

# EUROPEAN SCHOOL OF GENETIC MEDICINE

## EUROPEAN SOCIETY OF HUMAN GENETICS

### 27<sup>th</sup> 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.

**Fees:** The Registration fee (€ 350,00) includes tuition, course material and transportation from Bologna airport to Bertinoro on May 10<sup>th</sup> and back on May 15<sup>th</sup>

Accommodation fee (€ 390,00) includes full board accommodation in double room to be shared with another participant from May 10<sup>th</sup> to 15<sup>th</sup> (5 nights) at the University Residential Centre of Bertinoro or in hotels nearby the course venue.

**For info** please write to: [rpartisani@ceub.it](mailto:rpartisani@ceub.it).

#### **Fellowships:**

The European Society of Human Genetics offers fellowships covering the registration fee and full board accommodation . Applicants for fellowships should submit their request together with CV and a reference letter to [esgm.fellowships@eshg.org](mailto:esgm.fellowships@eshg.org) , [serena.paterlini2@unibo.it](mailto:serena.paterlini2@unibo.it) and to [rpartisani@ceub.it](mailto: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 19<sup>th</sup>, 2014 18.00 (Bruxelles Time)

**COURSE PROGRAMME:**

**Directors:**

H.G. Brunner (Nijmegen and Maastricht, the Netherlands)

G. Romeo ( Bologna, Italy)

B. Wirth (Cologne, Germany)

**Faculty:**

H.G. Brunner (Nijmegen and Maastricht, 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 (Antwerp, 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 <b>G. Romeo</b>
9.30 – 10.15	Medical Genetics Today <b>D. Donnai</b>
10.15 – 11.00	Genotypes & phenotypes <b>H. Brunner</b>
11.00 – 11.30	<b>Coffee Break</b>
11.30 – 12.15	NGS <b>J. Veltman</b>
12.15 – 13.00	How to deal with next generation sequencing output. <b>C. Gilissen</b>
13.10 – 14.00	<b>Lunch Break</b>

**Afternoon Session:**

14.00 – 14.30	Poster Viewing Session
14.30 – 16.00	Concurrent Workshops
16.00-16.30	<b>Coffee Break</b>
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 Detection and functional impact of mitochondrial DNA mutations in cancer progression  
**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**