UNIVERSITY OF CAMBRIDGE

Institute of Continuing Education

# Fundamentals of Genetics in Precision Medicine

**Start date** 11 June 2019 **End date** 12 June 2019

**Venue** Madingley Hall
Madingley
Cambridge

**Tutor** Dr Manuel Corpas
and guest lecturers

**Course code** 1819NDX407

**Director of Programmes** Dr Corinne Boz

**To book** See: www.ice.cam.ac.uk

**Tutor biography: Manuel Corpas PhD**

Manuel is an experienced researcher and trainer. He is Chief Scientist at Cambridge Precision Medicine, helping clinicians interpret genomics. Previously, he was the Scientific Lead at Repositive, a genome data sharing platform company. Before Repositive, Manuel was a 2016 fellow of the Software Sustainability Institute and Project Leader at The Genome Analysis Centre (now Earlham Institute). Previous roles also included Technical Coordinator ELIXIR-UK, a distributed infrastructure for life science information and coordinator of the BioJavaScript open source community. He has also been chair at the Technical Committee for the Global Organisation for Bioinformatics Learning, Education and Training and a member of the board of directors at the International Society for Computational Biology.

Manuel holds a Ph.D. in bioinformatics from the University of Manchester, UK, and did his postdoctoral work in clinical genomics at the Wellcome Trust Sanger Institute, in Cambridge. Manuel has authored more than fifty scientific publications and the 'Perfect DNA' book; a speculative futuristic novel exploring the ethical and social implications of personal genetic testing in society.

## Guest lecturers

**Karyn Megy**
Karyn has more than fifteen years' experience of bioinformatics and genetics. For the past four years she has been Clinical Feedback Lead at the Haematology Department, University of Cambridge, analysing the genome of patients with rare disorders and feeding back results to clinicians. Previously, she was a scientific programmer at the European Bioinformatics Institute (EMBL-EBI), annotating the genomes of mosquitoes and other vectors of human pathogens.

Karyn has over 40 peer reviewed publications in the field of genetics and holds a PhD in Bioinformatics, with a focus on Genetics and Developmental Biology, from the University of Aix-Marseille, France.

**Antonio Metastasio**
Antonio has a special interest in personalised medicine and genomics and in their application to psychiatry and addictions; he has been pioneering the use of genetic tests in both research and clinical settings.

Antonio works as Consultant Psychiatrist in London at the Camden and Islington NHS Foundation Trust. He has been a senior clinical Tutor of the University of Cambridge and he is currently teaching medical students of the University College London. He also runs a private clinic at Cognacity 54 Harley Street, London and in Cambridge. He is a specialist in General Adult Psychiatry and in Addiction Psychiatry.

Antonio holds a Masters degree in Genomic Medicine from the University of Cambridge and trained in Psychiatry in Cambridge. He completed a Masters degree in Neuroscience at the Institute of Psychiatry, King's College London, having graduated in Medicine Magna cum Laude at the University of Perugia in Italy.

**Edmund Lehmann**
Ed set up and runs his own fund, investing in start-up ventures where he has a particular interest and relevant experience to offer. He is Chief Executive of Cambridge Precision Medicine, a start-up company delivering genetic analysis to clinicians in an actionable and relevant way. The company one of his fund's investments.

He is an active member of the London Business Angels Network for individual investors. Ed spent most of his career in the energy industry, in commodities trading and business development. He's greatly enjoyed entrepreneurial roles he's been lucky enough to hold, and developed expertise in adding the required commercial insights to technical teams. Ed holds the bilingual Masters in Business Administration from IESE Business School in Spain.

**Course programme**

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|---|---|
| **Tuesday 11 June** | |
| 10:00 - 11:00 | 1. Introduction to precision medicine: individualised data driven diagnosis |
| 11:00 | Coffee |
| 11:30 - 13:00 | 2. From sample to results: genetic analysis and bioinformatics |
| 13:00 | Lunch |
| 14:00 - 15:30 | 3. Clinical genomics: identification of relevant variants |
| 15:30 | Tea |
| 16:00 - 17:30 | 4. An example of precision medicine in practice: rare diseases (guest lecturer Karyn Megy) |
| 17:30 - 18:30 | 5. The wider context: the potential of risk management in healthcare (guest lecturer Edmund Lehmann) |
| 18:30 | Dinner |
| 20:00 - 21:30 | Terrace bar open for informal discussion |
| **Wednesday 12 June** | |
| 09:30 - 11:00 | 6. Genetic risk: estimating the potential impact of genetic variations |
| 11:00 | Coffee |
| 11:30 - 13:00 | 7. An application of the concepts so far: the Corpasome |
| 13:00 | Lunch |
| 14:00 - 15:30 | 8. Introduction to Pharmacogenomics: how the genome affects drug reactions |
| 15:30 - 16:00 | Tea |
| 16:00 - 17:30 | 9. Application of Precision Medicine to Mental Health and Pharmacogenomics (guest lecturer Antonio Metastasio) |
| 17:30 – 18:00 | Wrap-up and close |

## Course Description

Advances in our understanding of the human genome and the use of technology and big data are having a fundamental impact on the way we approach healthcare. This facilitates preventative medicine, more accurate diagnosis and more precise treatment.

The course will provide a solid introduction to precision medicine, with a particular focus on genetics. Topics covered include an overview of technologies and methods required for genetic analysis, insight into the interpretation and application of results, an introduction to genetic risk and the interaction between drugs and genetics. These are illustrated by practical examples and case studies.

**Aims:**
The objective of this two day course is to:

- Develop an understanding of precision medicine, with a focus on genetics
- Become familiar with technologies and techniques required to analyse genetic information
- Learn about applications of precision medicine in real life situations

**Target audience:**
This course offers an overview and introduction and as such is widely applicable, for instance to those who would like to enter the precision medtech, biopharma, healthcare and clinical research areas.

**Extended Syllabus**

1. Introduction to precision medicine: individualised data driven diagnosis
    a. Context: present vs future medicine
    b. How information flows in living systems: Genes, DNA, RNA, Proteins
    c. Chromosomes, genomes and exomes
    d. Phenotype, Genotype
    e. Mutations: somatic vs germline; SNPs, InDels, structural variants
    f. Mendelian diseases vs complex diseases

2. From sample to results: genetic analysis and bioinformatics
    a. Sanger sequencing
    b. Arrays
    c. Next generation sequencing techniques
    d. Data processing

3. Clinical genomics: identification of relevant variants
    a. Sources of data: population scale initiatives (1000genomes, UK Biobank, Gnomad) and curated datasets (Omim, ClinVar, dbSNP, Decipher)
    b. Variant interpretation with artificial intelligence
    c. Secondary findings

4. An example of precision medicine in practice: rare diseases
    a. Introduction to the NIHR Rare Disease project and Bioresource
    b. Variant interpretation in affected individuals
    c. Feeding back results

5. The wider context: the potential of risk management in healthcare
    a. Population trends and the pressure on healthcare systems
    b. The benefits of being preventative
    c. Managing risk vs treating disease
    d. The impact on the delivery of healthcare

6. Genetic risk: estimating the potential impact of genetic variations
    a. Genetic vs environmental causes
    b. Genome wide association studies
    c. Odds ratio, relative risk and betas
    d. Population specific risks

7. An application of the concepts: the Corpasome
    a. Implementation of technologies
    b. Use of databases
    c. Visualisation and interpretation of results
    d. Ethical considerations and communication

8. Introduction to Pharmacogenomics: how the genome affects drug reactions
    a. The concept of drug-gene interaction
    b. Metaboliser status
    c. Critical genes: cytochrome P450
    d. An example: warfarin

9. Application of Precision Medicine in Mental Health
    a. The genetics of major psychiatric disorders (depression, bipolar affective disorder, schizophrenia) and of resilience to mental illness
    b. The importance of the environment in the genesis of mental ill health
    c. The role of pharmacodynamics and pharmacogenetics testing
    d. Clinical applications of genomic in Psychiatry and future developments

**Presentation of the course:**

The course will consist of a mixture of tutor led PowerPoint-illustrated lectures, whole group discussions and question and answer sessions.

**Reading and resources list**

Listed below are a number of texts that might be of interest for future reference, but do not need to be bought (or consulted) for the course.

| **Author** | **Title** | **Publisher and date** |
|---|---|---|
| Michael Snyder | Genomics and Personalised Medicine | Oxford University Press, 2016 |
| Eric Topol | The Patient Will See You Now | Basic Books, 2016 |
| Sally Davies | Generation Genome | Chief Medical Officer annual report 2017 |
| Bertalan Meskó | The Future of Medicine | Webicina Kft., 2014 |
| Peter Robinson<br>Rosario Piro<br>Marten Jäger | Computational Exome and Genome Analysis | CRC Press, 2018 |

**Note** Students of the Institute of Continuing Education are entitled to 20% discount on books published by Cambridge University Press (CUP) which are purchased at the Press bookshop, 1 Trinity Street, Cambridge (Mon-Sat 9am – 5:30pm, Sun 11am – 5pm). A letter or email confirming acceptance on to a current Institute course should be taken as evidence of enrolment.

*Information correct as of:* 10 April 2019