Peripartum management of congenital SEPN1 muscular dystrophy and rigid spine syndrome

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Case Presentation

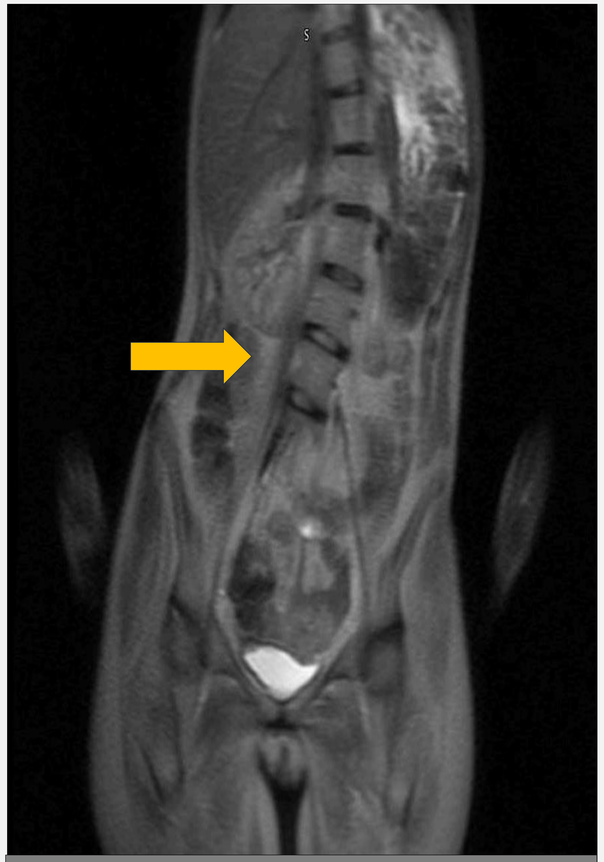
25-year-old Primigravid
SEPN1 muscular dystrophy
Severe scoliosis with rigid spine syndrome
Booking weight 32kg (BMI 15.6)

Worsening mobility due to fatigue and dyspnoea
Can walk 100m on flat but difficult to manage stairs
Uses two walking sticks for short distances and mobility scooter for longer distances

On non-invasive ventilation with Bilevel positive pressure ventilation (BiPAP) overnight and for short periods during the day if fatigued
Spirometry showed a restrictive respiratory pattern (FEV1 0.84L: 34% predicted & FVC 1.04L: 36% predicted)

Management of other health issues:
Dietician support for optimum nutrition intake
Close monitoring of renal function (history of recurrent renal stones)
Placed on antenatal enoxaparin as high VTE risk

Admitted to hospital at 30 weeks pregnant due to respiratory deterioration whereby she became dependent on BiPAP. Decision was made for her to have a category three caesarean delivery at 30+5 weeks



MRI of lumbar and sacral spine done pre-pregnancy showing scoliosis of lumbar spine concave to the right

Management

Considerations

- Ideally should have regional anaesthesia but expected difficult neuraxial and unpredictable spread due to severe scoliosis→ need to be ready for general anaesthesia
- Weight adjusted calculations of medications, anaesthetic drug dosages and trigger points for PPH management

Care delivered

- Category 3 LSCS at 30+5 weeks
- Multiprofessional involvement
- Semi-recumbent position with patient placed on BiPAP in theatre with ventilator on standby for additional oxygenation if needed
- Initial anaesthetic technique: arterial line monitoring, combined low dose spinal with slow epidural top-up
- Due to pain on surgical testing, converted to general anaesthetic using total intravenous anaesthesia
- Uneventful caesarean section, delivery of live neonate and minimal blood loss
- Extubated onto CPAP before returning to baseline nocturnal BiPAP and was discharged home within one week

Discussion

- SEPN1-related myopathy is a rare congenital neuromuscular disorder due to mutations in SELENON gene
- Symptoms include progressive central muscle weakness, subsequent difficulty with ventilation and severe spinal scoliosis
- Neuromuscular disorders can worsen during pregnancy, requiring careful antenatal planning and delivery management
- In this patient, neuraxial anaesthesia was prioritised to reduce risks of pulmonary complications from ventilation, but severe scoliosis contributed to inadequate anaesthetic spread
- Total intravenous anaesthesia was favoured to reduce risks associated with volatile agents in neuromuscular disease
- Post-operatively, the respiratory nurses were key in supporting the midwives on delivery suite to care for the patient on BiPAP

Key Learning Points

Awareness of neuromuscular disease in pregnancy and its effect on other systems → Importance of multiprofessional collaboration in patient care → Having a flexible anaesthetic strategy for these patients

REFERENCES

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