="#">Stephan Sanders, University of Oxford, UK</a>
11:55	12:20	Leveraging long-read RNA-sequencing for the development of RNA targeting therapies <a href="#">Mina Ryten, University of Cambridge, UK</a>
12:20	13:05	Genetic Hearing Loss Clinic and Gene Therapy <a href="#">Manohar Bance, University of Cambridge, UK</a>
<b>13:05</b>	<b>13:15</b>	<b>Closing remarks</b>
13:05	13:15	<b>Scientific Programme Committee:</b> <a href="#">Helen Firth, Cambridge University Hospitals Trust, UK</a> <a href="#">Julia Foreman, EMBL-EBI, UK</a> <a href="#">James Fasham - University of Exeter, UK</a>
13:15	14:00	Lunch and departures
14:00		Coach departure for Cambridge train station and city centre