

**“GENOMICS, PROTEOMICS AND BIOINFORMATICS APPLIED TO THE
STUDY OF RARE DISEASES”**

Dates: 3 and 4 May 2011

Place: “*Edificio del Rectorado Universidad de Málaga*”

University of Malaga (<http://www.uma.es/>). Malaga, Spain.

Organizers: Isabel Varela-Nieto and Francisca Sanchez-Jimenez

With the support of CIBERER.

Tuesday 3 May 2011

17.00-18.00.- Registration

18.00-18.20.- Welcome and Introduction. Francesc Palau (IBV CSIC, CIBERER, Spain)

18.20-19.10.- Inaugural conference. Orphanet: an Integrative Tool for the Study of Rare Diseases. Ana Rath (Orphanet, France)

Wednesday 4 May 2011

Session 1. Functional Genomics in Rare Diseases.

Chair: Isabel Varela-Nieto.

09.00-09.30.- International networks for the generation and study of mutant mice: applications in the rare diseases field Yann Herault (Institut Clinique de la Souris, Orléans, France)

09.30-10.00.- Genotype-phenotype analyses: is there a need for novel computational models? Isabel Varela-Nieto (IIBM CSIC-UAM, CIBERER, Spain).

10.00-10.30.- Mouse phenomics as a tool to identify biosignatures for predisposition, prodromal period and expressed disease Mara Dierssen (Genomic Regulation Center, CIBERER, Spain).

10.30-11.00.- Discussion.

11.00-11.30.- Coffee break.

Session 2. Proteomics in Rare Diseases

Chair: Francisca Sánchez-Jiménez.

11.30-12.00.- A technical proposal to advance in the integration of information on rare diseases (Jose F. Aldana Montes, UMA, Spain).

12.00-12.30.- The usefulness of combining in silico and “omics” technologies applied to rare/orphan diseases (Francisca Sánchez, UMA, CIBERER, Spain).

12.30-13.00.- Decipher: Database of chromosomal imbalance and phenotype in humans (Manuel Corpas, Wellcome Trust Sanger Institute, UK).

13.00-13.30.- Discussion.

13.30-15.00.- Lunch.

Session 3. Bioinformatics in Rare Diseases

Chair: Joaquín Dopazo.

15.00-15.30.- Introduction to systems biology in the study of rare diseases (speaker to be confirmed).

15.30-16.00.- Bioinformatic analysis from proteomic data (speaker to be confirmed).

16.00-16.30.- The role of bioinformatics in the integration of -omic data for gene prioritization (Joaquín Dopazo, CIPF, CIBERER, Spain).

16.30-17.00.- Discussion.

17.00-17.30.- Coffee break.

17.30-18.00.- Conclusions, next steps and closure.