



International symposium

Novel approaches to study rare genetic diseases

November 12th 2021

Aula de graus & streaming

Contact jornada.UBCIBERER@ub.edu for link

- 10h Interdisciplinary approaches to identify the genetic causes of ultra rare diseases
Dr. Roser Urreizti
Hospital Sant Joan de Déu, CIBERER, ISCIII, Barcelona
- 11h Third-generation sequencing: A journey to the world of RNA modifications
Dr. Eva Novoa Pardo (online talk)
Center for Genomic Regulation, Universitat Pompeu Fabra, Barcelona
- 12h Structural variants create new topological-associated domains and ectopic retinal enhancer-gene contact in dominant retinitis pigmentosa
Dr Suzanne de Bruijn (online talk)
Dept. of Human Genetics, Radboudumc, Nijmegen, the Netherlands
- 13h Artificial and patient-derived cellular systems to model rare diseases and assess therapeutics
Dr. Alex Garanto (online talk)
Radboudumc, Nijmegen, the Netherlands

Organizers: Susanna Balcells, Daniel Grinberg, Gemma Marfany, Roser Corominas

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