Genetic Diseases

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Main Lines of Research

- Study of the clinical heterogeneity of genetic autosomal recessive transmission of waist dystrophy and autosomal dominant transmission.
- Spinal atrophy and SMN genes:
  - 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 breast cancer and BRCA mutations:
  - Identification of mutations and genetic variants.
  - Molecular characterization of circulating tumor cells (CTCSS) through expression profiles in patients with breast cancer.
  - Analysis of free circulating tumor DNA (cfDNA) as a predictor of response to the treatment of breast cancer.
- Pharmacogenetics: adverse reactions to medications.
- Congenital coagulopathies: molecular pathology of haemophilias.
- Duchenne and Becker muscular dystrophy: molecular pathology of DMD gene.

Challenges

Pharmacogenetics

- Colorectal and lung cancer treatments.
- Chronic inflammatory disease treatment.
- HIV infection treatment.

Muscular Dystrophies

- Post-transcriptional regulation of the dystrophin gene using nonsense-mediated decay analysis in DNA from patients with Duchenne muscular dystrophy.
- Genes associated with new phenotypic forms of limb girdle muscular dystrophy.
- Workflow development for the analysis of DMD gene by NGS techniques.

Hereditary Breast/Ovarian Cancer

- DNA studies to classify DNA variants found in the BRCAs genes as pathogenic or neutral.
- CTCs as prognostic markers in patients with locally advanced and disseminated breast cancer.
- Workflow development for the analysis of BRCA1 and BRCA2 genes by NGS techniques.

Hereditary Motor Neurone Diseases

- Development of neuromuscular junction studies in health and disease.
- Broadening of the spectrum of motor neuron diseases to include bulbar and spinal muscular atrophy, distal muscular atrophies and amyotrophic lateral sclerosis.

Awards

- Ana Sebio García. Young Investigator Award. Gastrointestinal Group of the European Organisation for Research and Treatement of Cancer.

Grants Awarded in 2017

- Maria Pia Gallano Petit. Implementació de la medicina personalitzada basada en la genòmica en malalties minoritàries neurològiques no diagnosticades. SLT002/16/00174, Departament de Salut. Duration: 2017-2019. 12,000.00 €.

Note: Total amount granted to PI. It does not include indirect costs.

Active Grants

- Ana Sebio García. Contratos Juan Rodés 2014. JR14/00006. Instituto de Salud Carlos III. Duration: 2015-2018. 135,000.00 €.
- Lidia González Quereda. Implementación de la secuenciación masiva en el estudio de las Miopatías Congénitas y los Síndromes Miasténicos congénitos: un modelo de investigación traslacional en enfermedades raras. FMM 2015. Fundación Mutua Madrileña. Duration: 2015-2018. 74,200.00 €.
- Maria Pia Gallano Petit. Implementación de la secuenciación masiva en el estudio de Miopatías Congénitas y Síndromes Miasténicos congénitos: un modelo de investigación traslacional en enfermedades raras. PI15/01898. Instituto de Salud Carlos III. Duration: 2016-2018. 56,500.00 €.
- Pau Riera Armengol. Marcadors farmacogenètics de resposta/resistència a les teràpies biològiques (anti-EGFR) en càncer colorectal. 2016 FI_B 00368. Agència de Gestió d'Ajuts Universitaris i de Recerca. Duration: 2016-2019. 40,806.00 €.
- David Paez López-Bravo. Estudio farmacogenético fase II randomizado para evaluar eficacia y seguridad del esquema FOLFIRI con altas dosis de irinotecán (FOLFIRIAD) en pacientes con cáncer colorrectal metastásico de acuerdo con el genotipo UGT1A 1. EC11-336. Ministerio de Sanidad y Política Social. Duration: 2015-2017. 13,965.97 €.

Note: Total amount granted to PI. It does not include indirect costs.

***TIF: 52.877 **MIF: 5.287**

Scientific Production

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Paez D., Salazar R., Tabernero J., DPYD genotype-guided fluoropyrimidines dose: Is it ready for prime time? (2017) ANN ONCOL, 28 (12), 2913-2914.
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Sullivan I., Salazar J., Arqueros C., Andres M., Sebio A., Majem M., Szafranska J., Martinez E., Paez D., Lopez-Pousa A., Baiget M., Barnadas A., KRAS genetic variant as a prognostic factor for recurrence in resectable non-small cell lung cancer (2017) CLIN TRANSL ONCOL, 19 (7), 884-890.
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Vidal S., Brandi N., Pacheco P., Gerotina E., Blasco L., Trotta J.-R., Derdak S., Del Mar O'Callaghan M., Garcia-Cazorla A., *et al*. The utility of Next Generation Sequencing for molecular diagnostics in Rett syndrome (2017) SCI REP-UK, 7 (1).
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*Total Impact Factor **Mean Impact Factor