Epilepsy

Group leader

Boronat Guerrero, Susana (FGS)

Researchers

Aledo Serrano, Angel (FGS)
Gil-Nagel Rein, Antonio (IR)
Sierra Marcos, Alba (FGS)
Turon Viñas, Eulalia (FGS)
Turon Viñas, Marc (FGS)

DESCRIPTION

This is a multidisciplinary group including the fields of epileptology, pediatric neurology, neuropsychology, and clinical genetics. We study several aspects of Epilepsy, mainly genotype-phenotype correlation of genetic epilepsy, electroclinical phenotyping, status epilepticus and neuropsychological comorbidities. Our research includes patients of all ages, children and adults, to increase knowledge about the natural history of epilepsy. We also offer new treatments for refractory patients with no other therapeutic options, providing them access to clinical trials.

MAIN LINES OF RESEARCH

- Genotype-phenotype correlation in genetic epilepsy (Susana Boronat, Asun Díaz, and Victoria Ros).
- Electroclinical and neuroimaging phenotyping (Alba Sierra).
- Neuropsychological comorbidities (Marc Turon).
- Emergency management of acute seizures and status epilepticus (Laia Turon).

SCIENTIFIC CHALLENGES

- Increase knowledge about deep phenotyping of genetic epilepsies, including electroclinical phenotyping and neuroimaging features.
- Description of new epileptic genetic syndromes.
- Improving therapeutic options for acute seizures, status epilepticus, and neuropsychological comorbidities.

ACTIVE GRANTS

- Boronat Guerrero, Susana. VISOR EEG (dispositiVo regIStro hOlter seRas EEG). CPP2021-008311. Ministerio de Ciencia e Innovación (MICINN). Duration: 2022-2025. 121.887,00 €.
- Boronat Guerrero, Susana. ENVISION: Estudio de la evolución natural de lactantes y niños con encefalopatías epilépticas y del desarrollo (EED). ETX-DS-001. Natural History Study of Infants and Children with SCN1A-positive Dravet Syndrome. Encoded Therapeutics INC. Non-competitive. Duration: 2021-2023. 9.000,00 €.

GRANTS AWARDED

- Boronat Guerrero, Susana. Study of genomic disease burden to complement Cause of Death investigation in Sub-Saharan African: a pilot study from Child Health and Mortality Prevention Surveillance (CHAMPS) Network. OPP1126780 GATES. Gates Foundation. Duration: 2023-2025. 252.118,96 €.

SCIENTIFIC PRODUCTION

- Esmel R, Valenzuela I, Riaza L, Rodríguez B, Rosés F, Boronat S, Sabaté A. Arterial tortuosity syndrome: Phenotypic features and cardiovascular manifestations in 4 newly identified patients. European Journal of Medical Genetics. 2023; 66(9):104823. DOI:10.1016/j.ejmg.2023.104823. PMID:37619836. IF:1,900 (Q4/8D). Document type: Article.
- Iznardo H, Bernal S, Boronat S, Roé E. Sclerotic Bone Lesions as a Clue in the Diagnosis of Three Generations of Tuberous Sclerosis Complex: Case Report and Review of Literature. PEDIATRIC NEUROLOGY. 2023; 148DOI:10.1016/j.pediatrneurol.2023.07.022. PMID:37634327. IF:3,800 (Q1/2D). Document type: Article.
- Perry MS, Scheffer IE, Sullivan J, Brunklaus A, Boronat S, Wheless JW, Laux L, Patel AD, Roberts CM, Dlugos D, Holder D, Knupp KG, Lallas M, Phillips S, Segal E, Smeyers P, Lal D, Wirrell E, Zuberi S, Brünger T, Wojnaroski M, Maru B, O'Donnell P, Morton M, James E, Vila MC, Huang NR, Gofshteyn JS, Rico S. Severe communication delays are independent of seizure burden and persist despite contemporary treatments in SCN1A+ Dravet syndrome: Insights from the ENVISION natural history study. EPILEPSIA. 2023; DOI:10.1111/epi.17850. PMID:38049202. IF:5,600 (Q1/2D). Document type: Article.
- Turón E, López I, Coca E, Badell I, Sierra A, Turón M, Ribosa R, Boronat S. Seizures in children undergoing stem cell transplantation. PEDIATRIC TRANSPLANTATION. 2023; :e14619. DOI:10.1111/petr.14619. PMID:37803946. IF:1,300 (Q4/9D). Document type: Article.