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Agenesis of the epiglottis and false vocal folds with maxillary hypoplasia in an adult

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Abstract
Hypoplasia or absence of the epiglottis in an adult is a rarely reported congenital anomaly that usually occurs in association with congenital anomalies of other organ systems. Most epiglottic anomalies usually present in infancy and early childhood with respiratory and feeding problems and the affected individual dies shortly after birth due to multiple congenital anomalies. We present a case of congenital absence of the epiglottis and false vocal folds with hypoplastic maxillae in an adult.

Key words: Abnormalities; Epiglottis; Maxilla; Adult

Case report

A 20-year-old male presented with hoarseness of voice, frequent clearing of the throat and aspiration. His voice was hoarse and reduced in volume. He could not shout causing him social embarrassment. He gave no history of dysphagia, stridor nor respiratory infection.

On head and neck examination, both maxillae were hypoplastic (Figure 1) and the laryngeal prominence was not palpable (Figure 2). Other physical examinations were normal. Laryngeal telescopy showed total absence of the epiglottis and false vocal folds. The aryepiglottic folds were underdeveloped. The arytenoids were slightly injected but

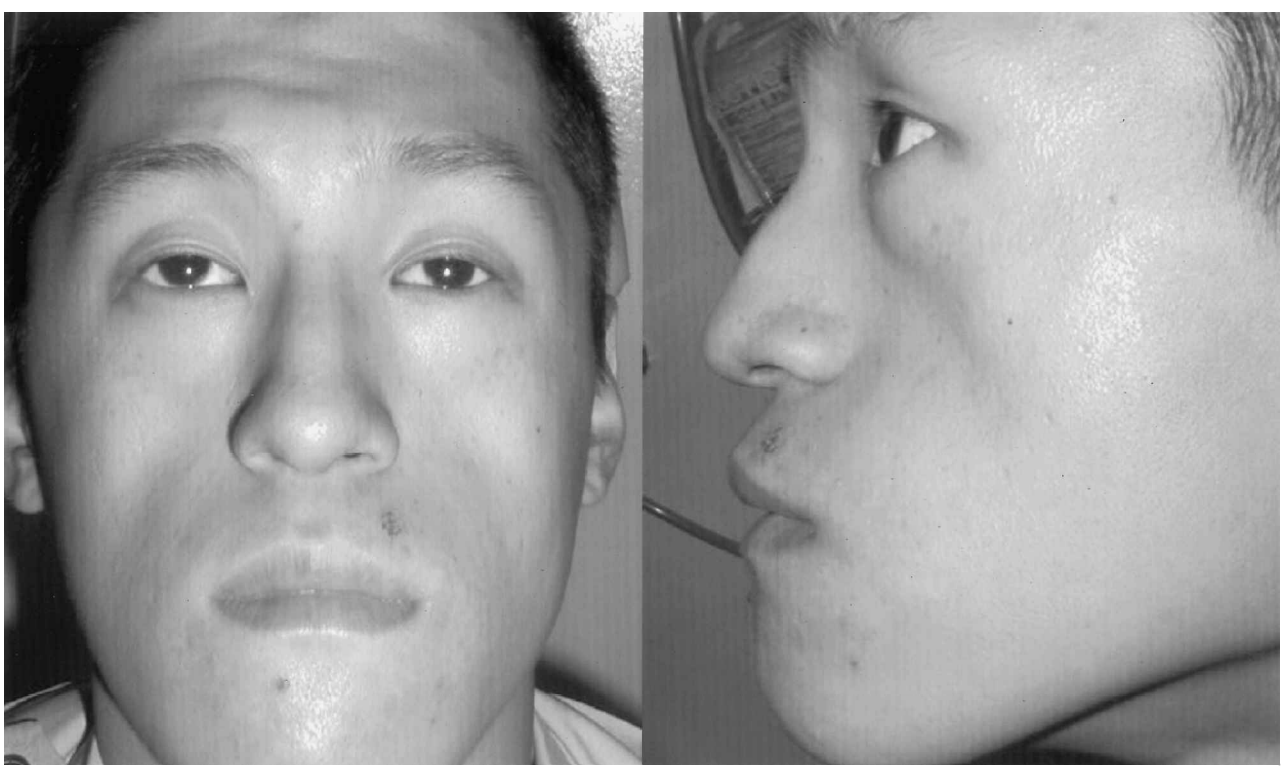

Fig. 1
Hypoplastic maxillae.

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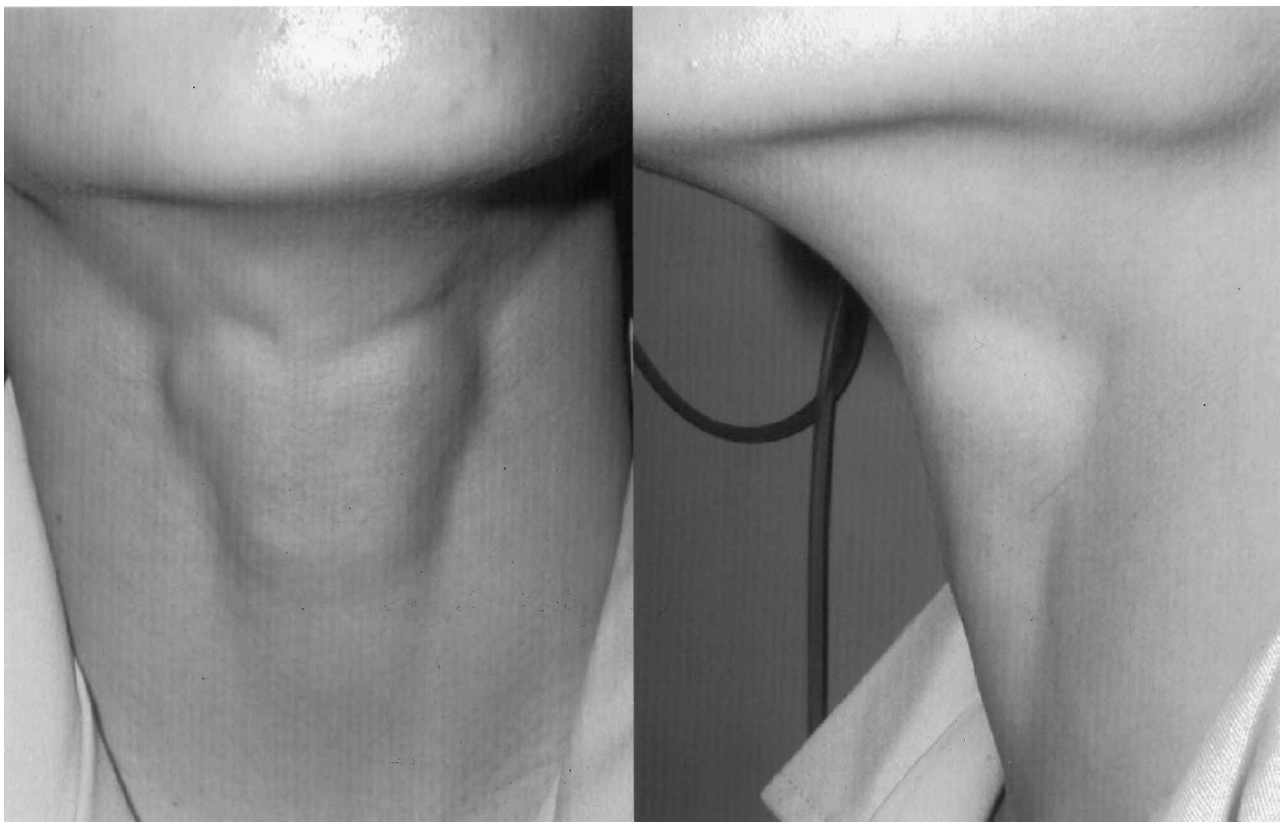

FIG. 2
Absent laryngeal prominence.

normal in size, adducting and abducting in a repetitive manner during phonation and swallowing. The true vocal folds were shortened and there was some salivary pooling (Figure 3).

Full blood count, ESR and autoimmune antibody screen were normal. Chromosomal studies revealed a normal male karyotype. Chest X-ray was normal. A barium swallow study was performed and a very small amount of barium was recognized in the larynx. His hearing was normal. At that time, no recognizable clinical syndrome was diagnosed.

A computed tomography (CT) scan revealed absence of the epiglottis and hypoplastic maxillae (Figure 4).

He had no further evaluation or treatment and was discharged.

Discussion

The congenital anomalies of the larynx include atresia, webs and stenosis. They usually present in infancy and early childhood with respiratory[1,2] and feeding problems.[3] They are often incompatible with life unless the condition is recognized at once at birth and immediate steps are taken to establish an airway.

Embryologically, the larynx begins to form during the third or fourth week of development and is usually completed at approximately nine weeks.[4] Noxious agents affecting the developing embryo at this time may cause malformations of the larynx. Although the embryology of the larynx has been well established, controversy continues regarding the development of the epiglottis. It is thought that the epiglottis arises as a swelling on the hypobranchial eminence approximately during the 32nd day of intrauterine life. During this time, arytenoid swelling and aryepiglottic folds also become apparent. It is interesting to note that the hard palate also develops during this time period. It is thought that interruption of growth at any time before this period results in the reported epiglottic anomalies, ranging from total absence, to hypoplasia, to a bifid epiglottis.[5]

Hypoplasia or absence of the epiglottis is a rarely reported congenital abnormality. Holinger *et al.*[6] reported a case of an 'extremely small' epiglottis occurring in association with a supraglottic web, but did not state

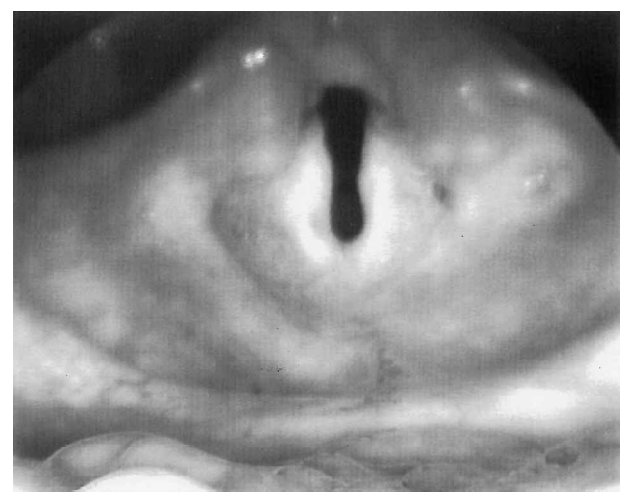

FIG. 3
Endoscopic view of the larynx demonstrating the total absence of epiglottis and false vocal fold and shortened true vocal fold.

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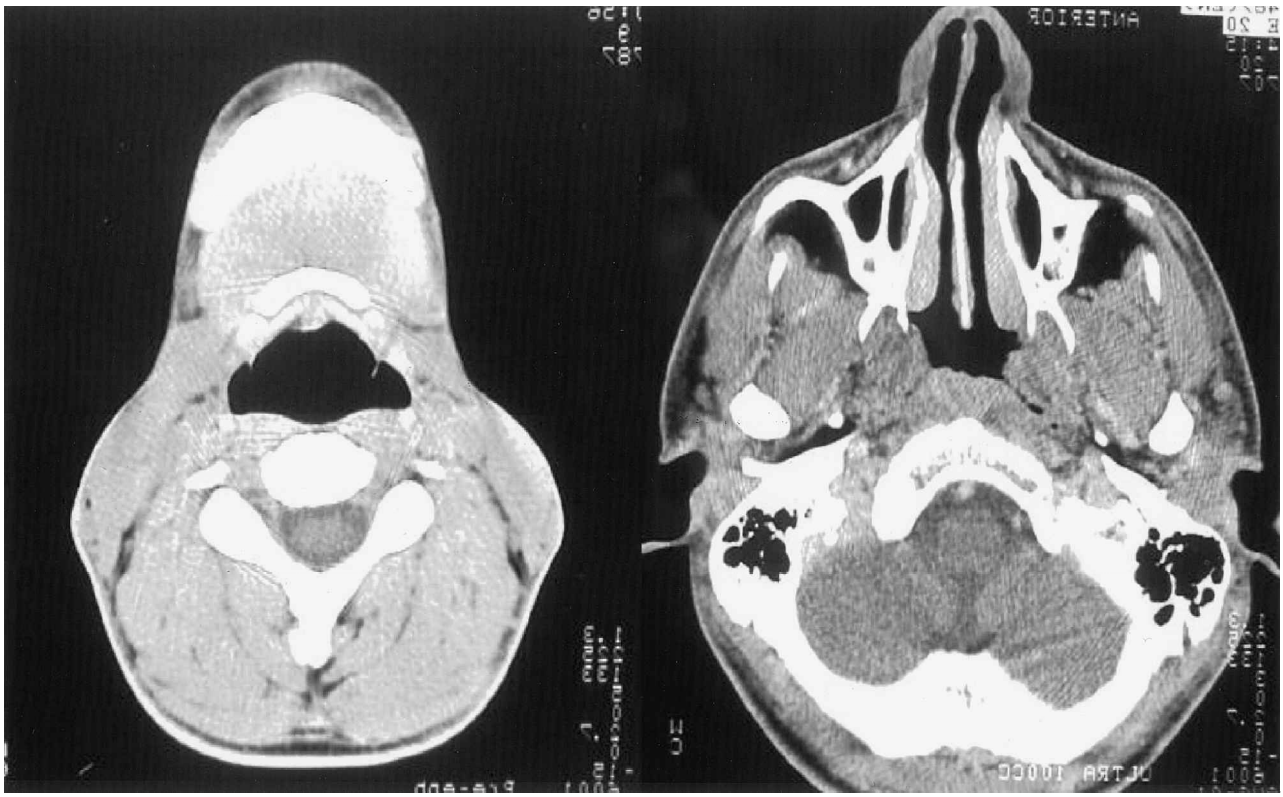

FIG. 4
Axial CT scans showing absence of the epiglottis and hypoplastic maxillae.

whether this abnormality caused aspiration or contributed to the terminal pneumonia. Bonilla *et al.*[7] described a case of aplasia of the epiglottis not associated with a recognizable syndrome or other severe laryngeal anomalies. The patient had multiple aspiration episodes that resolved in early childhood without permanent sequelae.

Absence or hypoplasia of the epiglottis has also been described in association with short rib polydactyly syndrome (SRPS) of the Majewski type.[8,9] Chen *et al.*[8] described four newborn infants with shortened limbs and ribs, polydactyly, median cleft lip, ambiguous genitalia, and hypoplastic lungs and epiglottis. Since then three other forms of SRPS have been described but abnormalities of the larynx have been described only in the Majewski type and are usually mentioned in passing merely as 'hypoplastic larynx'.[9] Knapp *et al.*[10] describe a case of Majewski syndrome illustrated by a photograph of the larynx removed at autopsy. The infant died of 'respiratory causes' and the epiglottis was severely hypoplastic; the remainder of the laryngeal anatomy appeared to be normal. Our case, with normal digits and limbs, does not fit into this category.

Our patient presented at the age of 20, with total absence of the epiglottis and hypoplastic maxillae. Despite the severity of his laryngeal anomaly, he had only a few symptoms, hoarseness of voice and frequent clearing of his throat due to aspiration. The epiglottis is functionally important in preventing aspiration. However, both the true and false vocal folds act as major factors in preventing aspiration and lethal complications of aspiration. In our case aspiration was not severe even without the presence of the false vocal folds.

Conclusion

We report a rare case of total absence of the epiglottis and false vocal folds and hypoplastic maxillae not associated with a recognizable syndrome. Total absence of the epiglottis and false vocal folds are compatible with reasonably normal basic functions of the aerodigestive tract such as airway protection and swallowing although highly specialized processes, such as the quality of voice may be affected.

- **Rare case of congenital absence of the epiglottis and false vocal folds with hypoplastic maxillae in an adult**
- **No treatment was recommended**

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