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

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

2nd Pirepred Scientific Meeting
The bench to bedside pathway: where are we now?

19 January 2018.

Salón de Actos, planta 10

Àrea General Hospital Universitari Vall d'Hebron

9:00-9:20h. *Welcome address*
Javier Sancho (Pirepred Coordinator, BIFI, University of Zaragoza).
Xavier de la Cruz (Scientific organizer, ICREA-VHIR).

MORNING SESSIONS

SESSION 1.
THE BIOMEDICAL/CLINICAL APPROACH TO RARE DISEASES. AN OVERVIEW

Chairmans: Xavier de la Cruz (ICREA-VHIR) and Javier Sancho (BIFI, University of Zaragoza)

9:20–10h. "*Triangles in rare diseases: a multidisciplinary approach to diagnosis, follow up/transition and therapeutic options*". Eduardo Tizzano (HUVH, VHIR).

10:00–10:40h. "*Neonatal screening in France and new variants responsible for familial congenital hypothyroidism*". Frédérique Savagner / Isabelle Olivier (CHU Toulouse).

10h40 - 11h10. "*Improving the newborn screening programs in Spain through the application of next generation sequencing*". Jairo Rodríguez (QGenomics, SL).

11:10–11:25h. "*Philanthropy and Biomedical Research*". Alejandra Manau (Unidad de Mecenazgo, VHIR).

11:25-11:55h. Coffee break

11:55–12:25h. "*Genetic Diagnosis of Primary Immunodeficiencies Using Next Generation Sequencing Technologies: Targeted gene panels vs WES*". Roger Colobran (Hospital Universitari Vall d'Hebron).

12h25 – 12h55. "*Effect of combined familial mutations on PAH activity in cell culture*". Marie-Lise Maddelein (IPBS, Toulouse).

12:55–13:25h. "*Cost: an (un)expected link between basic research and clinical applications?*". Xavier de la Cruz (ICREA-VHIR).

AFTERNOON SESSIONS

15:15-15:55h. "*The Sant Joan de Déu healthcare model for rare diseases: among medicine and biology*". Francesc Palau (Hospital Sant Joan de Déu).

SESSION 2.
A STUDY CASE: THE COMPONENTS OF TRANSLATION IN MITOCHONDRIAL DISEASE

Chairman: Juan Fernández-Recio (IBMB-CSIC, BSC)

15:55–16:25h. "*Mitochondrial proteins at the atomic level: a basis for functional studies in health and disease*". Maria Solà (Maria de Maeztu Unit of Excelence, IBMB-CSIC).

16:25-16:45 h. "*The structural landscape of damaging variants in mitochondrial proteins: consequences for pathogenicity prediction programs*". Elena Álvarez de la Campa (VHIR).

16:45–17:15h. "*Enhancing dNTP synthesis as a therapeutic approach for treating POLG deficiency*". Yolanda Cámara (Laboratori de Patologia Neuromuscular i Mitocondrial, VHIR; CIBERER).

17:15-17:45 h. Coffee break

SESSION 3.
UNVEILING THE MOLECULAR BASIS OF DISEASE WITH COMPUTATIONAL TOOLS

Chairman: Xavier de la Cruz (ICREA-VHIR)

17:45–18:15h. "*Simulating point mutations in enzymes related to metabolic disorders*". Juan José Galano, (BIFI, University of Zaragoza).

18:15–18:45h. "*Protein-protein docking can help the interpretation of genetic variants in rare diseases*". Juan Fernández-Recio (IBMB-CSIC, BSC).

18:45-19h. Closing remarks. Xavier de la Cruz (Scientific organizer, ICREA-VHIR).