Genetics in Neurodegenerative Diseases

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Main Lines of Research

- Use of state-of the-art genomic strategies to study the genetic architecture of complex diseases caused by neurodegenerative processes, including Alzheimer disease and other dementias, Parkinson disease, and amyotrophic lateral sclerosis (ALS), among others.
- Cloning and evaluation of novel genes associated with neurodegenerative disorders through genomic analyses on families with Mendelian pattern of inheritance.
- Study of the relationship between biomarkers and endophenotypes at the individual genetic background.
- Evaluation of the effect of novel genes related to neurodegenerative disorders in the Spanish population through multicentre collaborative studies.

Challenges

- Assemble a research team of excellence in the international field of genetics of neurodegenerative disorders.
- Increase our capacity of funding through EU support.
- Develop a comprehensive genomic database from well-characterized Spanish patients of European origin suffering from neurodegenerative disorders.

Collaborations

Collaborations with other IIB Sant Pau Groups

- Neurobiology of Dementia.
- Parkinson Disease and Movement Disorders.
- Neuromuscular Diseases.

External Collaborations

National and international consortiums

- European Early-Onset Dementia Consortium (EU-EOD)
- European Alzheimer Disease Initiative (EADI)
- International Genomics of Alzheimer Project (IGAP)
- International Parkinson's Disease Genetic Consortium (IPDGC)
- International Frontotemporal Dementia Genomics Consortium (IFGC)
- Dementia Genetics Spanish Consortium (DEGESCO)
- CIBER-Neurodegenerative Diseases (CIBERNED)

Collaborations with international centres

- Dr. John Hardy, University College of London, UK.
- Dr. Andrew Singleton, National Institutes of Health, USA.
- Dr. Jean-Charles Lambert, Directeur de recherche Inserm chez INSERM. Lille, France.
- Dr. Alfredo Ramírez, University of Bonn. Germany.
- Dr. Ekaterina Rogaeva, University of Toronto, Canada.
- Dr. Mikko Hiltunen, Kuopio University, Finland.
- Dr. Martin Ingelsson, Uppsala University, Sweden.
- Dr. Liana Fidani, Aristotle University of Thessaloniki, Greece.
- Dr. Christine Van Broekhoven, University of Antwerp, Belgium.

Grants Awarded in 2017

- Oriol Dols Icardo. Estudi del paper de les variants genètiques rares en l'arquitectura genètica de la malaltia d'Alzheimer. SLT002/16/00040. Departament de Salut. Duration: 2017-2019. 88.376,57 €.
- Jordi Clarimon Echavarria, Estudio del splicing alternativo en ARN de muestras con diagnóstico neuropatológico de ELA asociada a TDP-43. FUNDELA 2017. Fundació Espanyola per al Foment de la Investigació de l'Esclerosi Lateral Amiotròfica. Duration: 2017-2018. 26.087,00 €.

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

Active Grants

- Jordi Clarimon Echavarria. Estudio del perfil de expresión de microRNA exosomal en biofluídos para la identificación de biomarcadores de uso diagnóstico en la demencia frontotemporal. PI15/00026. Instituto de Salud Carlos III. Duration: 2016-2018. 101,500.00 €.
- Jordi Clarimon Echavarria. Unitat genètica malalties neurodegeneratives. IR16-P7. Fundació Privada Hospital de la Santa Creu i Sant Pau. Duration: 2017-2017. 7,000.00 €.
- Jordi Clarimon Echavarria. A comprehensive genomic analyis of patients with motor neuron disease and frontotemporal dementia to disentangle the missing genetic architecture of amyotrophic lateral sclerosis TODOS SOMOS RAROS 2014. Federación Española de Enfermedades Raras / Fundación Isabel Gemio / Federación Española de Enfermedades Neuromusculares. Duration: 2015-2017. 90,748.00 €.
- Jordi Clarimon Echavarria. Grup de Recerca en Demècies. 2014 SGR 235. Agència de Gestió d'Ajuts Universitaris i de Recerca. Duration: 2014-2017. 18,000.00 €.
- Jordi Clarimon Echavarria. Contratos Miguel Servet 2013 (II). MSII13/00005. Instituto de Salud Carlos III. Duration: 2014-2017. 101,250.00 €.
- Jordi Clarimon Echavarria. Grup de Recerca en Demències: Sant Pau. 2017 SGR 547. Agència de Gestió d'Ajuts Universitaris i de Recerca. Duration: 2017-2019. 35,200.00 €.

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

*TIF: 122.507 **MIF: 7.6566

Scientific Production

Alcolea D., Vilaplana E., Suarez-Calvet M., Illan-Gala I., Blesa R., Clarimon J., Llado A., Sanchez-Valle R., Molinuevo J.L., Garcia-Ribas G., Compta Y., Marti M.J., Pinol-Ripoll G., Amer-Ferrer G., Noguera A., Garcia-Martin A., Fortea J., Lleo A., CSF sAPPβ, YKL-40, and neurofilament light in frontotemporal lobar degeneration (2017) NEUROLOGY, 89 (2), 178-188. **IF: 7.180**

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Colom-Cadena M., Grau-Rivera O., Planellas L., Cerquera C., Morenas E., Helgueta S., Munoz L., Kulisevsky J., Marti M.J., Tolosa E., Clarimon J., Lleo A., Gelpi E., Regional overlap of pathologies in lewy body disorders (2017) J NEUROPATH EXP NEUR, 76 (3), 216-224. **IF: 3.271**

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*Total Impact Factor **Mean Impact Factor