

Curating the Clinical Genome 23-25 May 2018

Wellcome Genome Campus,
Hinxton, Cambridge, UK

Draft Conference Programme

Wednesday 23 May 2018

- 12:00-13:00** **Registration with lunch**
- 13:00-13:15** **Welcome and introduction**
Helen Firth
Cambridge University Hospitals, UK
- 13:15-15:00** **Session 1: Data Sharing**
Chair: Heidi Rehm, Harvard Medical School, USA
- 13:15 The future of health and research data in genomics
Ewan Birney
EMBL-EBI, UK
- 13:45 Scientific data sharing and participant data access: is it ethical to have one without the other?
Barbara Evans
University of Houston, USA
- 14:15 Short talk selected from abstracts
- 14:30 Short talk selected from abstracts
- 14:45 Discussion
- 15.00-15:30** **Afternoon tea**
- 15:30-17:15** **Session 2: Variant Guidelines and Resources**
Chair: Heidi Rehm, Harvard Medical School, USA
- 15:30 A systematic framework for the interpretation of copy number variants
Christa Martin
Geisinger, USA

16:00 Talk title TBC
Eimear Kenny
Mount Sinai, USA

16:30 Short talk selected from abstracts

16:45 Short talk selected from abstracts

17:00 Discussion

17:15-17:45 **Lightning talks**
Chair: Marc Williams, Geisinger, USA

17:45-19:15 **Poster session 1 (odd numbers) with drinks reception**

19:15 **Dinner**
Hall Restaurant

Thursday 24 May 2018

08:30-10:15 **Session 3: Variant Interpretation**
Chair: Christa Martin, Geisinger, USA

08:30 Disease-specific optimisation of variant interpretation
Nicola Whiffin
Imperial College London, UK

09:00 Common and rare genetic variants and the risk of breast cancer
Antonis Antoniou
University of Cambridge, UK

09:30 Short talk selected from abstracts

09:45 Short talk selected from abstracts

10:00 Discussion

10:15-10:45 **Morning coffee**

10:45-12:30 **Session 4: Somatic Variation**
Chair: TBC

10:45 Interpreting the cancer genome
Serena Nik-Zainal
University of Cambridge, UK

11:15 Cancer genome interpreter annotates the biological and clinical
relevance of tumor alterations
David Trembrero
UPF/IRB/Karolinska, Spain

11:45 Short talk selected from abstracts

12:00 Short talk selected from abstracts

12:15 Discussion

12:30-14:00 Lunch and meet the speakers

Hall Restaurant

14:00-15:45 Session 5: Next Generation Phenotyping

Chair: Helen Firth, Cambridge University Hospitals, UK

14:00 Phenotypic space around comparable genotypes: The beginning of the end for disease entities

David FitzPatrick

University of Edinburgh, UK

14:30 Talk title TBC

Speaker TBC

15:00 Short talk selected from abstracts

15:15 Short talk selected from abstracts

15:30 Discussion

15:45-16:15 Afternoon tea

16:15-17:45 Session 6: Gene Curation

Chair: David FitzPatrick, University of Edinburgh, UK

16:15 Lessons learned in the race to discover causative genes: gene curation for an inherited arrhythmia syndrome

Michael Gollob

University of Toronto, Canada

16:45 Short talk selected from abstracts

17:00 Short talk selected from abstracts

17:15 Short talk selected from abstracts

17:30 Discussion

17:45-18:15 Lightning talks

Chair: Marc Williams, Geisinger, USA

18:15-19:45 Poster session 2 (even numbers) with drinks reception

19:45 Dinner

Hall Restaurant

Friday 25 May 2018

09:00-10:45

Session 7: Renanalysis

Chair: Caroline Wright, University of Exeter, UK

09:00 Clinical whole-exome sequencing for the diagnosis of rare disorders:
interest of prospective annual reanalysis

Sophie Nambot

Orphanomix Physicians' Group8, France

09:30 GenomeConnect: Sharing individual level data through patient registries

Juliann Savatt

Geisinger, USA

10:00 Short talk selected from abstracts

10:15 Short talk selected from abstracts

10:30 Discussion

10:45-11:15

Morning coffee

11:15-12:15

Session 8: Considerations for Population Testing

Chair: Gert Matthijs, KU Leuven, Belgium

11:15 Balancing the sensitivity and specificity of variant classification for
healthy populations

Peter Kang

Counsyl, USA

11:45 Talk title TBC

Joep Defesche

Academisch Medisch Centrum, The Netherlands

12:15 Panel session/open discussion

12:45-13:00

Closing remarks

Helen Firth

Cambridge University Hospitals, UK

Heidi Rehm

Harvard Medical School, USA

13:00-13:15

Take away lunch

13:15

Coaches to Cambridge and Heathrow Airport via Stansted Airport depart