Clinical Genomics and NGS

Bertinoro (Italy), April 29 – May 4, 2018

31st 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 28th and back on May 5th.
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. Single rooms will be assigned if available.

A limited number of fellowships covering both registration fees and accommodation will be sponsored by 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 countries.

Applicants for fellowships should submit their request together with CV, motivation letter and a reference letter to rpartisani@ceub.it. Closing date for fellowship applications: February 15, 2018. A communication of acceptance (or non-acceptance) will be sent by mail within February 24.

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. The usable surface on the poster board will be 90 cm width x 150 cm height (approx. 35 x 59 inches)
Deadline for sending Abstracts for posters: March 31st, 2018. Each abstract should consist in principle of 1-2 typewritten pages including references. 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); Luis Galietta (Naples, Italy); Giovanni Germano (Milan, Italy); Eva Klopocki (Wurzburg, Germany); Michael Nothnagel (Cologne, Germany); Tobias Rausch (Heidelberg, Germany); Andrew Read (Manchester, UK); Augusto Rendon (Cambridge, UK); Kaitlin Samocha (Cambridge, UK); Lea Starita (Seattle, USA); Karen Temple (Southampton, UK); Thierry Voet (Leuven, Belgium); Janneke Weiss (Amsterdam, the Netherlands)

SATURDAY APRIL 28TH Arrival and dinner

SUNDAY APRIL 29TH

Morning Lectures:	**Medical Genetics concepts and principles**
8:30 – 9:00	Participants Registration
9:00 – 9:15	Introduction to the course – **G. Romeo**
9:15 – 10:00	Genomic Medicine – **D. Donnai**
10:00 – 10:45	Phenotype to genotype – **H. Brunner**
10:45 – 11:00	Coffee break
11:00 – 11:45	Cytogenetics and arrays – **E. Klopocki**
11:45 – 12:30	Complex disorders and classical gene identification – **A. Read**
12:30 – 13:15	Discussion of the morning lectures
13:30 – 14:30	Lunch Break

Afternoon Workshops

Session I (14:30 – 16:00)	Mutation patterns – **H. Brunner**
	Interpreting CNVs for beginners – **E. Klopocki** Computer room
	Dysmorphology – **D. Donnai**
16:00 – 16:30	Coffee break
Session II (16:30 – 18:00)	Mutation patterns – **H. Brunner**
	Interpreting CNVs for beginners – **E. Klopocki** Computer room
	Dysmorphology – **D. Donnai**

MONDAY APRIL 30TH

Morning Lectures:	**Basics of NGS for Mendelian disorders**
09:00 – 09:45	Basics of next generation sequencing technology – **A. Hoischen**
09:45 – 10:30	Basics of NGS bioinformatics – **C. Gilissen**
10:30 – 11:00	Coffee break
11:00 – 11:45	NGS in the clinic – **H. Brunner**
11:45 – 12:30	Long-read sequencing – **E. E. Eichler**
12:30 – 13:15	Discussion of the morning lectures
13:30 – 14:30	Lunch Break

Afternoon Workshops

Session I (14:30 – 16:00)	NGS Bioinformatics Basics – **C. Gilissen & T. Pippucci** Computer room
	Targeted NGS approaches – **A. Hoischen**
	Clinical Considerations for NGS **– H. Brunner**
16:00 – 16:30	Coffee break
Session II (16:30 – 18:00)	NGS Bioinformatics Basics - **C. Gilissen & T. Pippucci** Computer room
	How to set up a NGS lab? – **A. Hoischen**
	Copy number variations **– E. E. Eichler**

TUESDAY MAY 1ST

Morning Lectures:	**Therapy and prenatal diagnostics in the NGS era**
09:00 – 09:45	Therapy and cancer – **J. Burn**
09:45 – 10:30	Novel Cancer immunotherapy approach – **G. Germano**
10:30 – 11:00	Coffee break
11:00 – 11:45	SMA: From gene and modifier to therapy – **B. Wirth**
11:45 – 12:30	The therapy for cystic fibrosis as a paradigm for other genetic diseases **– L. Galietta**
12:30 – 13:15	Discussion of the morning lectures
13:30 – 14:30	Lunch Break

Afternoon Workshops

Session I (14:30 – 16:00)	NGS Bioinformatics, variant interpretation – **C. Gilissen & T. Pippucci** Computer room
	Rarity in the clinic – **J. Burn**
16:00 – 16:30	Coffee break
Session II (16:30 – 18:00)	NGS Bioinformatics, variant interpretation – **C. Gilissen & T. Pippucci** Computer room
	Ethics of medical genetics –**A. Read**
	From your newly discovered candidate gene to its function – **B. Wirth**
18:00	Poster viewing session with aperitif (session I)

WEDNESDAY MAY 2ND

Morning Lectures:	**Complex mechanisms of disease**
09:00 – 09:45	Molecular inversion probes and mosaicism – **A. Hoischen**
09.45 – 10:30	Epigenetics, imprinting, clinical – **K. Temple**
10:30 – 11:00	Coffee break
11:00 – 11.45	Non-coding mutations/long-range effects – **E. Klopocki**
11:45 – 12:30	Massively parallel functional assays – **L. Starita**
12:30 – 13:15	Discussion of the morning lectures
13:30 – 14:30	Lunch Break

Afternoon Excursion

THURSDAY MAY 3RD

Morning Lectures:	**Novel NGS applications**
09:00 – 09:45	Discovering structural variants in cancer using NGS data - **T. Rausch**
09:45 – 10:30	Phenotype and NGS integration/HPO benefits – **D. Fitzpatrick**
10:30 – 11:00	Coffee break
11:00 – 11:45	GWAS with NGS **- M. Nothnagel**
11:45 – 12:30	Data integration – **L. Franke**
12:30 – 13:15	Discussion of the morning lectures
13:30 – 14:30	Lunch Break

Afternoon Workshops

Session I (14:30 – 16:00)	Discovering structural variants in cancer using NGS data, par. 1 – **T. Rausch** Computer room
	Genetic Imprinting – **K. Temple**
	Multiplexed functional assays/variant interpretations – **L. Starita**
16:00 – 16:30	Coffee break
Session II (16:30 – 18:00)	Discovering structural variants in cancer using NGS data, par. 2 – **T. Rausch** Computer room
	How to do GWAS – **M. Nothnagel**
	How to do RNASeq – **L. Franke**
18:00	Poster viewing session with aperitif (session II)

FRIDAY MAY 4TH

Morning Lectures:	**Large scale NGS**
09:00 – 09:45	Patterns of rare variation contributing to disease – **K.Samocha**
09:45 – 10:30	Genomics England – **A. Rendon**
10:30 – 11:00	Coffee break
11:00 – 11:45	Non-invasive prenatal testing – **J. Weiss**
11:45 – 12:30	Single cell sequencing and applications to PGD – **T. Voet**
12:30 – 13:15	Discussion of the morning lectures
13:30 – 14:30	Lunch Break

Afternoon Workshops

Session I (14:30 – 16:00)	Discovering structural variants in cancer using NGS data, part. 1 - **T. Rausch** Computer room
	Interpretation of rare variants – **K. Samocha**
	Practical considerations for NIPT – **J. Weiss**
16:00 – 16:30	Coffee break
Session II (16:30 – 18:00)	Discovering structural variants in cancer using NGS data, part. 2 – **T. Rausch** Computer room
	Large genomics projects – **A. Rendon & D. Fitzpatrick**
	How to do single cell genomics? – **T. Voet**
18.00 – 18.30	Best poster presentations

Social dinner and farewell party

SATURDAY MAY 5TH

Departure