

Genomic Practice for Genetic Counsellors

Wellcome Genome Campus
Hinxton, Cambridge, UK

28 - 30 January 2019

Programme

Monday 28 January

- 11:00 - 11:45 **Registration with coffee**
- 11:45 - 12:00 Welcome and introduction
Anna Middleton
Head of Society and Ethics Research Connecting Science, Cambridge, UK
- 12:00 – 13:30 **Session 1: Refresher Session on Molecular Genetics**
Chair: Anna Middleton
- 12:00 - 12:45 Molecular genetics: DNA, genes, proteins
Andrew Read
University of Manchester, UK
- 12:45 - 13:30 Types of mutation and their impact
Andrew Read
University of Manchester, UK
- 13:30 - 14:30 **Lunch**
- 14:30 – 16:00 **Session 1 continued**
- 14:30 - 15:15 Genome interpretation: terminology you need to know
Julia Foreman
Wellcome Sanger Institute
- 15:15 - 16:00 An overview of sequencing technologies
Steve Scott
Public Engagement, Wellcome Genome Campus, UK
- 16:00 – 17:00 Campus Tour (optional)
- 17:00 – 17:15 **Afternoon tea**
- 17:15 – 18:15 Consolidating learning for the day Open Q+A, discussion
Chair: Anna Middleton
Led by: Andrew Read
- 19:00 **Dinner**

Tuesday 29 January

- 09:00-10.30 **Session 2: Sequencing and bioinformatics: what does the genetic counsellor need to know?**
Chair: Anna Middleton
- 09:00 Variant interpretation: going from millions to one of interest that could be the answer
Helen Firth
Cambridge University Hospitals, UK
- 09:45 Introduction to a genome browser
Gemma Chandratillake
University of Cambridge, UK
- 10:15 – 10:30 **Group Photo**
- 10:30 – 11:00 **Coffee**
- 11:00 - 12.30 **Session 3: The role of genomics in healthcare**
Chair: Anna Middleton
- 11:00 Genomics in England
Simon Ramsden
Consultant Clinical Scientist, Manchester, UK
- 11.45 Genomics Internationally
Gemma Chandratillake
University of Cambridge, UK
- 12:30 – 13:30 **Lunch**
- 13:30 – 15:00 **Session 4: Workshop on variant interpretation: Decipher**
Chair: Anna Middleton
Using real case studies to explore pathogenicity
Led by: Julia Foreman and Decipher team
Small group discussion, bring own laptops (not iPads)
- 15:00 – 15:30 **Afternoon Tea**
- 15:30 - 17:00 **Workshop on cancer variants**
Heather Pierce, Addenbrookes Hospital, UK
- 18:30 - 19:00 **Pre-dinner drinks**
- 19:00 **Dinner**

Wednesday 30 January

- 07:30 - 09:00 **Breakfast**
- 09:00 - 10:00 **Session 5: Testing in the real world**
Chair: Chris Patch
- 09:00 Variant classification
Simon Ramsden
St Mary's Hospital Manchester, UK
- 09 :30 Genomic Counselling within the MDT and
explanation of workshop
Georgie Hall and Simon Ramsden
Manchester, UK
- 10:10 - 10:40 **Morning Coffee**
- 10:40 - 12:30 **Session 6: Workshop on variant interpretation: eye disorders as an
example**
Using real case studies to explore pathogenicity
Led by: Georgie Hall / Simon Ramsden / Other from Manchester
Small group discussion, bring own laptops (not iPads)
- 12:30 - 13:30 **Lunch**
- 13:30 - 14:00 **Genetic Counselling within large sequencing programmes**
Chris Patch
Genomics England, UK
- 14:00 - 15:00 **Next steps: applying what you have learned on this course**
Panel discussion Q&A session
Led by Anna Middleton
Connecting Science, Cambridge, UK
(A question box will be available during the course ready for this session)
- 15:00 **Course wrap-up, closing comments and end of workshop**
Faculty
- 15:45 **Coach depart to Cambridge City Centre (Downing Street) via Train
Station**