Severe Congenital Hypoglossia: A Case Report

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Hypoglossia is referred to a small volume and/or size of the tongue. It is a rare congenital condition caused by failed embryogenesis of the lateral lingual swellings and tuberculum impar during the fourth to eighth weeks of gestation. The anomaly has often occurred in association with limb abnormalities and various syndromes, and it affects facial and mandibular growth. The present report describes a case of severe congenital hypoglossia in a female infant, her systemic and dentofacial features, and the initial management .
Keywords: *Congenital microglossia, hypoglossia, aglossia, child.*

INTRODUCTION

Congenital microglossia, or hypoglossia, is a rare condition caused by failed embryogenesis of the lateral lingual swellings and tuberculum impar, and it is manifested by the presence of a small tubercle or a rudimentary tongue. This anomaly can be present either isolated or, more commonly, associated with diverse systemic abnormalities or syndromes, including a wide spectrum of oromandibular and upper and lower limb hypoplasias.[1] Most reports of tongue anomalies have described its occurrence together with other malformations, particularly hypodactyly or adactyly.[2,3] Hall classified these entities as oromandibular-limb hypogenesis syndrome (OLHS), having five different groups, and he described a wide range of afflictions exhibiting hypoglossia in common.[3] In the case of hypoglossia, the lower third of the face is underdeveloped, resulting in a retrusive profile. In more severe cases, infants exhibit important complications, especially feeding problems, with repetitive episodes of life-threatening bronchoaspiration. This condition is uncommon, and most cases are sporadic in nature without gender preference or known etiology.[4] Diagnosis is based only on clinical findings. The purpose of the present report is to describe a case of severe congenital hypoglossia in a female infant, her systemic and consequent dentofacial features, and the initial delivered management.

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CASE REPORT

A 3-day-old female of Mexican origin presented to our department. Patient was the firstborn of a healthy 14-year-old girl; the mother mentioned past history of a brother with unilateral microtia-atresia and two paternal cousins with unilateral cleft lip and palate. The father was a 28-year-old drug addict, with a long-standing involvement to marijuana, tobacco, alcohol, and inhalants. The mother reported no drug ingestion during gestation, but was exposed to the second-hand marijuana and tobacco smoke of her partner. Besides, the mother had urinary infection during her first trimester, and ampicillin was prescribed. Gestation lasted 39.4 weeks, delivery was vaginal; the infant weighed 2,480 g, and she had a head circumference of 35 cm. Apgar and Silverman-Anderson tests scores were 8 and 0-0, respectively. The patient cried at birth; on physical examination, some clinical features were noticed: dolichocephalic skull, slightly imbricated sutures, normotensive anterior fontanel, broad nose with permeable nostrils, low-set and permeable ears, and remarkable micrognathia (Figure 1). Upper and lower limbs were normal. X-rays, neurological and heart, and CT assessments did not exhibit any pathology. Intraoral examination showed a number of findings: normal hard palate; short soft palate with some deviation to the right and fusion to the tonsillar pillar; velopharyngeal insufficiency; on the posterior zone of the mouth´s floor, a small tubercle resembling a tongue of only 1/3 normal size was fused to the alveolar process (Figure 2); besides, the patient exhibited loss of vestibular sulcus. Initial management of the child was addressed to maintain nutrition and an airway. Immediately after birth, the girl started feeding with formula milk, but she exhibited difficulties suckling and swallowing: in each attempt the liquid was expelled through the nostrils, accompanied by severe cyanosis. Therefore, patient was fed via nasogastric tube. Fifteen hours later, hypoglycemia of 27 mg/dL was detected, so 10% glucose solution was administered. After 25 days, the girl was discharged, maintaining the formula feeding through the nasogastric tube; a gastrostomy was performed in order to replace the first tube. Chromosomal analysis of

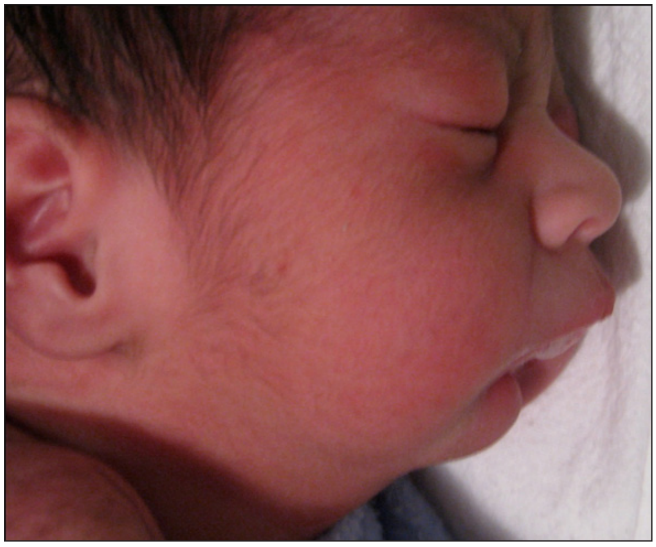

Figure 1. Lateral view of the infant revealing severe micrognathia, dolichocephalic skull, slightly imbricated sutures, low-set and permeable ears.

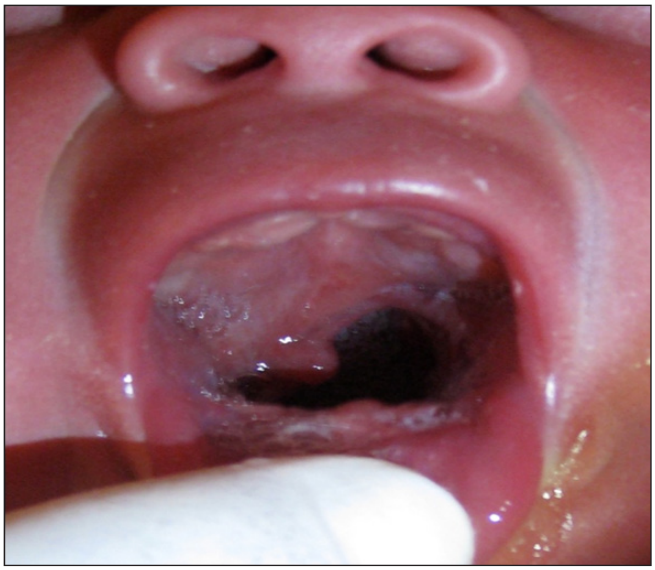

Figure 2. Intraoral examination showing normal hard palate, short soft palate with slight deviation to the right and fused to the tonsillar pillar, velopharyngeal insuffiency, and loss of vestibular sulcus.

the peripheral lymphocytes using G-T-C high resolution banding technique, revealed normal 46,XX karyotype. Metabolic screening and complete thyroid tests were normal. At nine months, the girl showed aspiration pneumonia which resolved satisfactorily. Psychomotor development was normal until then. Subsequently lost to follow up.

DISCUSSION

Congenital malformations involving the tongue are considered rare and may be associated with several syndromes of variable spectra, which can cause important comorbidities in the affected patient. Since the first publication in 1718,[5] the medical and dental literature has reported only a few cases of severe hypoglossia. In the present case, a severe hypoglossia with other associated abnormalities is reported.

The term *aglossia* has been applied to the complete absence of the tongue, whereas the terms *microglossia* and *hypoglossia* are used to describe a rudimentary tongue or small tubercular appendix. However, all these terms have been indistinctly employed to describe the same condition, which makes the review of reported cases difficult. We consider the term *aglossia* misapplied, since some embryologic tongue remnants are always present, either rudimentary or tuberculate, localized in the genioglossus process zone; that is why we believe *hypoglossia* to be the correct term. Furthermore, Salles *et al* stated that complete aglossia is practically incompatible with life, because nourishment is not possible without the suckling reflex.[3] Although its etiology is unknown, it has been mentioned that hypoglossia is frequently associated with genetic defects,[6] in which neural development is especially vulnerable to significant structural and functional alterations. This is particularly true during the neurogenesis and maturity stages, even when exposure to external or environmental influences is brief.[7-9] Other risk factors have been suggested, such as intrauterine traumatism, as in obstetric chorionic villus sampling, which may seriously damage fetal blood vessels and hemorrhagic injuries, especially when these samples are obtained between the 56th and 66th days of gestation.[1,10] These vascular lesions are most likely to occur in the distal zones of the tongue or the limbs, and occasionally in some parts of the brain.[11] Ingestion of teratogenic, antimicrobial, abortive, or sedative drugs during gestation predisposes to this anomaly, as does radiation exposure, for example, in the Moebius syndrome or arthrogryposis.[4,12] Another finding reported in the literature is that in most of these cases, the mothers are primigesta, less than 18 years old, and significantly younger than the fathers. Recently, Kantaputra and Tanpaiboon reported a case of hypoglossia related to hypothyroidism.[13] In the present case, we speculated that hypoglossia could be associated with marijuana addiction of the father, because there was not a significant familiar past history and her genetic studies (metabolic and neonatal thyroid screenings, and karyotype) did not show any evidence of abnormalities. On the other hand, several syndromes have been related to hypoglossia, for instance, Hanhart syndrome and aglossia-adactylia syndrome;[14-16] these conditions were discarded during diagnosis because of the absence of limb malformations. Mishima *et al* reported a case of micrognathia and rudimentary tongue, with loss of the anterior alveolar process of the mandible and low-set ears, findings coincident with the present case.[17] Thorp *et al* described a five-case series of extreme hypoglossia with micrognathia, respiratory difficulties, and feeding complications, two requiring nasogastric tube-feeding.[18] This was later converted to gastrostomy tube-feeding, as in our patient.

The literature contains information about the different classifications of oromandibular-limb hypogenesis. Among those classifications are those suggested by Hall; based on this classification, we consider our patient to belong to type I.[2] Likewise, Gorlin *et al* stated that it is almost impossible to categorize all these conditions separately, because many cases exhibit a mixture of diverse clinical features characteristic of two or more syndromes.[18]

Other reports and findings are presented in Table 1.

The prognosis of these cases is definitely negative, because hypoglossia and micrognathia may impair maxillofacial growth. Other associated conditions that impact on prognosis are microstomy, cleft lip and palate, partial or total anodontia, atrophy or fusion of alveolar ridges, pseudohypertrophy of salivary glands, palpebral defects, facial asymmetry, and cranial nerve injuries; mental disability is uncommon. Fundamental functions like speech, mastication, and swallowing are

Table 1. Some features Associated with Hypoglossia or Aglossia

	Condition	Associated Features
Gathwala *et al*[16]	Hypoglossia	Severe micrognathia, retrognathia, low-set ears, cleft palate, hypodactyly, micropenis, and cryptorchidism
Elaloui *et al*[15]	Hypoglossia	Hypodactyly, dextrocardia, septal defect, unique ventricle, and atrial visceral situs solitus
Ramirez-Cheyna *et al*[11]	Aglossia	Oromandibular hypogenesis, facial paralysis, deformity of the extremities
Kantapura *et al*[13]	Aglossia	Micrognathia, microstomy, microcephaly, and hypothyroidism
Amor *et al*[4]	Hypoglossia	Situs inversus
Mandai *et al*[14]	Aglossia	Jejunoilial atresia
Grippaudo and Kennedy[10]	Aglossia	Oromandibular and limb hypogenesis with signatia
Higashi and Edo[19]	Aglossia	Esophageal atresia, esophagus epiglottis, palpebral ptosis, and deafness
Mishima *et al*[17]	Hypoglossia	Micrognathia, lip attached to the mandibular gingiva, hypoplastic tongue, defect in right elbow, syndactyly of right foot, and brachydactyly of left foot
Arshad and Goh[20]	Hypoglossia	Severe micrognathia with anterior maxillomandibular fusion. Bilateral absence of forearm, absence of big toe of the right foot and the medial 4 toes of the left foot
Elzay and Van Sickels[21]	Hypoglossia	Hypognathia, microstomia, left hand absent, right hand with only first and fifth digits, both feet with total adactyly
Hoggins[22]	Aglossia	Fusion of the jaws, deformity of the lips, syndactyly of left hand, gross failure of development of right hand and congenital amputation of the foot

usually impaired, especially during infancy and adolescence. Moreover, hypoglossia affects facial aesthetics and thereby may curtail psychological and social development; many patients exhibit a high adaptive capacity for their anomaly.

Current dental management involves a corrective occlusal approach by means of mandibular distraction, palatal expanders for mandibular expansion, and surgical procedures.[3] Fixed orthodontic appliances permit treatment of micrognathia and mandibular hypoplasia, as well as the severe narrowing and transverse space deficit of the mandibular arch, thus improving the long-term dentofacial prognosis.

CONCLUSION

Congenital hypoglossia is a rare condition reported in the medical and dental literature. In general, involvement of the pediatric dentist in a multidisciplinary team committed to full rehabilitation is an important resource to achieve good quality life for the patients and their families. It is necessary an appropriate early management for this abnormality, thus avoiding or reducing life-threatening complications.

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