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