2nd Pirepred Scientific Meeting

The bench to bedside pathway: where are we now?

19 January 2018

Salón de Actos, planta 10
Área 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

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


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).


11:25-11:55h. Coffee break


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

AFTERNOON SESSIONS


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

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


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).


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