

UB-CIBERER Mini Symposium on Genetic Rare Diseases

November 7th 2022

Aula de Graus, Facultat Biologia UB & streaming

Free registration at <https://tinyurl.com/ciberer-preinscripcion-curso>



10:00 h *The relevance of microphenotyping in rare diseases: diagnosis, monitoring and research*

Dr. Mercedes Serrano

Neurology Dpt Hospital Sant Joan de Déu; Institut de Recerca Sant Joan de Déu; CIBERER; Barcelona

10:45 h *Challenges of integrating exome sequencing into the clinical practice: from genetic counseling to its analysis and interpretation*

Dr. Marta Codina

Medicina Genética, Institut de Recerca Vall Hebron; Barcelona

11:30 h Coffe Break

12:00 h *ONLINE TALK: iPSCs and Genome Editing to Investigate Neuronopathic Gaucher Disease*

Dr. Isaac Canals

Stem Cells, Aging and Neurodegeneration Group, Department of Clinical Sciences, Neurology, Lund Stem Cell Center, Lund University, Lund, Sweden

12:45 h *Searching for a treatment in rare neurological diseases: the case of Lafora disease*

Dr. Pascual Sanz

Instituto de Biomedicina de Valencia, CSIC; CIBERER; Valencia

13:30 h Round table with the four speakers

14:00 h Closing

Organizers: Susanna Balcells, Gemma Marfany, Raquel Rabionet, Roser Corominas
U718-U720-U735. Departament de Genètica Microbiologia i Estadística, Universitat de Barcelona

Collaborators: CIBERER, Master in Genetics and Genomics (UB), Genetics Doctoral Program (UB), Càtedra UB de Enfermedades Raras (UB)