Clinical Genomics and NGS

Bertinoro (Italy), April 30 – May 5, 2017

30th course jointly organized by ESGM, ESHG AND CEUB

Target Audience: This course is for those young professionals in Clinical and Medical genetics who want to learn about Clinical Genomics in the era of NGS. The course is based on the experience established by the European School of Genetic Medicine (ESGM) since 1988 with its 29 yearly courses in Medical Genetics and more recently with its NGS courses, which are now merged.
Starting from the basic notions of medical genetics and bioinformatics covered during the first two days, the course will offer a more specialized training in the following four days, designed for young clinicians and clinical laboratory specialists (in training) and for PhD or postdoc trainee scientists.
Morning plenary lectures are followed by afternoon workshops conducted by 1-2 Faculty with groups of 15-25 students, for more in-depth discussion of the morning's topics.
Bioinformatic workshops (with a choice of basic and advanced) are taught using computers and databases. Clinically-oriented workshops will use an interactive discussion format.

Venue: The University Residential Center of Bertinoro (www.ceub.it)

Fees: The Registration fee for the entire course (€780,00) includes tuition, course material, meals, coffee breaks, social dinner, party, and transportation from Bologna airport to the course venue on April 29th and back on May 6th.
The Registration fee for part of the course (€150,00/day) includes tuition, course material, meals, and coffee breaks.

Accommodation: You'll be lodged at the Course Venue (or hotels nearby the course venue if necessary).
The rate for a double room occupancy (to be shared with another participant) is €35,00/night and includes bed and breakfast.

A limited number of fellowships covering both registration fees and accommodation will be sponsored by the ESHG. Following the guidelines established by ESHG, students from economically less favoured countries are strongly encouraged to apply for ESHG fellowships: see list here https://www.eshg.org/fileadmin/eshg/countries/Collective_Members_Country_List_from_01_01_2017.pdf
However fellowships are not limited to these groups of countries.

Applicants for fellowships should submit their request together with CV and a reference letter to rpartisani@ceub.it. Closing date for fellowship applications: March 10th, 2017. A communication of acceptance (or non acceptance) will be sent by mal within March 17th.

Applicants without fellowships can register at the following link: www.ceub.it
Closing date for registration will be when all places are taken.

Poster submission:
All participants are encouraged to present a poster. All abstracts will be accepted.
Deadline for sending Abstracts for posters: March 31th, 2017. Please send your abstract to **rpartisani@ceub.it**

Directors:

Han Brunner (Nijmegen and Maastricht, the Netherlands); Christian Gilissen (Nijmegen, the Netherlands); Alexander Hoischen (Nijmegen, the Netherlands); Tommaso Pippucci (Bologna, Italy); Giovanni Romeo (Bologna, Italy); Brunhilde Wirth (Cologne, Germany)

Faculty:

John Burn (Newcastle, UK); Dian Donnai (Manchester, UK); Evan E Eichler (Seattle, USA); David Fitzpatrick (Edinburgh, UK); Lude Franke (Groningen, the Netherlands); Caterina Garone (Cambridge, UK); Nicholas Katsanis (Durham, USA); Eva Klopocki (Wurzburg, Germany); Jonathan O'Halloran (Newcastle, UK); Anita Rauch (Zürich, Switzerland); Tobias Rausch (Heidelberg, Germany); Andrew Read (Manchester, UK); Augusto Rendon (Cambridge, UK); Carlo Sidore (Cagliari, Italy); Karen Temple (Southampton, UK); Thierry Voet (Leuven, Belgium).

COURSE PROGRAM

SATURDAY APRIL 29TH Arrival and dinner

SUNDAY APRIL 30TH

Morning Lectures:	**Medical Genetics concepts and principles**
8:30 – 9:00	Participants Registration
9:00 – 10:00	Genomic Medicine - **Dian Donnai**
10:00 – 11:00	Phenotype to genotype - **Han Brunner**
11:00 – 12:00	Cytogenetics and arrays - **Eva Klopocki**
12:00 – 13:00	Complex disorders and classical gene identification - **Andrew Read**
13:10 – 14:00	Lunch Break

Afternoon Workshops

Session I (14:30 – 16:00)	Mutation patterns - **Han Brunner**
	Interpreting CNVs for beginners - **Eva Klopocki** Computer room
	Dymorphology - **Dian Donnai**
Session II (16:30 – 18:00)	Mutation patterns - **Han Brunner**
	Interpreting CNVs for beginners - **Eva Klopocki** Computer room
	Dymorphology - **Dian Donnai**

MONDAY MAY 1ST

Morning Lectures:	**Basics of NGS for Mendelian disorders**
9:00 – 10:00	Basics of next generation sequencing technology - **Alexander Hoischen**
10:00 – 11:00	Basics of NGS bioinformatics - **Christian Gilissen**
11:00 – 12:00	NGS in the clinic - **Anita Rauch**
12:00 – 13:00	Future NGS technologies - **Jonathan O'Halloran**
13:10 – 14:00	Lunch Break
Afternoon Workshops	
Session I (14:30 – 16:00)	NGS Bioinformatics Basics - **Christian Gilissen & Tommaso Pippucci** Computer room
	Targeted NGS approaches - **Alexander Hoischen**
	How to do GWAS - **Carlo Sidore**
Session II (16:30 – 18:00)	NGS Bioinformatics Basics - **Christian Gilissen & Tommaso Pippucci** Computer room
	Clinical considerations for NGS - **Anita Rauch**
	NGS Technologies of the future - **Jonathan O'Halloran**

TUESDAY MAY 2ND

Morning Lectures:	**Therapy and prenatal diagnostics in the NGS era**
9:00 – 10:00	Therapy and cancer - **John Burn**
10:00 – 11:00	SMA: From gene and modifier to therapy - **Brunhilde Wirth**
11:00 – 12:00	Non-invasive prenatal testing - **tbd**
12:00 – 13:00	Mithocondrial pathologies - **Caterina Garone**
13:10 – 14:00	Lunch Break
Afternoon Workshops	
Session I (14:30 – 16:00)	NGS Bioinformatics, variant interpretation – **C. Gilissen & T. Pippucci** Computer room
	Rarity in the clinic - **John Burn**
	Setting up NIPT and ethical concerns - **tbd**
Session II (16:30 – 18:00)	NGS Bioinformatics, variant interpretation – **C. Gilissen & T. Pippucci** Computer room
	Ethics of medical genetics - **Caterina Garone & Andrew Read**
	From your newly discovered candidate gene to its function – **B. Wirth**
18:00	Poster viewing session with aperitif (session I)

WEDNESDAY MAY 3RD

Morning Lectures:	**Complex mechanisms of disease**
9:00 – 10:00	NGS and structural variants - **Tobias Rausch**
10:00 – 11:00	Epigenetics, imprinting, clinical - **Karen Temple**
11:00 – 12:00	Non-coding mutations/long-range effects - **Eva Klopocki**
12:00 – 13:00	Oligogenic diseases - **Nicholas Katsanis**
13:10 – 14:00	Lunch Break

Afternoon Excursion

THURSDAY MAY 4TH

Morning Lectures:	**Novel NGS applications**
9:00 – 10:00	Molecular inversion probes and Saturation Genome editing – **A. Hoischen**
10:00 – 11:00	Long-read sequencing - **Evan E Eichler**
11:00 – 12:00	GWAS with NGS - **Carlo Sidore**
12:00 – 13:00	Data integration - **Lude Franke**
13:10 – 14:00	Lunch Break
Afternoon Workshops	
Session I (14:30 – 16:00)	NGS diagnostic variant interpretation – **C. Gilissen & T. Pippucci** Computer room
	Genetic Imprinting - **Karen Temple**
	Genomic exotica - **Nicholas Katsanis**
	Mechanisms for non-coding mutations - **Eva Klopocki**
Session II (16:30 – 18:00)	NGS diagnostic variant interpretation – **C. Gilissen & T. Pippucci** Computer room
	How to set up a NGS lab? - **Alexander Hoischen**
	Copy number variations - **Evan E Eichler**
18:00	Poster viewing session with aperitif (session II)

FRIDAY MAY 5TH

Morning Lectures:	**Large scale NGS**
9:00 – 10:00	Presentations of best poster from students
10:00 – 11:00	Genomics England - **Augusto Rendon**
11:00 – 12:00	Phenotype and NGS integration / HPO benefits - **David Fitzpatrick**
12:00 – 13:00	Single cell sequencing and applications to PGD - **Thierry Voet**
13:10 – 14:00	Lunch Break

Afternoon Workshops

Session I (14:30 – 16:00)	CNV detection in NGS data - **Tobias Rausch** Computer room
	Duplications and evolution of human gene innovation - **Evan E Eichler**
	How to do RNASeq - **Lude Franke**
Session II (16:30 – 18:00)	CNV detection in NGS data - **Tobias Rausch** Computer room
	Large genomics projects - **Augusto Rendon & David Fitzpatrick**
	How to do single cell genomics? - **Thierry Voet**

Social dinner and farewell party

SATURDAY MAY 6TH
Departure

For more information visit:

www.ceub.it

www.eshg.org