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Case Report

A Pediatric Case of Neuromuscular Scoliosis Caused by Nemaline Myopathy

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Abstract

Nemaline myopathy is one of the most common congenital myopathies known of at this time. Weakness of the paraspinal muscle can lead to spinal deformities such as scoliosis. We present a case of neuromuscular scoliosis caused by nemaline myopathy. As we were unable to identify any case requiring surgery for scoliosis caused by nemaline myopathy in the existing literature, we believe our case is the first report of such.

Keywords: *Neuromuscular Scoliosis; Nemaline Myopathy; Scoliosis Surgery*

Introduction

Nemaline myopathy is one of the most common congenital myopathies and is characterized by hypotonia, weakness, and an absence of deep tendon reflexes. The condition can be either autosomal dominant or recessive and may be either severe, moderate, or mild in nature. Severely affected patients may demonstrate weakness of the respiratory muscles and respiratory failure. Moderate disease causes progressive weakness in the muscles of the face, neck, trunk, and feet, but these individuals' life expectancy may be nearly normal. In contrast, mild disease is nonprogressive and associated life expectancy is normal. Causative mutations have been identified in 10 genes and all are related to the production of thin-filament proteins [1].

The defining feature of nemaline myopathy is the presence of fuchsinophilic proteins observed as sarcoplasmic rod-shaped structures (nemaline bodies) in muscle biopsy [2-5].

Patients who suffer from nemaline myopathy generally have weak paraspinal muscles. In effect, support for their spine weakens such that their spine may curve over time. Significant weakness of the paraspinal muscles could lead to more notable spinal deformities such as scoliosis. Scoliosis is one of the most common problems in neuromuscular diseases [6-8].

Aim of the Study

The aim of the present study was to evaluate an orthopedic case of neuromuscular scoliosis caused by nemaline myopathy.

Case Presentation

A six-year-old girl presented to our orthopedic clinic due to neuromuscular scoliosis. She had a degree of spinal curvature (72°) due to muscle weakness caused by nemaline myopathy, which is a congenital, genetic disorder (Figure 1). Initial radiograph was taken in sitting position.

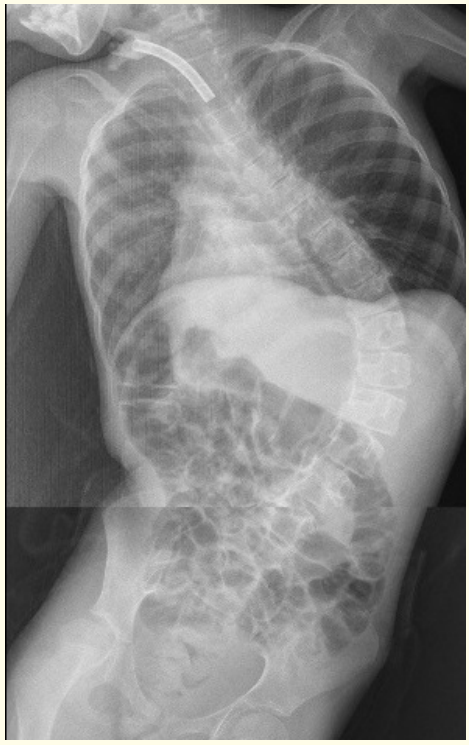

Figure 1

Throughout her early life, no problem had been observed. She was a normal baby during the perinatal period. However, a few months later, muscle problems began to be noticed by her parents. Her parents expressed concern specifically about her not having head control. Her sickness was eventually diagnosed by a geneticist and she was subsequently seen by a physiotherapist for five years to strengthen her muscles.

When she was four years of age, she developed pneumonia and her respiratory muscles became weak. Subsequently, she had to undergo a tracheostomy and use a home ventilator.

When the patient was six years of age, her parents noticed her spinal curvature. She was brought to our orthopedic clinic for a consultation with a pediatrician.

Upon examination, it was found that she had coronal and sagittal imbalance. There were also disruptions of sitting balance. The purpose of the surgery was to improve her lung capacity and to help sitting without support.

The T2-iliac sliding growing rod technique, which is a growth-friendly technique, was performed [9] (Figure 2). She stayed in the intensive care unit for one week and then underwent rehabilitation for one month. There were no complications that presented postoperatively. The Cobb's angle was measured to be 25. The post-surgical Cobb's angle measured in sitting position. After surgery, she could sit without support and walk with support. During the follow-up period, It was observed that the sliding growing rod technique was working well, as the patient could walk with support and her sitting balance had improved (Figure 3). The follow up period was six months. At follow up we have observed that pneumonia caused by lung compression occur post-surgically.

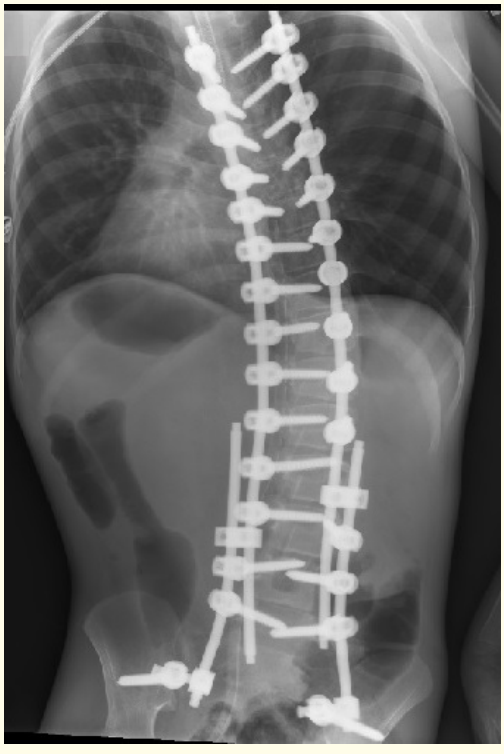

Figure 2

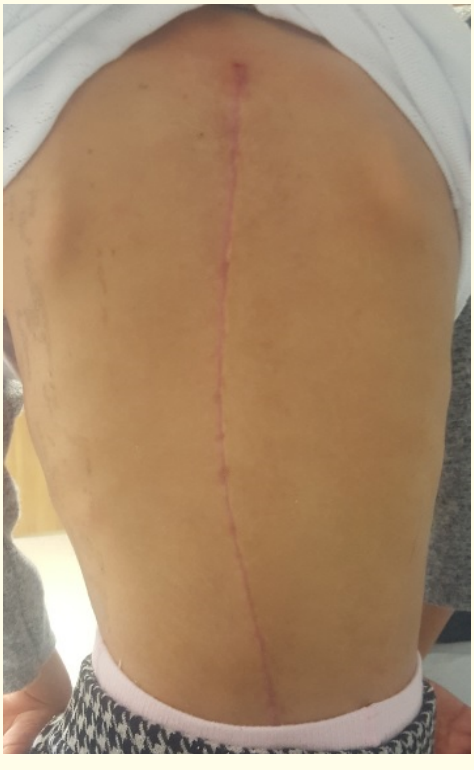

Figure 3

Discussion

We reported a case of scoliosis caused by nemaline myopathy and some complications related to spinal curvature in a six-year-old female patient.

Nemaline myopathy is commonly associated with dysfunction of the skeletal muscles. Depending on the progression and subtype of the condition, different muscle groups can be affected. Those patients whose respiratory muscles are affected often become ventilator-dependent [10,11]. Our case was also ventilator-dependent and underwent a tracheostomy.

In a prior study, other patients with nemaline myopathy reported the onset of sudden cardiac arrest [12]. However, cardiac involvement in this patient population rarely occurs, and our case did not experience cardiac problems.

Patients who have neuromuscular disease commonly suffer from scoliosis. Neuromuscular scoliosis differs from idiopathic scoliosis in several ways including further progression [13] and the use of conservative and surgical treatment [6]. In addition, there is a higher rate of postoperative complications [13].

Spinal deformities are not commonly caused by muscle weakness. Indeed, trunk muscle hypertonia causes scoliosis more than paralysis [6]. However, in our case, scoliosis was related to paraspinal muscle weakness.

Lung function could be compromised by progressive scoliosis, which produces lung compression [14].

In a study by Mukherjee*., et al.* considering 122 children, 46% of patients with pleural infections showed thoracic scoliosis on admission, while 71% of them showed such during subsequent stages of the illness [15]. Our case experienced pneumonia caused by lung compression.

In the postoperative period of scoliosis surgery, complications can be seen. In a study involving 236 cases, the rate of major medical complications was 7.2%, while that of wound complications was 1.7% [16].

Scoliosis surgery may be associated with a high risk of morbidity and mortality in children with non-idiopathic scoliosis [17,18].

In a study by Daniel., *et al.* It was reported that there were 29.019 cases of neuromuscular scoliosis with 1,385 complications from 2004 to 2015 [18].

Some complications can be fatal, such as superior mesenteric artery syndrome or acute pulmonary embolism, while others could simply decrease quality of life (e.g., neurological complications).

In our case, there were no postoperative complications.

Conclusion

To the best of our knowledge, our report is the first case evaluating the outcomes of surgery for scoliosis caused by nemaline myopathy. Neuromuscular scoliosis surgery is a very demanding condition.

Bibliography

1. North KN and Ryan MM. "Nemaline Myopathy". In: Adam MP, Ardinger HH, Pagon RA., *et al*. editors. Seattle (WA): University of Washington, Seattle 1993-2017.

2. Moreno CAM., *et al*. "Clinical and Histologic Findings in ACTA1-Related Nemaline Myopathy: Case Series and Review of the Literature". *Pediatric Neurology* 75 (2017): 11-16.

3. E Malfatti and NB Romero. "Nemaline myopathies: State of the art". *Revue Neurologique* 172.10 (2016): 614-619.

4. Wallgren-Pettersson C., *et al*. "Nemaline Myopathies". *Seminars in Pediatric Neurology* 18.4 (2011): 230-238.

5. Ennis J., *et al*. "Congenital Nemaline Myopathy: The Value of Magnetic Resonance Imaging of Muscle". *Canadian Journal of Neurological Sciences* 42.5 (2015): 338-340.

6. Vialle R., *et al*. "Neuromuscular scoliosis". *Orthopaedics and Traumatology: Surgery and Research* 99.1 (2013): 124-129.

7. Halawi MJ., *et al*. "Neuromuscular Scoliosis: Current Concepts". *Orthopedics* 38.6 (2015): e452-e456.

8. Miladi L., *et al*. "Minimally Invasive Surgery for Neuromuscular Scoliosis: Results and Complications in a Series of One Hundred Patients". *Spine (Phila Pa 1976)* 43.16 (2018): E968-E975.

9. Enercan M., *et al*. "Sliding-Growing Rod Technique for Management of Early-Onset Scoliosis". *Spine Deformity* 2.6 (2014): 499.

10. de Winter JM and Ottenheijm CAC. "Sarcomere Dysfunction in Nemaline Myopathy". *Journal of Neuromuscular Diseases* 4.2 (2017): 99-113.

11. North KN., *et al*. "Nemaline Myopathy: current concepts". *Journal of Medical Genetics* 34.9 (1997): 705-713.

12. Marseglia L., *et al*. "Sudden cardiac arrest in a child with nemaline myopathy". *Italian Journal of Pediatrics* 41 (2015): 20.

13. Montero CS., *et al*. "Outcomes and complications of S2 alar iliac fixation technique in patients with neuromuscular scoliosis: experience in a third level pediatric hospital". *Journal of Spinal Surgery* 3.4 (2017): 519-524.

14. Keskinen H., *et al*. "The lifetime risk of pneumonia in patients with neuromuscular scoliosis at a mean age of 21 years: the role of spinal deformity surgery". *Journal of Children's Orthopaedics* 9.5 (2015): 357-364.

15. Mukherjee S., *et al*. "Incidence and outcome of scoliosis in children with pleural infection". *Pediatric Pulmonology* 42.3 (2007): 221-224.

16. Tang H., *et al*. "Risk factors for postoperative complication after spinal fusion and instrumentation in degenerative lumbar scoliosis patients". *Journal of Orthopaedic Surgery and Research* 9.1 (2014): 15.

17. Khirani S., *et al*. "Non-invasive positive pressure ventilation to facilitate the post-operative respiratory outcome of spine surgery in neuromuscular children". *European Spine Journal* 23.4 (2014): S406-S411.

18. Cognetti D., *et al*. "Neuromuscular scoliosis complication rates from 2004 to 2015: a report from the Scoliosis Research Society Morbidity and Mortality database". *Neurosurgery Focus* 43.4 (2017): E10.

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