cnag
centre nacional d'anàlisi genòmica
centro nacional de análisis genómico

CRG
Centre for Genomic Regulation

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 *Overview on Rare Diseases*
10:10	**Belen Pérez**, Assoc. Prof. Biochemistry and Molecular Biology, CEDEM – UAM *Value of genetic analysis for confirming inborn errors of metabolism and for implementation of personalized therapies*
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) *Standardised genome-phenome analysis for personalised medicine on rare diseases*
12:00	**Sara Pasalodos**, EBMG and GCRB registered Genetic Counsellor and Nagen Project Manager *Navarra 1000 Genomes Project (NAGEN 1000)*
12:40	**Mark Caulfield**, Chief Scientist for Genomics England and Co-Director of the William Harvey Research Institute, Queen Mary University of London *The 100,000 Genomes Project*
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 *Sample to Answer: Developing a robust and cost-effective exome workflow for the investigation of rare diseases*
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-ISCIII *How genomic medicine is revolutionizing health care*
15:40	**Hanns Lochmüller**, Dept Neuropediatrics & Muscle Disorders, Med Center UniFreiburg & CNAG-CRG *Data analysis and data sharing in Rare Disease with RD-Connect*
16:20	**Lluís Montoliu**, Dept of Molecular & Cellular Biology, CNB-CSIC and CIBERER ISCIII *Towards universal diagnosis and future therapies in albinism: combining genome analysis and genome edition*
17:00	**CLOSURE**

IIS FJD INSTITUTO DE INVESTIGACIÓN SANITARIA FUNDACIÓN JIMÉNEZ DÍAZ

Hospital Universitario Fundación Jiménez Díaz Grupo quirónsalud

ciberer Centro de Investigación Biomédica en Red Enfermedades Raras

Agilent Trusted Answers