

JARCHO-LEVIN SYNDROME

INTRODUCTION

Jarcho-Levin Syndrome (JLS) is a rare congenital disorder, occurring in 1 in 40,000 births. It is characterised by abnormal development of the spine and ribcage, leading to short stature as a result of a shortened neck and trunk, and a small thoracic cavity - features with significant anaesthetic implications.^{1,2}

CASE REPORT: 20 YEARS OLD PRIMIGRAVIDA

One of only two women in the UK known to have this condition, presented at 20 weeks for delivery planning. She had no previous anaesthetic history. Her pre-operative assessment is outlined below.



- A** Short neck but good movements. Mallampati III. Thyromental distance > 5cm. Mouth opening > 3cm. No glottic abnormalities on nasoendoscopy.
 - B** 300m exercise tolerance. Restrictive lung defect: FVC 55% and FEV1 53% predicted. Floating ribs and absent sternum.
 - C** Spontaneously closed ASD. Normal echocardiogram.
 - D** Significant kyphoscoliosis. MRI Spine
 - Midline fusion defects
 - Hemivertebrae
 - Lower lumbar-sacral fusion
 - E** Reflux and swallowing difficulties. Severe anxiety and needle phobia.
- Allergies to pineapples, bananas, lactose, gluten and almonds.

JLS in the parturient presents further challenges with timing of delivery as the pregnancy progresses and can cause cardio-respiratory insufficiency. This has implications for timing and mode of delivery, choice of anaesthesia and management of airway and ventilation.

PRE-OPERATIVE

The patient was assessed pre-operatively at 20 weeks during a joint multidisciplinary team (MDT) meeting that included maternal medicine, anaesthetists, obstetricians, and ENT specialists.

A decision was made to perform a caesarean delivery at 33 weeks in view of worsening respiratory function and increasing musculoskeletal pain, balanced against the risks of prematurity.

INTRA-OPERATIVE

Regional anaesthesia was deemed unsuitable due to fused vertebrae, and patient unable to tolerate lying down in supine position due to musculoskeletal pain and shortness of breath.

An asleep intubation was chosen due to the patient's extreme anxiety around an awake fibreoptic procedure. Two consultant anaesthetists and an ENT surgeon were present at induction.

INDUCTION



An Oxford HELP pillow was used to position the patient for intubation.



General anaesthesia was induced with total intravenous anaesthesia (TIVA). BIS monitoring was used throughout surgery.



A Glidescope LoPro S3 hyperangulated videolaryngoscopy was used with a bougie. This provided a full view of the glottis.



A range of endotracheal tubes, sizes 4 through 7, was prepared. A size 6 endotracheal tube was ultimately inserted.



Bag-mask ventilation was uncomplicated, with peak ventilation pressures ranging from 16 to 18 cm H2O and a PEEP set at 3 cm H2O.

OUTCOMES

An arterial line was inserted following the delivery of the baby. Bilateral transversus abdominis plane blocks were injected at the end of surgery and a PCA oxycodone was set up for analgesia.

Cell salvage was used during the case due to limited allowable blood loss based on the patient's size. Total blood loss was 600 ml, with 261 ml returned via cell salvage. Her haemoglobin decreased from 109 g/L pre-operatively to 100 g/L.

Baby was admitted to NICU for CPAP after birth, however was spontaneously breathing on air after arrival. APGAR scores were 7 at 1 minute, 8 at 5 minutes and 10 at 10 minutes.

The mother was admitted to the maternal enhanced care room within the central delivery unit after an uneventful extubation.

She mobilised on day 1 post-surgery and was discharged on day 4 without post-operative complications.

This report emphasises the benefits of a early coordinated multidisciplinary approach to improve outcomes for mother and baby in a medically complex case.

References

- S. Sabaretnam, J. Chin, P. Kajekar, Anaesthetic considerations in a parturient with Jarcho-Levin syndrome, International Journal of Obstetric Anaesthesia, Volume 41, 125 – 126
- Ibrahim MA, Almetwally RR. Anaesthetic management of a parturient with spondylocostal dysostosis (Jarcho-Levin syndrome) for caesarean delivery. Anaesth. pain intensive care 2023;27(6):780–782