‘Clinical Application of MAVE Data’ Workshop
Hinxton Hall Conference Centre, UK
12 July 2023

This face-to-face workshop is jointly organised by the scientific programme committees of the Curating the Clinical Genome (10-12 July 2023) and the Mutational Scanning Symposium (13-14 July 2023) meetings.

Programme

The workshop will take place on Wednesday, 12 July 2023, between 14.00 and 18.40 in Hinxton Hall Conference Centre, Wellcome Genome Campus, Hinxton, UK.

This interactive workshop will consider the question, “What are the requirements for MAVE data to be used for clinical variant interpretation?”. The programme will start with brief introductory sessions on:

- Introduction to MAVE experiments: design and QC
  - Speaker: Greg Findlay – Francis Crick Institute, UK
- Overview of ACMG variant classification and evidence weighting
  - Speaker: Leslie Biesecker – NIH-NHGRI, USA
- BRCA1, TP53, PTEN and CDH1: what have we learnt so far from correlating MAVE data with clinical observations
  - Speaker: Shawn Fayer - University of Washington, USA

These will be followed by facilitated breakout table group works and panel discussions on the following topics:

- Evidence for linking assays to disease/phenotype
- Variant truthsets and standards for variant curation
- Requirements/standards for assay validation
- Standards for peer review/quality control of MAVE data ahead of clinical application
- Platforms for sharing MAVE data: formats/annotation/metadata servicing the different needs of clinical and research communities.

Organisers

David Adams - Wellcome Sanger Institute, UK
Leslie Biesecker - NIH-National Human Genome Research Institute, USA
Julia Foreman - EMBL-EBI, UK
Lara Muffley - University of Washington, USA
Lea Starita - University of Washington, USA
Clare Turnbull - Institute of Cancer Research, UK
Speakers and facilitators

George Burghel - Manchester University NHS Foundation Trust, UK
Fergus Couch - Mayo Clinic, USA
Miranda Durkie - Sheffield Children’s NHS Foundation Trust, UK
Shawn Fayer - University of Washington, USA
Greg Findlay - Francis Crick Institute, UK
Doug Fowler - University of Washington, USA
Alice Garrett - St George's University Hospitals NHS Foundation Trust/the ICR, UK
Sarah Hunt - EMBL-EBI, UK
Serena Nik-Zainal - University of Cambridge, UK
Tina Pesaran - Ambry Genetics, USA
Fritz Roth - University of Toronto, Canada
Alan Rubin - University of Melbourne, Australia
Eamonn Sheridan - University of Leeds, UK
Matthew Wakefield - The Walter and Eliza Hall Institute of Medical Research, Australia
Andrew Wilkie - University of Oxford, UK
Caroline Wright - University of Exeter, UK

How to apply?

Application deadline: 16 May 2023

Applications will open soon. Places are limited and will be awarded on merit. We will be in contact with the result of your application shortly after the deadline.

If you have any problems with the online application process, please contact the event organiser.

Confirmed attendees requiring a letter to support a visa application should contact the event organiser.

Cost

The workshop fee will be announced shortly. The fee will be requested once acceptance is confirmed.