Unmasking the Genome

Integrating Whole Genome Sequencing, AI and Functional Genomics

In partnership with *Google DeepMind* and the *UK Human Functional Genomics Initiative*

12:00 1st September – 16:00 4th September 2026

Peter Chalk Centre, University of Exeter, United Kingdom

Subject to change

Day 1 – Tuesday 1st September

Introduction to Whole genome sequencing, Variant Calling, and Quality Control

Time	Session	Facilitator(s)
12:00-13:00	*Lunch*	-
13:00-13:05	Welcome	Dr Gareth Hawkes
13:05-14:00	**Keynote** Genomic Medicine: Past, Present & Future	Prof. Caroline Wright
14:00-14:55	**Practical** Variant Detection of SNVs, indels and structural variants with Quality Control (Part 1)	Dr Matthew Wakeling Dr Kartik Chundru
14:55-15:05	*Comfort Break*	-
15:05-16:00	**Practical** Variant Detection of SNVs, indels and structural variants with Quality Control (Part 2)	Dr Matthew Wakeling Dr Kartik Chundru
16:00-16:20	*Coffee Break*	-
16:20-17:00	Attendee Lightning Introduction Talks	Dr Gareth Hawkes
17:00-18:00	**Keynote** From variants to mechanisms: integrating protein structure, population data and functional assays to improve interpretation of genetic variation in human disease	Prof. Joe Marsh

Evening Meal: Holland Hall

Day 2 – Wednesday 2nd September

Variant annotation and interpretation using AlphaGenome and the Alpha-Suite

Time	Session	Facilitator(s)
09:00-10:00	**Seminar** Introduction to AlphaGenome	Dr Amanda Stafford
10:00-10:15	*Coffee Break*	-
10:15-11:15	**Practical** Genome Track Predictions for SNPs and Indels	Dr Amanda Stafford
11:15-12:00	**Seminar** Introduction to Variant Scorers & Worked Example Locus	Dr Amanda Stafford
12:00-13:00	*Lunch*	-
13:00-14:00	**Seminar** Introduction to AlphaMissense	Dr Joshua Pan (Virtual)
14:00-15:00	**Practical** Scoring the functional impact of variants and indels	Dr Amanda Stafford
15:00-15:15	*Coffee Break*	-
15:15-16:15	**Seminar** Quantifying functional effects across the genome	Dr Amanda Stafford
16:15-17:20	**Practical** Mini Group Projects & Presentations	Dr Amanda Stafford
17:20-18:00	**Practical** Free time on AlphaGenome with support	Dr Amanda Stafford

Evening Meal: Explore Exeter!

Day 3 – Thursday 3rd September

Identifying causal non-coding loci for population genetics and rare disease

Time	Session	Facilitator(s)
09:00-09:30	**Seminar** Introduction to Population-Scale Rare-Variant Testing	Dr Gareth Hawkes
09:30-10:30	**Practical** Annotating and testing rare variants at population scale	Dr Robin Beaumont Mr Harrison Wright
10:30-11:45	**Practical** Interpreting Rare Variant loci	Dr Robin Beaumont Mr Harrison Wright
11:45-12:00	*Comfort Break*	-
12:00-13:00	**Keynote** The STAAR Pipeline for population-scale rare-variant association testing	Prof. Xihong Lin

13:00-14:00	*Lunch*	-
14:00-14:30	**Seminar** Rare Disease & Variant Pathogenicity Overview	TBC
14:30-15:10	**Practical** Non-coding variants impacting gene splicing	TBC
15:10-15:40	Coffee Break	-
15:40-16:20	**Practical** Variants in Regulatory Elements	TBC
16:20-17:00	**Debate**	TBC
17:00-17:15	*Comfort Break*	-
17:15-18:00	**Seminar** Insights into Non-coding Variants	Dr Matthew Johnson Prof. Nick Owens

Evening Gala Dinner: Reed Hall

Day 4 – Friday 4th September

Functional Validation and Investigation of Variant Effects

Time	**Session**	**Facilitator(s)**
09:00-10:00	**Keynote** Introduction to Functional Genomics & UK Functional Genomics Initiative	Prof. Jonathan Mill
10:00-10:30	*Coffee Break*	-
10:30-12:00	**Practical** Title TBC	Prof. Matthew Child Dr Akash Das
12:00-13:00	*Lunch*	-
13:00-14:00	**Seminar** Arrayed CRISPR screening to translate biology into the clinic	Dr. Nicola McCarthy
14:00-14:45	**Seminar** Integrating computational approaches for variant pathogenicity	Prof. Ewan Birney Dr Joe Leslie
14:45-15:00	*Coffee Break*	-
15:00-16:00	**Keynote** Genomics, Imaging and AI; three technologies which are changing our perspective on research from basic biology through to clinical translation	Prof. Ewan Birney