‘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
Matt Hurles - Wellcome Sanger Institute, UK
Lara Muffley - University of Washington, USA
Lea Starita - University of Washington, USA
Clare Turnbull - Institute of Cancer Research, UK
How to apply?

Application deadline: 18 April 2023

Please complete the online application form. Places are limited; therefore, early application is recommended. 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 is £20. The fee will be requested once acceptance is confirmed.