



**SSIEM course on next generation sequencing and clinical genomics
18th to 20th March 2019, Barcelona**

| 18th March | |
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| 10:00-14:00 | Registration |
| 11:30-11:40 | Welcome . Aims of the course <i>Antonia Ribes. Hospital Clínic, CIBERER, Barcelona</i> |
| 11:40 - 12:30 | Review of basic genetic concepts. <i>Gert Matthijs. University Hospital Leuven, Leuven</i> |
| 12:30 – 13:00 | Introduction to NGS technologies (from Sanger to targeted or /WES/WGS sequencing). <i>Joan Anton Puig. Hospital Clínic, CIBERER, Barcelona</i> |
| 13:00-14:00 | Lunch |
| 14:00- 14:30 | Bioinformatics and NGS data analysis pipeline (from raw data to variant annotation). <i>Joaquin Dopazo. CIBERER, Sevilla</i> |
| 14:30- 15:30 | Prioritization of variants and genes (filtering steps). Impact of variants (in silico predictors and ACMG criteria) <i>Frederic Tort. Hospital Clínic, CIBERER, Barcelona</i> |
| 15:30 - 16:00 | Coffee break |
| 16:00 – 16:30 | Hands on work: example of exome sequence alignment with reference genome and variant calling. <i>Javier Perez Florido. CIBERER, Sevilla</i> |
| 16:30 -17:30 | Hands on work: IVA-ACCI platform. <i>Joaquin Dopazo and Javier Perez Florido (CIBERER, Sevilla)</i> |
| 17:30-18:00 | HPO terminology in the clinical practice, hands on work. <i>David Gómez Andrés ,Hospital Vall d’Hebrón, Barcelona</i> |
| 19:30 | Dinner |

| 19th March | |
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| 9:00- 9:45 | Usefulness of NGS for the diagnosis of rare diseases. <i>Holger Prokisch, Institute of Human Genetics, Muenchen</i> |
| 9:45 – 10:30 | European platforms for the study of rare diseases: RD-Connect/RD-Cat links to different data types - omics (e.g. genomics), clinical information (HPO), patient registries and biobanks . <i>Sergi Beltran. CNAG-CRG, Barcelona</i> |
| 10:30- 11:00 | Coffee break |
| 11:00 – 11:30 | Software requirements: handling, storage, annotation and data bases. <i>Sergi Beltran. CNAG-CRG, Barcelona</i> |
| 11.30 – 12:15 | RNaseq data analysis and interpretation. <i>Holger Prokisch, Institute of Human Genetics, Muenchen</i> |
| 12:15- 13:00 | Lunch and networking |
| 13:00 -14:00 | Hands on work with a user friendly genomics platform. <i>Leslie Matalonga. Gemma Bullich. CNAG-CRG, Barcelona</i> |
| 14:00- 15:30 | Practical cases to be solved by the students with their own computer: easy cases Helping participants: <i>Laura Gort/ Frederic Tort/ Leslie Matalonga/Gemma Bullich/Judit Amstrong/Delia Yubero/Blai Morales</i> |
| 15:30-16:00 | Coffe break. |
| 16:00- 18:00 | Practical cases to be solved by the students with their own computer: difficult cases, cases solved by reanalysis of data. Helping participants: <i>Laura Gort/ Frederic Tort/ Leslie Matalonga/Gemma Bullich/Judit Amstrong/Delia Yubero/Blai Morales</i> |
| 19:30 | Dinner |

| 20th March | |
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| 9:00-10:00 | Analysis of mtDNA by NGS technology. <i>Elena Garcia Arumí. Hospital Vall d'Hebrón, Barcelona</i> |
| 10:00-11:00 | Functional studies for pathogenic confirmation. Laboratory approaches. <i>Richard J Rodenburg. Radboud university Medical Center, Nijmegen</i> |
| 11:00-11:30 | Coffee break |
| 11:30 -12:30 | System biology approaches. <i>Juan Antonio Garcia Ranea, University of Malaga, CIBERER, Malaga</i> |
| 12:30-12:40 | Concluding remarks <i>Antonia Ribes and Frederic Tort. Hospital Clínic, CIBERER, Barcelona</i> |
| 12:40 | Farewell lunch |