Clinical Genomics and NGS

Bertinoro - Italy
May 4 - 10, 2024

36th Course jointly organized by ESHG AND CEUB

Course Directors

Christian Gilissen (Nijmegen, the Netherlands); Alexander Hoischen (Nijmegen, the Netherlands); Tommaso Pippucci (Bologna, Italy); Malte Spielmann (Lübeck, Germany)

SATURDAY MAY 4TH

Arrival and dinner

SUNDAY MAY 5TH

Morning Lectures:	**Medical Genetics concepts and principles**
08:30 – 09:00	Participants Registration
08:50 – 09:00	Introduction to the course – **T. Pippucci**
09:00 – 09:45	Why Medical Genomics Matters – **D. Donnai**
09:45 – 10:30	The architecture of monogenic disease – **H. Brunner**
10:30 – 11:00	Coffee break
11:00 – 11:45	Cytogenetics and arrays – **E. Klopocki**
11.45 – 12.30	Syndromology in the NGS era **- S. Douzgou Houge**
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
	Clinical Genetics in an NGS era **- S. Douzgou Houge**
	Optical genome mapping – **A. Hoischen**
15:30 – 16:00	Coffee break
Session II (16:00 – 17:30)	Mutation patterns – **H. Brunner**
	Interpreting CNVs for beginners – **E. Klopocki** Computer room
	Clinical Genetics in an NGS era **- S. Douzgou Houge**
	Copy number variants from exome sequencing – **T. Pippucci**

MONDAY MAY 6TH

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	Long read sequencing technologies – **E. Eichler**
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
	Recessive Carriers and PGT – **H. Brunner**
	NGS in a clinical setting: pitfalls and possibilities **– L. Snijders Blok**
	Long read sequencing – **E. Eichler**
15:30 – 16:00	Coffee break
Session II (16:00 – 17:30)	Bioinformatics Basics **- C. Gilissen & T. Pippucci** Computer room
	Recessive Carriers and PGT – **H. Brunner**
	NGS in a clinical setting: pitfalls and possibilities **– L. Snijders Blok**
	Long read sequencing – **E. Eichler**

TUESDAY MAY 7TH

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	Boundaries between rare and common disease – **A. Reymond**
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 Counselling what's the point? – **J. Burn**
	Non-coding mutations - **M. Spielmann**
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**
	Genetic Counselling what's the point? – **J. Burn**
	Non-coding mutations - **M. Spielmann**
17:30	Poster viewing session with aperitif (session I)

WEDNESDAY MAY 8TH

Morning Lectures:	**Cell-free DNA and Mosaicism**
09:00 – 09:45	Somatic mutations and cancer – **T. Rausch**
09.45 – 10:30	Cell-free DNA and its applications **– E. Sistermans**
10:30 – 11:00	Coffee break
11:00 – 11.45	RNASequencing for Mendelian disease - **N. Whiffin**
11.45 – 12.30	Ethics in genetics – **A. Lucassen**
12:30 – 13:00	Discussion of the morning lectures
13:00 – 14:00	Lunch Break

Afternoon Excursion

THURSDAY MAY 9TH

Morning Lectures:	**New Technologies and Big Data**
09:00 – 09:45	Patterns of rare variation contributing to disease – **K. Samocha**
09.45 – 10:30	Genetic Imprinting – **K. Temple**
10:30 – 11:00	Coffee break
11:00 – 11.45	Complex disorders and classical gene identification – **M. Nothnagel**
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**
	GWAS analysis for beginners – **M. Nothnagel**
	Genetic Imprinting – **K. Temple**
	Population genomic screening – **D. Donnai & A. Lucassen**
15:30 – 16:00	Coffee break
Session II (16:00 – 17:30)	
	Interpretation of rare variants – **K. Samocha**
	GWAS analysis for beginners – **M. Nothnagel**
	Functional data in variant interpretation – **L. Starita**
	Population genomic screening – **D. Donnai & A. Lucassen**
17:30	Poster viewing session with aperitif (session II)

Social dinner and farewell party

FRIDAY MAY 10TH

Morning Lectures:	**Highlights of the 2024 course**
09:00 – 10:00	McKusick-Romeo Lecture: **S. Nik-Zainal**
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