European School of Genetic Medicine

Course in Next Generation Sequencing for rare and common genetic disorders

LOCATION

EuroMediterranean University Centre of Ronzano
Via di Gaibola 16, Bologna
14th -17th April 2011

Thursday, 14 April 2011

Morning Session: General Introduction

9.00 – 9.15	Registration to the course
9.15 – 9.30	Introduction to the course G. Romeo
9.30 – 10.30	Introduction in Next Generation Sequencing technologies and applications J. Veltman
10.30 – 11.00	Coffee Break
11.00 – 12.00	The power of population based genome sequencing M. Hurles
12.00 – 13.00	Next generation sequencing in rare genetic disorders P. Robinson
13.00 – 14.30	Lunch Break and tour of the Eremo di Ronzano

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Afternoon Session: Learning NGS details part 1

14.30 – 16.00	Workshop: 1-2 instructors will discuss details of the different NGS technologies and explain different approaches such as targeted sequencing and detection of structural genomic variation. Goal: to learn how to differentiate technologies (short vs long reads, targeted versus whole genome sequencing, single molecule sequencing)
16.00 – 16.30	Coffee Break
16.30 – 18.00	Workshop: Data-analysis hands-on using data from 454, solid, Illumina; learning to identify variants, discriminate true variants from noise

Friday, 15 April 2011

Morning Session: Applications

9.00 – 09.50	Rare Genetic Variants in Health and Disease (UK10k: http://www.uk10k.org/) also to discuss 1000 genomes project. M. Hurles
09.50 – 10.40	Predicting the pathogenicity of genetic variation (bioinformatics) R. Casadio
10.40 – 11.10	Coffee Break
11.10 – 12.00	Problems, pitfalls and limits P. Robinson
12.00 – 12.50	What's next: High-throughput functional follow-up of genetic variation M. Dermitzakis
12.50 – 14.00	Lunch Break

Afternoon Session: Learning NGS details part 2

14.00 – 15.30	Workshops

15.30 – 16.00	Coffee Break
16.00 – 17.30	Workshops
17.30 – 18.00	Plenary Discussion

Workshops: Data-analysis hands-on workshop using data from 454, solid, Illumina focusing more on prioritizing variants for pathogenicity using control data, various genomic features and prediction tools, followed by discussion on the validity of these predictions, the importance of control data etc.

Saturday, 16 April 2011

Morning Session: Diagnosing genetic disorders

9.00 – 10.10	Impact of next generation sequencing on clinical diagnostic practice G. Matthijs
10.10 – 11.20	Targeted versus whole genome sequencing in the clinic J. Veltman
11.20 – 11.50	Coffee Break
11.50 – 13.00	The changing role of clinical geneticists in the NGS era R. Hennekam
13.00 – 14.00	Lunch Break

Afternoon Session: Other NGS approaches/applications

14.00 – 15.30	Workshop on Consanguinity and NGS Workshop NGS for structural variation
15.30 – 16.00	Coffee Break
16.00 – 17.30	Poster Session
17.30 – 18.00	Plenary Discussion

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Sunday, 17 April 2011

Morning Session: Ethics and discussion

9.00 – 10.10	The personal genome project J. Lunshof
10.10 – 11.20	Ethical and legal frameworks applying for personal genome data A. Cambon-Thomsen
11.20 – 11.50	Coffee Break
11.50 – 13.00	Presentation of the best students posters and wrap up session organized by students
13.00 – 14.00	Lunch Break and Departure

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