Genetic interpretation of rare diseases:
from mutation to patient
6-7 July 2017
Salón de Actos
BioCruces. Hospital Universitario Cruces.
Plaza de Cruces 12, 48903. Barakaldo, Bizkaia (País Vasco)

July 6th
16:00-16:30h. Welcome address
Mª Luz del Valle (Managing Director, BioCruces)
Juana Mª Sáenz (Delegada FEDER País Vasco)
Javier Sancho (Pirepred Coordinator, BIFI, University of Zaragoza)
Juan Fernández-Recio (Scientific organizer, IBMB-CSIC, Barcelona Supercomputing Center)

SESSION 1:
GENETIC BASIS OF RARE DISEASES: MUTATION, ANALYSIS AND PREDICTION
Chairman: Xavier de la Cruz (VHIR)
16:30-17:00h. The genomic and biological landscape of rare and undiagnosed diseases.
Francesc Palau (Hospital Sant Joan de Deu).
17:00-18:00h. From measuring mutation impact to diagnostics yield: the path of translational research.
Xavier de la Cruz (VHIR).

18:00-18:30 h. Coffee break

18:30-19:00h. Computerization of Newborn Screening: Process and Procedures.
Tomás A. Pérez Fernández (UPV-EHU)
19:00-19:30h. Improving the newborn screening programs in Spain through the application of next generation sequencing.
Jairo Rodríguez (QGML).
19:30-19:45. Reporting mutation impact: encoding complex information to simplify clinical decisions.
Elena Álvarez (VHIR).
19:45-20:15. Genetic Bases of Rare Diabetes.
Luis Castaño. (BioCruces, Hospital Universitario Cruces, UPV/EHU, CIBERDEM, CIBERER).

July 7th
9:30-10:00h. Pharmacological chaperones as a novel therapeutic intervention line for congenital erythropoietic porphyria.
Oscar Millet (CIC-BioGUNE).
10:00-10:40h. Neonatal screening in France and new variants responsible for familial congenital hypothyroidism.
Frédérique Savagner / Isabelle Olivier (CHU Toulouse).
10:40-11:00h. Expanded newborn screening for metabolic disorders: experience in Aragon.
Inmaculada García (Hospital Miguel Servet).
11:00-11:20h. Mutations in proteins: MD for diagnostics and chaperones for therapy.
Javier Sancho (BIFI. Univ. Zaragoza).
11:20-12:00h. Coffee break
12:00-12:20. Structural modeling of pathological mutations involved in protein interactions.
Juan Fernández-Recio (CSIC,BSC)
12:20-12:45. In cellulo studies of new chemical drugs on PAH activity.
Marie-Lise Maddelein (IPBS).
12:45-13:00. The performance of standard pathogenicity predictors in metabolic disease.
Josu Aguirre (VHIR).
13:00-13:15. Strategies for MD simulation of reported mutations.
Juan José Galano (Univ. Zaragoza).

13:15-13:30 h. Closing remarks
Juan Fernández-Recio (Scientific organizer, IBMB-CSIC, Barcelona Supercomputing Center)