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 Klopacki (Wurzburg, Germany); Michael Nothnagel (Cologne, Germany); Tobias Rausch (Heidelberg, Germany); Andrew Read (Manchester, UK); Augusto Rendon (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. Kloocki

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. Kloocki
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. Kloocki
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. Galletta

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 Discovering structural variants in cancer using NGS data - T. Rausch
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. Klopopcki
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 Molecular inversion probes and mosaicism – A. Hoischen
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
Mechanism for non-coding mutations - E. Klopopcki

18:00 Poster viewing session with aperitif (session II)
**FRIDAY MAY 4TH**

<table>
<thead>
<tr>
<th>Time</th>
<th>Event</th>
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<tbody>
<tr>
<td>09:00 – 09:45</td>
<td>Presentations of best poster from students</td>
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<tr>
<td>09:45 – 10:30</td>
<td>Genomics England – <strong>A. Rendon</strong></td>
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<td>10:30 – 11:00</td>
<td>Coffee break</td>
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<tr>
<td>11:00 – 11:45</td>
<td>Non-invasive prenatal testing – <strong>J. Weiss</strong></td>
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<tr>
<td>11:45 – 12:30</td>
<td>Single cell sequencing and applications to PGD – <strong>T. Voet</strong></td>
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<tr>
<td>12:30 – 13:15</td>
<td>Discussion of the morning lectures</td>
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<tr>
<td>13:30 – 14:30</td>
<td>Lunch Break</td>
</tr>
</tbody>
</table>

**Afternoon Workshops**

**Session I (14:30 – 16:00)**

- Discovering structural variants in cancer using NGS data, part. 1 - **T. Rausch**  
  Computer room
- How to do RNASeq – **L. Franke**
- 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**

**Social dinner and farewell party**

**SATURDAY MAY 5TH**

**Departure**