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NAVIGATING THE AIRWAYS: STICKLER SYNDROME AND DIFFICULT AIRWAY MANAGEMENT: A CASE REPORT

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ABSTRACT **Background:** Stickler syndrome is a rare genetic connective tissue disorder characterized by craniofacial anomalies, joint hypermobility, ocular abnormalities, and hearing loss. A significant perioperative concern in these patients is difficult airway management, particularly in paediatric populations, due to micrognathia, retrognathia, and cleft palate. Effective anaesthetic planning is essential to mitigate airway-related complications and improve perioperative outcomes. **Case Report:** We present the case of an 11-year-old male with Stickler syndrome who underwent elective retinal detachment repair. The patient exhibited multiple craniofacial abnormalities, retrognathia, and a high-arched palate, classifying him as an anticipated difficult airway (ADA). Preoperative airway assessment using ultrasonography facilitated optimal endotracheal tube selection and localization of the cricothyroid membrane for potential emergency front-of-neck access (eFONA). Induction was performed using sevoflurane with spontaneous ventilation, followed by successful intubation with a C-MAC video laryngoscope. The perioperative period remained uneventful, and the patient was discharged without complications. **Conclusion:** Given the high likelihood of difficult intubation in Stickler syndrome, anaesthesiologists must adopt a structured, individualized airway management plan. Incorporating airway ultrasonography and advanced intubation techniques can significantly improve perioperative safety and outcomes in these patients.

KEYWORDS : Stickler syndrome, Difficult airway, Paediatric anaesthesia, Airway ultrasound

INTRODUCTION

Stickler syndrome is a rare, genetically linked multisystem connective tissue disorder primarily inherited in an autosomal dominant manner, with pathogenic variants in procollagen genes such as COL2A1 and COL11A1.(Mortier, 1993) The syndrome is characterized by a spectrum of clinical manifestations, including ocular abnormalities (myopia, cataracts, and retinal detachment), hearing loss (both conductive and sensorineural), joint hypermobility, and craniofacial anomalies such as midfacial hypoplasia and cleft palate.(Varandas et al., 2017) The estimated prevalence ranges from one to nine per 10,000 individuals, with an incidence of approximately one in 7,500 to 9,000 live births.(Mortier, 1993)

A significant clinical concern in Stickler syndrome is the potential for airway complications, particularly due to associated craniofacial abnormalities. Given the high-risk nature of these cases, careful perioperative planning is essential to mitigate complications related to ventilation, oxygenation, and intubation.

Despite the recognized challenges in the anaesthetic management of patients with Stickler syndrome, particularly in paediatric populations, literature on optimal airway management strategies remains scarce. Reports highlight an increased risk of anticipated difficult airway (ADA) scenarios, yet guidance on best practices is limited. This case report aims to contribute to the existing body of knowledge by detailing the anaesthetic considerations and challenges encountered in a paediatric patient with Stickler syndrome undergoing elective surgery. By sharing our clinical experience, we aim to enhance awareness and preparedness, particularly in paediatric surgical centers with limited exposure to genetic disorders, ultimately improving patient safety and perioperative outcomes.

CASE REPORT

An 11-year-old male, weighing 18 kg, presented with painless right eye vision loss secondary to retinal detachment and was scheduled for right eye scleral buckle and pars plana vitrectomy at our institution. He had a history of snoring, and his physical examination was significant for craniofacial anomalies, including low-set ears, left eye proptosis, a flattened nasal bridge, a small nose (Figure 1), pectus carinatum (Figure 2), a cleft between the third and fourth fingers, and bilateral hypoplastic fifth fingers (Figure 3). A systemic examination revealed no other abnormalities.

The preoperative anaesthetic evaluation identified an anticipated difficult airway (Figure 1), with a Mallampati grade III classification, cervical mobility <90 degrees, retrognathia, an adequate mouth opening, and a high-arched palate (Figure 4). The patient was classified as ASA Physical Status II. The potential risk of difficult airway management was thoroughly discussed with the patient's parents, and informed consent was obtained.

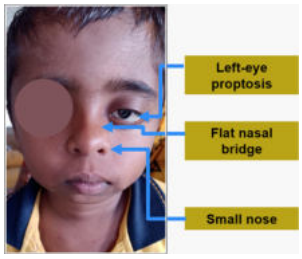

Figure 1: Craniofacial anomalies

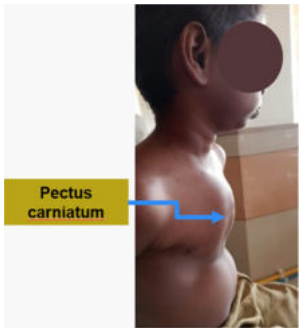

Figure 2: Inspection of chest revealing Pectus Carniatum

Figure 3: Cleft between the third and fourth fingers, and bilateral hypoplastic fifth fingers

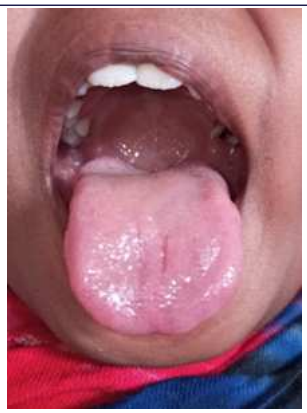

Figure 4: High arched palate

On the day of surgery, the patient was fasting for six hours for solids and two hours for clear fluids. A difficult airway cart was kept ready in the operating room. Upon arrival in the operating room, standard ASA monitors were applied. Inhalational induction was performed with 5% sevoflurane, and intravenous (IV) access was secured. The sevoflurane concentration was then reduced to 2% to maintain spontaneous ventilation. A pre-intubation airway ultrasound was conducted using a linear probe to assess the tracheal diameter (6.6 mm) for appropriate endotracheal tube (ETT) selection. The cricothyroid membrane was also marked in case emergency front-of-neck access (eFONA) became necessary. An otorhinolaryngologist was kept on standby for airway management support.

Premedication was administered with IV glycopyrrolate (0.05 mg/kg) and midazolam (0.03 mg/kg). The depth of anaesthesia was deepened with IV propofol (1 mg/kg) and fentanyl (3 µg/kg) to facilitate an initial laryngoscopic assessment. Check laryngoscopy revealed a Cormack-Lehane grade IIIA view. After confirming successful manual ventilation, IV atracurium (0.5 mg/kg) was administered, and after three minutes, intubation was performed using a C-MAC video laryngoscope with a paediatric D blade. A 6.5-mm internal diameter (ID) endotracheal tube was successfully placed, with correct positioning confirmed via auscultation and end-tidal carbon dioxide waveform analysis.

General anaesthesia was maintained with a 50:50 nitrous oxide-oxygen mixture, sevoflurane, and intermittent atracurium (0.1 mg/kg). To prevent intraocular pressure elevation, measures were taken to avoid using suxamethonium, excessive external ocular pressure from surgical drapes, a head-down position, and intraoperative hypercarbia. Hemodynamic parameters remained stable throughout the procedure. Smooth extubation was performed after the patient regained full consciousness. Neuromuscular blockade was reversed with IV glycopyrrolate (0.04 mg/kg) and neostigmine (0.05 mg/kg). The postoperative period was uneventful, and the patient was discharged on the fifth postoperative day without complications.

DISCUSSION

Stickler syndrome is a multisystem connective tissue disorder first described by Dr. Gunnar Stickler in 1960. It is characterized by ocular abnormalities (myopia, cataracts, and retinal detachment), sensorineural and conductive hearing loss, midfacial hypoplasia, cleft palate, joint hypermobility, and progressive osteoarthritis.(Heisnam et al., 2012) The syndrome is classified into three subtypes, with Stickler syndrome type 1 being the most prevalent. The inheritance pattern for types 1 and 2 follows an autosomal dominant pattern, conferring a 50% risk of transmission to offspring.(Mortier, 1993) Despite high penetrance, the clinical manifestations outside the ocular system remain highly variable.

One of the most significant perioperative concerns in patients with Stickler syndrome is airway management, primarily due to craniofacial anomalies such as micrognathia, retrognathia, cleft palate, and the Pierre Robin sequence (PRS). PRS presents with a triad of micrognathia, glossoptosis, and airway obstruction, contributing to difficult airway scenarios. Although airway obstruction associated with PRS typically improves with age, the risk of a difficult airway remains a key consideration during anaesthesia, particularly in paediatric patients.(Jones Mbbs et al., 1998) Large case series on Stickler syndrome and anaesthesia have demonstrated that most patients can undergo anaesthesia safely using standard airway management techniques; however, certain individuals present significant challenges requiring advanced airway strategies.

Studies report that approximately 40% of type 1 Stickler syndrome patients have a history of cleft repair, while PRS occurs in 13% of cases. (Zimmermann et al., 2021) Airway management is particularly challenging with Stickler syndrome as they exhibit intrinsic mandibular hypoplasia, characterized by a concave mandibular depression and a shortened ramus. Over time, both the maxilla and mandible remain proportionally underdeveloped, though mandibular hypoplasia persists.(Sadewitz, 1992; Shprintzen, 1992)

The approach to securing the airway in patients with Stickler syndrome must be meticulously planned. Inhalational induction has been widely recommended for managing difficult paediatric airways, with halothane historically used in craniofacial anomaly cases.(COOPER & MURRAY-WILSON, 1987; Johnson & Sims, 1994) Sevoflurane has since emerged as a superior alternative, providing smooth and rapid induction while allowing swift recovery in the event of airway compromise. In our case, we employed inhalational induction with sevoflurane and maintained spontaneous ventilation, ensuring the ability to assess airway patency before administering muscle relaxants. Although we did not encounter difficulties during mask induction, preparation for airway compromise was paramount, including the availability of a laryngeal mask airway (LMA) as a rescue device. LMAs have been established as effective tools for airway maintenance in patients with difficult intubation secondary to retrognathia, with reports supporting their use as a conduit for fiber optic-guided intubation.(COOPER & MURRAY-WILSON, 1987; Muraika et al., 2003; Officer et al., 2005)

While awake fiberoptic intubation is considered the gold standard for difficult airway management, it is often impractical in paediatric patients due to poor cooperation. Alternative techniques, such as retrograde intubation, remain invasive and distressing, particularly in young children.(Heisnam et al., 2012) The use of deep inhalational induction to assess airway visualization before administering neuromuscular blockade is a well-documented strategy. In our patient, initial laryngoscopy following deep inhalational induction revealed a Cormack-Lehane grade III A view, permitting safe intubation after administration of muscle relaxants. Similar approaches have been successfully employed in other craniofacial syndromes, including Treacher-Collins syndrome and multiple pterygium syndrome.(Muraika et al., 2003)

Advancements in airway ultrasonography (POCUS) have provided anaesthesiologists with valuable tools for preoperative airway evaluation. Point-of-care ultrasound has proven effective in predicting airway dimensions, guiding endotracheal tube (ETT) selection, differentiating tracheal from oesophageal intubation, and identifying the cricothyroid membrane for potential emergency front-of-neck access (eFONA).(Jagannathan et al., 2016; Veiga et al., 2023)In this case, ultrasound examination allowed us to determine the optimal ETT size, an essential consideration when using reinforced tubes with larger external diameters. Additionally, preoperative localization of the cricothyroid membrane ensured readiness for emergency airway intervention, highlighting the growing utility of airway ultrasound in difficult paediatric airways.

Ultimately, this case underscores the importance of meticulous preoperative preparation, particularly in patients with anticipated difficult airways. The integration of airway ultrasound, video laryngoscopy, and rescue airway devices significantly enhances the safety and efficacy of airway management in children with craniofacial anomalies. Our experience reinforces the necessity of a structured airway management strategy, emphasizing the role of advanced airway assessment techniques in optimizing perioperative outcomes.

CONCLUSION

This case highlights the importance of thorough preoperative assessment and preparation in paediatric patients with anticipated difficult airways. By leveraging modern airway management techniques, including point-of-care ultrasonography, anaesthesiologists can optimize airway control, reduce complications, and ensure safe perioperative care for patients with Stickler syndrome.

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