

Título del Proyecto	Translational research, experimental medicine and therapeutics on Charcot-Marie-Tooth disease
Nº de expediente asignado	IR11/TREAT-CMT
Abstract	<p>Charcot-Marie-Tooth disease (CMT), characterised by a motor and sensory neuropathy, is the most frequent hereditary neuromuscular disorder with an estimated prevalence of 17-40/100,000, but it is still considered a rare disease. All mendelian inheritance patterns can occur in CMT, which shows a high degree of genetic heterogeneity. So far more than 40 genes have been described to be associated with CMT and related neuropathies; however, a number of CMT-causing genes remain unknown. In the last 20 years CMT research has rapidly moved from clinical description towards molecular medicine based on molecular genetics, cell biology and pathophysiology. Thus far, however, there is no effective drug or advanced therapy available for CMT. To move forward translation from research into clinical medicine of CMT we propose a Spanish CMT Consortium, TREAT-CMT, based on a multidisciplinary collaboration of twelve clinical and basic research groups, with a comprehensive and integrative approach. The aims and activities of the network are focused on three clinical and scientific goals: (1) natural history research, indepth phenotyping and clinical research tools, (2) translational genetic and genomic studies to discover new CMT genes and searching for biomarkers, and (3) pathophysiology and therapeutics of mitochondrialrelated CMT neuropathies. Based on preliminary data preclinical assays in mouse models will test modulation of calcium metabolism as a proof-of-concept of axonal neuropathies treatment. We have put emphasis on efficient coordination and management structure. In addition, a cross-cutting action will integrate scientific developments with societal and ethical issues, along with their dissemination to the community at large. We expect that TREAT-CMT becomes a strong tool to foster translational research of peripheral neuropathies in Spain and improve the quality of life of patients, with additional connection and collaboration with other European and international networking initiatives in the field of neuromuscular disorders, especially those focused on peripheral neuropathy.</p>
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