Epilepsy

Group leader

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Researchers

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Research technicians

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DESCRIPTION

This is a multidisciplinary group that includes 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 encompasses patients of all ages, including children and adults, to expand knowledge about the natural history of epilepsy. We offer also 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 (Boronat Guerrero, Susana; A Díaz Gómez, Maria Asunción; Ros Castello, Victoria).
- Electroclinical and neuroimaging phenotyping (Sierra Marcos, Alba).
- Neuropsychological comorbidities (Turon Viñas, Marc).
- Emergency management of acute seizures and status epilepticus (Turon Viñas, Eulàlia).

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 & AWARDED 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. 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 €
- Turon Viñas, Eulàlia. PReDICT: Ictus pediátrico asociado a enfermedades raras: Diagnóstico y tratamiento integrando Multi-ómica e inteligencia artificial. PMPER24/00021. Instituto de Salud Carlos III (ISCIII). Duration: 2025-2026. 39.764,37 €

SCIENTIFIC PRODUCTION

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- Esmel R, Miguel LDD, Artigas A, Turón E, Cuscó I, Díaz A, Panadés LPD, Rocamora R, Boronat S. Cardiovascular abnormalities in patients with SHANK3 pathogenic variants: Beyond neurodevelopmental disorders and epilepsy. European Journal of Medical Genetics. 2024; 71:104965. DOI:10.1016/j.ejmg.2024.104965. PMID:39094681. IF:1,600 (Q3/8D). Document type: Article.
- Giorgi S, Auvin S, Schoonjans AS, Turón E, Sánchez I, Gil A, Lagae L, Aibar JA. A tool for Dravet syndrome-associated neuropsychiatric comorbidities evaluation (DANCE). EPILEPSY & BEHAVIOR. 2024; 158:109958. DOI:10.1016/j.yebeh.2024.109958. PMID:39067307. IF:2,300 (Q2/5D). Document type: Article.
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