

# EUROPEAN SCHOOL OF GENETIC MEDICINE

## EUROPEAN SOCIETY OF HUMAN GENETICS

### 26<sup>th</sup> COURSE IN MEDICAL GENETICS

**Bertinoro di Romagna (Italy), May 12-16, 2013**

**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.

**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: [serena.paterlini2@unibo.it](mailto:serena.paterlini2@unibo.it) and to [rpartisani@ceub.it](mailto:rpartisani@ceub.it).

## **COURSE PROGRAMME:**

### **Directors:**

H. Brunner (Amsterdam, The Netherlands)

G. Romeo (Bologna, Italy)

B. Wirth (Cologne, Germany)

### **Faculty:**

A. Ballabio (Naples, Italy)

H. Brunner (Nijmegen, the Netherlands)

P. Chinnery (New Castle, UK)

D. Donnai (Manchester, UK)

C. Gilissen (Nijmegen, the Netherlands)

E. Klopocki (Wurzburg, Germany)

B. Loeys (Ghent, Belgium)

N. Katsanis (Durham, USA)

A. Read (Manchester, UK)

G. Romeo (Bologna, Italy)

K. Temple (Southampton, UK)

J. Veltman (Nijmegen, the Netherlands)

J. Vermeesch, (Leuven, Belgium)

B. Wirth (Cologne, Germany)

B. Wollnik (Cologne, Germany)

**Arrival: Saturday May 11**

## **Sunday, May 12**

### **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 – 10.10	Medical Genetics Today <b>D. Donnai</b>
10.10 – 10.50	Genotypes & phenotypes <b>H. Brunner</b>
10.50 – 11.20	<b>Coffee Break</b>
11.20 – 12.00	Arrays & CNVs <b>J. Vermeesch</b>
12.00 – 12.40	Molecular syndromology in the NGS-era: which phenotype, which family, which strategy? <b>B. Wollnik</b>
12.40 – 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 13**

### **Morning Session: Approaches to Clinical and Molecular Genetics**

9.00 – 9.50	Dysmorphology <b>D. Donnai</b>
9.50 – 10.40	Linkage and association in a conceptual and historic perspective <b>A. Read</b>

10.40 – 11.10	Coffee Break
11.10- 12.00	Complex disease genetics: GWAS and beyond <b>C. van Duijn</b>
12.00 – 12.50	Mitochondrial inheritance and disease <b>P. Chinnery</b>
12.50 – 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

**Tuesday, May 14**

**Morning Session: From Monogenic to Complex Genetic Disorders**

9.00 – 9.50	Aging phenotypes <b>B. Wollnik</b>
9.50 – 10.40	Oligogenic inheritance <b>N. Katsanis</b>
10.40 – 11.10	<b>Coffee Break</b>
11.10 – 12.00	Marfan syndrome, related diseases and therapy <b>B. Loeys</b>
12.00 – 12:50	Epigenetics and disease <b>K. Temple</b>
13:00 – 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	Coffee Break
16.30 – 18.00	Concurrent Workshops

**Wednesday, May 15**

**Morning Session: From Gene Identification and Gene Regulation to Therapy and Ethical Implications**

9.00 – 9.50	Gene identification for Intellectual Deficiencies (I.D.) <b>H. Brunner</b>
9.50 – 10.40	SMA: From gene and modifiers to therapy <b>B. Wirth</b>
10.40 – 11.10	Coffee Break
11.10 – 12.00	Long distance regulation in skeletal disorders <b>E. Klopocki</b>
12.00 – 12.50	Ethical Issues <b>A. Read</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	Coffee Break

16.30 – 18.00 Concurrent Workshops

## **Thursday, May 16**

### **Morning Session: Next Generation Sequencing**

9.00 – 9.50	NGS <b>J. Veltman</b>
9.50 – 10.40	How to deal with next generation sequencing output. <b>C. Gilissen</b>
10.40 – 11.00	<b>Coffee Break</b>
11.00 – 11.50	Best Posters Presentations by students
11.50	Wrapping up of the course
12.00	<b>Lunch</b>

### **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](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 , 2013 at 18.00 (Bruxelles Time)

### **Poster submission:**

**Deadline for sending Abstracts for posters:** April 19<sup>th</sup>, 2013 at 18.00 (Bruxelles Time)