

## **GENE THERAPY FOR NEUROMETABOLIC DISEASES**

### **ASSUMPCIÓ BOSCH**



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#### **PROFILE**

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## RESEARCH

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### RESEARCH INTERESTS/ STRATEGIC OBJECTIVES

The research activity of our group is focused on the development of gene therapy strategies for rare diseases affecting the nervous system, both central (lysosomal storage diseases, ALS, Wolfram Syndrome and MLC) and peripheral (genetic and acquired neuropathies) and on the elucidation of the molecular mechanisms implicated in the development of these pathologies combining the use of animal models, tissue cell culture and viral vectors.

### MAIN RESEARCH LINES

1. Study of the tropism of different gene therapy viral vectors in the central and peripheral nervous system. Combination of different serotypes of AAV vectors with specific cell promoters and route of administration to target defined structures or cell types.
2. Gene therapy in preclinical models for rare disease affecting the CNS: lysosomal storage diseases, amyotrophic lateral sclerosis, megalencephalic leukoencephalopathy with subcortical cysts (MLC), or Wolfram Syndrome.
3. Gene therapy in preclinical models for diseases affecting the peripheral nervous system: diabetic neuropathy and adrenomyeloneuropathy (AMN).

### LAB FEATURED PUBLICATIONS

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Leal-Julià M., Vilches J., Onieva A., Verdés S., Sánchez A., Chillón M., Navarro X., **Bosch A.** Proteomic quantitative study of dorsal root ganglia and sciatic nerve in type 2 diabetic mice. Molecular Metabolism, (2022) 55:101408.  
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Roig-Soriano J, Griñán-Ferré C, Espinosa-Parrilla JF, Abraham C, **Bosch A**, Pallàs M, Chillón M. AAV-mediated expression of secreted and transmembrane aKlotho isoforms rescue relevant aging hallmarks in senescent SAMP8 mice. *Aging Cell* (2022), 00:e13581. doi: 10.1111/acel.13581.

Mòdol-Caballero G, García-Lareu B, Herrando-Grabulosa M, Verdés S, López-Vales R, Pagès G, Chillón M, Navarro X, **Bosch A**. Specific expression of Glial-Derived Neurotrophic Factor in muscles as gene therapy strategy for Amyotrophic Lateral Sclerosis. *Neurotherapeutics* (2021); 18(2):1113–1126, doi.10.1007/s13311-021-01025-6.

Sanchez, A, García-Lareu B, Puig M, Prat E, Ruberte J, Chillón M, Nunes V, Estevez R, **Bosch A**. Cerebellar Astrocyte Transduction as Gene Therapy for Megalencephalic Leukoencephalopathy. *Neurotherapeutics* (2020); 17(4):2041-2053. doi:[10.1007/s13311-020-00865-y](https://doi.org/10.1007/s13311-020-00865-y). PMID: 32372403

Mòdol-Caballero G, Herrando-Grabulosa M, García-Lareu B, Solanes N, Verdés S, Osta R, Francos-Quijorna I, López-Vales R, Calvo AC, **Bosch A\***, Navarro X\*. Gene therapy for overexpressing Neuregulin 1 type I in skeletal muscles promotes functional improvement in the SOD1G93A ALS mice. *Neurobiology of Disease* (2020); 37:104793. doi.org/[10.1016/j.nbd.2020.104793](https://doi.org/10.1016/j.nbd.2020.104793). PMID: 32032731.

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Mòdol-Caballero G, García-Lareu B, Verdés S, Ariza L, Sánchez-Brualla I, Brocard F, **Bosch A**, Navarro X, Herrando-Grabulosa M. Therapeutic role of neuregulin 1 type III in SOD-linked amyotrophic lateral sclerosis. *Neurotherapeutics* (2020) 17, 1048-1060. doi:[10.1007/s13311-019-008811-7](https://doi.org/10.1007/s13311-019-008811-7). PMID: 31965551

Pagès G., Giménez-Llort L., García-Lareu B., Ariza L., Navarro M., Casas C., Chillón M., **Bosch A**. Intrathecal AAVrh10 corrects biochemical and histological hallmarks of mucopolysaccharidosis VII mice and improves behavior and survival. *Human Molecular Genetics* (2019); 28, 3610–3624. doi:[10.1093/hmg/ddz220](https://doi.org/10.1093/hmg/ddz220). PMID: 31511867

Massó A, Sánchez-Osuna A, **Bosch A**, Giménez-Llort L, Chillón M. Secreted-Klotho isoform administered into CNS protects against age-dependent memory deficits in middle-aged and aged animals. *Molecular Psychiatry* (2018); 3(9):1-11. doi: 10.1038/mp.2017.211. PMID: 29086766