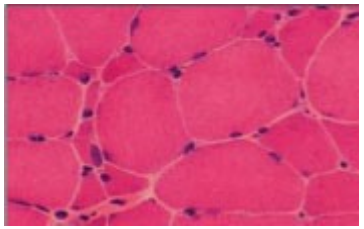

# PROGRESSIVE NEUROMUSCULAR SCOLIOSIS AND RESPIRATORY DISTRESS IN A CHILD WITH NEMALINE ROD MYOPATHY – A CASE REPORT

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# INTRODUCTION

- **Nemaline Rod Myopathy (NRM)** – inherited congenital myopathy [1]
- CHARACTERIZED BY :-
  - slow / non-progressive skeletal muscle weakness and
  - presence of rod shaped inclusion bodies/ fuchsinophilic proteins (nemaline rod) in muscle BIOPSY
- First described in 1963
- Derived from Greek word 'nema' meaning 'thread' [2]
- Though rare, it is the commonest non-dystrophic congenital myopathy

  (Estimated incidence worldwide - 0.02 / 100,000 live births)
- We hereby present such a case of severe rigid scoliosis in a child with NRM - clinical presentation and successful management by surgery.

# CASE HISTORY

## PRESENT HISTORY

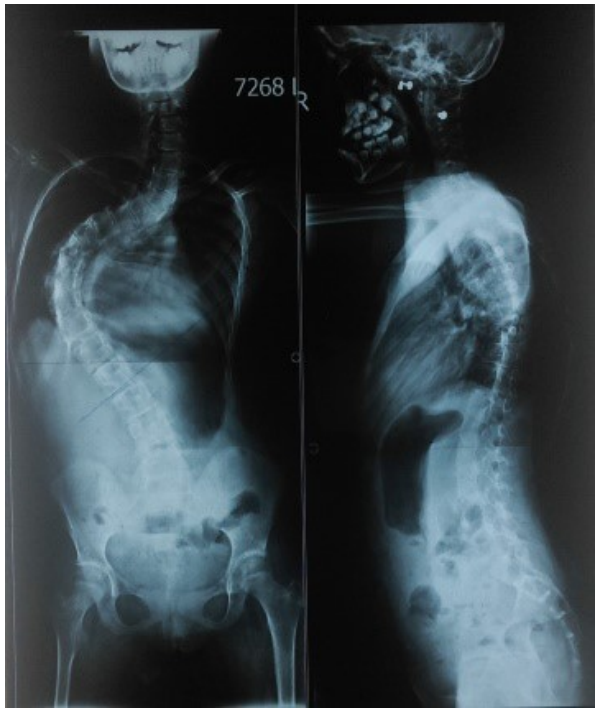
- 11-year-old wheelchair bound girl
- Progressive spinal deformity affecting sitting balance on wheelchair

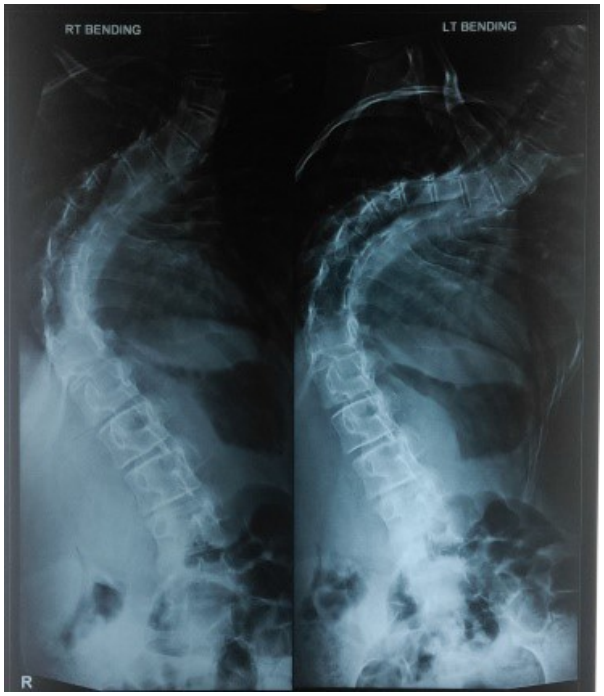
## PAST HISTORY

- Uneventful normal delivery to a non-consanguineously married couple
- Grew normally except for delayed motor milestones.
- From 3-years of age, developed repeated respiratory difficulties and nocturnal BiPAP was initiated.
- In view of generalized motor weakness and respiratory difficulty, muscle biopsy done → revealed NRM.

## EXAMINATION

- Diffuse muscular weakness with atrophy
- Face - decreased expressivity, prognathism and weak facial muscles.
- Right sided thoracic curve with a rib hump
- Motor examination revealed strength of MRC grade 3 in all muscle groups while sensations were intact.
- No muscle fasciculation/ pseudo-hypertrophy of muscles/ Gower's sign/ joint laxity



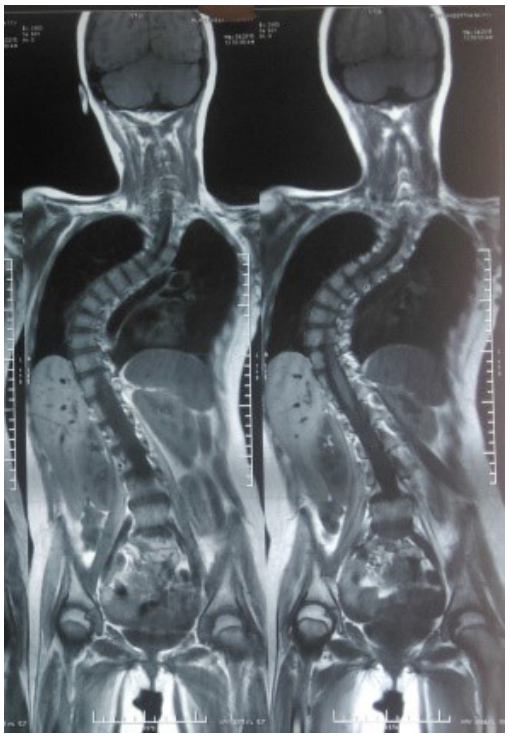

- Xray →

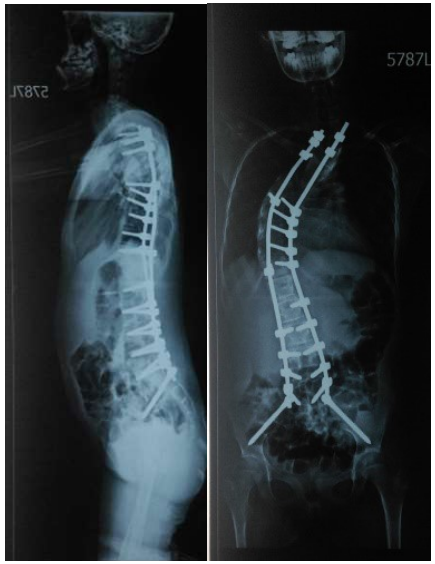
✓ Rigid Right Thoracic Scoliosis T3–L1 (Cobb angle 87°)

✓ Fixed pelvic obliquity 10°

- MRI screening →

✓ no intra-spinal abnormalities

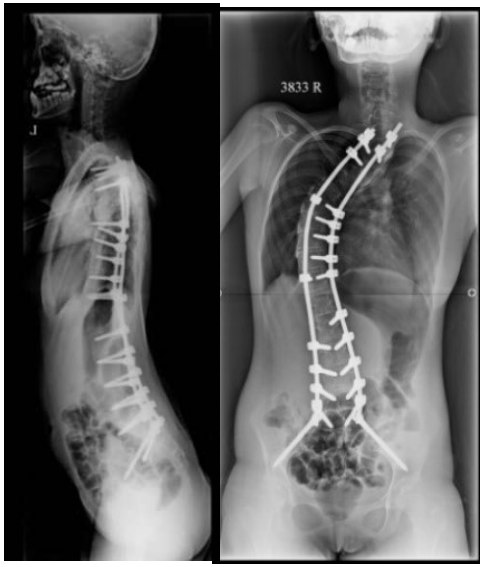




IMMEDIATE POST-OP

# RESULT

- Multi-modal intra-operative neuro-monitoring used [3]
- Multiple Ponte osteotomies
- Instrumented fusion T2-ilium
- Duration: 6hours
- Blood loss : 850 ml blood



AFTER 2 YEARS POST-OP

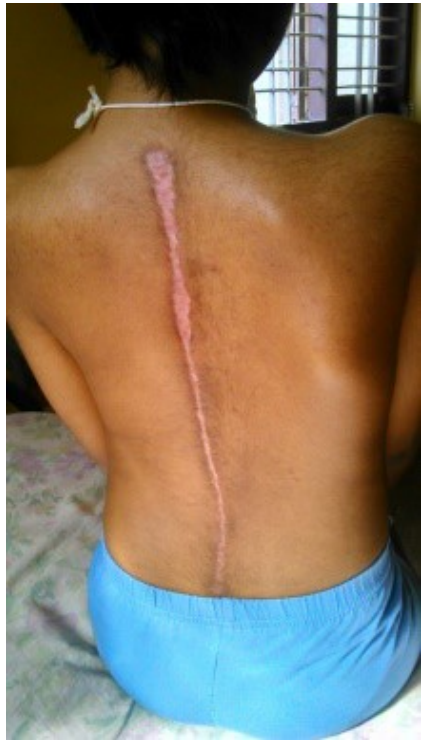
- Shifted to PICU for elective ventilation; gradually weaned off ventilator to continuous BiPAP support
- Able to sit independently and breathe better on the 4th post-op day
- Post-op radiograph → acceptable scoliosis correction (Cobb angle 30°) and good coronal & sagittal balance
- At 2-years follow up →

-- significant improvement in respiratory reserve and was out of ventilator support even at night

-- The X-ray showed the correction to be well maintained and fusion completed

# CONCLUSION

- Surgical management of these patients requires multi-disciplinary 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.



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