Neurología. 2017;xxx:xxx—xxx

SOCIEDAD ESPAÑOLA DE NEUROLOGIA

NEUROLOGÍA

www.elsevier.es/neurologia

REVIEW ARTICLE

Report by the Spanish Foundation for the Brain on the social impact of amyotrophic lateral sclerosis and other neuromuscular disorders☆

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Received 7 January 2015; accepted 7 February 2015

KEYWORDS
Dependence;
Epidemiology;
Neuromuscular diseases;
Amyotrophic lateral sclerosis;
Spain;
Economic impact;
Mortality

Abstract
Introduction: A thorough knowledge of the socioeconomic scope of neuromuscular disease is essential for managing resources and raising social awareness.
Development: Our group reviewed current data on the epidemiology, mortality and dependence rates, and socioeconomic impact of amyotrophic lateral sclerosis and neuromuscular diseases in Spain. We also recorded how neurological care for these patients is organised.
Conclusions: Neuromuscular disorders are a very heterogeneous group of diseases, and some are very rare. These disorders account for between 2.8% and 18% of the total motives for a neurological consultation. In Spain, prevalence and incidence figures for amyotrophic lateral sclerosis are similar to those in other countries; however, figures for patients with other neuromuscular diseases are not known. Since the diseases are chronic, progressive, and debilitating, they cause considerable disability and dependence, which in turn directly affects healthcare and social costs associated with the disease. The costs generated by one patient with amyotrophic lateral sclerosis or Duchenne disease have been calculated at about 50 000 euros per year. Neuromuscular disease shows aetiological, diagnostic, and prognostic complexity, and it requires multidisciplinary management. Follow-up for these patients should be entrusted to specialised units.

☆ Please cite this article as: Camacho A, Esteban J, Paradas C. Informe de la Fundación Del Cerebro sobre el impacto social de la esclerosis lateral amiotrófica y las enfermedades neuromusculares. Neurología. 2017. https://doi.org/10.1016/j.nrl.2015.02.003

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PALABRAS CLAVE
Dependencia;
Epidemiología;
Enfermedades neuromusculares;
Esclerosis lateral amiotrófica;
España;
Impacto económico;
Mortalidad

Informe de la Fundación Del Cerebro sobre el impacto social de la esclerosis lateral amiotrófica y las enfermedades neuromusculares

Resumen
Introducción: El conocimiento del alcance socioeconómico de la patología neuromuscular es esencial para la planificación de recursos y la concienciación social.
Desarrollo: Se ha realizado una revisión de los datos publicados hasta el momento sobre epidemiología, mortalidad, dependencia e impacto sociosanitario de la esclerosis lateral amiotrófica y las enfermedades neuromusculares en España. Además, se ha recogido cómo está organizada la atención neurológica en estos pacientes.
Conclusiones: La patología neuromuscular constituye un grupo muy heterogéneo de enfermedades, algunas de las cuales se consideran raras por su baja frecuencia. Esta patología supone entre el 2,8 y el 18% de los motivos de consulta en un Servicio de Neurología. En España, las cifras de prevalencia e incidencia de esclerosis lateral amiotrófica son similares a otros países; sin embargo, se desconoce el número de pacientes con otras enfermedades neuromusculares. Son enfermedades crónicas, progresivas y debilitantes, lo que condiciona una importante discapacidad y dependencia. Esto repercute directamente en los costes sanitarios y sociales asociados a la enfermedad. Se ha calculado que el coste de un paciente con esclerosis lateral amiotrófica o enfermedad de Duchenne se acerca a los 50.000 euros anuales. La patología neuromuscular tiene una gran complejidad etiológica, diagnóstica y pronóstica, y requiere un manejo multidisciplinar. Las Unidades especializadas deben ser las encargadas del seguimiento de estos pacientes.

Introduction

The purpose of the Foundation for the Brain is to raise awareness of the impact of neurological diseases on patients and their families, friends, and carers, with the ultimate goal of improving patients' and relatives' quality of life. To this end, the Foundation for the Brain has developed a series of initiatives in the fields of information, prevention, research, patient care, and social integration. As part of these activities, the Foundation for the Brain prepares reports on the societal impact of different neurological diseases in Spain.

This report focuses on neuromuscular disorders (NMD), a large, complex group of disorders that merit separate analysis in view of their individual characteristics. More specifically, this report addresses amyotrophic lateral sclerosis (ALS) and other relevant non-ALS neuromuscular disorders.

Neuromuscular disorders affect the peripheral nervous system, skeletal muscle, neuromuscular junctions, and spinal cord. This heterogeneous group includes a wide range of entities (Table 1), classified by lesion location: motor neuron diseases, radiculopathies, plexopathies, neuropathies, neuromuscular junction diseases, and muscle disorders. These conditions may be metabolic, infectious, toxic, immune-mediated, genetic, or neurodegenerative in origin. They may manifest during childhood or adulthood and may be acute, subacute, or chronic. NMDs require highly specialised diagnosis and treatment; the study and management of these disorders therefore constitutes a subspecialty within general neurology.

ALS, including its variants (primary lateral sclerosis, progressive muscular atrophy, and progressive bulbar palsy), is the most frequent motor neuron disease in adults. This disabling, progressive neurodegenerative condition affects both the upper (located in the precentral motor cortex) and the lower motor neurons (located in the motor nuclei of the brainstem and the anterior horn of the spinal cord). Although it is typically sporadic (an isolated case in a family), 5% to 10% of cases are clustered in families, most frequently following an autosomal dominant inheritance pattern (however, autosomal recessive and X-linked dominant inheritance patterns have also been reported). Around 5% to 10% of cases also exhibit dementia, normally frontotemporal dementia, which may appear before, after, or simultaneously with ALS onset. However, neuropsychological tests have found evidence of executive dysfunction in over 50% of patients with ALS.

ALS is a progressive, invariably fatal disease. Patients typically die within 5 years of onset as a consequence of progressive restrictive respiratory failure. There is still no curative treatment for ALS; current treatment approaches focus on providing palliative care to control symptoms from disease onset.

Though diverse, non-ALS NMDs share certain characteristic features with similar consequences, which explains why they are grouped together. The most common clinical manifestation of these disorders is loss of strength, which is usually progressive. Muscle weakness may cause a number of orthopaedic disorders, including rigidity and joint deformities, which are especially pronounced in paediatric patients. Other frequent motor symptoms include fatigue,