CASE REPORT

Emergency salpingectomy for ruptured ectopic pregnancy in patient with Melnick-Needles syndrome: a rare otopalatodigital syndrome

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SUMMARY

Melnick-Needles syndrome (M-NS) is a rare genetic disorder which primarily affects skeletal developments. M-NS may also affect the cardiorespiratory and renal systems. A 35 kg patient presented complaining of abdominal pain. Following a positive pregnancy test and ultrasound examination, she was diagnosed with a ruptured ectopic pregnancy. She had a significant background history of M-NS, obstructive sleep apnoea (with narrow gauge tracheostomy in situ), obstructive lung disease and scoliosis. She received fluid resuscitation, and the case was managed using an open salpingectomy and clot evacuation under combined spinal/epidural anaesthesia. Anticipated difficulty in securing a definitive airway was pivotal to choosing a regional anaesthetic technique. The operation was successful and the patient recovered well. This was a unique surgical and anaesthetic challenge due to abnormal facial, spinal and abdominal morphology compounded by the time-critical emergency nature of the case. Individually tailored perioperative management is frequently required for patients with rare syndromes.

narrow shoulders and short clavicles. A pectus excavatum is often observed in affected individuals in over 50% of instances.² Severe lung disease, recurrent pulmonary infections and pulmonary hypertension are described in affected individuals.³ Vertebral malformations such as wedged vertebrae frequently lead to scoliosis or kyphosis (figure 2). Short stature is characteristic and hip dislocation may occur. Cardiac manifestations can include mitral and/or tricuspid prolapse. Hydronephrosis may occur secondary to small ureters and renal hypoplasia.⁴

Inheritance is X-linked dominant, the condition being lethal in most males and only a few instances of male survival being recorded.⁵ The majority of patients are female but disease severity is greater in male who survive.⁵ The condition is caused by mutation in the Filamin A (*FLNA*) gene on Xq28, all affected individuals sharing mutations within exon 22 of the gene.⁴ There are several allelic disorders, caused by mutation elsewhere within the gene and some of which, such as otopalatodigital syndrome, share some of the bony features of M-NS. A characteristic M-NS mutation of the *FLNA* gene had been established in this patient, consistent with the clinical diagnosis.

BACKGROUND

Obstetric patients, although generally a healthy cohort, may have significant comorbidities. When a patient presents in the emergency setting with a rare, severe congenital syndrome that has significant surgical and anaesthetic implications, individualised tailoring of care must be undertaken to ensure safe and effective treatment. Here, we document the safe management of a patient with Melnick-Needles syndrome (M-NS) presenting in hypovolaemic shock due to a ruptured ectopic pregnancy requiring salpingectomy which was performed using an open approach under neuraxial anaesthesia.

M-NS is a rare genetic disorder. While primarily affecting bone development, it also has wider phenotypic manifestations affecting multiple body systems.¹ It is characterised by facial dysmorphic manifestations and radiological abnormalities. The characteristic facies is typified by micrognathia, small facial bones, exophthalmos and full cheeks (figure 1). Radiologically, there is bowing of the bones of upper and lower limbs, sometimes referred to as 'wavy bone appearance'. The effects of the syndrome on the chest is variable, but clinical and radiological evidence of chest involvement is common including irregular ribbon-like ribs,

CASE PRESENTATION

A 30-year-old woman with M-NS at 6 weeks gestation presented to the emergency department (ED) of a general hospital during on call hours at the weekend. She was complaining of severe lower abdominal pain. The onset of pain was 5 hours prior to admission, gradually increasing in severity and localising to her left iliac fossa. She had initially presented to her general practitioner, who, after confirming a positive pregnancy test, referred her urgently to the ED of the nearest general hospital. On arriving to the ED, she received intravenous fluid resuscitation and haemodynamic stability was achieved. A provisional diagnosis of an ectopic pregnancy was made. She was transferred via ambulance to our unit. This patient's previous medical notes were unavailable due to timing and urgency of presentation.

On arrival, she was unwell, tachycardic with severe abdominal pain associated with shoulder tip pain, nausea and vomiting.

The prior medical notes were not available.

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Figure 1 Facial appearance with tracheostomy tube visible.

She stated that her medical history was significant for M-NS and obstructive sleep apnoea (OSA). She was diagnosed with OSA in childhood and had had a tracheostomy inserted aged 5 following a failed course of nocturnal continuous positive airway pressure. Of note, she did not get recurrent pulmonary infections. She quoted her pulmonary function tests as having a forced

Figure 2 Back appearance.

expiratory volume in 1 s:forced vital capacity ratio of 30%. She had no known cardiac pathology. She was a non-smoker.

Surgical history was significant for previous mandibular osteotomy in addition to the tracheostomy procedure.

She was not taking any regular medications and had no known drug allergies.

Her family history revealed her mother and sister also had M-NS, however to a less severe degree.

General physical examination revealed a number of striking features including micrognathia, exophthalmos and protruding cheeks, a tracheostomy tube (figure 1) and short stature with scoliosis (figure 2). Her height at admission was 135 cm and her weight 35.8 kg. She had a long-term 3.5 mm uncuffed tracheostomy tube in situ.

Her vital signs showed a tachycardia of 120 beats per minute, her blood pressure was 119/86 mm Hg and she had an oxygen saturation of 100%. Urinary beta human chorionic gonadotropin (β hCG) was positive.

Abdominal examination revealed tenderness across the lower abdomen, most pronounced in the left iliac fossa. No rebound tenderness or guarding was elicited.

Airway assessment revealed reduced mouth opening, a small airway, a receding mandible and reduced lung volumes. Front of neck access was in situ via an uncuffed tracheostomy tube of 3.5 mm internal diameter (4.5 mm outer diameter).

Transvaginal ultrasound imaging revealed an empty uterus with a thickened endometrium of 19 mm. A pool of free fluid with a maximum depth of 2.7 cm was seen with an adnexal mass highly suspicious of an ectopic pregnancy on the left side. She had significant cervical excitation on vaginal examination.

A diagnosis of a suspected ruptured ectopic pregnancy was made. The on-call consultants for obstetrics and anaesthetics were informed, and the patient was prepared for emergency surgery.

INVESTIGATIONS

- ▶ Urinary β hCG.
- ▶ Pelvic ultrasound.

DIFFERENTIAL DIAGNOSIS

Ruptured ectopic pregnancy.

TREATMENT

The patient underwent an uncomplicated open salpingectomy under combined spinal/epidural anaesthesia using a low spinal dose of local anaesthetic (5.5 mg hyperbaric bupivacaine with 5 μ g of fentanyl) along with titrated epidural local anaesthetic (10 mL 2% lignocaine). A sensory level of T6 was achieved bilaterally and surgery was commenced. Surgical anaesthesia was adequate. Shoulder tip pain was treated with intravenous fentanyl boluses (0.3 μ g/kg). Paracetamol (15 mg/kg), diclofenac (1 mg/kg) and oxycodone (50 μ g/kg) were given intravenously for postoperative analgesia.

A total of 500 mL of clotted blood was removed from her abdomen and pelvis. The total blood loss was estimated at 860 mL. The patient received a total of 1500 mL of crystalloid intraoperatively and an infusion of crystalloid at 2 mL/kg/hour was commenced in the high-dependency unit (HDU) postoperatively. An epidural infusion of 0.125% levobupivacaine was commenced in the HDU along with regular, simple intravenous analgesia.

OUTCOME AND FOLLOW-UP

She had an uncomplicated postoperative course. Prior to discharge, she was started on a contraceptive pill. She recovered well and was

discharged on postoperative day 3. She was seen in the outpatient department and advised against future pregnancies.

DISCUSSION

This was a challenging case and careful consideration was given to deciding the optimum surgical and anaesthetic technique. Discussion between the anaesthetic and surgical teams was important as the anaesthetic technique would have implications as to the surgical technique employed and vice versa. There are no similar cases reported in the literature.

Current Royal College of Obstetricians and Gynaecologists guidelines recommend a laparoscopic salpingectomy in cases of a ruptured ectopic pregnancy in a haemodynamically stable patient.⁶

In our stand-alone obstetric unit, laparoscopic surgery under general anaesthesia is the preferred option for the treatment of suspected ruptured ectopic pregnancy. While it is possible to perform laparoscopic surgery under neuraxial anaesthesia, it was felt that this patient's compromised lung function would inhibit her ability to tolerate a pneumoperitoneum and Trendelenburg position to facilitate such a technique. In addition, owing to the patient's short stature and relatively small pelvic anatomy, it was anticipated a laparoscopic approach may be difficult. General anaesthesia, too, presented challenges in this woman as she was very likely to be difficult or impossible to bag-mask ventilate or intubate through her mouth or nose.

Initially, general anaesthesia was attempted using the in situ tracheostomy; however, the narrow tube diameter led to very small tidal volumes being achieved and a large leak was detected. Anaesthesia was not achieved. Furthermore, the patient's airway would not be protected during the case using this uncuffed tube. The long-standing in situ tracheostomy tract was fully epithelialised and dilating the tract to facilitate a larger/cuffed tube was likely to be difficult, traumatic and painful. Furthermore, our stand-alone obstetric unit has limited advanced airway specialist support during the out-of-hours period. Given the aforementioned issues and her unknown airway anatomy, unavailable prior notes, and we decided to use an open approach operation under combined spinal epidural anaesthesia. Neuraxial anaesthesia also presented challenges as she had a significant kyphoscoliosis and it may compound her haemodynamic instability.

Consideration was given to transferring the patient back to the general hospital owing to the complexity of the case; however, due the patient's deteriorating clinical condition, it was decided to proceed.

Otopalatodigital syndromes, such as M-NS, may present difficulties to anaesthetists and surgeons alike, particularly in the emergency setting. Abnormal anatomy, compromised physiology

and previous procedures can contribute to airway difficulties and complicate surgical procedures which may necessitate tailoring the perioperative management of these patients.

Learning points

- ▶ Obstetric patients may present with complex craniofacial and skeletal abnormalities.
- ▶ Standard surgical and anaesthetic care may need to be tailored for individual patient requirements.
- ▶ Good communication between clinical teams is important for delivering safe and effective care particularly in the emergency setting.
- ▶ Regional anaesthesia may be employed effectively when used with caution in patients with hypovolaemia.
- ▶ A tracheostomy tube in situ is not a panacea for delivering general anaesthesia.

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Competing interests None declared.

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