Genetic Diseases

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DESCRIPTION

The Genetic Diseases Group is a multidisciplinary group with an important clinical component in its research within the Genetics Department at the Hospital de Sant Pau. Its activities also have a translational orientation facilitated by the clinical environment in which it performs the laboratory work.

The primary objective of the Genetic Diseases Group is to advance the molecular diagnosis of genetic and rare diseases through genomic, transcriptomic, functional, and pharmacogenetic studies, thereby improving our understanding of the molecular mechanisms of described genes and identifying new ones. The results of these genetic studies allow for the establishment of different therapeutic options in many cases and to offer genetic counselling to the families. The fact that the group is made up entirely of Sant Pau Genetics Dept staff supports the clinical translational effect of their research work.

MAIN LINES OF RESEARCH

- Genetics of congenital myopathies, congenital and adult muscular dystrophies, including limb girdle muscle dystrophies, and congenital myasthenic syndromes.
- Spinal atrophy and SMN genes.
- Genetics of hereditary cancer.
- Pharmacogenetics.
- Cardiogenetics.
- Genetics in paediatrics-obstetrics.

SCIENTIFIC CHALLENGES

- Congenital myopathies, congenital and adult muscular dystrophies, including limb girdle muscle dystrophies, and congenital myasthenic syndromes.
  - Identification of mutations and genetic variants.
  - Studies of molecular pathology and disease mechanisms of neuromuscular disease-related genes.
  - Transcriptome analysis by RNA Seq technology.
  - Functional studies on the DMD genes to determine the pathogenic effect of the identified variants.
  - Long Read Sequencing by ONT technology to characterise long repeat expansions in size and composition.
- Spinal atrophy and SMN genes
  - To design new strategies for the study and improvement of the molecular diagnosis of atypical cases.
  - To perform functional studies for the characterisation of genetic variants of uncertain clinical effect to improve diagnosis.
  - Studies of molecular pathology, disease mechanisms, and SMN gene expression.
  - Identification of modifying genes.
  - Study of biomarkers for validation processing in spinal muscular atrophy.
  - Study of the neuromuscular junction in human development.
- Hereditary cancer
  - Development of Precision Medicine in hereditary syndromes of predisposition to cancer: an approach to the diagnosis, treatment and prevention of cancer through the identification of pathogenic variants in multigene assays.
  - Development of functional studies for the characterisation of genetic variants of uncertain clinical effects.
  - Analysis of biomarkers in liquid biopsy-free circulating tumor DNA (cfDNA) as a predictor of response to the treatment of breast cancer.
  - Contribution to the development of an open database of genetic variants included in Hereditary Cancer Genetic Testing Panels.
- Pharmacogenetics
  - Identification of biomarkers of response/toxicity to drugs used in cancer, rheumatology, inflammatory bowel diseases, cardiology, hematology.
  - Congenital coagulopathies: molecular pathology of hemophilia.
  - Duchenne and Becker muscular dystrophy: molecular pathology of the DMD gene.
  - Identification of the molecular alterations responsible for hereditary hearing loss.
  - Studies of the molecular pathology causing ataxias.
  - Clinical and Molecular diagnosis of complex diseases of pediatric-onset with special attention to Neurodevelopmental Disorders.
- Cardiogenetics
  - Clinical and Molecular studies of patients with cardiomyopathies (dilated, hypertrophic, arrhythmogenic, non-compacted, etc) to identify disease-associated genetic variants.
  - Study of clinically discordant families with the same pathogenic genetic variant to identify phenotype modifiers in cardiomyopathies.
  - Clinical and Molecular characterisation of patients with aortic diseases (Aortic Aneurysms, Aortic Valvular Disease, Aortic Occlusive Disease, Traumatic Aortic Disease, Aortic Dissections).
- Paediatrics-obstetrics
  - Clinical and Molecular studies of syndromic patients: Identification of responsible variants and discovery of new genes.
  - Clinical and Molecular studies of fetal anomalies detected during pregnancy.

ACTIVE & AWARDED GRANTS

- González Quereda, Lídia. Análisis integrado de datos genómicos y transcriptómicos para el avance en el conocimiento molecular y diagnóstico de las miopatías. PI22/01859. Instituto de Salud Carlos III (ISCIII). Duration: 2023-2025. 87.120,00 €
- González Quereda, Lídia. Application of novel sequencing technologies to solve the most complex dystrophinopathy cases. DUCHENNE ESPAÑA 2024. Asociacion Duchene Parent Project España. Duration: 2025-2026. 59.907,92 €
- Riera Armengol, Pau. Integración de la farmacogenética y la telemedicina mediante salud móvil para el manejo de los efectos adversos en pacientes con cáncer colorrectal. (ePGx study). PI21/01913. Instituto de Salud Carlos III (ISCIII). Duration: 2022-2024. 63.525,00 €
- Rodríguez Santiago, Benjamín. Catalan Interhospital web of genetic variants to improve genetic diagnosis in rare diseases. MARATO 202040-33. Fundació La Marató de TV3. Duration: 2021-2024. 26.625,00 €

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