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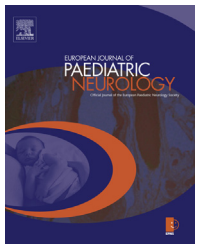

Original Article

Pain in adolescents with spinal muscular atrophy and Duchenne and Becker muscular dystrophy

Christina Lager [a,*], *Anna-Karin Kroksmark* [b]

[a] *Habilitation Center Malmö, Entrance 122, Skane University Hospital Malmö, SE-205 02 Malmö, Sweden*
[b] *University of Gothenburg, Institute of Neuroscience and Physiology, Occupational Therapy/Physiotherapy, Box 455, SE-405 30 Gothenburg, Sweden*

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ABSTRACT

Background/purpose: The purpose of this study was to explore the prevalence, nature and scope of pain in adolescents with spinal muscular atrophy and Duchenne and Becker muscular dystrophy and whether the pain differs between diagnostic groups or between adolescents with different ambulation status. Furthermore to study the consequences of pain and to identify pain-exacerbating and pain-relieving factors.

Methods: In a national survey, fifty-five adolescents with spinal muscular atrophy and dystrophinopathy completed a questionnaire assessing pain frequency, duration, location using a body map, intensity and discomfort using visual analogue scales, pain interference using a modified version of Brief Pain Inventory and factors exacerbating and relieving pain.

Results: Sixty-nine per cent of the adolescents reported pain during the past three months and 50% reported chronic pain. The pain prevalence did not differ significantly between diagnostic groups or between ambulators and non-ambulators. The average pain intensity was graded as mild and the worst pain as moderate. The pain typically occurred weekly, most frequently in the neck/back or legs. General activity and mood were the areas that were most affected by pain. Common pain-exacerbating factors were sitting, too much movement/activity and being lifted or transferred.

Conclusion: Pain is a frequent problem in adolescents with spinal muscular atrophy and dystrophinopathy. The assessments used enable an understanding both of the nature and scope of pain and of the impact of pain in everyday life. The study highlights the importance of assessing pain in a systematic manner and offering an individual approach to interventions designed to reduce pain in this population.

* *Corresponding author*. Tel.: +46 406730836; fax: +46 406730890.
E-mail addresses: christina.lager@skane.se (C. Lager), anna-karin.kroksmark@vgregion.se (A.-K. Kroksmark).
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1. Introduction

Pain has previously not been regarded as a prominent symptom in neuromuscular disorders (NMD). However, over the past decade, a growing number of studies have shown that chronic pain is a frequent symptom in many different types of NMD, but with great heterogeneity regarding prevalence.[1]

Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy (BMD) are X-linked recessive disorders caused by mutations in the dystrophin gene, leading to the total absence of or an abnormal but partly functional dystrophin protein. The lack of dystrophin results in progressive muscle weakness and the degeneration of muscle fibres, which are replaced by fat and connective tissue. DMD and BMD both belong to the group of dystrophinopathies.[2]

DMD is the most common NMD in children, with an incidence of one in 3500–4700 male births.[3–5] Without treatment, the boys lose ambulation before the age of 13. As permanent wheelchair users, most of the boys develop joint contractures and scoliosis. Cardiac and respiratory complications emerge, often necessitating mechanical ventilation. Boys with DMD have a shortened life expectancy. Improved multidisciplinary care in recent decades, with long-term corticosteroid treatment, respiratory care, spinal surgery and the treatment of cardiomyopathy, has resulted in prolonged life expectancy and improvements in function.[6,7] BMD is the milder phenotype of dystrophinopathy, with a later onset and slower progression of the disorder. The incidence is one in 18,500 male births.[8] In both DMD and BMD, a non-progressive cognitive dysfunction may be present.[6,9]

Spinal muscular atrophy (SMA) is an autosomal recessive NMD caused by a mutation of the survival motor neuron (SMN1) gene located on chromosome 5, resulting in muscular atrophy and weakness. SMA is one of the most common NMDs in children, with an incidence of one in 10,000–11,800 live births.[10–12] SMA is divided into subtypes based on the age of onset and the maximum motor function achieved. SMA I manifests before six months of age and most children die before two years of age, due to bulbar dysfunction and pulmonary complications. SMA II manifests before 18 months of age and the children achieve sitting function but are unable to stand or walk. In SMA III, the onset of weakness takes place after 18 months of age and these children achieve the ability to walk independently.[13,14] The disorder is slowly progressive.[15,16] Scoliosis and joint contractures often develop, especially in the weaker children and adolescents who are permanent wheelchair users. Recurrent respiratory infections and reduced pulmonary function are other common features, often necessitating respiratory care and mechanical ventilation.[14]

The causes of pain in neuromuscular disorders are multifactorial and could, for example, be due to severe contractures, osteoporoses and vertebral fractures, orthopaedic procedures or muscular overuse – all secondary consequences of severely decreased muscle strength and limited mobility or secondary to treatment.[7,14,17] Studies in adults with NMD have reported pain prevalence of more than 60%.[18–21] No study in adults has focused separately on SMA, DMD or BMD. In a study of adults with different NMDs, Tiffreau et al.[21] state that the DMD and BMD population reported the highest frequency of chronic pain.

In children and adolescents with NMD, the nature and scope of pain has not been well studied. Two studies have examined the prevalence of pain in a group of children and adolescents with different types of NMD.[22,23] Engel et al.[22] studied pain prevalence in a mixed group (age 8–20 years) with different types of NMD. Fifty-five per cent reported chronic pain and 83% reported that they used pain medication. A study of pain prevalence in boys (age 8–18 years) with DMD and BMD showed that 54% of boys with DMD and 80% of boys with BMD reported that they experienced pain at least once a week. The study also showed that pain was underrecognised by the treating physicians.[23]

The knowledge of pain in children and adolescents with NMD is limited, in terms of pain frequency, intensity, location and discomfort, as well as its impact on function and quality of life. This knowledge is important both for treatment options and for the understanding of the impact of pain on patient adherence and the outcomes of other rehabilitation interventions.

The purpose of the present study was to explore the prevalence, nature and scope of pain in adolescents with SMA, DMD and BMD and to determine whether the pain differs between diagnostic groups or between adolescents with different ambulation status. The purpose was also to study the consequences of pain in everyday life and to identify the factors that exacerbate and relieve pain.

2. Materials and methods

2.1. *Participants*

All known adolescents in Sweden with SMA, DMD and BMD who met the inclusion criteria were asked to participate in the study. The criteria included: (1) a confirmed diagnosis of SMA, DMD or BMD; (2) age span 12–18 years of age; (3) no significant cognitive impairment and attending normal school programmes and (4) speaking and reading the Swedish language.

The adolescents were recruited through a national Swedish network for NMD, which is a group of professionals from all over the country working with patients with different types of NMD. Physiotherapists in the network were asked to identify all eligible adolescents with SMA, DMD and BMD and their regular physiotherapists in their county.

The study was approved by the Regional Ethical Review Board in Gothenburg, Sweden, and informed consent to participate in the study was obtained from the adolescents and their parents or legal guardians.

2.2. *Measurements*

Demographic and clinical data, including gender, age and diagnosis, were collected. The adolescents were asked whether they had experienced any pain in the past three months other than occasional pain, such as headache or toothache. Those who answered "yes" to the latter question were asked to respond to the rest of the questionnaire.