

Development of an interest in spinal muscular atrophy and disabilities

As First Vice-President of the American Academy of Cerebral Palsy and Developmental Medicine (AAPDM), I have the privilege of writing the editorial for the September issue of *Developmental Medicine & Child Neurology (DMCN)* which coincides with the annual meeting of the Academy. My interest in spinal muscular atrophy (SMA) and children with disabilities was spurred by one patient, whom I met early in my career at Newington Children's Hospital, Connecticut, USA. Although *DMCN* has not published many articles on the subject of SMA in the past 10 years (8 of 1389 articles), I would like to take this opportunity to discuss my experience of this condition. This autosomal recessive disease, of which our knowledge has changed greatly over the past 30 years, is the second most common recessive disease in infancy and childhood.

My patient was 16 years of age in 1971. He taught me that SMA is a progressive disease. The literature at that time suggested that if there were no change in strength or function for 2 years then the condition would remain static.¹ At 18 months of age, he was diagnosed as having mental retardation*, as the cause of his motor and language delay. Not until age 5 was the correct diagnosis established, namely SMA. He was told that his condition would not worsen. At age 8, he could no longer ambulate with his braces, the exclamation being that he was 'too overweight'; a manual wheelchair was prescribed. At age 14 he needed a power wheelchair, as he could no longer push the wheelchair. By age 24, he had difficulty lifting his arms above his head. His story led us to review the charts of the SMA patients at Newington Children's Hospital. Subsequently, we published an article in 1983 noting that spinal muscular atrophy was a slowly but definitely progressive disease.² Others confirmed this in the mid-90s.³

Dramatic changes in knowledge about SMA occurred in the 1990s. The gene for this condition was identified in 1990⁴ and cloned in 1995⁵ leading to the development of a DNA test. If the diagnosis of SMA is suspected on the basis of the history and physical examination, only a diagnostic DNA test is necessary to confirm the diagnosis. Since the DNA test has been developed, it has now come to light that many other causes of muscle weakness secondary to anterior horn cell degeneration occur that are not linked to the gene on chromosome 5.⁶

The present classification is based on the clinical picture. Patients with SMA type I never sit independently; their lifespan is approximately 2 years of age. Patients with SMA type II never walk but may live into their 40s. Patients with SMA type III walk at least 5 steps independently; their lifespan is 'normal'.⁷ The phenotype-genotype correlation is emerging.⁸ A mouse model for SMA has been developed.⁹ In fact, there has been a recent suggestion that riluzole¹⁰ might be effective in this condition.

The cure for SMA may or may not be close at hand. Nevertheless, as my patient constantly reminded me: 'Sure, I want to see a cure, but help me live a better life.' We should strive to understand what we can do to improve function and quality of life within the limitations imposed by the disability. As Peter Rosenbaum wrote in his editorial when he was First Vice-President of the AAPDM, we must be concerned about 'activity, and functional adaptation, and participation in the life of family and the community, and the environment.'¹¹ I do not have to remind readers that treatment does not necessarily mean cure. Helping patients live a better life has provided me with a great deal of satisfaction and gives reason to go to work each morning with a smile and new sense of excitement and challenge.

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*UK usage: learning disability.