CASE REPORT

Rare presentation of bilobed posterior tongue in Goldenhar syndrome

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SUMMARY

A bilobed posterior tongue is a rare malformation with few reported cases in the literature. This anomaly has not been demonstrated in patients with Goldenhar syndrome. We report a case of a 5-month-old child with the classic signs of Goldenhar syndrome and laryngomalacia with an incidental finding of a bilobed posterior tongue. Careful assessment and monitoring are crucial, especially in syndromic babies.

BACKGROUND

Oculo-auriculo-vertebral syndrome, or also known as Goldenhar syndrome, is a rare congenital disorder. It involves the abnormality of the first and second branchial arches. Worldwide, the incidence is accounted to be from 1:3500 to 1:5600 cases. The predilection of male to female is 3:2.¹ This syndrome is linked to the incomplete development of unilateral facial components, which is associated with eye, ear and jaw anomalies. One-sided facial involvement represents almost 85% of cases. There are few related vertebral malformations too, such as cervical fusion, hemivertebrae and scoliosis. Other head and neck features that are linked to this syndrome are cleft lip and palate, macrostomia and micrognathia. In addition, malformations like webbing of the neck, short neck, tracheoesophageal fistula and also sternocleidomastoid muscle abnormalities are associated with the Goldenhar syndrome. Inguinal, urological anal and vaginal malformations have also been reported. The defects seen in the eyes are like coloboma, lipodermoids, epibulbar dermoids and microphthalmia. The prominent aural characteristics of Goldenhar syndrome are microtia, preauricular tragi and hearing loss.^{2,3}

Nevertheless, the exact cause of this syndrome is not well recognised. Both genetics and environmental risk factors are thought to be involved. Chromosomal abnormalities, as well as disruptions of the neural crest cells, are part of the aetiologies discussed in the literature. In addition, the involvement of influenza and rubella viruses during pregnancy are also associated with the development of Goldenhar syndrome. The rampant maternal diabetes is another related risk factor.⁴ Pregnant mothers who take medications and drugs like tamoxifen, isotretinoin, thalidomide and cocaine are generally susceptible to having newborns with congenital defects, including Goldenhar syndrome.⁵

The congenital bilobed posterior tongue is a rare entity and so far only one case has been reported

in current literature. In that case, a non-syndromic child has been found to have a bilobed posterior tongue with dysarthria.⁶ Complete or incomplete clefts involving anterior two-thirds of the tongue, however, have been reported numerously. This deformity can be isolated or related to syndromes like oro-facial-digital syndrome, median cleft syndrome and Klippel-Feil anomaly.⁷

We report a case of a bilobed posterior tongue in a 5-month-old child with Goldenhar syndrome and laryngomalacia, which may well be the first in Southeast Asia.

CASE PRESENTATION

A 3-month-old full-term baby was initially referred to us for suspected laryngomalacia. In retrospect, this child was born via spontaneous vaginal delivery with a weight of 1.78 kg. A preliminary diagnosis of Goldenhar syndrome was conferred in view of malformations of the left ear, absent right ear and small chin (figures 1 and 2). Mother is a 30-year-old, para 1. No significant maternal history is obtained. Her infective screening is negative. The child was then referred to our otorhinolaryngology team due to prolonged feeding time with airway obstruction. The child was initially on nasal continuous positive airway pressure (CPAP) which was failed to be weaned off. For feeding, the child was on Ryle's or nasogastric tube feeding throughout the stay in the hospital and tolerated the feeding well. Initially,

Figure 1 Facial dysmorphism showing small chin (frontal view).

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Figure 2 Facial dysmorphism showing small chin (side view).

the child was unable to be given oral feeding as the child developed desaturation episodes postfeeding and had episodes of prolonged feeding time. Child was given antireflux treatment too. No abnormal cry was observed.

INVESTIGATIONS

Direct laryngoscopy showed laryngomalacia features like tight aryepiglottic folds, floppy epiglottis and an incidental finding of a bilobed posterior tongue (figure 3 video 1). We proceeded with aryepiglottopexy, however, the tongue findings were managed conservatively. The child was monitored in paediatric intensive care unit (PICU) and required rigid/flexible bronchoscopy and supraglottopexy at the fifth month of age, which showed absent left upper bronchus with laryngomalacia and right bronchomalacia. Meanwhile, investigations of inborn errors of metabolism, chromosomal study and a pentad of congenital infections: TORCHE (Toxoplasmosis Rubella Cytomegalovirus Herpes Syphilis) were found to be normal.

DIFFERENTIAL DIAGNOSIS

Goldenhar syndrome is related by certain characteristics to other congenital abnormalities like oro-facial-digital syndrome, median cleft syndrome and Klippel-Feil anomaly. But none of

Figure 3 Direct laryngoscopic view showing posterior third tongue split.

Video 1 Video taken during direct laryngoscopy showing bilobed posterior tongue, floppy epiglottis, with laryngomalacia

these syndromes has reported a case of bilobed posterior tongue in literature, so far.

TREATMENT

We proceeded with aryepiglottopexy, however, the tongue findings were managed conservatively. The child was monitored in PICU and required rigid/flexible bronchoscopy and supraglottopexy at the fifth month of age. The child was put on CPAP postoperatively.

OUTCOME AND FOLLOW-UP

The child then recovered uneventfully and discharged home with CPAP. Currently, the child is 1 year old and is thriving well. CPAP was managed to be weaned off, and the patient is comfortable under room air. The child is also tolerating oral feeding now. Postoperatively, the Ryle's tube feeding was slowly weaned and child was allowed oral feeding at about 1 year old at a similar time as the CPAP was weaned off. A swallowing assessment done prior to that showed that the child was able to swallow. Trials of oral feeding were initiated. No aspiration signs were observed after oral feeding was commenced.

DISCUSSION

This deformity is a rare entity and has not been linked, by far, to any syndromes, unlike anterior bifid tongue. In this case, this child has been associated with Goldenhar syndrome, laryngomalacia and bronchomalacia.

A glance into the embryology of tongue reveals that tongue has contributions from all the pharyngeal arches. There are three swellings arising from the primitive pharynx at end of fourth week. The floor of the pharynx gives rise to the median tongue while the ventromedial first branchial arches form a pair of lateral tongue buds. The lateral tongue buds grow rapidly, fusing in the midline, developing into the anterior two-thirds of the tongue. The median tongue bud atrophies and forms no recognisable part of the adult tongue. The posterior third of the tongue forms from the copula which continues from the ventromedial region of the second branchial arches just below the foramen caecum.^{8,9} It is easy to postulate that this deformity occurs due to abnormal development of copula, but it is difficult to prove as the posterior one-third of a tongue is from a single medial swelling which does not fuse, unlike anterior tongue. The supraglottic region collapse is mainly due to the laryngomalacia changes seen during direct laryngoscopy and a video illustrating it is attached. Besides, the bilobed posterior tongue is a separate, incidental finding encountered during the procedure. Formation

of this deformity, however, would have also contributed to a floppy epiglottis.

The child needed CPAP more for the features of both laryngomalacia and bronchomalacia than the posterior tongue finding. The child had to be dependent on CPAP for nearly 5 months to improve gaseous exchange and to maintain good weight gain. Besides, tracheostomy was not required for any worsening airway obstruction as the child progressed well with non-invasive ventilation and surgical treatment of laryngomalacia. This case proves that a good recovery is still possible in a child with both laryngomalacia and the rare finding of posterior bilobed tongue as in other laryngomalacia babies.

Tongue lesions may be asymptomatic but can cause symptoms of haemorrhage, airway obstruction as in sleep apnoea and stridor, inadequate oral intake or cosmetic concerns. Bilobed anterior two-thirds of tongue can give rise to feeding difficulties like poor sucking reflex and speech disorders. But these lesions do not typically cause obstructive symptoms and have minimal morbidity after repair or excision.⁸ But a child with a bilobed posterior tongue is more prone to develop symptoms of respiratory distress and dysphagia. This is especially true if other associated congenital abnormalities are present, as in this case report. These posterior tongue changes can have higher morbidity than anterior lesions due to proximity with the upper aerodigestive tract potentially causing airway obstruction and swallowing difficulty.

Initially, the child had desaturations postoral feeding, which was the reason to be put on Ryle's tube feeding. Postoperatively, a swallowing test was done. The child has passed the assessment and was given trials of oral feeding. Thus, it is important for objective assessments such as swallowing test to be done prior to allowing oral feeding for a child with multiple congenital abnormalities. This is to detect any silent aspiration or dysphagia present.

Ryle's tube feeding alleviates any unwanted complications due to aspiration of milk. Therefore, these babies should be held upright and put on leaning position on the parents' or caretaker's chest. Proper feeding techniques must be explained and taught. As such, nursing in prone or lateral positions is important to displace the tongue anteriorly in cases of any airway obstruction. Another way to reduce airway compromise and hypoxia are nasopharyngeal intubation. The overall objective in this type of oropharyngeal abnormality like Goldenhar syndrome is to maintain a good respiratory effort and pattern. A more aggressive management is preferred for patients with severe or refractory upper airway compromise.

Besides the conservative therapies discussed, a few surgical options can also be considered in these types of syndromic babies. A tracheostomy is likely needed as part of the long-term management. It can be either weaned off once the patient makes a remarkable progress in terms breathing and feeding or as a permanent one. Other surgical interventions like tongue-lip

adhesion and mandibular distraction osteogenesis may be applied for refractory or rare cases.^{10 11} Will a surgery be able to give a better management option or will it do more harm than good? This question definitely needs to be justified by the overall medical conditions of the patient, especially the cardiorespiratory status.

Learning points

- ▶ Laryngomalacia is a common diagnosis encountered in paediatric settings, but not a bilobed tongue.
- ▶ A bilobed posterior tongue can be found both in a syndromic or non-syndromic child.
- ▶ Physicians should always maintain a high index of suspicion when attending to a syndromic child.
- ▶ Complications arising from a bilobed posterior tongue need to be dealt with, either conservatively or surgically.
- ▶ Proper nursing care is needed for these babies, or in severe cases, a surgical intervention would be beneficial.

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