International Journal of Pediatric Otorhinolaryngology, 19 (1990) 163–168
Elsevier

PEDOT 00630

Case Reports

Congenital aglossia and situs inversus

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(Received 4 August 1989)
(Accepted 30 September 1989)

Key words: Congenital anomaly; Tongue; Mandible; Situs inversus

Abstract

Lingual agenesis is a rare anomaly caused by failed embryogenesis of the lateral lingual swellings during the fourth and fifth gestational weeks. Most reported cases have been part of oromandibular limb hypogenesis syndromes. A review of the literature reveals two previously reported cases of congenital aglossia and situs inversus. A case of lingual agenesis associated with micrognathia and situs inversus is reported in a newborn presenting with upper airway obstruction at birth.

Introduction

The first reported case of lingual agenesis with situs inversus appears in the literature in 1925 [14]. No other cases are reported until 1971 with the report of a 6-year-old girl with microglossia, hypodontia, micrognathia, and situs inversus [6]. We report a case of complete lingual agenesis, micrognathia, and situs inversus in an infant presenting with upper airway obstruction.

Case report

The patient was a 3640 g black male infant born by normal spontaneous cephalic delivery to a gravida 2, para 2, abortus 0 mother following an uncomplicated gestation. The patient developed upper airway obstruction with cyanosis and bradycardia at birth. Apgar scores were 3 and 7 at 1 and 10 min respectively. The

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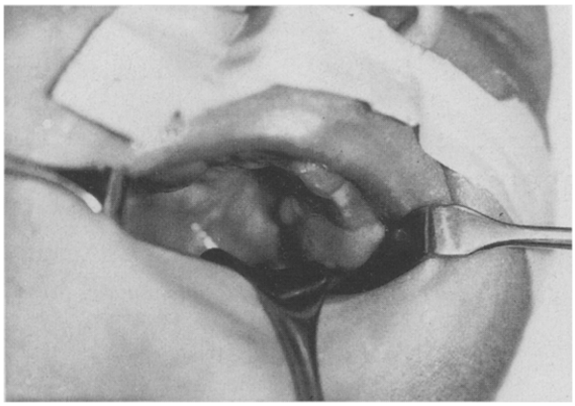


Fig. 1. Narrow palate with maxillary alveolar thickening.

airway was secured with a nasal endotracheal tube and the patient was transferred to the neonatal intensive care unit.

There was no family history of congenital anomalies and the patient had one normal sibling. The maternal history revealed no prenatal illness and there was no history of maternal drug usage.

Physical examination revealed a term male infant with a markedly underdeveloped lower face and micrognathia. The anterior mandible was shortened, narrowed, and wedge-shaped. The palate was arched with thickening of the maxillary alveolar ridge (Fig. 1). The floor of the mouth had a symmetrical mucosal lining with no evidence of an anterior tongue (Fig. 2). A small swelling was noted in the region of the normal tongue base.

The patient was taken to the operating room where laryngoscopy and bronchoscopy revealed an otherwise normal airway. Extubation was attempted at 3 days of age but the patient developed recurrent aspiration and upper airway obstruction requiring tracheotomy and gavage feeding.

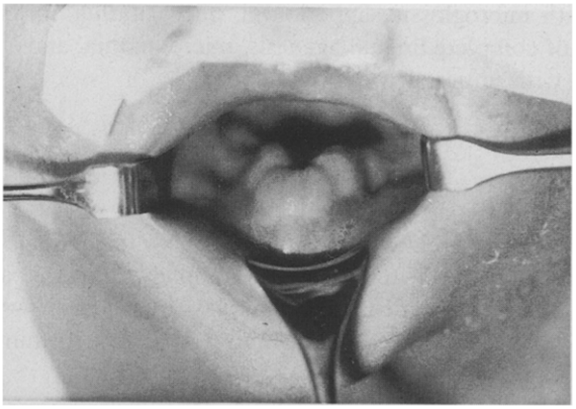


Fig. 2. Lingual agenesis and micrognathia.

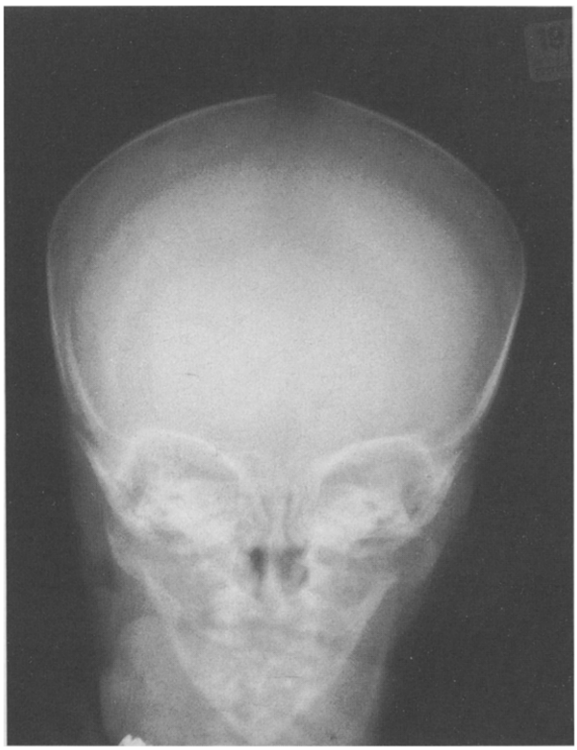
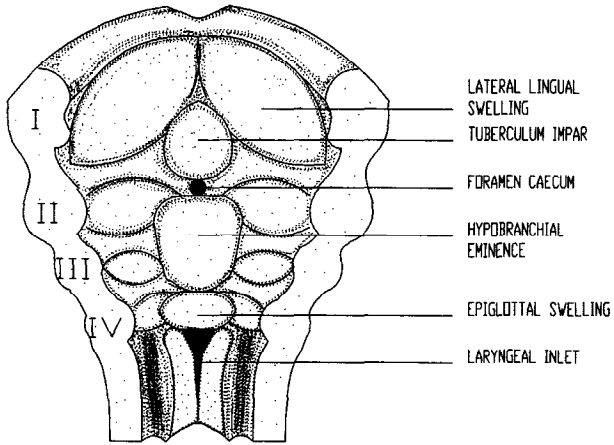


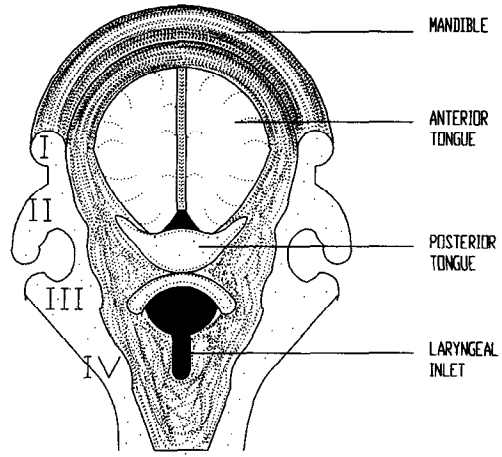
Fig. 3. Facial radiograph showing small mandible with polycystic bone.

Routine hematologic studies, thyroid panel, and urinalysis were within normal limits. Chest and abdominal series of X-rays revealed complete situs inversus and mandible radiographs showed a small arch with polycystic bone (Fig. 3). A barium esophagram revealed significant reflux with no evidence of tracheoesophageal fistula. Chromosomal analysis disclosed a 46, XY, 16 qh+ pattern. This was consistent with a normal male karyotype with one No. 16 chromosome having extra heterochromatin material and was not felt to be clinically significant.

Initially, following tracheotomy, the patient experienced relief of his airway obstruction. His gastroesophageal reflux was managed with small, frequent feedings and bethanechol. He was discharged at 3 weeks of age, tolerating nipple feedings, with his tracheotomy tube in place. He again required admission for pneumonia at age 5 weeks which resolved with antibiotic therapy. At age 6 months a developmental assessment revealed appropriate motor skills, though there was no evidence of vocalization or speech. The child passed behavioral auditory screening at birth and at age 6 months. Unfortunately, at age 10 months he experienced tracheotomy occlusion with a mucous plug while at home. He presented to the emergency room in complete respiratory arrest and died of asphyxiation. A postmortem examination was not performed.



5 th week



6 th week

Discussion

At approximately the fourth week of fetal gestation the tuberculum impar appears as the first evidence of lingual development. The lateral lingual swellings arising from the first branchial arch on each side of the tuberculum impar enlarge, fuse, and give rise to the anterior two-thirds of the tongue (Fig. 4). The tuberculum impar is overgrown by the lateral lingual swellings and does not contribute significantly to the adult tongue. The posterior one-third of the tongue arises from the hypobranchial eminence of the third and fourth branchial arches. Failure of development of the lateral lingual swellings results in the absence of the anterior two-thirds of the tongue, and is usually associated with other first arch anomalies including micrognathia [4].

Most reported cases of lingual hypoplasia have been associated with limb anomalies. Hall, in 1971, classified oromandibular limb hypogenesis syndrome (OLHS) and described 5 subtypes (Table I) [5]. Numerous references appear in the literature reporting the hypoglossia–hypodactylia syndrome, which Hall classifies as type II OLHS [1–3,7–10,12,13]. Type I OLHS includes lingual hypogenesis without associated limb anomalies, under which we would classify our current case and the two previously reported cases of aglossia–micrognathia associated with situs inversus.

Familial patterns have not been reported in previous cases of congenital aglossia with situs inversus and no maternal factors can be identified.

Several possible etiologic factors have been cited in the hypoglossia–hypodactylia syndrome (OLHS Type III) including drugs, intrauterine factors, and genetic factors [4,5,9,11,13]. Whether or not the various findings included in Hall's classification have a similar genetic basis is not clear, though it is likely that OLHS represents a spectrum of disorders.

TABLE I

Hall's classification of lingual hypogenesis

Type I	Type IV
A. Hypoglossia	A. Intraoral Bands
B. Aglossia	B. With Hypoglossia
	C. With Hypoglossia–Hypodactylia
Type II	D. With Hypoglossia–Hypomelia
A. Hypoglossia–Hypodactylia	E. With Hypoglossia–Hypodactylomelia
B. Hypoglossia–Hypomelia	
C. Hypoglossia–Hypodactylomelia	Type V
	A. Hanhart Syndrome
Type III	B. Charlie M. Syndrome
A. Glossopalatine Ankylosis	C. Piere Robin Syndrome
B. With Hypoglossia	D. Möbius Syndrome
C. With Hypoglossia–Hypodactylia	E. Amniotic Band Syndrome
D. With Hypoglossia–Hypodactylomelia	

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