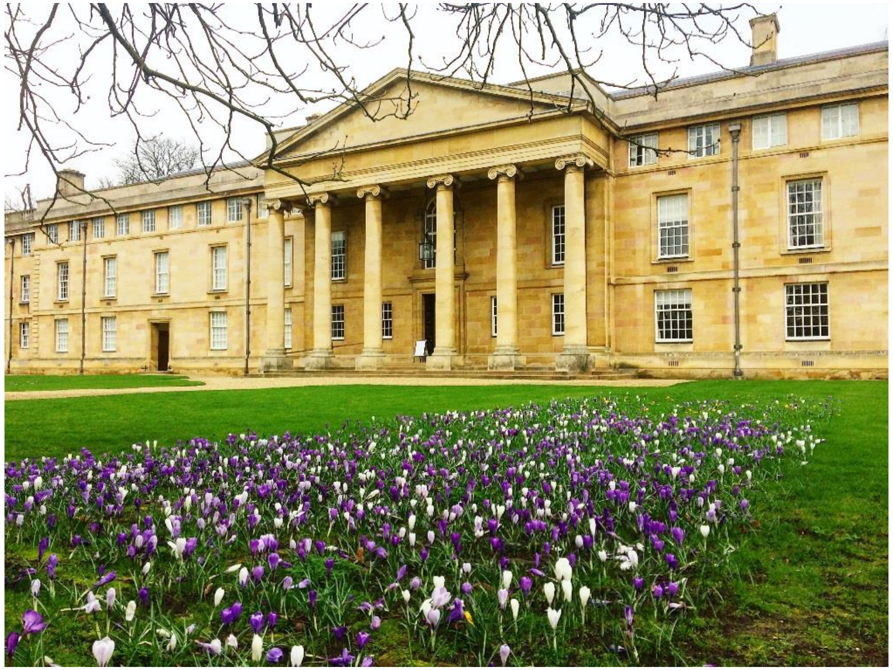
Monday 17th September - Thursday 20th September

# TrainMalta Workshop in Bioinformatics

Bioinformatics Training Facility
Craik Marshall Building
University of Cambridge
Downing Site
Downing Street
Cambridge, CB2 3EH



Train MALTA
Horizon 2020

# BEYOND THE GENOME

Dr Rosienne Farrugia

Analysis of whole genome data sets unearths a multitude of variants of different classes, which need to be filtered, annotated and validated to arrive at a causative variant for the disease. The current short length sequences, whilst being excellent at identifying single nucleotide variants (SNVs) and short insertions/deletions (indel), struggle to correctly map structural variants (SVs). Current findings also highlight the challenges of analysing the non-coding space and how to identify deleterious variants it may harbour; variants which are harder to validate and associate with functions. RNA sequencing and the integrated analysis of RNA and epigenetic data together with whole genome data, GWAS and eQTLs are essential in annotating the non-coding space. Furthermore, proteins generally do not act on their own but form part of pathways or networks, necessitating the analysis of genomic data in a pathway or network oriented approach. These are the next challenges in genomics.

This 3-day workshop will tackle these challenges through seminars, discussions and hands-on-training sessions on the following topics:

- Long read sequencing technologies
- Short read whole genome data: calling of variants
- Assigning function to the non-coding space
- Machine learning and AI to integrate genomic, transcriptomic and epigenomic data
- Protein–protein interactions: Pathways and Networks

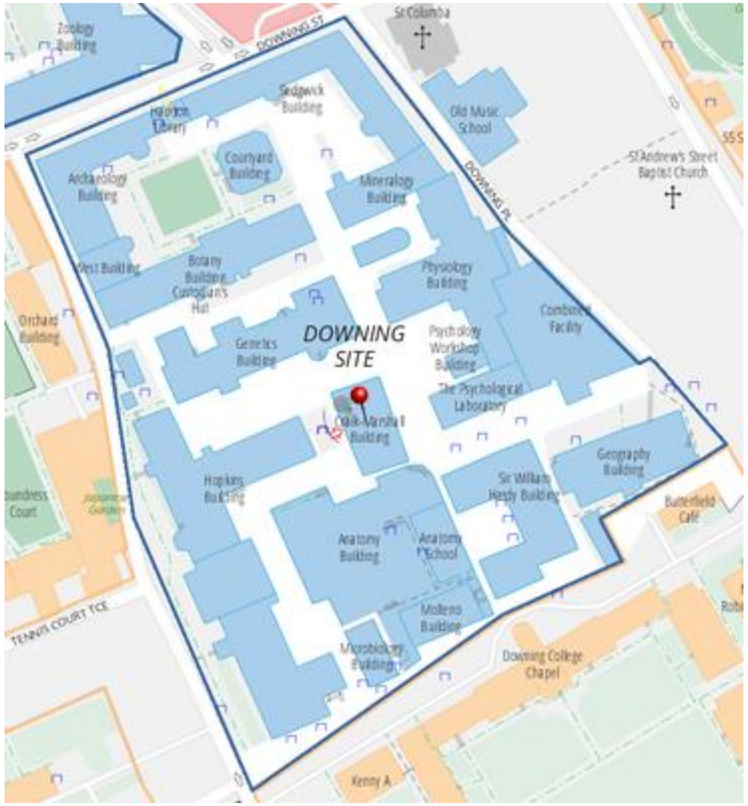
## Attendees

Rosienne Farugia
Rosalin Bonetta
Kate Burley
Alex Bye
Francesca Borg Carbott
Analisse Cassar
Janine Collins
Amanda Dalby
Lore De Kock
Melissa Formosa
Kathleen Freson
Jon Gibbins
Lucy Goudswaard
Jessica Heremans
Ingeborg Hers
Matthew Hindle
John Lambourne
Souradip Mookerjee
Andrew Mumford
Zoltan Nagy
Khalid Naseem
Vanessa Petroni Magri
Lidia Ryabova
Matt Sims
Rachel Stapley
Luca Stefanucci
Joep ten Wolde
Amanda Unsworth
Timo Voegtle
James Warland
Azra Zejnelagic

## Faculty:

William Astle
Kate Downes
Mattia Frontini
Cedric Ghevaert
Nick Gleadall
Daniel Greene
Jose Guerrero
Roman Kreuzhuber
Willem H Ouwehand
Pablo Porras Millan
Chris Penkett
Alba Sanchis-Juan
Denis Seyres
Olga Shamardina
Kathy Stirrups
Ernest Turro
Thawfeek Varusai
Joep ten Wolde

**Meeting Organisation: Mrs Lindsay Walker**,
*Phone: 01223 58 8183*
*Mobile: 07903 33 1271*
*e-mail: ouwehand-pa@medschl.cam.ac.uk*



## Monday 17th September - PM
## Long Read Sequencing - Oxford Nanopore platform
## Lead Trainers: Nick Gleadall, Alba Sanchis-Juan

| | |
|---|---|
| 14.00 | Welcome - *Willem H Ouwehand* |
| 14.20 | Oxford Nanopore *speaker - tbc* |
| 14.50 | Discussion |
| 15.00 | Structural variants - How does the analysis approach work |
| | *Nick Gleadall, Alba Sanchis-Juan* |
| 15.30 | Refreshments and Practical set up |
| 16.00 | Practical |
| | *Nick Gleadall, Alba Sanchis-Juan, ONP speaker* |
| 17.20 | Q&A Session |
| 17.45 | Close |
| 19.15 | **Social Dinner at Zizzi, Bene't Street, Cambridge** |

## Tuesday 18th September - AM

## The 100 000 Genomes Project - Rare Diseases Pilot

**Lead Trainers: Chris Penkett, Olga Shamardina, Kathy Stirrups**

**Trainers: Daniel Greene, Alba Sanchis-Juan, Ernest Turro**

| | |
|---|---|
| 09.00 | The NIHR BioResource Rare Diseases Pilot<br>*Kathy Stirrups* |
| 09.30 | From sequencer to IGV files<br>*Chris Penkett* |
| 10.00 | Refreshments and practical set up |
| 10.30 | Practical<br>*Daniel Greene, Chris Penkett, Olga Shamardina, Kathy Stirrups, Alba Sanchis-Juan* |
| 12.00 | Challenges with calling of variants<br>*Olga Shamardina* |
| 12.30 | Lunch |

## Tuesday 18th September - PM

## The Regulatory Space

**Lead Trainers: Denis Seyres, Ernest Turro**

**Trainers: Mattia Frontini, Daniel Greene, Nick Gleadall, Alba Sanchis-Juan, Joep ten Wolde**

| | |
|---|---|
| 13.30 | ChIP-seq to annotate the regulatory space<br>*Denis Seyres* |
| 14.00 | Cataloguing Regulatory Elements<br>*Ernest Turro, Joep ten Wolde* |
| 14.30 | Refreshments and practical set up |
| 15.00 | Practical<br>*Daniel Greene, Mattia Frontini, Denis Seyres,*<br>*Joep ten Wolde, Ernest Turro* |
| 16.30 | Break |
| 16.45 | Analytical challenges in deletion calling<br>*Daniel Greene, Alba Sanchis-Juan* |
| 17.15 | Practical<br>*Daniel Greene, Mattia Frontini, Denis Seyres*<br>*Joep ten Wolde, Ernest Turro* |
| 18.30 | Close |
| **19.00** | **Dinner – Graham Storey Room, Trinity Hall, CB2 1TJ**<br>**Dress code: Relaxed and informal** |

## Wednesday 19th September – AM
## Linking GWAS signals to target genes
**Lead Trainers: William Astle, Roman Kreuzhuber**
**Trainers: Kate Downes, Mattia Frontini**

| | |
|---|---|
| 09.00 | GWAS for platelet traits<br>*William Astle* |
| 09.30 | Practical session<br>*William Astle, Kate Downes, Mattia Frontini, Roman Kreuzhuber* |
| 10.45 | Refreshments |
| 11.15 | Identifying long-range interactions<br>*Mattia Frontini* |
| 11.35 | eQTL studies and KIPOI<br>*Roman Kreuzhuber* |
| 12.00 | Practical session<br>*William Astle, Kate Downes, Mattia Frontini, Roman Kreuzhuber* |
| 13.15 | Lunch |

## Wednesday 19th September – PM
## From genome to pathways and molecules
**Lead: Pablo Porras Millan, Thawfeek Varusai**
**Trainers: Jose Guerrero, Matthew Sims**

| | |
|---|---|
| 14.15 | INTACT - from Genome to Proteome<br>*Pablo Porras Millan* |
| 14.35 | REACTOME - Protein Networks and Pathways<br>*Thawfeek Varusai* |
| 15.00 | Practical session<br>*Jose Guerrero, Pablo Porras Millan, Matthew Sims, Thawfeek Varusai* |
| 16.15 | Refreshments |
| 16.45 | Generating new interaction data<br>*Jose Guerrero* |
| 17.05 | IPSC-derived megakaryocytes: a clue to diseases and developmental stages<br>*Cedric Ghevaert* |
| 17.45 | Summing up |
| 18.15 | Closing comments - *Rosienne Farrugia* |
| **18.45** | **Social event, Punting (weather dependent), or CB2** |

## Thursday 20th September – AM
## Round up session

| | |
|---|---|
| 09.00 | Time to have small group meetings and free time to explore Cambridge |