# The EUROPEAN SCHOOL OF GENETIC MEDICINE
# and the EUROPEAN SOCIETY OF HUMAN GENETICS
# organize the
# 5th COURSE IN NEXT GENERATION SEQUENCING
# Bertinoro di Romagna (Italy), May 4-7, 2016

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 3rd**

**Wednesday, May 4**

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

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|---|---|
| 9.00 – 9.15 | Introduction to the course<br>**Giovanni Romeo** |
| 9.15 – 10.45 | Next Generation Sequencing basics<br>**Joris Veltman** |
| 10.45 – 11.15 | **Coffee Break** |
| 11.00 – 12.30 | Bioinformatic basics<br>**Christian Gilissen** |
| 12.30 – 13.30 | Bioinformatic strategies & ontology's<br>**Peter Robinson** |
| 13.30– 14.30 | **Lunch Break** |

**Afternoon Session:**

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|---|---|
| 14.30 – 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<br>Computer practical: Variant identification (**C. Gilissen** & **T. Pippucci**)<br>Workshops by speakers |

**Thursday, May 5**

**Morning Session: Applications to disease gene identification & diagnostics**

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|---|---|
| 9.00 – 10.00 | Exome diagnostics in intellectual disability<br>**Anita Rauch** |
| 10.00 – 11.00 | Genomics England<br>**Katherine Smith** |
| 11.00 – 11.30 | **Coffee Break** |
| 11.30- 12.30 | De novo mutations in human genetic disease<br>**Joris Veltman** |
| 12.30 – 13.30 | WGS for non-coding mutations in congenital disorders<br>**Malte Spielmann** |
| 13.30 – 14.30 | **Lunch Break** |

**Afternoon Session:**

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|---|---|
| 14.30 –15.00 | Poster Viewing Session |
| 15.00 – 16.30 | Concurrent Workshops<br>Computer practical: Disease gene identification (**C. Gilissen** & **T.Pippucci**)<br>Workshops by speakers |
| 16.30 – 17.00 | **Coffee Break** |
| 17.00 – 18.30 | Concurrent Workshops<br>Computer practical: Disease gene identification (**C. Gilissen** & **T.Pippucci**)<br>Workshops by speakers |

**Friday, May 6**

**Morning Session: Applications to common disease & cancer**

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|---|---|
| 9.00 – 10.00 | Handheld diagnostics on nanowires<br>**Heather Murton** |
| 10.00 – 11.00 | Application of NGS for the diagnosis of neonatal diabetes<br>**Elisa de Franco** |
| 11.00 – 11.30 | **Coffee Break** |
| 11.30 – 12.30 | Cancer genomics<br>**Francesca Demichelis** |
| 12.30 – 13:30 | NGS data integration and statistics<br>**Lude Franke** |
| 13:30 – 14.30 | **Lunch Break** |

**Afternoon Session:**

| | |
|---|---|
| 14.30 –15.00 | Poster Viewing Session |
| 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 7**

**Morning Session: Genome technologies**

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|---|---|
| 9.00 – 10.00 | Single-cell genomics unveils genetic heterogeneity in health and disease, and enables novel clinical applications<br>**Thierry Voet** |
| 10.00 – 11.00 | The Sardinia project<br>**Carlo Sidore** |
| 11.00 – 11.30 | **Coffee Break** |
| 11.30 – 12.00 | Best Posters Presentations by students |
| 12.00 – 12.30 | Wrapping up of the course **(Christian Gilissen**) |
| 12.30 | **Lunch** |

## Departures