Spanish registry of patients with McArdle disease

Description

The Spanish registry of patients with McArdle disease was created with the purpose of analysing their genotypic and phenotypic characteristics.

In Spain, it is feasible to gather data on all Spanish McArdle patients due to the diagnostic protocol followed by the National Health System. When McArdle disease is suspected (typically based on hyper-CK-aemia and exercise intolerance), patient's blood samples together with medical history data (and biopsy specimens, when available) are sent to three 'reference' public hospitals (Hospital 12 de Octubre, Madrid; Hospital Val d'Hebron, Barcelona; and Hospital Meixoeiro, Vigo) for biochemical and molecular genetic analysis. The neuromuscular genetics laboratories of these hospitals have been responsible for confirming the disease from the time molecular genetics tools were available for PYGM genotyping in our countrydthat is, starting in 1998 for Hospital 12 de Octubre, in 1999 for Hospital Meixoeiro and in 2001 for Hospital Val d'Hebron. Furthermore, since 2005, Spanish McArdle patients have been offered the chance to perform aerobic capacity tests in the Universidad Europea de Madrid (Madrid).