Neuromuscular Diseases

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Main Lines of Research

Autoimmune neuromuscular diseases

- Characterization of new target antigens in Myasthenia Gravis (MG), and immune neuropathies (CIDP, GBS, MMN). Their use as diagnostic and therapeutic biomarkers.
- Pathogenesis of newly recognized antigens both in MG and in CIDP.
- Analysis of the innate immunity in inflammatory myopathies.
- Analysis of the impact of new immunomodulating therapies on IgG4 mediated Neuromuscular Diseases (NMD).
- Functional aspects of immune system cells (response to ligands, production of antibodies, etc).
- Phase 2 clinical trials in autoimmune neuromuscular diseases.
- IGOS data base for Guillain Barre syndrome.
- NMD-ES Spanish registry for neuromuscular diseases.

Muscular dystrophy, dysferlinopathy and distal myopathies

- Muscle MRI analysis as a biomarker of different muscular dystrophies.
- Role of PDGF in muscular dystrophies fibrosis. Study of nintedanib as an antifibrotic drug in muscular dystrophies.
- Natural history of Dysferlinopathies. The international COS-study Jain Foundation.
- Study of proteasome inhibitors in dysferlin myopathy.
- Clinical trials in muscular dystrophies and metabolic muscle diseases.

Amyotrophic lateral sclerosis

- Epidemiology of ALS in Catalonia.
- Gene profile of ALS patients in Spain.
- Biomarkers profile in different phenotypes of ALS.

Challenges

- Advance in the knowledge of the immunological mechanisms involved in the pathogenesis of autoimmune neuromuscular diseases (MG, CIDP, MMN).
- Advance in the knowledge of the pathogenetic mechanisms of inflammatory myopathies (DM).
- Search for new antigens and develop diagnostic tests with new biomarkers in immune-mediated neuropathies and Myasthenia Gravis.
- Implement new diagnostic and disease follow-up methods for muscular dystrophies resulting from dysferlinopathy and other myopathies research (biomarkers (miRNA, secretome), MRI, etc).
- Advance in the knowledge of the pathogenetic mechanisms involved in dysferlin deficiency and other muscular dystrophies.
- Search for new genes and biomarkers in the different phenotypes of ALS.
- Use of the NMD.ES registry to perform research in different NMD.

Collaborations

Collaborations with other IIB Sant Pau Groups

- Genetic Diseases
- Inflammatory Diseases

External Collaborations

- Jerome Devaux, investigador CNRS (Institut de Neurosciences de Montpellier, U1051R1). France.
- Silvere van der Maarel, Leiden, The Netherlands.
- Xavier Navarro. Departament de Medicina, Institut de Neurociències. Universitat Autònoma de Barcelona, Spain.
- R. Martí, Elena Garcia Arumí. Vall d'Hebron Research Institute, Barcelona, Spain. CIBERER CB 06/07/015.
- Vilchez JJ. Hospital La Fe Valencia. CIBERER CB 06/05/ 091.
- Al Chalabi A. King's College, London, UK.
- Benveniste, O. Hopital Salpetriere, Paris, France.
- Nagaraju, K/ Jaiswal, J. Children's Hospital, Washington D.C. USA.
- Mouly, V. Institut de Myologie, Paris, France.
- Pinal I. National Institute of Health, Bethesda, MD, USA.

Active Grants

- Maria Isabel Illa Sendra. Nuevas reactividades antigénicas y estudios de inmunidad innata en enfermedades neuromusculares autoinmunes. PI13/00937. Instituto de Salud Carlos III. Duration: 2014-2017. 160,250.00 €.
- Eduard Gallardo Vigo. Recerca en Malalties Neuromusculars. 2014 SGR 272. Agència de Gestió d'Ajuts Universitaris i de Recerca. Duration: 2014-2017. 30,000.00 €.
- Ricardo Rojas García. Biomarkers profile in different phenotypes of Motor Neuron Disease. MARATO 20143710. Fundació La Marató de TV3. Duration: 2015-2018. 120,500.00 €.
- Jorge Alberto Díaz Manera. PDGF como nuevo biomarcador y diana terapéutica en pacientes con distrofia muscular. PI15/01822. Instituto de Salud Carlos III. Duration: 2016-2018. 76,500.00 €.
- Eduard Gallardo Vigo. Influencia de factores inmunológicos y no inmunológicos en la patogenia de las miopatías inflamatorias PI15/01597. Instituto de Salud Carlos III. Duration: 2016-2018. 91,500.00 €.
- Ricardo Rojas Garcia. Identificación de biomarcadores diagnósticos en pacientes con Esclerosis Lateral Amiotrófica . PI15/01618. Instituto de Salud Carlos III. Duration: 2016-2018. 91,500.00 €.
- Luis Antonio Querol Gutiérrez. Identificación de autoanticuerpos en neuropatías inflamatorias y caracterización del fenotipo celular implicado en su desarrollo. PI16/00627. Instituto de Salud Carlos III. Duration: 2017-2019. 76,500.00 €.
- Maria Isabel Illa Sendra. Estudios moleculares y celulares en Miastenia Gravis: Correlaciones clínicas y terapéuticas PI16/01440. Instituto de Salud Carlos III. Duration: 2017-2019. 111,500.00 €.
- Elena Cortes Vicente. Ajuts per a contractes de formació "Río Hortega" 2016. CM16/00096. Instituto de Salud Carlos III. Duration: 2017-2018. 53,732.00 €.
- Maria Isabel Illa Sendra. Aproximaciones terápeuticas en distrofias musculares mediante modelos celulares y animales. GEMIO 2016. Fundació Isabel Gemio. Duration: 2016-2019. 160,908.99 €.
- Eduard Gallardo Vigo. El proteasoma com a diana terapèutica en distròfies musculars per dèficit de disferlina. IR16-P4. Fundació Privada Hospital de la Santa Creu i Sant Pau. Duration: 2017-2017. 7,000.00 €.
- Noemi de Luna Salva. Activación de la inmunidad innata en células deficientes en disferlina: nuevas dianas terapéuticas F. RAMÓN ARECES 2016. Fundació Ramón Areces. Duration: 2017-2019.
- Xavier Suarez Calvet. New therapeutic targets in dermatomyositis. Research Grant Application 2014. The Myositis Association. Duration: 2015-2017. 83,586.64 €.

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

Grants Awarded in 2017

- Elena Cortés Vicente. Antigen-specific regulatory networks to treat Myasthenia Gravis. MV17/00012. Instituto de Salud Carlos III. Duration: 2018. 11,960.00 €.

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

Awards

- Extraordinary PhD award to Alba Ramos Franci for the thesis Nuevos marcadores diagnósticos y factores pronósticos en miastenia Gravis.
- Award to the best presentation of a young researcher on the treatments in the muscular dystrophy of Duchenne. Patricia Piñol. Nintedanib as a new therapeutic agent for Duchenne Muscular Dystrophy: preclinical and in vivo studies. 22nd World Muscle Society Meeting, St Malo, France.
- Elsevier WMS Membership Award. Alicia Alonso. Magnetic Resonance Image in Oculopharyngeal muscle dystrophy. 22nd World Muscle Society Meeting, St Malo, France.

Patents

- RH Brown, MF Ho, I Illa, E Gallardo. Blood-Based Assay for Dysferlinopathies. US7172858B2. USA, Canada, Japan, 06/02/2007

Neuromuscular Diseases

Books or chapters with ISBN

Eduardo Gutiérrez-Rivas (ed). Manual de Enfermedades Neuromusculares. 2017. ISBN: 978-84-16732-35-2

- Chapter 17. Estudios inmunológicos. E Gallardo, X. Suárez, I. Illa. page 137
- Chapter 34. Neuropatías disinmunes. L. Querol, R. Rojas. page 275
- Chapter 45. Distrofinopatías. J Diaz, E. Gallardo. page 375
- Chapter 67. Panorama de la Investigación sobre las enfermedades neuromusculares en España. R. Torrón, I. Illa, et al. page 581

Medicina Interna Farreras Rozman (18th edition) ISBN 978-84-9022-996-5

- Chapter 174. Ataxias y paraparesias espásticas. Enfermedades de la neurona motora. E. Muñoz, R. Rojas Garcia. page 1409.
- Chapter 182. Enfermedades de los nervios periféricos. J. Casademont y R. Rojas Garcia. page 1470.
- Chapter 184. Enfermedades de la unión neuromuscular: Miastenia Gravis y síndromes miasténicos. JM Grau y I. Illa. page 1496.

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Scientific Production

Aragones J.M., Altimiras J., Roura P., Alonso F., Bufill E., Munmany A., Alfonso S., Illa I., Prevalence of myasthenia gravis in the Catalan county of Osona Prevalencia de miastenia gravis en la comarca de Osona (Barcelona, Cataluña) (2017) NEUROLOGIA, 32 (1), 1-5.
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Cortes-Vicente E., Pradas J., Marin-Iahoz J., De Luna N., Clarimon J., Turon-Sans J., Gelpi E., Diaz-Manera J., Illa I., Rojas-Garcia R., Early diagnosis of amyotrophic lateral sclerosis mimic syndromes: pros and cons of current clinical diagnostic criteria (2017) AMYOTROPH LAT SCL FR, 18 (5-6), 333-340.
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De Luna N., Suarez-Calvet X., Lleixa C., Diaz-Manera J., Olive M., Illa I., Gallardo E., Hypoxia triggers IFN-I production in muscle: Implications in dermatomyositis (2017) SCI REP-UK, 7 (1).
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Delmont E., Manso C., Querol L., Cortese A., Berardinelli A., Lozza A., Belghazi M., Malissart P., Labauge P., Taieb G., Yuki N., Illa I., Attarian S., Devaux J.J., Autoantibodies to nodal isoforms of neurofascin in chronic inflammatory demyelinating polyneuropathy (2017) BRAIN, 140 (7), 1851-1858.
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Garibaldi M., Tasca G., Diaz-Manera J., Ottaviani P., Laschena F., Pantoli D., Gerevini S., Fiorillo C., Maggi L., Tasca E., D'Amico A., Musumeci O., Toscano A., Bruno C., Massa R., Angelini C., Bertini E., Antonini G., Pennisi E.M., Muscle MRI in neutral lipid storage disease (NLSD) (2017) J NEUROL, 264 (7), 1334-1342.
IF: 3.783

Howard J.F., Utsugisawa K., Benatar M., Murai H., Barohn R.J., Illa I., Jacob S., Vissing J., Burns T.M., Kissel J.T., et al., Safety and efficacy of eculizumab in anti-acetylcholine receptor antibody-positive refractory generalised myasthenia gravis (REGAIN): a phase 3, randomised, double-blind, placebo-controlled, multicentre study (2017) LANCET NEUROL, 16 (12), 976-986.
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Illa I., ARTHUR ASBURY LECTURE: Chronic inflammatory demyelinating polyradiculoneuropathy: clinical aspects and new animal models of auto-immunity to nodal components (2017) J PERIPHER NERV SYST, 22 (4), 418-424.
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Martinez-Martinez L., Lleixa M.C., Boera-Carnicero G., Cortese A., Devaux J., Siles A., Rajabally Y., Martinez-Pineiro A., Carvajal A., Pardo J., Delmont E., Attarian S., Diaz-Manera J., Callegari I., Marchioni E., Franciotta D., Benedetti L., Lauria G., de la Calle Martin O., Juarez C., Illa I., Querol L., Anti-NF155 chronic inflammatory demyelinating polyradiculoneuropathy strongly associates to HLA-DRB15 (2017) J NEUROINFLAMM, 14 (1).
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Morenas-Rodriguez E., Camps-Renom P., Perez-Cordon A., Horta-Barba A., Simon-Talero M., Cortes-Vicente E., Guisado-Alonso D., Vilaplana E., Garcia-Sanchez C., Gironell A., Roig C., Delgado-Mederos R., Marti-Fabregas J., Visual hallucinations in patients with acute stroke: a prospective exploratory study (2017) EUR J NEUROL, 24 (5), 734-740.
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Pinol-Jurado P., Gallardo E., de Luna N., Suarez-Calvet X., Sanchez-Riera C., Fernandez-Simon E., Gomis C., Illa I., Diaz-Manera J., Platelet-Derived Growth Factor BB Influences Muscle Regeneration in Duchenne Muscle Dystrophy (2017) AM J PATHOL, 187 (8), 1814-1827.
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*Total Impact Factor **Mean Impact Factor