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Adult-onset spinal muscular atrophy: An update.

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Abstract

Spinal muscular atrophy (SMA) refers to a group of disorders affecting lower motor neurons. The age of onset of these disorders is variable, ranging from the neonatal period to adulthood. Over the last few years, there has been enormous progress in the description of new genes and phenotypes that throw new light on the molecular pathways involved in motor neuron degeneration. Advances in our understanding of the pathophysiology of the most frequent forms, SMA linked to SMN1 gene mutations and Kennedy disease, has led to the development of therapeutic strategies currently being tested in clinical trials. This report provides a general overview of the clinical features and pathophysiological mechanisms in adult-onset genetic SMA disorders in which the causative gene has been identified (SMN1-related SMA, Kennedy disease, CHCHD10, TRPV4, DYNC1H1 and BICD2). Sporadic lower motor neuron disease, also known as progressive muscular atrophy (PMA), is also discussed. The finding of TDP-43 aggregates in immunohistochemical studies of PMA strongly supports the idea that it is a phenotypic variant of amyotrophic lateral sclerosis (ALS).

KEYWORDS: Kennedy disease; Lower motor neuron syndrome; Progressive muscular atrophy; SMN1; Spinal muscular atrophy

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