A Case Report of Aplasia of the Epiglottis and the Hyoid Bone in a Pierre Robin Sequence Patient, an Underreported Association?

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ABSTRACT

•**Introduction:** Abnormalities of the epiglottis and hyoid bone have been described in association with Pierre Robin Sequence (PRS). Embryologically, the epiglottis stems between the third and fourth branchial arches, while the hyoid bone develops from the second and third arches. The body of the hyoid bone completes its development by 4 months of age, and in fact can be seen on imaging in the majority of children after 9 months of age.

•**Case:** We present a case of an 18-year-old male known for PRS, who underwent a tracheostomy within a week of life and multiple airway evaluations since. Complete absence of his epiglottis was noted early on. Numerous computed tomographies (CT's) preformed during his lifetime with three-dimensional reconstruction of facial bones; however, only reported bony anomalies of the mandible and zygomatic arches. In a retrospective review of imaging by his surgeons and the center's radiologist, the body of the hyoid bone was found to be clearly absent from the earliest CT at 2 years of age. His greater cornua; meanwhile, remain present leading into soft tissue and potentially cystic content in the midline.

•**Discussion:** These anomalies bring up the question of an underreported association within PRS patients considering their close embryological origins. Aplasia of the epiglottis is well reported independently, but there have been very few studies on hyoid anomalies in this population. A larger series is needed to evaluate the frequency as well as potential clinical impact of these abnormalities in unison within the PRS population.

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BACKGROUND

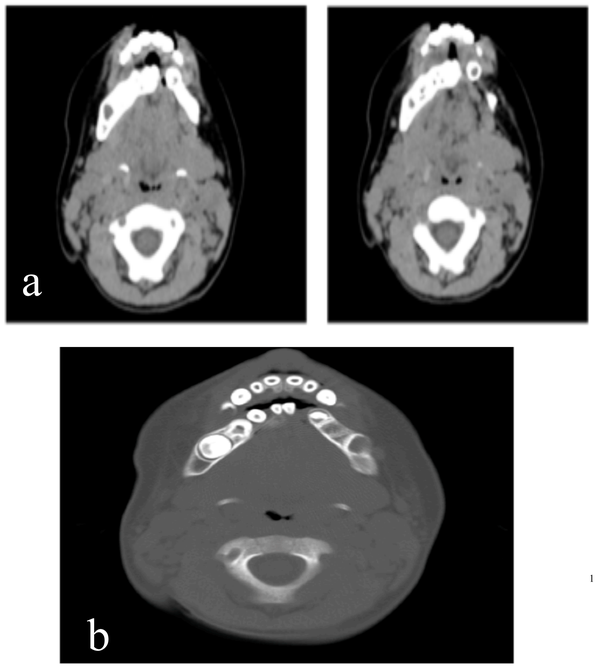
Pierre Robin Sequence (PRS) was first described by Pierre Robin in 1923 as a combination of micrognathia and glossoptosis. [1] In 1934 he revised the definition to include a triad of micrognathia, glossoptosis, and a U-shaped cleft palate[2]. Aplasia of the epiglottis is a rare anomaly, its incidence not known. Its first report within the PRS population was in 2011, in an infant that did not survive far past birth[3]. Hyoid anomalies were also reported in these patients using computed tomographies (CTs) as the body of the hyoid bone completes its development by 4 months of age, and can be seen on imaging in the majority of children after 9 months of age. One study found the majority of these anomalies occurring in the syndromic PRS patients[4].

In the embryological development of the larynx, the epiglottis stems between the third and fourth branchial arches, while the hyoid bone develops from the second and third arches[5]. In a population with various malformations, the close origin of these two laryngeal structures could indicate a more common association not yet discovered.

CASE DESCRIPTION

We present the case of an 18-year-old male known for PRS who received a tracheostomy and a gastrostomy tube (G-tube) within his first week of life. He underwent several other surgeries throughout his lifetime including multiple clubbed feet repairs, an iliac crest bone graft to his mandible at four years old, and bilateral mandibular distractions at twelve years old as well as a mandibular reconstruction at sixteen. Despite multiple jaw surgeries, he remains severely micrognathic with an element of glossoptposis and a grade IV view on repeat direct laryngoscopy evaluations as per the Cormack-Lehane system[6]. He remains tracheostomy dependent at this time, and tolerates a Passy-Muir valve during school hours with a strong voice.

Congenital absence of his epiglottis was noticed early on during in office as well as operative laryngoscopies (Figure.c-d). Despite this, repeated evaluations of feeding and swallowing, though reporting reduced mastication limiting solid intake, showed no evidence of penetration or aspiration into the airway.



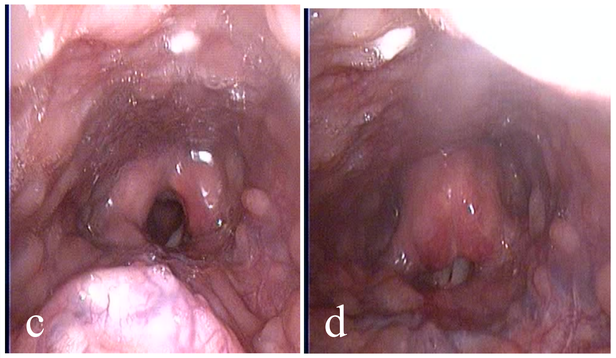


FIGURE a-b Axial CT images of the maxillofacial in soft tissue and bone windows displaying bilateral greater cornua and congenital absence of the hyoid bone.

c-d. In office flexible rhinolaryngoscopy demonstrating congenital absence of the epiglottis. Evidence of normal vocal fold functions through abduction (c) and adduction (d).

He maintains functional feeding skills with thin liquids and purees, but relies on the G-tube for complete nutrition.

He underwent multiple computed tomography's (CT's) during his lifetime with three-dimensional reconstruction of the facial bones for diagnostic and surgical planning purposes. Throughout these imaging's, absence of the zygomatic arches and hypoplasia of the mandible and bilateral anterior digastric muscles were reported. No mention was given to the nature of the hyoid bone. Going back to the earliest CT scan with our radiologist, when the patient was 2 years of age, a clear absence of the body of the hyoid bone could be appreciated (Figure a-b). The greater cornua were present bilaterally leading into soft tissue and potentially cystic content in the midline (Figure a). Remaining imaging over his lifetime were reviewed, confirming these same findings.

CONCLUSIONS

This case report describes aplasia of the epiglottis in a PRS patient, a finding mentioned independently in the literature. However, the coexisting hyoid anomaly begs the question of frequency of these coinciding findings as well as their potential clinical impact. A larger series within this population is needed to further evaluate and explore this possibly underreported association.

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