

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

Day 1 – Tuesday 1st September

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

Time & Location	Session	Facilitator(s)
12:00-13:00 <i>Peter Chalk Centre</i>	<i>Lunch</i>	-
13:00-13:05 <i>Green</i>	Welcome	Dr Gareth Hawkes
13:05-14:00 <i>Green</i>	Keynote Genomic Medicine: Past, Present & Future	Prof. Caroline Wright
14:00-14:55 <i>Green</i>	Practical Variant Detection of SNVs, indels and structural variants with Quality Control (Part 1)	Dr Matthew Wakeling Dr Kartik Chundru
14:55-15:05	<i>Comfort Break</i>	-
15:05-16:00 <i>Green</i>	Practical Variant Detection of SNVs, indels and structural variants with Quality Control (Part 2)	Dr Matthew Wakeling Dr Kartik Chundru
16:00-16:20	<i>Coffee Break</i>	-
16:20-17:00 <i>Green</i>	Attendee Lightning Introduction Talks	Dr Gareth Hawkes
17:00-18:00 <i>Green</i>	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
18:00-18:45 <i>Peter Chalk</i>	Drinks Reception	-
19:15 -	Dinner at 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 <i>Green</i>	Seminar Introducing AlphaGenome	Dr Amanda Stafford
10:00-10:15	<i>Coffee Break</i>	-
10:15-10:45 <i>Collaborative</i>	Seminar AlphaGenome in Action	Dr Amanda Stafford
10:45-12:00 <i>Collaborative</i>	Practical Predicting and interpreting the effects of a variant using AlphaGenome	Dr Amanda Stafford
12:00-13:00	<i>Lunch and Poster Viewing</i>	-
13:00-13:30 <i>Collaborative</i>	Seminar Introduction to AlphaMissense	Dr Joshua Pan (Virtual)
13:30-14:00 <i>Collaborative</i>	Practical Analysing multiple variants in a locus	Dr Amanda Stafford
14:00-14:30 <i>Collaborative</i>	Practical Group Task: Applying AlphaGenome to variant effect analysis (Part 1)	Dr Amanda Stafford
14:30-15:00	<i>Coffee Break</i>	-
15:00-16:30 <i>Collaborative</i>	Practical Group Task: Applying AlphaGenome to variant effect analysis (Part 2)	Dr Amanda Stafford
16:30-17:40 <i>Collaborative</i>	Practical Group Task Presentations	Dr Amanda Stafford
17:40-18:00 <i>Collaborative</i>	Practical Free time on AlphaGenome with support	Dr Amanda Stafford

Attendees are encouraged to visit the city centre on the second evening of the conference to find dinner. Please find a list of recommendations in the “Local Information” attachment in the information email

Day 3 – Thursday 3rd September

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

Time	Session	Facilitator(s)
09:00-09:30 <i>Collaborative</i>	Seminar Introduction to Population-Scale Rare-Variant Testing	Dr Gareth Hawkes
09:30-10:30 <i>Collaborative</i>	Practical Testing rare variants at population scale	Dr Robin Beaumont Mr Harrison Wright
10:30-11:45 <i>Collaborative</i>	Practical Interpreting Rare Variant loci	Dr Robin Beaumont Mr Harrison Wright

11:45-12:00	<i>Coffee & Comfort Break</i>	-
12:00-13:00 <i>Green</i>	Keynote Accelerating Trustworthy Discovery in Genomic Health by Harnessing Synthetic Data from Generative AI	Prof. Xihong Lin
13:00-14:00	<i>Lunch and Poster Viewing</i>	-
14:00-14:30 <i>Collaborative</i>	Seminar Rare Disease & Variant Pathogenicity Overview	Dr Matthew Johnson
14:30-15:10 <i>Collaborative</i>	Practical Non-coding variants impacting gene splicing	Prof Elisa De Franco
15:10-15:40	Coffee Break	-
15:40-16:20 <i>Collaborative</i>	Practical Variants in Regulatory Elements	Prof Sarah Flanagan
16:20-17:00 <i>Collaborative</i>	Debate	Dr Matthew Johnson
17:00-17:15	<i>Comfort Break</i>	-
17:15-18:00 <i>Green</i>	Seminar Insights into Non-coding Variants	Dr James Russ-Silsby Prof. Nick Owens
19:00- <i>Reed Hall</i>	Gala Dinner	-

Day 4 – Friday 4th September

Functional Validation and Investigation of Variant Effects

Time	Session	Facilitator(s)
09:00-10:00 <i>Green</i>	Keynote Introduction to Functional Genomics & UK Functional Genomics Initiative	Prof. Jonathan Mill
10:00-10:30	<i>Coffee Break</i>	-
10:30-12:00 <i>Collaborative</i>	Practical How to Engineer a Genome	Prof. Matthew Child Dr James Stephenson
12:00-13:00	<i>Lunch and Poster Viewing</i>	-
13:00-14:00 <i>Green</i>	Seminar Arrayed CRISPR screening to translate biology into the clinic	Dr. Nicola McCarthy
14:00-14:45 <i>Green</i>	Seminar Integrating computational approaches for variant pathogenicity	Prof. Ewan Birney Dr Joe Leslie
14:45-15:00	<i>Coffee Break</i>	-
15:00-16:00 <i>Forum Alumni Auditorium</i>	Keynote and Close Genomics, Imaging and AI; three technologies which are changing our perspective on research from basic biology through to clinical translation	Prof. Ewan Birney