

CNAG-CIBERER Symposium on Genomic Medicine Applied to Rare Diseases

Madrid 5th July 2018

Aula Magna - Fundación Jiménez Díaz (Entreplanta)
 Avda. de los Reyes Católicos 2, 28040 Madrid

8:30	REGISTRATION
9:00	Opening and Presentation Jesús Fernandez Crespo , Director, Instituto de Salud Carlos III Ana Leal , Subdirectora Médica del Hospital Universitario, FJD Carmen Ayuso , Director, Biomedical Research Institute, FJD Ivo Gut , Director, Centro Nacional de Análisis Genómico (CNAG-CRG) Pablo Lapunzina , Scientific Director, CIBERER
	Session 1 – Chairperson: Carmen Ayuso, Director, Biomedical Research Institute, FJD
9:30	Pablo Lapunzina , Scientific Director, CIBERER <i>Overview on Rare Diseases</i>
10:10	Belen Pérez , Assoc. Prof. Biochemistry and Molecular Biology, CEDEM – UAM <i>Value of genetic analysis for confirming inborn errors of metabolism and for implementation of personalized therapies</i>
10:50	COFFEE BREAK
	Session 2 – Chairperson: Ivo Gut, Director, Centro Nacional de Análisis Genómico (CNAG-CRG)
11:20	Sergi Beltran , Head Bioinformatics Unit, Centro Nacional de Análisis Genómico (CNAG-CRG) <i>Standardised genome-phenome analysis for personalised medicine on rare diseases</i>
12:00	Sara Pasalodos , EBMG and GCRB registered Genetic Counsellor and Nagen Project Manager <i>Navarra 1000 Genomes Project (NAGEN 1000)</i>
12:40	Mark Caulfield , Chief Scientist for Genomics England and Co-Director of the William Harvey Research Institute, Queen Mary University of London <i>The 100,000 Genomes Project</i>
13:20	LUNCH
	Session 3 – Chairperson: Pablo Lapunzina, Scientific Director, CIBERER
14:20	Chitra Kotwaliwale , Senior Global Product Manager - NGS Technology Group, Diagnostics and Genomics, Agilent Technologies <i>Sample to Answer: Developing a robust and cost-effective exome workflow for the investigation of rare diseases</i>
15:00	Encarna Guillén , Jefa de Sección de Genética Médica. Servicio de Pediatría, Hospital Clínico Universitario Virgen de la Arrixaca. IMIB-Arrixaca. Universidad de Murcia. CIBERER-ISCIIII <i>How genomic medicine is revolutionizing health care</i>
15:40	Hanns Lochmüller , Dept Neuropediatrics & Muscle Disorders, Med Center UniFreiburg & CNAG-CRG <i>Data analysis and data sharing in Rare Disease with RD-Connect</i>
16:20	Lluís Montoliu , Dept of Molecular & Cellular Biology, CNB-CSIC and CIBERER ISCIIII <i>Towards universal diagnosis and future therapies in albinism: combining genome analysis and genome edition</i>
17:00	CLOSURE