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Introduction

- SOX proteins play a crucial role in brain development and SOX-11 mutations are associated with neurodevelopmental anomalies, influencing autonomic and sensory nerve function and present with features of Coffin-Siris syndrome (CSS).
- These include, hypotonia, intellectual disability, craniofacial abnormalities, scoliosis/kyphosis, chronic respiratory infection, cardiac abnormalities and gastroesophageal reflux [1].

Case Report

- A 34-year-old primigravida with known SOX-11 mutation presented for induction of labour due to pregnancy induced hypertension.
- She was of short stature at 145cms with small digits and learning difficulties
- She mobilised with a stick due to knee osteoarthritis, denied spinal abnormalities or neurological symptoms
- She had no distinguishable craniofacial abnormalities and on airway examination was Mallampati I with good mouth opening and full range of neck movement.

- On examination, her spine was in the midline with easily to palpate spinous processes with no overlying skin lesions.
- Spinal ultrasound examination did not demonstrate any obvious spinal abnormalities with a thecal sac depth of approximately 6cm and clear visualisation of lumbar interspaces.
- In the event she was to require a neuraxial block she was counselled about the possibility of abnormal spinal anatomy, risk of nerve damage and potential for high block, failure and conversion to GA.
- Despite the risks, if a caesarean section was required neuraxial anaesthesia was deemed preferable to a general anaesthetic given increased risk of respiratory complications and altered drug sensitivity. There were no cardiorespiratory abnormalities.
- The patient required an emergency caesarean section under spinal anaesthetic.
- The procedure was straightforward at L3/4 interspace using 25G 80mm Whitacre spinal needle with no complications.
- A reduced dose of 2.2ml of 0.5% heavy Marcaine with 400mcg of diamorphine was used – resultant block to T2 bilaterally.

- The Caesarean section was performed with no complications and without any prolonged spinal blockade.

Discussion

- SOX-11 mutation results in autosomal dominant inheritance of CSS.
- This syndrome complicates anaesthesia for caesarean section effecting haemodynamic stability, nerve function, potential for difficult spinal or epidural placement, a potentially difficult airway and high aspiration risk.
- These patients require individualised anaesthetic management dependent on phenotype.
- Regional techniques are often favoured to avoid airway complications and to minimise systemic drug effects.
- Reduced doses recommended due to heightened sensitivity and altered nerve conduction.
- Continuous monitoring and vasopressor availability advised to mitigate likely autonomic dysfunction [2].

References

1. Hempel A, Pagnamenta AT, Blyth M et al. Deletions and de novo mutations of SOX11 are associated with a neurodevelopmental disorder with features of Coffin-Siris syndrome. J Med Genet. 2016 Mar;53(3):152-62
2. Fornet I, Morillas P, Lopez MA et al. Emergency caesarean in a patient with Coffin-Siris syndrome. Rev Esp Anesthesiol Reanim. 2007 Nov;54(9):563-5