

European School of Genetic Medicine
European Society of Human Genetics
COURSE IN NEXT GENERATION SEQUENCING
Bertinoro di Romagna (Italy), May 17-20, 2013

Abstract: This course aims to give students insight into the use of next generation sequencing technology for genetic studies in humans. Aimed at researchers and diagnostics personnel with a background in genetics, biology, biomedical & molecular sciences and/or computational biology.

Topics: Next generation sequencing basics: targeted/exome/genome sequencing, data analysis and interpretation, clinical applications.

Venue: The University Residential Centre of Bertinoro ([Ce.U.B.](#)) is located on the hilltop of the Romagna area, between the cities of Forlì and Cesena. The Centre is set in three historical buildings within easy reach:

the Bishop's Fortress

- the Rivellino
- the former Episcopal Seminary.

Bertinoro is easily reachable from *Bologna* and the international airport of Bologna (BLQ) is 80 Km far from the University Centre.

Fee: The Registration fee includes tuition, course material, lunches, dinners, coffee breaks, accommodation and transportation from Bologna to Bertinoro and back.

For info please write to: serena.paterlini2@unibo.it and to rpartisani@ceub.it.

COURSE PROGRAMME:

Director:

J. Veltman (Nijmegen, The Netherlands)

Speakers and Instructors

M. Bamshad (Seattle, USA)

E. Capriotti (Birmingham , USA)

P. Froguel (Lille, France)

C. Gilissen (Nijmegen, The Netherlands)

M. Hurles (Cambridge, UK)

J. Korbel (Heidelberg, Germany)

J. Lundeberg (Stockholm, Sweden)

P. Robinson (Berlin, Germany)

T. Pippucci (Bologna, Italy)

A. Rauch (Zürich, Switzerland)

G. Romeo (Bologna, Italy)

N. Shomron (Tel Aviv, Israel)

I. Tomlinson (Oxford, UK)

Arrival: Thursday May 16

Friday, May 17

Morning Session: Introduction to Next Generation Sequencing

9.00 – 9.15 Introduction to the course
G. Romeo

- 9.15 – 10.15 Next Generation Sequencing basics
J. Veltman
- 10.15 – 10.45 **Coffee Break**
- 10.45 – 11.45 Detecting all variation in a genome; Are we getting there?
M. Hurles
- 11.45 – 12.45 Bioinformatic challenges in next generation sequencing
P. Robinson
- 12.45– 13.30 **Lunch Break**

Afternoon Session:

- 13.30 –14.00 Poster Viewing Session
- 14.00 – 16.00 Concurrent Workshops:
Computer practical: Variant identification (**C. Gilissen & T. Pippucci**)
Workshops by speakers
- 16.00-16.30 **Coffee Break**
- 16.30 – 18.00 Concurrent Workshops
Computer practical: Variant identification (**C. Gilissen & T. Pippucci**)
Workshops by speakers

Saturday, May 18

Morning Session: Next generation disease gene identification

- 9.00 – 10.00 Next generation disease gene identification
M. Bamshad
- 10.00 – 11.00 Next generation consanguinity studies
G. Romeo
- 11.00 – 11.30 Coffee Break
- 11.30- 12.30 Exome sequencing to study rare and common variation in diabetes
P. Froguel
- 12.30 – 13.30 Cancer genome sequencing
I. Tomlinson
- 13.30 – 14.30 **Lunch Break**

Afternoon Session:

- 14.30 – 16.00 Concurrent Workshops
Computer practical: Disease gene identification (**C. Gilissen & T.Pippucci**)
Workshops by speakers
- 16.00-16.30 **Coffee Break**
- 16.30 – 18.00 Concurrent Workshops
Computer practical: Disease gene identification (**C. Gilissen & T.Pippucci**)
Workshops by speakers

Sunday, May 19

Morning Session: Diagnostic next generation sequencing

- 9.00 – 10.00 Targeted breast cancer diagnostics
t.b.d
- 10.00 – 11.00 Exome diagnostics in intellectual disability
A. Rauch
- 11.00 – 11.30 **Coffee Break**
- 11.30 – 12.30 Improving the prediction of disease-related variants using protein structure
E. Capriotti
- 12.30 – 13:30 Non-invasive prenatal genome sequencing
M. Bamshad
- 13:30 – 14.30 **Lunch Break**

Afternoon Session:

- 14.30 –15.00 Poster Viewing Session (or CLCbio/Cartegenia demonstration?)
- 15.00 – 16.30 Concurrent Workshops
Computer practical: Diagnostic NGS (**C. Gilissen & T. Pippucci**)
Workshops by speakers
- 16.30-17.00 Coffee Break
- 17.00 – 18.30 Concurrent Workshops
Computer practical: Diagnostic NGS (**C. Gilissen & T. Pippucci**)
Workshops by speakers

Monday, May 20

Morning Session: New frontiers in next generation sequencing

9.00 – 10.00	Single cell genome sequencing J. Lundeberg
10.00 – 11.00	Understanding the non-coding genome N. Shomron
11.00 – 11.30	Coffee Break
11.30 – 12.00	Best Posters Presentations by students
12.00 – 12.30	Wrapping up of the course (J. Veltman)
12.30	Lunch

Departure

Fellowships:

The European Society of Human Genetics offers some fellowships covering the registration fee.

Applicants for fellowships should submit their request together with CV and a reference letter to esgm.fellowships@eshg.org, serena.paterlini2@unibo.it and to rpartisani@ceub.it.

Deadline for sending application: April 5th , 2013 at 18.00 (Bruxelles Time)

Poster submission:

Deadline for sending Abstracts: April 19th, 2013 at 18.00 (Bruxelles Time).