Anaesthetic Management for Elective Caesarean section in a pregnant woman with Pycnodysostosis

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INTRODUCTION

Pycnodysostosis is a rare autosomal-recessive condition with an incidence of 1.7 per 1 million births. It is caused by the mutation of cathepsin K gene. The affected individuals are an anaesthetic challenge due to their short stature with multiple cranio-facial and spinal abnormalities. We describe a case of pycnodysostosis in a parturient posted for elective caesarean section.

CASE REPORT



A 37-year-old gravida 2 with previous spontaneous miscarriage was posted for elective caesarean section at 37+1weeks. Her BMI was 34.7 with height of 144cm and weight of 72kgs. Due to significant background history of Pycnodysostosis she initially presented to our high-risk anaesthetic antenatal clinic for review. She had a history of multiple fractures until the age of 15 years including bilateral femur, right tibia, and skull fractures. She was on established continuous positive airway pressure therapy for her obstructive sleep apnea. In the past, she had an elective surgery for mal-aligned jaw with planned tracheostomy. Clinical examination showed typical features of Pycnodysostosis such as short stature, frontal bossing, and small hands. Airway examination confirmed a small mouth opening, Mallampati 3, reduced thyromental distance, limited neck extension, minimal jaw protrusion and a previous tracheostomy scar. Spine examination showed accentuated lordosis with slight scoliosis and narrow intervertebral spaces. Her baseline haemoglobin was 114g/dl and we crossmatched 2 units of blood. Her placenta was anterior, not low and baby growth was below 90th centile. A Multi-disciplinary team (MDT) input was sought for the management of her delivery including advice from the critical care, ENT, neonatal and obstetric team. She was deemed suitable to be delivered at our district general hospital. After discussion, our plan was to do a single shot spinal as plan A and in case of failure to convert to general anaesthetic with THRIVE and videolaryngoscopy as plan B. Spinal was administered with 2.2ml of 0.5% bupivacaine and 0.3 mg diamorphine. Block test post spinal confirmed a good motor block bilaterally and loss of cold sensation to level of T4 bilaterally. Patient was comfortable and pain free throughout surgery and baby was delivered with good apgar scores. Estimated blood loss was 900mls. Overall, we managed the patient successfully during her elective caesarean section at 37 weeks of gestation under regional anaesthetic technique and postoperatively transferred her to the labour ward for close monitoring. In the post operative period, care was taken to avoid weight bearing until full recovery was made. She was discharged on day 2 post caesarean section and had no re-admissions for any complications.



DISCUSSION

Pycnodysostosis (the Toulouse-Lautrec syndrome) is a rare autosomal-recessive disorder of osteoclast dysfucction.[1] Patients with Pycnodysostosis have characteristic abnormalities which include frontoparietal bossing, hypoplastic paranasal sinuses, high arched palate, mandibular hypoplasia, spinal abnormalities, short stature, and large protruding tongue. [2] The craniofacial abnormalities may predispose to difficulty during intubation with risk of cervical spine fracture during laryngoscopy and neck flexion. Such patients may also pose a significant challenge to the anaesthetist performing a central neuraxial block due to their spinal abnormalities and short stature. While considering our options for regional technique, we preferred a spinal anaesthetic over combined spinal epidural. In our opinion, a spinal anaesthetic would give good quality block, be of reduced anaesthetic challenge in the background of spinal abnormalities and would be adequate due to virgin abdomen, nil placental concerns and a senior surgeon. Our detailed preoperative assessment, MDT involvement and frank discussion with the patient about her needs and expectations led to a successful and safe outcome for both the baby and the mother.

REFERENCES

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