



Clinical Genomics and NGS

**Bertinoro - Italy
May 8-13, 2022**

34rd 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 MAY 7TH

Arrival and dinner

SUNDAY MAY 8TH

Morning Lectures:

Medical Genetics concepts and principles

08:30 – 09:00	Participants Registration
08:50 – 09:00	Introduction to the course – H. Brunner – with video of G. Romeo
09:00 – 9:45	Phenotype to genotype – H. Brunner / D. Donnai
09:45 – 10:30	Cytogenetics and arrays – E. Klopocki
10:30 – 11:00	Coffee break
11:00 – 11:45	Complex disorders and classical gene identification – A. Read
11.45 – 12.30	GWAS and follow-up - K. Ludwig
12.30 – 13.00	Discussion of the morning lectures
13:00 – 14:00	Lunch Break

Afternoon Workshops

Session I (14:00 – 15:30)	Mutation patterns – H. Brunner
	Interpreting CNVs for beginners – E. Klopocki Computer room
	GWAS analysis for beginners – K. Ludwig
	Ethics for Medical Genetics – A. Read
15:30 – 16:00	Coffee break
Session II (16:00 – 17:30)	Mutation patterns – H. Brunner
	Interpreting CNVs for beginners – E. Klopocki Computer room
	GWAS analysis for beginners – K. Ludwig

MONDAY MAY 9TH

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 patient care – L. Snijders Blok
11:45 – 12:30	The new era of clinical genetics – K. Boycott
12:30 – 13:00	Discussion of the morning lectures
13:00 – 14:00	Lunch Break

Afternoon Workshops

Session I (14:00 – 15:30)	Bioinformatics Basics – C. Gilissen & T. Pippucci Computer room
	Targeted NGS approaches – A. Hoischen
	Clinical Considerations for NGS – L. Snijder Blok
15:30 – 16:00	Coffee break
Session II (16:00 – 17:30)	Bioinformatics Basics - C. Gilissen & A. Hoischen Computer room
	Clinical Considerations for NGS – L. Snijder Blok

TUESDAY MAY 10TH

Morning Lectures:

Therapy and novel disease mechanisms in the NGS era

09:00 – 09:45	Non-coding mutations/long-range effects - M. Spielmann
09:45 – 10:30	Epigenetics, imprinting, clinical – K. Temple
10:30 – 11:00	Coffee break
11:00 – 11:45	SMA: From gene and modifier to therapy – B. Wirth
11:45 – 12:30	Therapy and cancer – J. Burn
12:30 – 13:00	Discussion of the morning lectures
13:00 – 14:00	Lunch Break

Afternoon Workshops

Session I (14:00 – 15:30)	Variant interpretation – C. Gilissen & T. Pippucci Computer room
	Genetic Imprinting – K. Temple
15:30 – 16:00	Coffee break
Session II (16:00 – 17:30)	Variant interpretation – C. Gilissen & T. Pippucci Computer room
	From genes to function – B. Wirth
17:30	Poster viewing session with aperitif (session I)

WEDNESDAY MAY 11TH

Morning Lectures:

Cell-free DNA and Mosaicism

09:00 – 09:45

Cell-free DNA and its applications – **F. Erger**

09.45 – 10:30

Noninvasive prenatal testing – **J. Weiss**

10:30 – 11:00

Coffee break

11:00 – 11.45

Somatic mutations in neurodevelopmental disease – **S. Baldassari**

11.45 – 12.30

Somatic mutations in cancer – **S. Nik-Zainal**

12:30 – 13:00

Discussion of the morning lectures

13:00 – 14:00

Lunch Break

Afternoon Excursion

THURSDAY MAY 12TH

Morning Lectures:

New Technologies and Big Data

09:00 – 09:45	Patterns of rare variation contributing to disease – K. Samocha
09.45 – 10:30	Boundaries between rare and common disease – A. Reymond
10:30 – 11:00	Coffee break
11:00 – 11.45	Long read sequencing technologies – E. Eichler
11.45 – 12.30	Massively parallel functional assays – L. Starita
12:30 – 13:00	Discussion of the morning lectures
13:00 – 14:00	Lunch Break

Session I (14:00 – 15:30)

Interpretation of rare variants – **K. Samocha**

Long read sequencing – **E. Eichler**

15:30 – 16:00

Coffee break

Session II (16:00 – 17:30)

Interpretation of rare variants – **K. Samocha**

Long read sequencing – **E. Eichler**

17:30

Poster viewing session with aperitif (session II)

Social dinner and farewell party

FRIDAY MAY 13TH

Morning Lectures:

09:00 – 10:30

10:30 – 11:30

11:30 – 12:00

12:00 – 13:00

Departure after lunch

Highlights of the 2022 course

New McKusick-Romeo Lecture: ???

Quiz on: “What have you learned”?

Best poster pitches by students

Lunch Break