



WELLCOME  
GENOME  
CAMPUS

ADVANCED  
COURSES AND  
SCIENTIFIC  
CONFERENCES

# CURATING THE CLINICAL GENOME

22-24 JUNE 2016

# ABSTRACT BOOK

**Thursday, 23 June**

09:00-10:15

**Session 3: Evolving guidelines/resources to support variant assessment continued**

*Chair: Matt Hurles*

- 09:00 Integration of diverse data sources to aid clinical genome interpretation  
*Michael Simpson*  
*Genomics plc, UK*
- 09:15 Comprehensive interpretation guidelines for clinical genomics; a UK clinical science perspective  
*Dominic McMullan*  
*Birmingham Womens Hospital, UK*
- 09:30 Classification and clinical management of Variants of Uncertain Significance in high penetrance cancer predisposition genes  
*Christi van Asperen*  
*Leiden University Medical Center, The Netherlands*
- 09:45 Identification of novel pathways linked to Bardet-Biedl Syndrome (BBS) using a multi-omic strategy (HIGH5 Project)  
*Phil Beales*  
*UCL, UK*

10:15-10:45

**Morning Coffee**

10:45-12:45

**Session 4: Patient reported data**

*Chair: Beverly Searle*

- 10:45 Prospective Registry of Multiplex Testing (PROMPT): a web-based platform for patients, families, and researchers seeking to assess cancer risk of genomic variants  
*Mark Robson*  
*Prompt, USA*
- 11:15 Attitudes towards genomic data sharing: Your DNA, Your Say  
*Anna Middleton*  
*Wellcome Trust Sanger Institute, UK*
- 11:45 GenIDA (Genetics of Intellectual Disabilities and Autism spectrum disorders): an online registry and clinical database for patients, families and professionals  
*Florent Colin*  
*IGBMC, France*
- 12:15 Smart phone phenotyping  
*Euan Ashley*  
*Stanford University, USA*

12:45-14:00

**Lunch**