Clinical Genomics and NGS

Bertinoro - Italy
April 28 – May 3, 2019

32nd Course jointly organized by ESGM, ESHG AND CEUB

University Residential Centre
Via Frangipane, 6 – Bertinoro

Course Directors:

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

COURSE PROGRAM

SATURDAY APRIL 27TH   Arrival and dinner

SUNDAY APRIL 28TH

Morning Lectures: **Medical Genetics concepts and principles**

8:30 – 9:00	Participants Registration
9:00 – 9:15	Introduction to the course – **H. Brunner – with video of 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 – **M. Spielman**
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 – **M. Spielman**
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 – **M. Spielman**
Computer room

Dysmorphology – **D. Donnai**

MONDAY APRIL 29TH

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

Rarity in the clinic – **J. Burn**

Copy number variations **– E. E. Eichler**

TUESDAY APRIL 30TH

Morning Lectures: **Therapy and novel disease mechanisms in the NGS era**

09:00 – 09:45   Therapy and cancer – **J. Burn**
09:45 – 10:30   SMA: From gene and modifier to therapy – **B. Wirth**
10:30 – 11:00   Coffee break
11:00 – 11:45   Molecular inversion probes and mosaicism – **A. Hoischen**
11:45 – 12:30   Epigenetics, imprinting, clinical – **K. Temple**

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

Genetic Imprinting – **K. Temple**

New workshop tbd **– David Fitzpatrick**

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 1ST

Morning Lectures:	**Prenatal testing in the NGS era and complex mechanisms of disease**
09:00 – 09:45	Non-invasive prenatal testing – **J. Weiss**
09.45 – 10:30	Single cell sequencing and applications to PGD – **T. Voet**
10:30 – 11:00	Coffee break
11:00 – 11.45	Non-coding mutations/long-range effects – **M. Spielman**
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 2ND

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	Patterns of rare variation contributing to disease – **tbd**
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

Multiplexed functional assays/variant interpretations – **L. Starita**

How to do single cell genomics? – **T. Voet**

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, par. 2 – **T. Rausch**
Computer room

How to do GWAS – **M. Nothnagel**

Interpretation of rare variants – **tbd**

18:00   Poster viewing session with aperitif (session II)

Social dinner and farewell party

FRIDAY MAY 3RD

Morning Lectures: **Highlights of the 2019 course**

09:30 – 10:30   New McKusick-Romeo Lecture: INVITED SPEAKER NN
10:30 – 11:30   Quiz on: "What have you learned"?
11:30 – 12:00   Best poster pitches by students

12:00 – 13:00   Lunch Break

Departure after lunch