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CRG
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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	**Jesús Fernandez Crespo**, Director, Instituto de Salud Carlos III **Carmen Ayuso**, Director, Biomedical Research Institute Fundación Jimenez Diaz **Ivo Gut**, Director, Centro Nacional de Análisis Genómico **Pablo Lapunzina**, Scientific Director, CIBERER *Opening and Presentation*
9:30	**Pablo Lapunzina**, Scientific Director, CIBERER *Overview on Rare Diseases* (title TBA)
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**
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**
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. of Neuropediatrics and Muscle Disorders, Medical Center - University of Freiburg and Centro Nacional de Análisis Genómico (CNAG-CRG) *Data analysis and data sharing in Rare Disease with RD-Connect*
16:20	**Lluís Montoliu**, Department of Molecular and Cellular Biology, CNB-CSIC and CIBERER ISCIII *Towards universal diagnosis and future therapies in albinism: combining genome analysis and genome edition*
17:00	**CLOSURE**

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