



WELLCOME  
GENOME  
CAMPUS

ADVANCED COURSES AND  
SCIENTIFIC CONFERENCES

## Translational Bioinformatics

Wellcome Genome Campus  
Hinxton, Cambridge, UK

12 – 13 June 2017

Draft Programme

### Monday, June 12

- 09:00-10:15 Registration with refreshments
- 10:15-10:30 Welcome and Introduction
- 10:30-11:30 **Keynote speaker:** Big data goes translational: opportunities and challenges  
*Ewan Birney*  
*EMBL-EBI, UK*
- 11:30-13:00 **Session 1: Rare and common disease: Using the power of genomics to understand and advance drug targets**  
Chair: *Clare Turnbull, Genomics England, UK*
- 11:30 Integrative Informatics for Cancer Drug Discovery and Precision Therapy  
*Bissan Al-Lazikani*  
*Institute of Cancer Research, UK*
- 12:00 High Response-rate Precision Medicine beyond 'one-gene one-drug'  
*Yatin Mundkur*  
*Cellworks, USA*
- 12:30 The Cancer Genome Interpreter: a tool to annotate the biological and clinical relevance of tumor alterations  
*David Tomborero*  
*Universitat Pompeu Fabra, Spain*
- 12.45 Use of Gold Standards for supervised machine learning: applications to drug target prioritization  
*Bogdan Knezevic*  
*WTCHG, University of Oxford, UK*
- 13:00-14:00 Lunch

- 14.00-15:15 **Session 2: Bioinformatic tools and resources for biomedical research**  
Chair: *Paul Flicek, EMBL-EBI, UK*
- 14:00 Designing bioinformatic software for healthcare scientists: The Alamut experience  
*André Blavier*  
*Interactive Biosoftware, France*
- 14:30 Genome-based discovery and diagnostic technologies  
*Nick Lench*  
*Congenica, UK*
- 14:45 Scalable detection of complex de novo variants in pedigrees  
*Sergii Ivakhno*  
*Illumina, UK*
- 15:00-15:30 Afternoon Tea
- 15:30 -16:45 **Session 3: Community engagement: Using bioinformatics to help centralise and organise medical knowledge**  
Chair: *Fiona Cunningham, EMBL-EBI, UK*
- 15:30 Evaluation and comparison of current in silico drug repurposing studies  
*Katjusa Koler*  
*University of Sheffield, UK*
- 15:45 Bioinformatic technologies to improve rare disease diagnosis  
*Helen Firth*  
*Cambridge University Hospitals Trust, UK*
- 16:15 RareConnect + PhenoTips: integrating a patient-facing portal into the daily practice of genomic medicine  
*Michael Brudno*  
*University of Toronto, Canada*
- 16:45 -18:00 **Panel discussion on the needs of the healthcare system**  
Chair: *Elliott Margulies*
- *Clare Turnbull, Genomics England, UK*
  - *Jeff Barrett, Open Targets, UK*
  - *Helen Firth, Cambridge University Hospitals Trust, UK*
  - *André Blavier, Alamut, France*
- 18:00-19:30 Poster Session I & 2 (odd & even numbers) with Drinks Reception
- 19:30 Dinner

**Tuesday, June 13**

09.00-10:30 **Session 4: Exciting applications of bioinformatics in the clinical setting**

Chair: *Jeff Barrett, Open Targets, UK*

09:00 Mutational signatures and human cancers

*Serena Nik-Zainal*

*Wellcome Trust Sanger Institute, UK*

09:30 Development of analytical processes to optimise genomic analysis

*Vincent Plagnol*

*Inivata, UK*

10:00 The MedBioinformatics project: Facilitating the use of omics knowledge by non-bioinformaticians

*Ferran Sanz*

*MedBioinformatics*

10:15 DisGeNET discovery platform 5.0: illuminating the study of human diseases

*Laura Furlong*

*Fundació IMIM (GRIB), Spain*

10:30-11:00 Morning Coffee

11:00-12:15 **Workshop on starting a company**

Chair: *Paul Flicek*

- Abel Ureta-Vidal, Eagle Genomics, UK
- Fiona Nielsen, DNAdigest and Repositive, UK
- Jelena Aleksic, Gene Adviser, UK
- Nick Lench, Congenica, UK

12:15-14:00 Lunch

14:00-15:30 **Session 5: Disease studies: Using the power of large-scale studies to gain insights into the genomics of disease**

Chair: *Elliott Margulies, Illumina, USA*

14:00 The European Variation Archive

*Cristina Yenyxe Gonzalez Garcia*

*EMBL-EBI, UK*

14:15 The NHGRI-EBI Genome-Wide Association Studies (GWAS) Catalog - imposing structure on unstructured data

*Aoife, McMahon*

*EMBL-EBI, UK*

14:30 European Genome-phenome Archive: Facilitating biomedical research

*Dylan Spalding*

*EMBL-EBI, UK*

14:45 Accurate genomic variant annotation in VEP using RefSeq transcript models  
*Will McLaren*  
*EMBL-EBI, UK*

15:00 Precision Oncology Decision Support: Getting the Right Drug(s) to the Right Patient(s) at the Right Time(s)  
*Kenna M. Shaw*  
*MD Anderson Cancer Centre, USA*

15:30 – 15:45 Closing remarks

15:45 **Conference close**

16:00 **Transport departs for Cambridge, Stansted and Heathrow**