



Clinical Genomics and NGS

Virtual edition

Bertinoro - Italy

May 6-11, 2021

33rd 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)



Lectures guidelines:

- 30min talk, 15min Q&A (with several questions by the speaker to students), or: Every talk should at least two interactive question parts
- Every speaker has to prepare 3 multiple choice questions prior to the course for quiz (exam for certificate)

THURSDAY MAY 6th

Medical Genetics concepts and principles

| <i>Time</i> | <i>Title</i> | <i>Speaker & moderator</i> |
|---------------|--|-------------------------------------|
| 09:00 – 09:15 | Introduction to the virtual course | H. Brunner & T. Pippucci |
| 09:15 – 10:15 | Phenotype to genotype | Dian Donnai and H. Brunner |
| 10:15 – 11:00 | Complex disorders and classical gene identification | A. Read |
| 11.00 –11.30 | Coffee break | |
| 11.30 – 12.15 | Cytogenetics and arrays | E. Klopocki |
| 12.15 – 13.00 | GWAS and follow-up | K. Ludwig |
| 13.00 – 13.15 | Coffee break | |
| 13.15 – 14.00 | Reflection of day 1 (Q&A and quiz) (<i>all speakers present</i>) | |

FRIDAY MAY 7TH

NGS basics and applications in clinical genetics

| <i>Time</i> | <i>Title</i> | <i>Speaker & moderator</i> |
|---------------|--|-----------------------------------|
| 13:00 – 13:15 | Introduction day 2 | T. Pippucci & B. Wirth |
| 13:15 – 14:00 | Basics of next generation sequencing technology | A. Hoischen |
| 14:00 – 14:45 | Basics of NGS bioinformatics | C. Gilissen |
| 14.45 – 15.15 | Coffee break | |
| 15.15 – 16.00 | NGS in patient care | L. Snijders Blok |
| 16.00 – 16.45 | The new era of clinical genetics | K. Boycott |
| 16.45 – 17.15 | Coffee break | |
| 17.15 – 18.00 | Reflection of day 2 (Q&A and quiz) (<i>all speakers present</i>) | |
| 18.00 – 19.00 | Breakout session: fireside chat with faculty | |

WEEKEND OFF

MONDAY MAY 10TH

Beyond the exome, rare disease therapy

| <i>Time</i> | <i>Title</i> | <i>Speaker & moderator</i> |
|---------------|--|-----------------------------------|
| 13:00 – 13:15 | Introduction day 3 | B. Wirth & C. Gilissen |
| 13:15 – 14:00 | Non-coding mutations/long-range effects | M. Spielmann |
| 14:00 – 14:45 | Epigenetics, imprinting, clinical | K. Temple |
| 14.45 – 15.15 | Coffee break | |
| 15.15 – 16.00 | SMA: From gene and modifier to therapy | B. Wirth |
| 16.00 – 16.45 | Therapy and cancer | J. Burn |
| 16.45 – 17.15 | Coffee break | |
| 17.15 – 18.00 | Reflection of day 3 (Q&A and quiz) (<i>all speakers present</i>) | |
| 18:00 – 19:00 | Evening lecture: Long read genomes | Evan Eichler |

TUESDAY MAY 11TH

New technologies and big data

| <i>Time</i> | <i>Title</i> | <i>Speaker & moderator</i> |
|---------------|--|--------------------------------------|
| 13:00 – 13:15 | Introduction day 4 | C. Gilissen & A. Hoischen |
| 13:15 – 14:00 | Patterns of rare variation contributing to disease | K. Samocha |
| 14:00 – 14:45 | Boundaries between rare and common disease | A. Reymond |
| 14.45 – 15.15 | Coffee break | |
| 15.15 – 16.00 | Non-invasive prenatal testing | J. Weiss |
| 16.00 – 16.45 | Single-cell genomics | J. Shendure |
| 16.45 – 17.15 | Coffee break | |
| 17.15 – 18.00 | Reflection of day 4 (Q&A and quiz) (<i>all speakers present</i>) | |
| 18:00 – 19:00 | Romeo-McKusick lecture | H. Clevers |