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

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

19th March	
9:00- 9:45	Usefulness of NGS for the diagnosis of rare diseases. *Holger Prokisch, Institute of Human Genetics, Muenchen*
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 . *Sergi Beltran. CNAG-CRG, Barcelona*
10:30- 11:00	Coffee break
11:00 – 11:30	Software requirements: handling, storage, annotation and data bases. *Sergi Beltran. CNAG-CRG, Barcelona*
11.30 – 12:15	RNAseq data analysis and interpretation. *Holger Prokisch, Institute of Human Genetics, Muenchen*
12:15- 13:00	Lunch and networking
13:00 -14:00	Hands on work with a user friendly genomics platform. *Leslie Matalonga. Gemma Bullich. CNAG-CRG, Barcelona*
14:00- 15:30	Practical cases to be solved by the students with their own computer: easy cases Helping participants*: Laura Gort/ Frederic Tort/ Leslie Matalonga/Gemma Bullich/Judit Amstrong/Delia Yubero/Blai Morales*
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*: Laura Gort/ Frederic Tort/ Leslie Matalonga/Gemma Bullich/Judit Amstrong/Delia Yubero/Blai Morales*
19:30	Dinner

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