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 activity 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 the organization and management of sample collections.

MAIN LINES OF RESEARCH

- Autoimmune neuromuscular diseases (myasthenia, neuropathies, and myopathies).
  - Characterization 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 recognized antigens both in MG and in CIDP.
  - Collaborators of the IGOS database for Guillain Barre syndrome.
  - Coordinators of the INCBase (international database for CIDP).
  - NMD-ES Spanish registry for neuromuscular diseases.
- Muscular dystrophies.

- 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.
- Pathogenic mechanisms in inflammatory myopathies (DM, necrotizing myopathies).
- Clinical, pathological, and molecular characterization of rare myopathies.

- Amyotrophic lateral sclerosis.
  - Gene profile of ALS patients in Spain.
  - Biomarkers profile in different phenotypes of ALS.

SCIENTIFIC CHALLENGES

- Advance 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 2D and 3D cell-based assays to understand the effect of autoantibodies in Myasthenia gravis.
- To develop mouse models to demonstrate the pathogenic effect of antibodies to new antigens in MG.
- Advance in the knowledge of the pathogenetic mechanisms of inflammatory myopathies (DM, necrotizing myopathies).
- Search for new antigens and develop diagnostic tests with known and new biomarkers in immune-mediated neuropathies, myasthenia gravis, and inflammatory myopathies.
- Implement new diagnostic and disease follow-up methods for muscular dystrophies resulting from dysferlinopathy and other myopathies research (biomarkers (miRNA, proteome), MRI, etc).
- Advance in the knowledge of the pathogenetic mechanisms involved in muscular dystrophies.
- Search for new genes and biomarkers in the different phenotypes of ALS.
- Advance in the knowledge of the pathogenetic mechanisms involved in ALS.
- Use of the NMD.ES registry to perform research in different NMD.
- Search for new myopathy causative genes.
- Characterization of new muscle disorders.

ACTIVE GRANTS

- Gallardo Vigo, Eduard & Diaz 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 & Cortés 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 €.
- E.Gallardo. Validació de l'eficàcia dels PS-Liposomes-haAChR-EP per el tractament de la Miastènia Gravis: estudi de generació de tolerancia antígen específica en un model murí de Miastènia Gravis. Ahead Therapeutics. Non-competitive. Duration: 2020- 2024. 50.000 €.
- 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. IMAGiNe study. IMAGiNe. Foundation for Peripheral Neuropathy . Duration: 2022-2023. 12.654,00 €.

- Querol Gutierrez, Luis Antonio. Precision medicine in chronic inflammatory neuropathy assessment. GBS/CIDP 2020. GBS CDIP Foundation International. Duration: 2020-2023. 50.245,70 €.
- Querol Gutierrez, Luis Antonio. Nuevos marcadores serológicos en enfermedades neuroinmunes raras. ER22PA2C762. CIBERER. Duration: 2023-2024. 45.000,00 € (CIBER).
- Querol Gutierrez, Luis Antonio. Medicina de precisión en neuropatías autoinmunes. PI22/00387. Instituto de Salud Carlos III (ISCIII). Duration: 2023-2025. 135.520,00 €.
- 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 €.

GRANTS AWARDED

- 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 €.
- Roja 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€.

DOCTORAL THESES DEFENDED

- Alonso Pérez, Jorge. Caracterización clínica, genética y de las vías de degeneración muscular de la distrofia muscular por déficit de sarcoglicano. 22/05/2023. Universitat Autònoma de Barcelona. Supervisors: Díaz Manera, Jorge Alberto; Suárez Calvet, Xavier; Querol Gutiérrez, Luis Antonio. http://hdl.handle.net/10803/690666
- Álvarez Velasco, Rodrigo. Detección de marcadores clínicos e inmunológicos para diagnóstico y pronóstico en miastenia gravis. 21/09/2023. Universitat Autònoma de Barcelona. Supervisors: Gallardo Vigo, Eduard; Querol Gutiérrez, Luis Antonio.

SCIENTIFIC PRODUCTION

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