

## Branchio-otic syndrome: An opportunity to reassess the paediatric anaesthetists' approach to the difficult syndromic airway

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Branchio-oto-renal spectrum disorders are rare genetic entities with variable penetrance and concurrently display a wide phenotypic variation. A common issue to syndromic children is a propensity for difficult bag-mask ventilation, intubation or both. Unfortunately, there is no uniformity of this challenge, assessment strategy or management plan. This case with features of branchio-otic syndrome provided the opportunity to examine several aspects of paediatric anaesthetic airway management. The child was booked for branchial cyst removal but appeared to have other features of abnormal branchial cleft development. An outline of the executed anaesthetic plan is presented and discussed. Three different techniques were sequentially tried in this patient before the airway was secured. Various case reports in the literature inconsistently describe easy to very difficult airway management in children with branchio-oto-renal spectrum disorders. Branchial arch dysgenesis is almost always associated with difficult direct laryngoscopy. There is undoubtedly no singular way to ideally manage a child with a difficult airway. Many tools for difficult airways are available. However, despite the improvement of difficult paediatric airway equipment, it would appear that for the anaesthetist the flexible bronchoscope remains an indispensable tool.

**Keywords:** Branchio-otic syndrome, paediatric, difficult airway, flexible bronchoscopy

### Introduction

Astute airway management skill is inherent to the safe practise of anaesthesia. Craniofacial malformations independently predict difficult airways and are common to a large group of paediatric syndromes.<sup>1</sup> The nature of the airway management difficulty relates to the anatomical site involved in the specific syndrome. Developmental aberrations impacting the airway may occur at the level of the nasopharynx, mandible, maxilla, oral cavity, larynx, pharynx, trachea and cervical spine.<sup>1</sup> Difficulty in airway management may manifest with unsuccessful mask ventilation, laryngoscopy, supraglottic rescue or infraglottic rescue, either in isolation or combination depending on the underlying developmental deformity.

The embryological origins of the human face and airway structures derive from a series of pharyngeal arch pairs. Abnormalities of pharyngeal (branchial) arch development occur in a subset of congenital syndromes. Oculo-auriculo-vertebral spectrum (OAVS), also known as "First and Second Branchial Arch Syndrome", encompasses three related disorders existing on a severity spectrum: oculo-auriculo-vertebral disorder (OAVD) (mild), hemifacial microstomia (intermediate) and Goldenhar syndrome (severe).<sup>2</sup> Estimates of OAVS rates of occurrence as high as one in 3000 live births have been reported.<sup>3</sup>

Branchio-oto-renal spectrum disorders are related to OAVS disorders but are encountered less frequently. The spectrum includes branchio-oto-renal syndrome, branchio-otic syndrome, branchio-oto-ureteral syndrome, branchio-oculo-facial syndrome, and oto-facio-cervical syndrome.<sup>4</sup> These syndromes are generally inherited in an autosomal dominant fashion but with variable penetrance. Hence the clinical characteristics

differ considerably between affected individuals. Branchio-oto-renal (BOR) syndrome is characterised by a specific phenotype. Individuals with branchio-otic (BO) syndrome are distinguished from the prior by the absence of renal anomalies.<sup>5</sup> The estimated prevalence of the BOR/BO syndrome is 1 in 40 000, with males and females equally affected.<sup>5</sup> Branchio-ocular-facial syndrome is exceedingly rare with approximately 50 documented cases in the medical literature.<sup>6</sup>

Clinical diagnosis of BOR/BO syndrome rests on identifying key features. The five cardinal features include branchial defects, hearing loss, preauricular pits, external ear abnormalities, and renal malformations. At least two of the five features must be present in an individual with two or more affected family members, or three features are present in an individual with no affected family members.<sup>5</sup> Molecular genetic testing for mutations in the *EYA1*, *SIX5* and *SIX1* genes is available to confirm a clinical diagnosis of BOR/BO syndrome. However, demonstrable genetic mutations may be absent in up to 50% of cases.<sup>5</sup> The case described below fulfils criteria for branchio-otic syndrome on clinical grounds.

Owing to the overlap in clinical presentation of these spectra disorders and their composite syndromes, confusion about distinction of disease entities exists.<sup>4,6</sup> Moreover, there is discrepancy in the literature regarding the frequency of difficult airways in children with branchio-oto-renal spectrum disorder.<sup>7,8</sup> Embryological distortion of branchial cleft development will invariably impact direct laryngoscopy. International branchio-oto-renal registries exist,<sup>5</sup> but the prevalence in South Africa is not known. The experience with a child falling within the branchio-oto-renal spectrum provided the opportunity to review

several aspects of paediatric anaesthetic airway management. An outline of the executed anaesthetic plan is presented and discussed.

### Case presentation

A two-year-old boy, weighing 12 kg, presented to the otolaryngology department at Charlotte Maxeke Johannesburg Academic Hospital with recurrent infections of a right sided branchial cleft cyst. He appeared to have severe craniofacial abnormalities and was also known to attend the genetics, plastics, audiology and speech therapy clinics of the hospital. A previous ophthalmology review had excluded ocular pathology. Perinatal history was unremarkable and currently no obstructive sleep apnoea features were noted. Apart from hearing loss and delayed speech, remaining gross and fine motor milestones had been attained. Previous general anaesthesia at age two-months for radiological imaging of the branchial cyst was uneventful except for difficulty in establishing intravenous access. The airway was maintained with a size 1 laryngeal mask airway.

On examination, the child was small-for-age and displayed craniofacial abnormalities. These include hemi-facial microsomia, right hypoplastic jaw and a small malformed right ear (Figures 1 and 2). Airway assessment was limited as mouth opening and mallampatti scoring could not be elicited. However, neck movements were within the normal range. Auscultation of the heart and lung fields was within normal limits. Due to the abnormal facial development suggestive of branchial arch dysgenesis, extensive genetic testing was done. However, multiplex ligation-dependant probe amplification (MLPA) testing for microdeletion syndromes and subtelomeric deletions or duplications did not confirm a diagnosis. Renal function was fully investigated (urinalysis, blood tests and ultrasound) and was found to be normal. Audiology testing could not be performed



Figure 1: Facies of child with clinical diagnosis of branchio-otic syndrome illustrating hemifacial microstomia and right hypoplastic lower jaw.



Figure 2: Profile of child post-intubation showing small abnormal right ear, pre-auricular pit and indurated skin overlying an infected branchial cleft cyst.

on the abnormal right ear. On the left side, a type B tympanogram trace was elicited, indicative of middle ear pathology.

A potentially difficult airway was anticipated. Anaesthetic safety checks were completed and a variety of difficult airway adjuncts were available, including a video laryngoscope with a size 2 blade, oro- and naso-pharyngeal airways and a size 2 laryngeal mask airway. Senior anaesthetic staff and the surgical consultant were present for the induction. The surgeon had a suspension laryngoscope (Figure 3) as well as a tracheostomy set in theatre in case of difficult bag mask ventilation, inability of the anaesthetic team to visualise the vocal cords or to perform infraglottic or supraglottic rescue.

No premedication was prescribed. Under monitored conditions, an inhalation induction was provided with 8% sevoflurane in oxygen via a Jackson-Reese circuit. Once induced, the volatile concentration was immediately decreased to 2% and intravenous access attempted. Intravenous access was once again found to be difficult with initial cannulation of an aberrant radial artery on the dorsum of the hand. Eventual access was attained on the contralateral forearm. Spontaneous breathing was maintained throughout but it was possible to override respiration with an oro-pharyngeal airway and bag mask ventilation. Prior to airway instrumentation a bolus dose of propofol (25 mg) with fentanyl (25 mcg) was administered which rendered the child apnoeic.

Direct laryngoscopy with a Macintosh blade was attempted and a Cormack-Lehane grade 3 view was obtained with marked anatomical deviation of pharyngeal structures to the right. A video laryngoscope (IntuBrite™ VSL 8800) was then used and the vocal cords were visualised. It was not possible to intubate the trachea, despite using an introducer to navigate the endotracheal tube, as the vocal cords were markedly anterior. The surgeon then tried a straight rigid direct laryngoscope (suspension laryngoscope) and with varying degrees of external cricoid manipulation was able to eventually pass a 4,5 uncuffed endotracheal tube. The position of the tube was checked, found to be correct and secured. A 4,5 cuffed endotracheal tube was initially selected but subglottic dimensions failed to allow the deflated cuff to pass into the trachea.



Figure 3: Illustration of the assembly and use of the suspension laryngoscope.

Anaesthesia was maintained with appropriately titrated sevoflurane and the child was switched to the ventilator on pressure support. Adequate analgesia was given and muscle relaxants were omitted to allow intraoperative facial nerve conduction testing. A ventilatory leak around the endotracheal tube persisted but without effect on respiratory gas tensions. Following uneventful surgery, the child was woken up and the trachea extubated with the child fully awake. Postoperatively, the child was discharged to the ward. No complications occurred in the postoperative period. The child has since been seen at his first follow-up visit and is clinically well.

## Discussion

Infants and children present with anatomical, physiological and psychological challenges that differ from adults. The airways of syndromic children are deemed inherently difficult. In accordance with the ASA Task Force, a difficult airway is described as "the clinical situation in which a conventionally trained anesthesiologist experiences difficulty with facemask ventilation of the upper airway, difficulty with tracheal intubation, or both."<sup>9</sup>

Pragmatically, airway difficulties can be further stratified to identify the cause of unsuccessful airway management. Four arms need address: mask ventilation, laryngoscopy, supraglottic rescue and infraglottic rescue. Various case reports in the literature inconsistently describe easy to very difficult airway management in children with branchio-oto-renal spectrum disorders.<sup>7,8</sup> This case described difficulty with both laryngoscopy and infraglottic rescue in a child with a branchio-otic syndrome.

In this case, airway management adhered to the recommendations of a multicentre study of 1018 children with difficult airways, where it was shown that more than two direct laryngoscopy attempts was associated with high failure rates and an increased incidence of severe complications.<sup>10</sup> Guidelines stemming from this study urge that the number of attempts at direct laryngoscopy should be minimised and airway adjuvants should be considered to provide continuous oxygenation during airway attempts and aid with eventual intubation.<sup>10</sup>

As a supraglottic airway had been used successfully in a previous anaesthetic, it was available as a potential rescue or conduit

device. Even though failure rates with supraglottic devices are less likely to occur in infants and children as compared to adults, Mathis et al. highlight that congenital airway abnormalities, otolaryngologic procedures and prolonged surgical duration are factors causing the failure of supraglottic airways in the paediatric population.<sup>11</sup> Nonetheless, international authors have successfully used supraglottic airways in children with difficult airways.<sup>12,13</sup>

In this case, a failed attempt at direct laryngoscopy was followed by video laryngoscopy and when this was not successful, the surgeon managed to intubate the child with the suspension laryngoscope. Between the three attempts, bag mask ventilation was maintained. Had the third attempt not succeeded, two options could have been considered: to insert a supraglottic airway and ventilate through the airway for the duration of the procedure; or to intubate through the supraglottic device with a flexible bronchoscope.

It would appear that even for children, flexible bronchoscopy remains the gold standard tool for difficult airway management. Unfortunately, its use necessitates a steep learning curve and requires practice to maintain the skill. Furthermore, flexible bronchoscopy can only be performed in anaesthetised children and since airway tone is known to be attenuated post-induction, visualisation of the vocal cords may be more difficult. Various techniques have been described to compensate for this loss of tone: bronchoscopy through a supraglottic airway, concomitant use of nasopharyngeal airways and, most importantly, application of firm jaw thrust with all the above-mentioned techniques.<sup>12-14</sup>

In this case, a difficult airway was suspected. The necessary adjuvant airway equipment was brought into theatre and the otolaryngologist was informed to be prepared and ready to intervene. A suggested approach for future surgeries for this child where the otolaryngologist may not be present would be to proceed directly to intubate via a flexible bronchoscopy, without initial attempts at direct or video laryngoscopy.

## Conclusion

The syndromic child may present with extreme anatomic, physiologic and psychological challenges. Craniofacial abnormalities, notably branchial arch anomalies, should alert the anaesthetist that the airway may be difficult. There is undoubtedly no singular way to ideally manage the airway of these children. Continual advancement in specialised paediatric airway equipment is ongoing and it is incumbent on the anaesthetist to keep abreast of these improved devices. The outlined contingency plan in this case moved from preliminary airway manoeuvres to the use of specialised equipment. A dearth of literature highlights the role of flexible bronchoscopy in difficult paediatric airway management. To this end, flexible bronchoscopy should be available and practised as it is likely to remain the gold standard.

### Disclaimer

Parental consent for the publication was granted provided patient anonymity is maintained at all times. Ethical approval for publication of anonymised information was obtained from the University of the Witwatersrand Human Research Ethics Committee (Medical). Clearance certificate number M1810107.

### Declaration of Interest

None declared.

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