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Dr. Vedhika Shanker
Junior Consultant,
Department of Anaesthesia,
NH-SRCC Children's Hospital,
Mumbai, Maharashtra, India

Dr. Nandini Dave
Senior Consultant and Head,
Department of Anaesthesia,
NH-SRCC 2 Children's
Hospital, Mumbai,
Maharashtra, India

Corresponding Author:
Dr. Vedhika Shanker
Junior Consultant,
Department of Anaesthesia,
NH-SRCC Children's Hospital,
Mumbai, Maharashtra, India

Case series of the anaesthesia management in two children with Desbuquois Syndrome and review of literature

Vedhika Shanker and Nandini Dave

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Abstract
Background: We report the anaesthesia management of two children with Desbuquois syndrome. Both had characteristic features – flat-face, micrognathia, bell-shaped thorax, hyper-flexible joints, clinodactyly, short neck and dwarfism, and genetic testing confirming the syndrome. Less than 50 cases are documented, with only 2 reporting anaesthesia management in children: this case series a first documenting the course of 2 patients over 4 surgical procedures.
Cases: Both the patient had challenging airways, managed with endotracheal intubations due to failed LMA placement. We also postulate an element of subglottic stenosis as despite good view of the glottis, the age-appropriate endotracheal tube could not be negotiated, necessitating the use of a smaller tube in both patients. Post-operative high dependency care was needed in both patients for respiratory insufficiency.
Conclusion: Anticipate a difficult airway, difficulty in the placement of regional blocks and have low threshold for intensive care following anaesthesia as respiratory insufficiency is the leading cause of fatality.

Keywords: Desbuquois syndrome, paediatric anaesthesia, difficult airway, dwarfism, general anaesthesia

Introduction

Desbuquois syndrome has an incidence of less than 1/1,000,000 live births. The disorder was first described in 1966 by Desbuquois *et al* and less than 50 cases have been reported in literature, with wide heterogeneity in characteristics. It has a mortality of > 33%, attributed mostly to respiratory causes, with peak incidence of death being in infancy [1]. Our case series is one of three describing the anaesthesia management in 2 children with genetically proven (CANT1-gene anomaly) Desbuquois syndrome.

Case Series: Our first patient was a 21-month male child with Desbuquois syndrome undergoing bilateral leg deformity correction for knee subluxation. The child weighed 5 kg with a height of 67 cm, and the second was a 4-year male, weighing 6.4 kg and 84cm – both less than the 3rd percentile for weight and height. On examination, both displayed typical features associated with the syndrome; round flat face, microstomia, micrognathia, midface hypoplasia, depressed nasal bridge, anteverted nares and very short neck – multiple markers of a difficult airway - additionally showed ligamentous laxity, bilateral hip and knee subluxations, narrow, bell-shaped thorax, radial deviation of fingers and delayed milestones (Figure 1). Written informed consent was obtained from the patient's family prior to publication of the images. Both had hypotonia and history of multiple lower respiratory tract infections, needing hospitalization.
Pre-anaesthetic evaluation revealed potential difficulties with obtaining intravenous access, positioning, difficult airway and difficulty in placing regional blocks.

Case 1: Following premedication with nasal midazolam, inhalational anaesthesia was administered with sevoflurane and an intravenous access obtained. Our plan-A airway management was laryngeal mask airway(LMA) placement avoiding neuromuscular blockade. However, a 1.5 size i-gel LMA was ineffective due to inadequate seal. Our Plan-B was endotracheal tube (ETT) placement - Laryngoscopy was performed, CL IIb view was

obtained but a 4.0 uncuffed tube met with resistance on attempting to pass through the cords, despite a good view of the glottis. We ultimately intubated the child with a 3.5 uncuffed ETT railroaded over an 8Fr FROVA™.

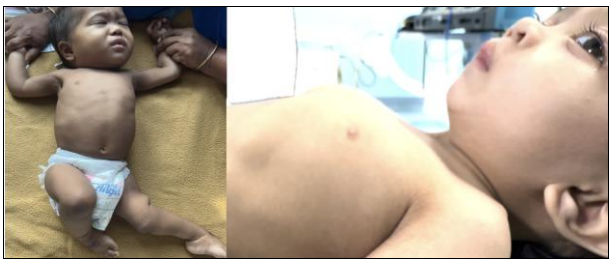

Fig 1: Typical phenotypic elements of Desbuquois syndrome – (left) Knee subluxation, Narrow bell-shaped thorax, short neck, Inward deviated toes, ligamentous laxity (right) profile view - Flattened nasal bridge, upturned nares, retrognathia.

Post intubation, a landmark guided caudal block was performed which was challenging due to the absence of classical landmarks. The surgeons faced difficulties at their end as well; tourniquets could not be used as the child's limbs were short and malrotated. The intraoperative course was uneventful and post procedure the child was shifted to the ward.

This child returned 1-month later for hip deformity correction for which a general anaesthetic with ETT intubation (3.5 uncuffed) and a caudal block was administered. This was a longer procedure; the child had respiratory distress post extubation and was shifted to the ICU where he received supplemental oxygen and nebulisation. Remainder of the post-operative period was uneventful, and child was discharged 3 days later.

Case 2: Based on our prior experience our Plan-A airway management was endotracheal intubation. Child was induced with inhalational anaesthesia and muscle relaxation achieved using Inj. Cisatracurium. Direct laryngoscopy was done, which had a CL III view; a 3.5 microcuff ETT railroaded over an 8 Fr FROVA™ was used for intubation. We opted for a smaller for age tube, based on our experience, and found it had a snug fit, so we avoided cuff inflation and checked cuff pressures (20 cmH_2O). Analgesia was provided with an ultrasound guided femoral, Lateral femoral cutaneous and anterior sciatic nerve blocks. The placement of blocks was challenging due to hip and knee dislocations causing rotational anatomical variations.

Tourniquets could not be used due to the short limb stature. The procedure went on uneventfully and the child was extubated post operatively.

Post extubation, the child was tachypnoeic with audible stridor and retractions. Despite nebulisation with adrenaline and bronchodilators, he continued to remain tachypnoeic, needing supplementary oxygen support. Hence, child was shifted to the high dependency unit.

This child was brought for the other leg deformity correction 3 weeks later and since we knew what to expect with the airway, we intubated using a 3.5 microcuff ETT railroaded over an 8 Fr FROVA™. The procedure was uneventful, and the child was moved to the high dependency unit overnight for observation.

Discussion

The challenge with anaesthetising any patient with rare syndromes lies in the paucity of information. The anaesthesiologist must rely on clinical acumen and have an alternate plan of management. In our literature search for patients with Desbuquois syndrome undergoing anaesthesia, we found only two case reports in children [2, 3]. Both reported airway difficulties - one needing to resort to an LMA as a rescue in a neonate [3] the other, had an LMA provide inadequate seal [2]. Okutani *et al* report attempting securing the airway with a Proseal LMA which failed, necessitating endotracheal intubation [2]. The table (Table 1) below documents the available literature on Desbuquois syndrome, and how it compares to our cases.

Table 1: Literature review of the anaesthesia management in patients with Desbuquois Syndrome and its clinical implication

Paper	Finding	Our experience	Clinical relevance
Lethality in Desbuquois dysplasia: three new cases [1]	Case series of 3 infants with Desbuquois syndrome all of whom died in early infancy - high rate (33%) of lethality. 10 of the 36 cases in literature who died, mostly did so between birth and 7 months due to respiratory problems	Our children both had history of repeated respiratory infections, and needed monitoring for respiratory distress post anaesthesia	Patients should be monitored closely as to their pulmonary status. Consider high dependency unit/Paediatric ICU monitoring.
Anaesthetic management in a child with Rolland-Desbuquois type dyssegmental dysplasia [2]	Case report of the anaesthesia management of a 17-month-old with Rolland-Desbuquois type of dyssegmenta Dysplasia. The report the inadequacy of seal with a Proseal LMA 2.0, and needing to intubate over a fiberoptic bronchoscope, using a 4.0 uncuffed endotracheal tube. Post surgery, tube was removed but child had needed repeated suctioning to clear airway secretions.	We also faced a similar issue with the seal of an i-gel LMA being inadequate, needing to proceed with intubation. However, the difference in our cases was the need for a small-for-age endotracheal tube in both patients.	A supraglottic airway device does not seem to provide adequate seal enough to ventilate effectively and hence, the primary plan to perform endotracheal intubation might be the most prudent airway management strategy.
Use of CobraPLATM for airway management in a neonate with Desbuquois syndrome. Case report and anaesthetic implications [3]	The case report discusses airway management in a neonate with Desbuquois syndrome where the authors avoided endotracheal intubation by using a 0.5 size CobraPLA to manage the airway. They had attempted an intubation through the LMA but due to the acute angle of access into the larynx, the tube could not be placed. The child had an uneventful post-operative course	In both our intubations, we had found the larynx to be quite anterior which corroborates with the view obtained through the LMA in this neonate.	Expect an anterior larynx and have the endotracheal tube pre-loaded onto an intubating assist to facilitate passage into the anterior larynx

Post-anaesthesia respiratory distress can be an issue when anatomical airway abnormalities, history of recurrent respiratory infections and multiple airway instrumentations all act in concert. Both our patients needed supplemental oxygen and high dependency care post operatively, and it was the opinion of the intensive care and anaesthesia teams that the hypotonia alongside the residual anaesthetic effects was the cause for the respiratory distress; this settled with time and round-the-clock nebulisation. Case reports also mention excessive secretions [2] managed with Inj. Glycopyrrolate as the antisialogogue.

Reports mention cervical spine instability; hence, neck radiographs were done which showed no cervical vertebral anomalies [5]. We maintained neutral neck position throughout the anaesthetic.

We believe an element of subglottic stenosis complicated both patients' airway; this could represent an unidentified component of Desbuquois syndrome – this information would be invaluable to any anaesthetist working with this syndrome. Thomas *et al* documents hypospadias in both their Desbuquois patients in a similar manner [4]. A formal airway assessment, however, could not be done due to cost related reasons.

Conclusion

This is a rare syndrome with implications for the anaesthesiologist in being difficult airways with a probable element of subglottic stenosis, post-operative respiratory compromise, and difficult regional anaesthesia. Additionally, these children will likely require multiple surgical procedures and anaesthetics, during their treatment course.

Conflict of Interest

Not available.

Financial Support

Not available.

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