

EUROPEAN SCHOOL OF GENETIC MEDICINE

EUROPEAN SOCIETY OF HUMAN GENETICS

27th COURSE IN MEDICAL GENETICS

Bertinoro di Romagna (Italy), May 11-15, 2014

Programme

Abstract: Medical Genetics is a postgraduate level course addressed to both researchers and clinicians seeking an up-to-date overview of the field of medical genetics today. Morning sessions will provide an overall view of the clinical developments which are taking place in the major application fields of modern genetics in different medical specialties while afternoon concurrent workshops will consist in groups with one or more faculty and up to 15 students discussing specific research topics and research projects.

The morning sessions of the present edition will cover: Introduction to Human Genome Analysis, Clinical and Molecular Genetics, From monogenic to complex genetic disorders, Therapy and Gene Regulation, NGS, Mitochondria and cancer.

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

- the Bishop's Fortress
- the Rivellino
- the former Episcopal Seminary.

Bertinoro, is easily reachable from Bologna and its international airport (BLQ), located 80 Km away from the University Centre.

Fee: The Registration fee (740 Euro) includes tuition, course material, lunches, dinners, coffee breaks, accommodation and transportation from Bologna to Bertinoro and back.

For info please write to: rpartisani@ceub.it.

Fellowships:

The European Society of Human Genetics offers 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, 2014 at 18.00 (Bruxelles Time)

Poster submission:

Deadline for sending Abstracts for posters: at April 19th, 2014 18.00 (Bruxelles Time)

COURSE PROGRAMME:

Directors:

H. Brunner (Amsterdam, The Netherlands)

G. Romeo (Bologna, Italy)

B. Wirth (Cologne, Germany)

Faculty:

H. Brunner (Nijmegen, the Netherlands)

J. Burn (Newcastle, UK)

F. Cucca (Cagliari, IT)

D. Donnai (Manchester, UK)

G. Gasparre (Bologna, Italy)

C. Gilissen (Nijmegen, the Netherlands)

E. Klopocki (Berlin, Germany)

B. Loeys (Ghent, Belgium)

N. Katsanis (Durham, USA)

A. Read (Manchester, UK)

G. Romeo (Bologna, Italy)

E. Rugarli (Cologne, Germany)

K. Temple (Southampton, UK)

J. Veltman (Nijmegen, the Netherlands)

J. Vermeesch (Leuven, Belgium)

B. Wirth (Cologne, Germany)

C. Wijmenga (Groningen, Netherlands)

B. Wollnik (Cologne, Germany)

Arrival: Saturday May 10

Sunday, May 11

Morning Session: Introduction to Human Genome Analysis

9.00 – 9.15	Registration to the course
9.15 – 9.30	Introduction to the course G. Romeo
9.30 – 10.15	Medical Genetics Today D. Donnai
10.15 – 11.00	Genotypes & phenotypes H. Brunner
11.00 – 11.30	Coffee Break
11.30 – 12.15	NGS J. Veltman
12.15 – 13.00	How to deal with next generation sequencing output. C. Gilissen
13.10 – 14.00	Lunch Break

Afternoon Session:

14.00 – 14.30	Poster Viewing Session
14.30 – 16.00	Concurrent Workshops
16.00-16.30	Coffee Break
16.30 – 18.00	Concurrent Workshops

Monday, May 12

Morning Session: Approaches to Clinical and Molecular Genetics

9.00 – 9.50	Linkage and association in a conceptual and historic perspective A. Read
9.50- 10.40	Arrays & CNVs J. Vermeesch
10.40 – 11.10	Coffee Break
11.10 – 12.00	Dysmorphology D. Donnai
12.00 – 12.50	Molecular syndromology in the NGS-era: which phenotype, which family, which strategy? B. Wollnik
13.10 – 14.00	Lunch Break

Afternoon Session:

14.00 –14.30	Poster Viewing Session
14.30 – 16.00	Concurrent Workshops
16.00-16.30	Coffee Break
16.30 – 18.00	Concurrent Workshops

Tuesday, May 13

Morning Session: From monogenic to complex genetic disorders

9.00 - 9.50	Marfan syndromes, related diseases and therapy B. Loeys
9.50 – 10.40	Aging Phenotypes B. Wollnik
10.40 – 11.10	Coffee Break
11.10 – 12.00	Oligogenic inheritance N. Katsanis

12.00 – 12.50 Complex genetics
C. Wijmenga

13:10 – 14.00 **Lunch Break**

Afternoon Session:

14.00 –14.30 Poster Viewing Session

14.30 – 16.00 Concurrent Workshops

16.00-16.30 Coffee Break

16.30 – 18.00 Concurrent Workshops

Wednesday, May 14

Morning Session: Therapy and Gene Regulation

9.00 – 9:50 Inherited cancer and prospects for therapy
J. Burn

9.50 – 10.40 SMA: From gene and modifiers to therapy
B. Wirth

10.40 – 11.10 Coffee Break

11.10- 12.00 Epigenetics and disease
K. Temple

12.00-12.50 Long Distance Regulation
E. Klopocki

13:10 - 14.00 **Lunch Break**

Afternoon Session:

14.00 –14.30 Poster Viewing Session

14.30 – 16.00 Concurrent Workshops

16.00 – 16.30 Coffee Break

16.30 – 18.00 Concurrent Workshops

Thursday, May 15

Morning Session: Mitochondria and evolution

9.00 – 9.50 Mitochondrial inheritance and disease
Rugarli E.

9.50 – 10.40 Mitochondria and cancer
G. Gasparre

10.40 – 11.00 **Coffee Break**

11.00 – 11.50 Population genetics in the genome era: the Sardinia project
F. Cucca

11:50-12:40 Best Posters Presentations by students

12:40 Wrapping up of the course

13.00 **Lunch**

Departure