PROYECTO COFINANCIADO POR EL FONDO EUROPEO DE DESARROLLO REGIONAL (FEDER)

CE PROJET EST COFINANCÉ PAR LE FONDS EUROPÉEN DE DÉVELOPPEMENT RÉGIONAL (FEDER)

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)

July 6th

SESSION 1:
GENETIC BASIS OF RARE DISEASES: MUTATION, ANALYSIS AND PREDICTION

Chairman: Xavier de la Cruz (VHIR)

16:30-17:30h. *The genomic and biological landscape of rare and undiagnosed diseases.*
Francesc Palau (Hospital Sant Joan de Deu).

17:30-18:00h. *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

SESSION 2:
INTERPRETING MUTATIONS AT MOLECULAR LEVEL: TOWARDS IMPROVED DIAGNOSIS AND THERAPEUTICS

Chairman: Olivier Cuvillier (IPBS)

9:30-10:20h. *Pharmacological chaperones as a novel therapeutic intervention line for congenital erythropoietic porphyria.*
Oscar Millet (CIC-BioGUNE).

10:20-10:40h. *Neonatal screening in France and new variants responsible for familial congenital hypothyroidism.*
Frèdèrique 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).

July 7th

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