

Delivery challenges of Bethlem myopathy, a rare congenital muscular dystrophy

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Introduction

- Bethlem myopathy (BM) is a rare, autosomal dominant inherited muscular dystrophy.
- It is slowly progressive with hypotonia in the first or second decade, joint abnormalities and more rarely respiratory and/or cardiac involvement. [1]
- Information regarding the anaesthetic management during pregnancy and delivery is limited. We present the case of a primigravida.**

Case Report

- A 30 year old primigravida was seen in the specialist obstetric anaesthesia clinic at 33 weeks gestation and a history of BM was noted.
- The patient's baseline functioning was walking with a “waddling gait” and climbing stairs slowly due to upper leg weakness.



- Her sleep study revealed a mean SpO₂ 90% with lowest recorded 72%, **uncovering occult nocturnal hypoventilation. Overnight non-invasive ventilation (NIV) was established to optimise oxygen delivery to the baby as the pregnancy progressed.**
- Lying FEV₁ and FVC were 46% and 43% of predicted. (Pre-pregnancy 84% and 86% respectively). Her FVC dropped by 13% on lying down indicating diaphragmatic impairment. Her ECHO was normal.
- A significant scoliosis was noted on examination, and the spinous processes in the lumbar region were very difficult to feel.
- An early epidural was recommended and the increased risk of an inadequate block with severe scoliosis was discussed.**
- For operative delivery a slow epidural top-up was planned if the labour epidural was working bilaterally. Alternatively a low dose spinal CSE or general anaesthetic if the patient was unable to lie flat or bilateral block was not achieved.
- Full dose spinal was thought to be contraindicated due to loss of intercostal muscle function.
- She was admitted at 38+4 weeks for induction of labour and had an epidural inserted by a consultant anaesthetist in theatre under ultrasound guidance. This was maintained via patient controlled epidural and gave a bilateral block with no impact on respiratory function.
- Subsequently a decision was made to perform a caesarean section due to a pathological CTG.
- The epidural topped up with 15mls of 2% lidocaine with 1:200,000 adrenaline and 100 mcg fentanyl resulting in a good block for delivery.
- She had a further 4mls of 0.5% levobupivacaine and 2mg morphine via the epidural catheter towards the end of the procedure.
- She was cared for in labour ward HDU and discharged home 36 hours later.
- Respiratory review four months after delivery notes discontinuation of NIV.

Discussion

- This was a challenging case due to limited data on the management of BM in pregnancy.
- There were two case reports available – in one the patient required a wheelchair from 35 weeks gestation and went on to have a vacuum extraction delivery at 38 weeks with return to pre-pregnancy functional state afterwards. The other described a patient with deterioration in left ventricular function necessitating delivery at 36+2 weeks via caesarean birth and resolution of echocardiographic findings postnatally.
- Neither case comments on the anaesthetic management.
- The principles of management involved are a multi-disciplinary approach and optimisation of patient condition in the antenatal period.**
- The effective early epidural together with the lack of respiratory decompensation in labour were key in allowing a safe epidural top up in theatre.**

References

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