Neuromuscular Diseases

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DESCRIPTION

The Neuromuscular Diseases Unit is a multidisciplinary unit with extensive experience in the diagnosis, treatment and research of neuromuscular diseases. It is a team in which clinical and research activities are perfectly overlapped and in which clinical problems are addressed in the laboratory to find new biomarkers and diagnostic tools, new pathological mechanisms of disease and new therapeutic targets. The Unit is composed of neurologists, biologists and laboratory technicians who are all experts in basic and translational research and in the organisation and management of sample collections.

MAIN LINES OF RESEARCH

- Autoimmune neuromuscular diseases (myasthenia, neuropathies, and myopathies). (Querol Gutiérrez, Luis Antonio; Cortes Vicente, Elena; Gallardo Vigo, Eduard).
  - Characterisation of new target antigens in Myasthenia Gravis (MG), and immune neuropathies (CIDP, GBS, MMN). Their use as diagnostic and therapeutic biomarkers.
  - Nerve damage biomarkers.
  - Pathogenesis of newly recognised antigens both in MG and in CIDP.
  - Collaborators of the IGOS database for Guillain Barre syndrome.
  - Coordinators of the INCBase (international database for CIDP).

    - GENRARE Spanish registry for neuromuscular diseases.
- Muscular dystrophies. (Olive Plana, Montserrat).
    - Muscle MRI analysis as a biomarker of different muscular dystrophies.
    - Natural history of Dysferlinopathies. The international COS-study Jain Foundation.
    - Development of cell models of NMD diseases to study pathogenic mutations.
    - Clinical, pathological and molecular characterisation of rare myopathies.
- Amyotrophic lateral sclerosis. (Rojas García, Ricardo).
    - Gene profile of ALS patients in Spain.
    - Biomarkers profile in different phenotypes of ALS.

SCIENTIFIC CHALLENGES

- Advances in the knowledge of the immunological mechanisms involved in the pathogenesis of autoimmune neuromuscular diseases (MG, CIDP, MMN).
- Evaluate the use of NFL and myelin proteins as biomarkers of nerve damage in inflammatory neuropathies.
- To develop mouse models to demonstrate the pathogenic effect of antibodies to new antigens in MG.
- Advances in the knowledge of the pathogenetic mechanisms of inflammatory myopathies (DM, necrotising myopathies).
- Search for new antigens and develop diagnostic tests using both known and novel biomarkers in immune-mediated neuropathies, myasthenia gravis, and inflammatory myopathies.
- Advances in the knowledge of the pathogenetic mechanisms involved in muscular dystrophies.
- Search for new genes and biomarkers in the different phenotypes of ALS.
- Advances in the knowledge of the pathogenetic mechanisms involved in ALS.
- Use of the GENRARE registry to perform research on different NMDs.
- Search for new genes that cause myopathy.
- Characterisation of new muscle disorders.

ACTIVE & AWARDED GRANTS

- Cortes Vicente, Elena. Validación de una plataforma tecnológica basada en la reprogramación del sistema inmunológico en humanos; leading asset en Miastenia Gravis. CPP2023-010523. Ministerio de Ciencia, Innovación y Universidades. Duration: 2024-2027. 906.021,71 €
- Gallardo Vigo, Eduard & Díaz Manera, Jorge Alberto. Aproximaciones terapéuticas en distrofias musculares mediante modelos celulares y animales. GEMIO 2019. Fundacion Isabel Gemio para la investigación de distrofias musculares y otras enfermedades raras. Duration: 2019-2024. 150.000,02 €
- Gallardo Vigo, Eduard & Cortes Vicente, Elena. Estudio exhaustivo de la miastenia "seronegativa": de la clínica al descubrimiento de nuevos autoantígenos y su validación in vitro e in vivo. PI22/01786. Instituto de Salud Carlos III (ISCIII). Duration: 2023-2025. 135.520,00€
- Gallardo Vigo, Eduard. Valorización de una terapia antígeno-específica para el tratamiento disruptivo de la diabetes tipo 1; esclerosis múltiple, miastenia gravis, enfermedad celíaca y artritis reumatoide. CPP2021-008475. Ministerio de Ciencia e Innovacion (MICINN). Duration: 2023-2025. 138.873,88 €
- Gallardo Vigo, Eduard. Characterization and optimization of myasthenia gravis care. OptiMyG. Competitiu. EJPR23-104 European Joint Programme of Rare Diseases. AC23_2/00030 EJP-RD ISCIII. 2024-2026. 174.918,00 €
- Olive Plana, Montserrat. Abordaje multidisciplinar para el diagnóstico y caracterización de enfermedades musculares raras. PI21/01621. Instituto de Salud Carlos III (ISCIII). Duration: 2022-2024. 123.420,00 €

- Pascual Goñi, Elba. Biomarkers in the diagnosis and follow-up of CIDP. GBS-CDIP-ELBA. GBS CDIP Foundation International. Duration: 2022-2025. 337.563,00 €
- Pascual Goñi, Elba. Caracterización inmunológica de las neuropatías de fibra fina idiopáticas. PI24/00336. Instituto de Salud Carlos III (ISCIII). Duration: 2025-2027. 77.500,00€
- Querol Gutiérrez, Luis Antonio. Medicina de precisión en neuropatías autoinmunes. PI22/00387. Instituto de Salud Carlos III (ISCIII). Duration: 2023-2025. 135.520,00 €
- Querol Gutiérrez, Luis Antonio. Red Española de Neuropatías Autoinmunes. PMPER24/00018. Instituto de Salud Carlos III (ISCIII). Duration: 2025-2026. 1.634.710,00 €
- Rojas García, Ricardo. Validación de biomarcadores candidatos para el diagnóstico y la progresión de la Esclerosis Lateral Amiotrófica y estudio de las vías fisiopatogénicas implicadas. PI19/01543. Instituto de Salud Carlos III (ISCIII). Duration: 2020-2024. 123.420,00 €
- Rojas García, Ricardo. Caracterización del perfil transcriptómico en el espectro completo de Esclerosis Lateral Amiotrófica-Demencia frontotemporal (ELADFT) para la investigación y validación de biomarcadores en biofluidos. PI23/00845. Instituto de Salud Carlos III (ISCIII). Duration: 2024-2026. 127.500,00€

SCIENTIFIC PRODUCTION

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- Bolano C, Verdú J, González A, Fitzsimmons S, Veeranki G, Straub V, Díaz J. Magnetic resonance imaging-based criteria to differentiate dysferlinopathy from other genetic muscle diseases. NEUROMUSCULAR DISORDERS. 2024; 34DOI:10.1016/j.nmd.2023.11.004. PMID:38007344. IF:2,700 (Q2/4D). Document type: Article.
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