

# 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	<b>REGISTRATION</b>
9:00	<b>Jesús Fernandez Crespo</b> , Director, Instituto de Salud Carlos III <b>Carmen Ayuso</b> , Director, Biomedical Research Institute Fundación Jimenez Diaz <b>Ivo Gut</b> , Director, Centro Nacional de Análisis Genómico <b>Pablo Lapunzina</b> , Scientific Director, CIBERER <i>Opening and Presentation</i>
9:30	<b>Pablo Lapunzina</b> , Scientific Director, CIBERER <i>Overview on Rare Diseases (title TBA)</i>
10:10	<b>Belen Pérez</b> , 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	<b>COFFEE BREAK</b>
11:20	<b>Sergi Beltran</b> , Head Bioinformatics Unit, Centro Nacional de Análisis Genómico (CNAG-CRG) <i>Standardised genome-phenome analysis for personalised medicine on rare diseases</i>
12:00	<b>Sara Pasalodos</b> , EBMG and GCRB registered Genetic Counsellor and Nagen Project Manager <i>Navarra 1000 Genomes Project (NAGEN 1000)</i>
12:40	<b>Mark Caulfield</b> , 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	<b>LUNCH</b>
14:20	<b>Chitra Kotwaliwale</b> , 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	<b>Encarna Guillén</b> , Jefa de Sección de Genética Médica. Servicio de Pediatría, Hospital Clínico Universitario Virgen de la Arrixaca. IMIB-Arrixaca. Universidad de Murcia. CIBERER-ISCIII <i>How genomic medicine is revolutionizing health care</i>
15:40	<b>Hanns Lochmüller</b> , Dept. of Neuropediatrics and Muscle Disorders, Medical Center - University of Freiburg and Centro Nacional de Análisis Genómico (CNAG-CRG) <i>Data analysis and data sharing in Rare Disease with RD-Connect</i>
16:20	<b>Lluís Montoliu</b> , Department of Molecular and Cellular Biology, CNB-CSIC and CIBERER ISCIII <i>Towards universal diagnosis and future therapies in albinism: combining genome analysis and genome edition</i>
17:00	<b>CLOSURE</b>