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CASE REPORT

Management of Progressive Neuromuscular Scoliosis and Respiratory Distress in a Child with Nemaline Rod Myopathy

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ABSTRACT

CASE

Eleven-years old wheelchair bound girl presented with progressive breathing difficulty, deformity of back, and worsening sitting balance of 2-year duration. The child from the age of 3-years started developing repeated respiratory difficulties and was started on nocturnal BiPAP ventilator support. In view of generalized motor weakness and respiratory difficulty, the muscle biopsy done revealed NRM. The child presented to spine care center for the management of progressive spinal deformity affecting the sitting balance on wheelchair.

CONCLUSION

To the best of our knowledge this is the rare case of a severe rigid scoliosis in a child with NRM managed surgically successfully. Surgical management of these patients requires multidisciplinary approach for successful outcome in view of respiratory failure. Deformity correction will improve the respiratory reserve of child and may even obviate the need of nocturnal mechanical ventilation support.

KEYWORDS

Nemaline rod myopathy; Progressive neuromuscular scoliosis; Respiratory distress

1. INTRODUCTION

Nemaline Rod Myopathy (NRM) is a neuromuscular disorder characterized by slowly progressive skeletal muscle weakness and the presence of rod-shaped inclusion bodies (nemaline rod) in affected muscle fibers on histological examination [1]. It was first described in 1963 and the name derived from Greek word ‘nema’ which means ‘thread’ [2]. The estimated incidence worldwide is 0.02 per 100,000 live births [3]. Respiratory muscle weakness leading to respiratory failure is typical and most devastating complication of NRM [4].

2. CASE REPORT

Eleven-years old wheelchair-bound mild mentally retard girl who was dependent on caregivers for her activities of daily living presented with progressive breathing difficulty, deformity of back, and worsening sitting balance of 2-years duration. The child from the age of 3-year started developing multiple episodes of respiratory worsening, admission to ICU and was started on nocturnal BiPAP ventilator support. The muscle biopsy done revealed NRM.

Physical examination revealed diffuse axial and peripheral muscular weakness, decreased facial expressivity, prognathism, and weak facial muscles. She had rigid right sided thoracic curve with a rib hump. X-ray of whole spine showed rigid right thoracic scoliosis T3-L1 (Cobb angle 87°) and fixed pelvic obliquity of 10° (Figure 1).

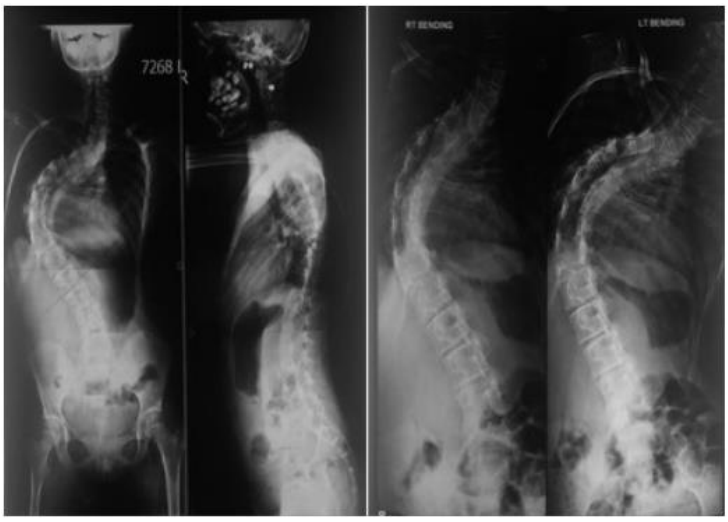

Figure 1: X-ray of whole spine in AP, lateral and side-bending position showed rigid right thoracic scoliosis T3-L1 (Cobb angle 87°) and fixed pelvic obliquity of 10°.

In view of progressive scoliosis deformity, poor sitting balance and respiratory difficulty, surgical correction of scoliosis was contemplated. Multidisciplinary evaluation was done and cleared for surgery under high risk in view of chronic type 2 respiratory failure. Surgery was performed successfully with multiple Ponte osteotomies and instrumented fusion T2-ilium. The surgery lasted for 6-hours with the blood loss of 850 ml. In PICU, she was gradually weaned off the ventilator to continuous BiPAP support (Table 1). She was able to sit independently and breathe better on the 4th post-operative day. A post-operative radiograph showed acceptable scoliosis correction (Cobb reduced to 30°) with good coronal and sagittal balance. She was discharged on 10th post-operative day with nocturnal BiPAP only.

Table 1: Respiratory function evaluation tests.

Tests	FVC	FEV	PaO2	BIPAP requirement	AHI
Before Surgery	64	59	70	26/10 (Throughout Day)	36
After Surgery	65	64	84	14/5 (Night Only)	18
After 2-Years	71	71	95	No Requirement	6

FVC: Forced Vital Capacity; FEV: Forced Expiratory Volume; PaO2: Arterial Oxygen Partial Pressure; BiPAP: Bilevel Positive Airway Pressure; AHI: Apnea Hypopnea Index

At 2-years follow-up, she was found to have significant improvement in her respiratory reserve and was out of even nocturnal ventilator support. The X-ray showed the correction to be well maintained and fusion completed (Figure 2).

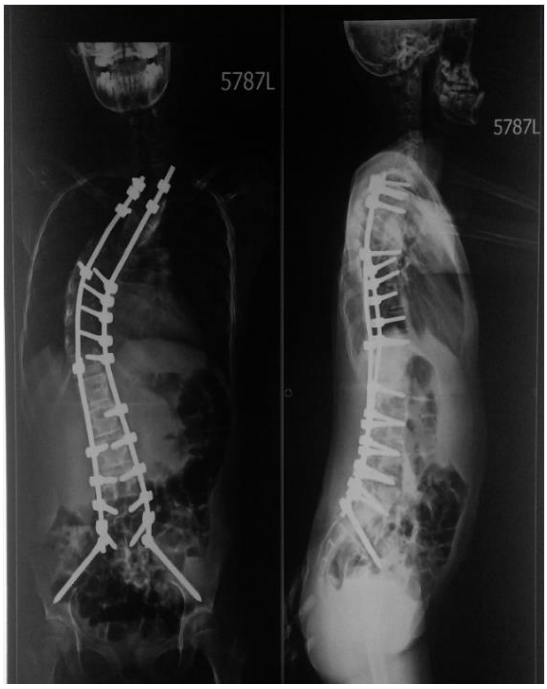

Figure 2: At 2-years follow up X-ray showed the correction to be well maintained and fusion completed.

3. DISCUSSION

Children with NRM have generalized muscle weakness with predilection to neck, face, axial, and proximal extremity muscles [4]. Respiratory muscle weakness affects the overall prognosis and survival. Even ambulant and mildly affected patient may develop unsuspected hypoxia especially during night and hence requires assisted mechanical ventilation [5]. The risk of developing scoliosis in neuromuscular disorders ranges from 80-100% in non-ambulatory patients [6].

Our reported child was wheelchair bound with decreased expression, prognathism, generalized muscle weakness and weak facial muscles (features typical of NRM). He was on nocturnal BiPAP ventilator support since the age of 3-years due to multiple episodes of respiratory infections and worsening respiratory reserve secondary to progressive scoliosis.

Scoliosis with rigid, severe curve requires long complicated surgery with risk of neurological deterioration, blood loss and risk of surgical site infection [7]. Surgical correction has become less risky with preoperative non-invasive ventilation even in patients with a vital capacity as low as 40%. Early surgical correction can slow down the progressive pulmonary dysfunction [8].

We corrected deformity successfully. Following surgery, she was able to sit independently, able to breathe better and requiring less BiPAP support (Figure 3). At 2-years follow up, her breathing reserve improved significantly and not even requiring nocturnal ventilator support.

Figure 3: Following surgery, she was able to sit independently, able to breathe better and requiring less BiPAP support.

4. CONCLUSION

To the best of our knowledge this is the rare case of a severe rigid scoliosis in a child with NRM managed surgically successfully. Surgical management of these patients requires multidisciplinary approach for successful outcome in view of respiratory failure. Deformity correction will improve the respiratory reserve of child and may even obviate the need of nocturnal mechanical ventilation support.

5. CONFLICT OF INETEREST

The authors declare that there are no conflicts of interest.

REFERENCES

1. Sanoudou D and Beggs AH (2001) Clinical and genetic heterogeneity in nemaline myopathy-a disease of skeletal muscle thin filaments. Trends in Molecular Medicine 7(8): 362-368.
2. Shy GM, Engel WK, Somers JE et al. (1963) Nemaline myopathy. A new congenital myopathy. Brain 86:793-810.
3. Wallgren-Pettersson C and Laing NG (2000) Report of the 70th ENMC international workshop: Nemaline myopathy, 11-13 June 1999, Naarden, The Netherlands. Neuromuscular Disorders 10(4): 299-306.
4. Wallgren-Pettersson C, Sewry CA, Nowak KJ et al. (2011) Nemaline myopathies. In Seminars in Pediatric Neurology 18(4): 230-238.
5. Heckmatt JZ, Loh L, Dubowitz V (1989) Nocturnal hypoventilation in children with nonprogressive neuromuscular disease. Pediatrics 83(2): 250-255.
6. Canavese F, Rousset M, Le Gledic B et al. (2014) Surgical advances in the treatment of neuromuscular scoliosis. World Journal of Orthopedics 5(2): 124-133.
7. Cunliffe M and Burrows FA (1985) Anaesthetic implications of nemaline rod myopathy. Canadian Anaesthetists' Society Journal 32(5): 543-547.
8. Gill I, Eagle M, Mehta JS et al. (2006) Correction of neuromuscular scoliosis in patients with preexisting respiratory failure. Spine 31(21): 2478-2483.