# The EUROPEAN SCHOOL OF GENETIC MEDICINE
# and the EUROPEAN SOCIETY OF HUMAN GENETICS
# organize the
# 3rd COURSE IN NEXT GENERATION SEQUENCING
# Bertinoro di Romagna (Italy), May 6-10, 2014

**Abstract**: This course aims to give students insight into the use of next generation sequencing technology for genetic studies in humans. Aimed at researchers and diagnostics personnel with a background in genetics, biology, biomedical & molecular sciences and/or computational biology. **Topics**: Next generation sequencing basics: targeted/exome/genome sequencing, data analysis and interpretation, clinical applications.

## Arrival: Tuesday May 6

## Wednesday, May 7

**Morning Session: Introduction to Next Generation Sequencing**

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| 9.00 – 9.15 | Introduction to the course<br>**Giovanni Romeo** |
| 9.15 – 10.15 | Next Generation Sequencing basics<br>**Joris Veltman (Nijmegen)** |
| 10.15 – 10.45 | **Coffee Break** |
| 10.45 – 11.45 | Bioinformatic basics<br>**Christian Gilissen (Nijmegen)** |
| 11.45 – 12.45 | Bioinformatic strategies & ontology's<br>**Peter Robinson (Berlin)** |
| 12.45– 13.30 | **Lunch Break** |

**Afternoon Session:**

| | |
|---|---|
| 13.30 –14.00 | Poster Viewing Session |
| 14.00 – 16.00 | Concurrent Workshops:<br>Computer practical: Variant identification (**C. Gilissen** & **T. Pippucci**)<br>Workshops by speakers |
| 16.00-16.30 | **Coffee Break** |

16.30 – 18.00 Concurrent Workshops
Computer practical: Variant identification (**C. Gilissen** & **T. Pippucci**)
Workshops by speakers

## Thursday, May 8

**Morning Session: Application disease gene identification & diagnostics**

9.00 – 10.00 Targeted breast cancer diagnostics
**Gert Matthijs (Leuven)**

10.00 – 11.00 Exome diagnostics in intellectual disability
**Anita Rauch (Zürich)**

11.00 – 11.30 **Coffee Break**

11.30- 12.30 De novo mutations in human genetic disease
**Joris Veltman (Nijmegen)**

12.30 – 13.30 Prenatal sequencing
**Dagan Wells (Oxford)**

13.30 – 14.30 **Lunch Break**

**Afternoon Session:**

14.30 – 16.00 Concurrent Workshops
Computer practical: Disease gene identification (**C. Gilissen** & **T.Pippucci**)
Workshops by speakers

16.00-16.30 **Coffee Break**

16.30 – 18.00 Concurrent Workshops
Computer practical: Disease gene identification (**C. Gilissen** & **T.Pippucci**)
Workshops by speakers

## Friday, May 9

**Morning Session: Application in common disease & cancer**

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|---|---|
| 9.00 – 10.00 | NGS in population genetics and complex diseases<br>**Paul de Bakker (Utrecht)** |
| 10.00 – 11.00 | The DDD and UK10K project<br>**t.b.d.** |
| 11.00 – 11.30 | **Coffee Break** |
| 11.30 – 12.30 | Cancer genome sequencing<br>**Ian Tomlinson (Oxford)** |
| 12.30 – 13:30 | Exome sequencing to study rare and common variation in diabetes<br>**Amélie Bonnefond (Montpellier)** |
| 13:30 – 14.30 | **Lunch Break** |

**Afternoon Session:**

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|---|---|
| 14.30 –15.00 | Poster Viewing Session (or CLCbio/Cartegenia demonstration?) |
| 15.00 – 16.30 | Concurrent Workshops<br>Computer practical: Diagnostic NGS (**C. Gilissen** & **T. Pippucci**)<br>Workshops by speakers |
| 16.30-17.00 | Coffee Break |
| 17.00 – 18.30 | Concurrent Workshops<br>Computer practical: Diagnostic NGS (**C. Gilissen** & **T. Pippucci**)<br>Workshops by speakers |

## Saturday, May 10

**Morning Session: Genome technologies**

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|---|---|
| 9.00 – 10.00 | Handheld diagnostics on nanowires<br>**Jonathan O'Halloran (Newcastle)** |
| 10.0 – 11.00 | NGS-based detection of somatic mutations in human disease<br>**t.b.d.** |
| 11.00 – 11.30 | **Coffee Break** |
| 11.30 – 12.00 | Best Posters Presentations by students |
| 12.00 – 12.30 | Wrapping up of the course **(J. Veltman**) |
| 12.30 | **Lunch** |

## Departure