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Unilateral vocal fold agenesis: an unusual cause of dysphonia

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Abstract. *Unilateral vocal fold agenesis: an unusual cause of dysphonia.* Unilateral vocal fold agenesis is a rare congenital laryngeal anomaly. To our knowledge, there have been no published reports about this clinical entity to date. A 6-year-old female patient was referred to our practice for persistent hoarseness of voice since birth. On physical examination, her voice was hoarse and reduced in volume. She had facial features typical of Down syndrome and macroglossia. Laryngeal examination revealed the absence of the right vocal fold. She had no further evaluation or treatment.

Case report

A 6-year-old female patient was referred to our practice for persistent hoarseness of voice since birth. Her mother showed no abnormalities during the pregnancy and the family history was unremarkable. Shortly after birth, the patient was noted to have the characteristic facial features of Down syndrome and chromosomal analysis confirmed the diagnosis of Down syndrome. Her parents reported no history of stridor, dysphagia, aspiration, or recurrent respiratory infections at any point in her life.

On physical examination, her voice was hoarse and reduced in volume. She was mentally retarded and had difficulty speaking intelligibly. She had facial features typical of Down syndrome and macroglossia. Other physical examination findings were normal.

The patient could not tolerate the direct endoscopic examination and laryngeal examination was performed by fiber optic laryngoscopy in the operation room under sedation with intravenous ketamine. Fiber optic examination revealed the absence of right vocal fold (Figure 1). Epiglottis, aryepiglottic folds, and vestibular folds were normal. Arytenoids were larger than normal and mobile (Figure 2). There was some salivary pooling. She had no further evaluation or treatment.

Discussion

Many congenital conditions that cause dysphonia are apparent at birth or shortly thereafter.[1] It is important to determine whether the onset of hoarseness occurred at birth or later. Congenital causes of dysphonia include glottic web, saccular cyst, and laryngomalacia. Other rare causes of congenital dysphonia have also been reported: agenesis of the epiglottis,[2] bifid epiglottis,[3] and laryngeal hypoplasia.[4] Although numerous publications have been devoted to congenital causes of dysphonia, to our knowledge, only one case of bilateral vocal fold agenesis has been reported in the literature; this case occurred in a patient with tuberous sclerosis.[5] This patient had presented with life-long aphonia and the laryngeal examination revealed the vocal cords to be represented by white ridges on the lateral walls of the larynx and showed no movement on electrical stimulation, suggesting bilateral vocal cord agenesis.

The embryology of the human larynx is well studied, and new findings have been reported recently. The larynx begins to develop during stage 11 of embryogenesis (the fourth embryological week). In this stage, the laryngotracheal sulcus and pulmonary sac develop from the foregut.[4,6] These two structures are the progenitors of the various elements of the respiratory system.[7]

The vocal folds develop at the level of the laryngotracheal sulcus during the eighth week of embryogenesis.[8] At this level, the conus elasticus arises caudally from cricoid cartilage and inserts on the anterior aspect of the arytenoid cartilages posteriorly and the thyroid cartilage anteriorly. The free cephalic border stretches from the arytenoid cartilage to the thyroid cartilage, forming the vocal folds.[9] It is thought that

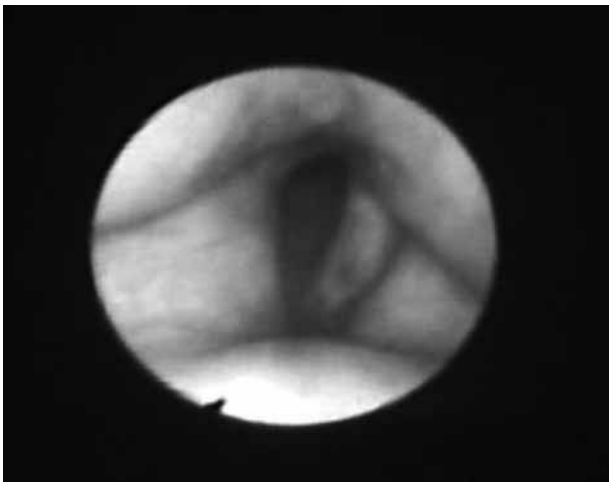

Figure 1
Fiber optic laryngeal examination revealed the absence of the right vocal fold.

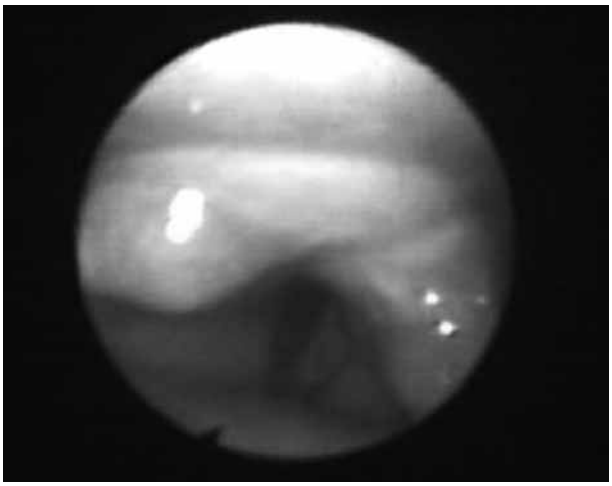

Figure 2
Laryngeal examination

interruption of growth at this period results unilateral or bilateral vocal fold agenesis.

Down syndrome is associated with numerous congenital anomalies. Upper airway anomalies occur with an increased frequency in children with Down syndrome, the most common anomaly is a decreased nasopharyngeal, oropharyngeal, or subglottic airway diameter.[10] Other upper airway anomalies associated with Down syndrome include laryngomalacia, subglottic stenosis, and tracheomalacia.[11] These abnormalities are usually associated with respiratory morbidity including stridor, recurrent pulmonary infection, respiratory failure, and sometimes with dysphonia.

Conclusion

Vocal fold agenesis is an unusual congenital laryngeal anomaly that should be considered when congenital dysphonia is encountered, especially in patients with Down syndrome.

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