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ABSTRACT

Introduction: Nemaline myopathy, described in 1950, is a rare syndrome, and among the possible alterations, scoliosis is a frequent alteration. This article describes the case of a 13-year-old patient with nemaline myopathy and scoliosis treated surgically. During the procedure, several unexpected surgical difficulties were encountered, such as fusion of posterior elements, difficulty in passing screws and liquor leaked out. Therefore, more studies are necessary and the presence of an experimental operation to help with surgical complications are prudent.

Keywords: scoliosis, nemaline myopathy, neuromuscular scoliosis.

RESUMO

Introdução: A miopatia nemalina, descrita em 1950, é uma síndrome rara e, entre as possíveis alterações, a escoliose é uma alteração frequente. Este artigo descreve o caso de um paciente de 13 anos com miopatia nemalina e escoliose tratada cirurgicamente. Durante o procedimento, várias dificuldades cirúrgicas inesperadas foram encontradas, como fusão de elementos posteriores, dificuldade em passar parafusos e vazamento de licor. Portanto, mais estudos são necessários e a presença de uma operação experimental para ajudar com complicações cirúrgicas é prudente.

Palavras-chave: escoliose, miopatia nemalina, escoliose neuromuscular.

1 INTRODUCTION

Nemaline myopathy (NM) was first identified in the 1950s in muscle biopsies from children with hypotonia, in microscopic vision were identified rod like or ovoid structures in muscle fibers. NM are a heterogenous group of congenital myopathies caused by mutation in the genes encoding skeletal muscle α -actin. Clinically, most patients presents with muscle weakness and hypotonia, but the spectrum of phenotypes is heterogeneous, presenting from severe neonatal presentations to onset of in childhood.(1)

Monique M et al in a retrospective study with 143 patients, 33 cases of scoliosis in patients with NM were identified. In another classic retrospective study, Waugren-Pettersson

followed up 13 patients with NM and the muscular weakness was constantly found, the flexors of the neck and trunk were one of the most affected groups. In the study 8 of the 13 patients developed scoliosis. The prognosis seems to be influenced not only by the intrinsic grade of disease activity, but also by whether or not severe scoliosis develops. (2)

2 MATERIAL AND METHODS

We report a case of a 13-year-old patient who was admitted on our service with neuromuscular scoliosis secondary to NM. Family members report the onset of the deformity in childhood, with worsening in recent years. Due to the underlying pathology, the patient was gastrostomized, tracheostomized, unable to walk, but was able to stand alone with the support of upper limbs.

On the panoramic radiograph, two curves were identified, the largest being thoracolumbar with a Cobb of 65 from T9 to L3 and a second proximal thoracic curve from T1 to T5 with 45 degrees. The thoracic segment between the curves, from T5 to T8 was rectified. Therefore, this curve is not classified by the Lenke classification

Research was made on PubMed and Google academic database for review studies and case reports about Nemaline myopathy and scoliosis.

3 RESULTS

Patient underwent surgery with T2-L4 arthrodesis. During the procedure, difficulties were encountered in identifying the anatomical structures, as multiple fusions of posterior elements were found. Secondary to this difficulty, during the passage of two screws, liquor leaked out. Furthermore, during the passage of the screw from T2 in the left, there was a drop in potential in the left lower limbs, with partial recovery at the end of the procedure.

4 CONCLUSION

Nemaline myopathy is a rare disease with lack of study. Little specific literature was found on the treatment of scoliosis in these patients. Despite the lack of literature, the prognosis seems to be influenced by the scoliosis and the restrictive lung disease secondary the deformity. The treatment is multidisciplinary with focus on maintenance of as much muscle power and function as possible, with emphasis on the treatment of scoliosis.

When opting for surgical treatment, care must be taken with the other repercussions of muscle weakness, such as respiratory disease. During the procedure, the surgeon must be prepared for multiple difficulties, secondary to deformity in the anatomy. Therefore, we

recommend that the surgical team include an experienced surgeon due to the high chances of intraoperative complications.

REFERENCES

- (1) Nemaline myopathies: a current view Caroline A. Sewry^{1,2} · Jenni M. Laitila^{3,4} · Carina Wallgren-Pettersson^{3,4}
- (2) Congenital nemaline myopathy A clinical follow-up study of twelve patients Carina WaUgren-Pettersson Children's Hospital, University of Helsinki, SF-00290 Helsinla" (Finland)
- (3) Nemaline Myopathy: A Clinical Study of 143 Cases Monique M. Ryan, MB, BS,
- (4) A Pediatric Case of Neuromuscular Scoliosis Caused by Nemaline Myopathy Furkan Selim Usta