

**The EUROPEAN SCHOOL OF GENETIC MEDICINE**  
**with the UNIVERSITY RESIDENTIAL CENTER OF BERTINORO**  
**and the EUROPEAN SOCIETY OF HUMAN GENETICS**  
**organize**  
**the 29<sup>th</sup> COURSE IN MEDICAL GENETICS**  
**Bertinoro (Italy), May 8-12, 2016**

**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 of students with one or more faculty and up to 15 students discussing specific research topics and research projects.

**Venue:** The University Residential Centre of Bertinoro ([www.ceub.it](http://www.ceub.it)) is located on one of the hilltops of the Romagna area, between the cities of Forlì and Cesena.

**Fees:** The Registration fee (€ 565,00) includes tuition, course material, lunches, coffee breaks, dinners and transportation from Bologna airport to Bertinoro on May 7<sup>th</sup> and back on May 12<sup>th</sup>.

**Accommodation:** You'll be lodged at the Course Venue or in case of overbooking in hotels nearby the Centre. The rate for a double room occupancy (to be shared with another participant) is €35,00/per night, including breakfast.

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

**Fellowships:**

The European Society of Human Genetics offers fellowships covering both registration fee and accommodation. Applicants for fellowships should submit their request together with CV and a reference letter to [jbazzoli@ceub.it](mailto:jbazzoli@ceub.it).

**Deadline for sending applications:** March 12<sup>th</sup>, 2016 at 18.00 (Bruxelles Time). You'll receive a notification of acceptance (or non acceptance) by mail within March, 31<sup>st</sup>.

**Poster submission:**

**Deadline for sending Abstracts for posters:** April 19<sup>th</sup>, 2016 at 18.00 (Bruxelles Time). Please send your abstract to [jbazzoli@ceub.it](mailto:jbazzoli@ceub.it)

All participants are encouraged to present a poster. All abstracts will be accepted.

## **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)

C. Gilissen (Nijmegen, the Netherlands)

A. Hoischen (Nijmegen, the Netherlands)

N. Katsanis (Durham, USA)

E. Klopocki (Würzburg, Germany)

B. Loeys (Antwerp, Belgium)

A. Read (Manchester, UK)

G. Romeo (Bologna, Italy)

L. Salviati (Padua, Italy)

K. Temple (Southampton, UK)

D. Wells (Oxford, UK)

B. Wirth (Cologne, Germany)

B. Wollnik (Göttingen, Germany)

## **Arrival: Saturday May 7**

### **Sunday, May 8**

#### **Morning Session: Introduction to Human Genome Analysis**

8:30-9:00	Registration to the course
9.00– 9.30	Introduction to the course <b>G. Romeo</b>
9.30 – 10.15	Genotypes & phenotypes <b>H. Brunner</b>
10.15 – 11.00	Arrays and CNVs <b>E. Klopocki</b>
11.00 – 11.30	<b>Coffee Break</b>
11.30 – 12.15	NGS <b>A. Hoischen</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 9**

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

- 9.00 – 9.50            Complex disorders  
**A. Read**
- 9.50- 10.40           Gene Targeting into the 21<sup>st</sup> Century: Mouse Models of Human Disease from Cancer to Neuropsychiatric Disorders  
**M. Capecchi**
- 10.40 – 11.10           **Coffee Break**
- 11.10 – 12.00           Molecular syndromology in the NGS-era: which phenotype, which family, which strategy? Applications to aging research  
**B. Wollnik**
- 12.00 – 12.50           Mitochondrial Disorders  
**L. Salviati**
- 13.10 – 14.00           **Lunch Break**

### **Afternoon Session:**

- 14.00 –14.30           Poster Viewing Session
- 14.30 – 16.00           Concurrent Workshops – including Ethics by Andrew Read
- 16.00-16.30           **Coffee Break**
- 16.30 – 18.00           Concurrent Workshops

## **Tuesday, May 10**

### **Morning Session: Gene regulation and complex genetic disorders**

- 9.00 - 9.50            Ethical aspects of Genomics  
**A. Read**
- 9.50 – 10.40           Epigenetics and disease  
**K. Temple**
- 10.40 – 11.10           **Coffee Break**
- 11.10 – 12.00           Long range effects  
**E. Klopocki**

12.00 – 12.50 Behavioral genetics  
**H. Brunner**

13:10 – 14.00 **Lunch Break**

**Afternoon Session:**

14.30 –16.00 Concurrent Workshops

16.00-16.30 **Coffee Break**

16.30-18.00 **J. Burn** - “Dealing with rarity in the clinic”  
Interactive discussion and debate

**Social Dinner**

**Wednesday, May 11**

**Morning Session: Using Protein Networks and Gene Modifiers to Develop Therapies**

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 Marfan syndromes, related diseases and therapy  
**B. Loeys**

12.00-12.50 Oligogenic Disease, and disease modelling  
**N. Katsanis**

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 Poster Viewing Session

## **Thursday, May 12**

### **Morning Session: The returns of Genomic Medicine**

- 9.00 – 9.45 Best Posters presentations by students
- 9.45-10.15 Screening in Iran  
**S. M. Akrami** (TUMS and Ministry of Health and Medical Education, Iran)
- 10.15 – 10.45 **Coffee Break**
- 10.45 – 12.15 PGD and PGS: technical and clinical aspects  
**D. Wells** (with discussant **L. Gianaroli**)
- 12:15 - 12:30 Wrapping up
- 12:30 Hand in evaluations; hand out certificates
- 12:45 **Lunch**
- 14:30 *Discussion groups with high school students and ESGM students (Guglielmo Project - see next page)*

### **Departure**

# THE GUGLIELMO PROJECT

LAUNCHED BY THE EUROPEAN SCHOOL OF GENETIC MEDICINE IN BERTINORO, ITALY  
FOR HIGH SCHOOL STUDENTS

Since 1988 thousands of young geneticists, especially physicians and biologists, have been trained by the European School of Genetics Medicine (ESGM). They came from every side of Europe, from the southern rim of the Mediterranean sea, from the Middle East and from several other countries.

Taking into consideration only the period 2001-2011 the participants to the ESGM advanced training courses have been 6000. Most of the courses took place at the Centro Universitario di Bertinoro di Romagna. ESGM and Bertinoro have thus become the most important European training center for young graduates in the medical genetic field and for this reason ESGM is recognized and sponsored with scholarships by many Universities across Europe and by the European Society of Human Genetics. In 2016 ESGM and Bertinoro will start a completely new experience for the communication in genetics.

Forty students of the last year of the High School from Forlì (Emilia-Romagna region) in addition to students from Sardinia and students from Iran will be hosted in Bertinoro to attend the last morning lecture of the 29<sup>th</sup> COURSE IN MEDICAL GENETICS on Thursday May 12<sup>th</sup> as well as some of the lectures of the previous days. One of the Faculty of the course, the Nobel Prize Prof. Mario Capecchi, will give a talk on the public understanding of genetics directed to the general public on May 9 at 5:00 pm in Forlì.

On May 12<sup>th</sup> the ESGM students of the European School of Genetics Medicine will become the "instructors" of the High School students for debates, discussions and questions regarding medical genetic research and practice in the contemporary world. In particular in the morning of May 12<sup>th</sup> the students will attend the lecture of Prof. Dagan Wells from Oxford on preimplantation genetic diagnosis. In the afternoon they will divide into different discussion groups to deal with topics like the thalassemias and others chosen by the students themselves.

Why Sardinian and Iranian students?

During the last 40-30 years several programs for thalassemia prevention have been successfully developed in Sardinia and Iran and it will be of great interest to discover the perception that Sardinian and Iranian students have about questions raised by their national prevention strategies.

Why the Project is named after Guglielmo?

Guglielmo Dall'Ongaro, a native of Rome, was a young brave man who died in August 2015, at the age of 23, after a long fight against a genetic disease which was diagnosed too late. Early diagnosis, might have saved him.

Which are the goals of the *Guglielmo Project*?

- To increase the knowledge about human genetics and genetic disease among the young
- To test new communication tools between physicians and users of genetic medicine
- To introduce new high-qualification courses for students from secondary school
- To strengthen the cooperation between university and school networks.

The promoters of the *Guglielmo Project* are:

- European School of Genetic Medicine and the Italian Society of Human Genetics
- The Fondazione Cassa di Risparmio di Forlì
- The Centro Residenziale Universitario di Bertinoro and the Bertinoro City Council Administration
- High Schools from Forlì (Italy), from Sardinia, from Teheran and from other regions of the world which intend to participate in the project.