Clinical Genomics and NGS

Bertinoro - Italy
April 28 – May 3, 2019

32nd Course jointly organized by ESHG AND CEUB

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)

SATURDAY APRIL 27TH

Arrival and dinner

SUNDAY APRIL 28TH

Morning Lectures:	**Medical Genetics concepts and principles**
08:30 – 09:00	Participants Registration
09:00 – 09:15	Introduction to the course – **H. Brunner – with video of G. Romeo**
09:15 – 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**
	Linux Basics **– C. Gilissen**
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**
	Annotation Tools **– T. Pippucci**

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 Variant interpretation – **C. Gilissen & T. Pippucci** Computer room
	Genetic Imprinting – **K. Temple**
16:00 – 16:30	Coffee break
Session II (16:30 – 18:00)	NGS Variant interpretation – **C. Gilissen & T. Pippucci** Computer room
	Ethics for Medical Genetics –**A. Read**
	From genes to 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	Introduction to cancer genomics and its caveats - **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 – **K. Samocha**
12:30 – 13:15	Discussion of the morning lectures
13:30 – 14:30	Lunch Break

Afternoon Workshops

Session I (14:30 – 16:00)	Introduction to cancer genomics NGS data analysis – **T. Rausch** Computer room
	Interpretation of rare variants – **K. Samocha**
	Single cell genomics? – **T. Voet**
	Reporting NIPT results, and ethical concerns – **J. Weiss**
16:00 – 16:30	Coffee break
Session II (16:30 – 18:00)	Analyzing the chromatin accessibility landscape using ATAC-Seq data – **T. Rausch** Computer room
	How to do GWAS – **M. Nothnagel**
	Multiplexed functional assays – **L. Starita**
	Using DECIPHER for Variant Interpretation and Pathogenicity Scoring **D. Fitzpatrick**
18:00	Poster viewing session with aperitif (session II)

Social dinner and farewell party

FRIDAY MAY 3RD

Morning Lectures:	**Highlights of the 2019 course**
09:00 – 10:30	New McKusick-Romeo Lecture: **A. Ballabio**
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