The Epidemiology of Neuromuscular Diseases

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KEYWORDS

• Neuromuscular disease • Epidemiology • Global health • Burden of disease
• Public health

KEY POINTS

- The goal of this article is to examine current understanding of the epidemiology and burden of neuromuscular diseases in different global regions, focusing on the neuromuscular diseases listed in Box 1, divided by anatomic origin.
- Neuromuscular diseases are a relatively rare heterogeneous group of disorders that affect the function of various components of the peripheral nervous system.
- Rapid advances in genome sequencing have elucidated the pathogenetic mechanisms of several neuromuscular diseases and permitted recognition of therapeutic targets for interventional clinical trials.
- Understanding epidemiologic trends across global regions can aid governments, nongovernmental organizations, and international institutions formulate policies focused on prevention and surveillance.

Video content accompanies this article at http://www.neurologic.theclinics.com.

ANTERIOR HORN

Spinal Muscular Atrophy

Spinal muscular atrophy (SMA) is a genetic disease involving degeneration of anterior horn cells in the spinal cord and motor neurons in the nuclei of the lower brainstem (**Box 1**).[1] The causative genetic mutation is usually inherited in an autosomal-recessive (AR) pattern, disrupting production of the survival motor neuron protein.[2] Although first described clinically and pathologically in infants with motor paralysis,[3] SMA disorders have a heterogeneous age of onset and clinical course, which provide the basis for its classification. SMA type I is the most common and severe form, affecting infants at birth or after a few months, resulting in severe weakness and ultimately death from respiratory failure by age 2. SMA type II is an intermediate form with onset of symptoms between 6

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Box 1
Neuroanatomical classification of neuromuscular diseases

Anterior horn

- Spinal muscular atrophy (SMA)
- Amyotrophic lateral sclerosis (ALS)
- Post polio syndrome (PPS)

Nerve

- Guillain-Barre syndrome (GBS)
- Chronic inflammatory demyelinating polyneuropathy (CIDP)

Neuromuscular junction

- Myasthenia gravis (MG)

Muscle

- Duchenne muscular dystrophy (DMD)
- Becker muscular dystrophy (BMD)
- Polymyositis (PM)
- Dermatomyositis (DM)
- Inclusion body myositis (IBM)

and 18 months of age; patients can sit independently but rarely can ambulate. With appropriate respiratory care, patients may survive into the third or fourth decade. SMA type III is a mild form with variable onset between late adolescence and childhood with ability to ambulate into the fifth decade and may live a life expectancy seen in a normal population.[4] SMA type IV is the rarest form of the disorder with onset in adulthood and slower rates of disability progression. All presentations of SMA have decreased or absent reflexes, weakness of proximal muscles, and progressive respiratory decompensation as shared clinical features.[5,6] Select global incidence and prevalence rates of SMA are summarized in **Table 1**.

Type I has the highest incidence of the SMA types, but because of early mortality, type II and type III are more prevalent.[16] The highest incidence and prevalence rates of SMA in the studies examined were reported by Burd and colleagues[8] in North Dakota, which included only type I patients. The investigators included 14 patients from retrospective review of birth and death certificates and a coded diagnosis; medical records were not found for 6 patients. Possibility of misclassification and heterogeneity may account for the high estimates. Ludvigsson and coworkers[11] studied the incidence of all SMA types from a national registry in Iceland during a 15-year period, with a combined incidence of 13.7 per 100,000 live births, which did not differ significantly from other studied populations.

SMA prevalence rates from UK,[7] Swedish,[9] Irish,[10] and English[13] populations show similar rates across Western Europe. MacMillan and Harper[7] retrospectively searched hospital inpatient records, genetic outpatient records, and electrodiagnostic testing records for their districts in South Wales and noted rates were likely underestimated because of ascertainment methods used to maximize diagnostic accuracy as well as possible variation in gene frequency between populations.[7] In contrast, Ludvigsson and colleagues[11] examined several neuromuscular diseases and used district and health insurance registries, communications with