Clinical Genomics and NGS

Bertinoro - Italy
May 8-13, 2022

34rd Course jointly organized by ESHG AND CEUB

ESHG
EUROPEAN SOCIETY OF HUMAN GENETICS

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)

CENTRO RESIDENZIALE
UNIVERSITARIO
Bertinoro

SATURDAY MAY 7TH

Arrival and dinner

SUNDAY MAY 8TH

Morning Lectures:	**Medical Genetics concepts and principles**
08:00 – 08:30	Participants Registration
08:30 – 08:45	Introduction to the course – **H. Brunner – with video of G. Romeo**
08:45 – 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 (recorded talk)**
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**
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: 'Tracking mutational footprints to study the origin of cancer' **– R. van Boxtel**
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 sequncing – **E. Eichler**
	NGS data visualisation with IGV – **F. Erger** Computer room
15:30 – 16:00	Coffee break
Session II (16:00 – 17:30)	
	Interpretation of rare variants – **K. Samocha**
	Long read sequncing – **E. Eichler**
	NGS data visualisation with IGV – **F. Erger** Computer room
17:30	Poster viewing session with aperitif (session II)

Social dinner and farewell party

FRIDAY MAY 13TH

Morning Lectures:	**Highlights of the 2022 course**
09:00 – 10:00	McKusick-Romeo Lecture: **Matthew Hurles (recorded talk, followed by live laudatio and live Q&A)**
10.00 – 10.30	Coffee break
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