Anaesthetic considerations in an orphan disease with skeletal anomalies

Sir,

A 12-year-old girl presented for surgical correction of bilateral genu valgus. She was a diagnosed case of cleidocranial dysplasia (CCD), karyotyping 46, XX normal, and a product of non-consanguineous marriage. Written informed parental consent for surgery and publication without identity disclosure was obtained.

The patient had frontal bossing, micrognathia, depressed nasal bridge, high-arched palate, multiple impacted supernumerary teeth [Figure 1], barrel-shaped chest, cup-shaped distal phalanges and down-curving nails. Mouth opening was adequate but Mallampati airway grade (MP) was indeterminate. Height (135 cm) was less than 3rd percentile and weight (30 kg) was 10th percentile for her age. She had mild intellectual developmental disability. Radiologically, hypoplastic clavicles and thickening of the occipital calvarium were observed [Figure 2]. Other systems and hormonal profile were within normal limits.

Standard anaesthesia procedure was followed. Pre-medication (injection glycopyrrolate 0.03 mg, fentanyl 50 μg and midazolam 1 mg intravenous [IV]) was administered in the pre-operative area of the operation theatre (OT) in the presence of her parents. Due to parental separation anxiety, her father accompanied the patient inside the OT. Induction was done with propofol 50 mg IV, and after ascertaining adequate mask ventilation, atracurium 15 mg was administered. Laryngoscopic Cormack and Lehane (CL) Grade III was converted to CL IIb by applying external cricoid pressure. Airway secured with a cuffed 5.0 mm endotracheal tube. Maintenance was done with oxygen and sevoflurane (1–1.5 minimum alveolar concentration). Single-shot caudal epidural was given for peri-operative pain relief with 12 mL of 0.25% bupivacaine and 10 μg of fentanyl. Care was taken while positioning to avoid excessive range of movements at all joints. Vitals were maintained. The patient was reversed with 1.5 mg neostigmine and glycopyrolate 0.18 mg. She had an uneventful recovery.

More than 500 cases have been reported worldwide of CCD, also called cleidocranial dysostosis or Marie Sainton syndrome. It is an orphan disease, genetically mapped to 6p21 but with considerable variability of genetic expression.[1,2] Facial dysmorphisms, skeletal

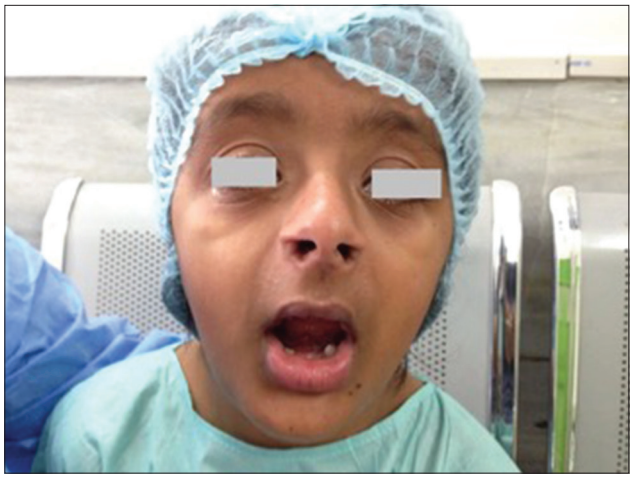

Figure 1: Cleidocranial dysplasia

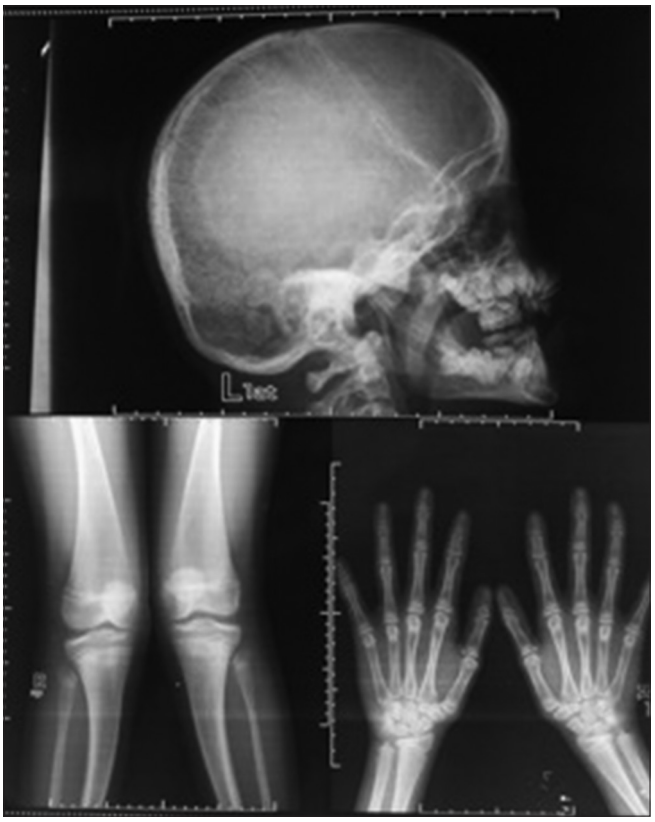

Figure 2: Thickening of the occipital calvarium

anomalies and recurrent respiratory tract infections in a background of near-normal general health, cognition and intellect present unique challenges for the anaesthesiologist. The clinical spectrum of this orphan disease can vary from mild dental and skeletal abnormalities to rare, serious afflictions such as syringomyelia. Bony abnormalities such as brachycephalic skull with mid-face retrusion, abnormal dentition (delayed eruption of secondary dentition, failure of primary teeth eruption, supernumerary teeth with dental crowding) and clavicular hypoplasia are present.[3] The facial dysmorphism, micrognathia, abnormal palatal shape and fragile teeth together may produce a 'cannot-ventilate, cannot-intubate' like situation and difficult airway cart should be ready. Other abnormalities include narrow pelvis with wide symphysis pubis and spina bifida occulta.

Features similar to CCD are also seen in various other disorders like congenital pseudoarthrosis of clavicle, which is usually unilateral; pyknodyostois, which does not have supernumary teeth and Yunus Varon Syndrome which is associated with low intelligence and failure to thrive.[3]

Although the difficult intubation was managed with external laryngeal manipulation in this case, cases with restricted mouth opening, higher MP and CL grades have been reported previously.[4,5] Difficult airways in these patients are usually manageable with external laryngeal manipulations and intubation aids.

Prior knowledge and preparation of potential anaesthetic challenges can further smoothen the anaesthetic course. Pre-operative evaluation for difficult airway, restrictive lung disease and kypho-scoliosis should be done. Ultrasound-guided central venous line and brachial plexus blocks are advised in these patients with clavicular hypoplasia. Positioning should be done carefully because of open skull sutures, increased joint laxity, hyper mobility and osteopenia. Pre-operative radiologic evaluation of odontoid process is recommended as atlantoaxial subluxation with consequent myelopathy has also been reported in these patients.[3]

Skeletal dysmorphism with near normal emotional intelligence can result in a sense of insecurity and heightened parental separation anxiety. They should be handled with compassion and pre-operative anxiolysis is important.

Although the anaesthetic challenges in CCD patients are usually not severe and readily manageable, a thorough pre-operative workup is essential. Detailed assessment of the airway, spine and pulmonary function should be done. Difficult airway cart should be kept handy. Careful positioning and use of ultrasound is for brachial plexus blocks and central venous line insertions are advised.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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