Anaesthesia for a patient of Hajdu Cheney syndrome scheduled for scoliosis surgery-A case study

Dear Editor,

Hajdu Cheney syndrome (HCS), a rare connective tissue disorder associated with the *NOTCH2* gene mutation,[1] presents unique challenges in anaesthetic management due to its multisystem involvement, including characteristic arthro-dento-osteodysplasia and various craniofacial abnormalities.[2-4] We report the perioperative anaesthetic management of a 17-year-old female patient diagnosed with HCS undergoing scoliosis correction and posterior instrumentation.

Our patient, exhibiting distinct clinical features such as osteoacrolysis, platybasia and Arnold–Chiari malformation Type 1, underwent comprehensive preoperative assessment and appropriate preparation. Physical examination showed a short-statured girl (146 cm) with low-set ears, a broad, puffy face, hypertelorism and abnormal dentition. Hyperlaxity of joints and short, stubby fingers with discoloured nails were noted [Figure 1]. Airway examination revealed a short neck, mandibular hypoplasia, multiple clustered teeth and Mallampati grade 2. There was a pronounced thoracolumbar scoliosis with right-sided convexity with Cobb's angle 90° [Figure 2]. Her exercise capacity was less than 4 metabolic equivalents with Modified Medical Research Council grade 1–2. Pulmonary function tests showed a severe restrictive pattern with forced expiratory volume in the first second 45% of the predicted, forced vital capacity 58% of the predicted and diffusion capacity 70% of the predicted. Preoperative arterial blood gas values were as follows: pH 7.42, partial pressure of oxygen (PO_2)- 70 mmHg, partial pressure of carbon dioxide (PCO_2)- 35 mmHg, bicarbonate (HCO_3)- 24 mEq/l, and haemoglobin (Hb)- 11 g/dL. Complete blood count, coagulation profile and electrolytes were within normal limits.

The case involved careful consideration of airway management due to micrognathia, mandibular hypoplasia and cervical instability.[2] A difficult airway cart, including small-sized endotracheal tubes [size 6.5- and 5.5-mm internal diameter (ID)], a bougie, a video laryngoscope and a paediatric fibreoptic bronchoscope, was kept ready. Anaesthetic induction was achieved with intravenous 90 μg fentanyl, 90 mg propofol and 90 mg suxamethonium to facilitate tracheal intubation. Given the patient's anatomical challenges, manual in-line cervical stabilisation and video laryngoscopy were utilised for intubation.[4] Laryngoscopy was performed using Karl Storz Video Macintosh laryngoscope, which revealed a Cormack Lehane grade 2b view. A 7-mm ID cuffed endotracheal tube (Portex, Hythe, Kent) could not be negotiated. So, a 6.5-mm ID cuffed tube was inserted. Intrathecal morphine 150 μg was administered, and the patient was then positioned prone on a bean bag and snugly encased. Needle electrodes were placed over the scalp, forearm and calf to monitor somatosensory evoked potentials and motor evoked potentials. A lung protective ventilation strategy with low tidal volume, high inspiratory time [inspiratory: expiratory (I:E) ratio 1:1] and adequate peak end-expiratory pressure

Figure 1: Osteoacrolysis

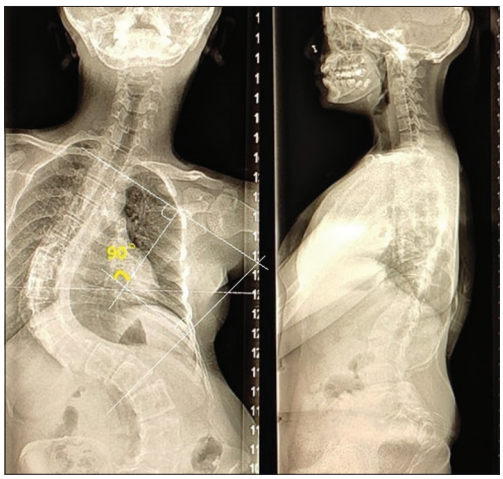

Figure 2: Anterior–posterior and lateral scoliosis plain films demonstrating severe scoliosis of 90°

were employed owing to severe lung restriction in preoperative testing. Ventilatory settings were adjusted based on intraoperative blood gas values. Bispectral index values of 40–60 were maintained during the surgery. Pre-extubation blood gas showed pH 7.38, PO_2 68 mmHg, PCO_2 45 mmHg, HCO^{3-} 21 mEq/l, and Hb 9 g/dL. Tracheal extubation was uneventful. The patient required two red blood cell transfusions during the surgery, and multiple platelet transfusions during her 3-day intensive care unit stay.

This report emphasises the need for a tailored anaesthetic approach in HCS patients, focusing on airway assessment, systemic organ function evaluation, meticulous positioning and preparedness for potential bleeding complications. Theoretically, *NOTCH2* mutations are known to be associated with bleeding risks.[5] HCS appears to be one of the most difficult airway-related syndromes because of the characteristics of skull and neck abnormality.[6] We advocate using a video laryngoscope or fibreoptic scope for tracheal intubation, smaller-size endotracheal tubes, assessing coagulation profiles preoperatively and ensuring adequate blood product availability. Osteoporosis and joint laxity mandate careful and gentle prone positioning with particular emphasis on the pressure points to avoid iatrogenic fractures. Care with settings of mechanical ventilation is required, bearing in mind the severe restrictive lung disease seen in these patients.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient consented to her images and other clinical information to be reported in the journal. The patient understands that her name and initials will not be published and due efforts will be made to conceal her identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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