Oromandibular and limb hypogenesis syndrome: treatment report

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The oromandibular and limb hypogenesis syndrome is characterized by aglossia or hypoglossia presenting with limb anomalies. In this case report, we describe congenital hypoglossia associated with glossopalatine ankylosis and middle finger hypomelia, a type III-D malformation in the Hall Classification. The orthodontic and surgical treatment consisted of 3 maxillary expansions, mandibular surgery, and dental alignment. This extensive treatment resulted in acceptable function and esthetic appearance; however, because of lingual hypoglossia, it did not effect perfect occlusion. (Oral Surg Oral Med Oral Pathol Oral Radiol 2013;116:e230-e236)

Tongue malformations are congenital structural changes that may vary in size, location, causation, and effect; they may be isolated or multiple.[1,2] According to Neville et al.,[3] hypoglossia is defined as an anomalous small tongue. It may be difficult to diagnose when it is mild in degree and occurs in isolation. Rarely, the tongue may appear to be virtually absent.

Often hypoglossia represents part of the oromadibular and limb hypogenesis syndrome (OLHS). Affected patients present absence of fingers and partial or total limb hypoplasia. The aim of this study is to report a different method of mandibular distraction osteogenesis for treating such patients with hypoglossia.[4,5]

CASE REPORT

At the Pediatric Otolaryngology Clinic at Federal University of São Paulo, Escola Paulista de Medicina, UNIFESP/EPM, a 15-year-old female patient was diagnosed with congenital hypoglossia associated with glossopalatine ankylosis and middle finger hypomelia, which was classified by Hall in 1971 as type III-D (Table I; Figures 1 and 2).[6]

The patient's father was an alcoholic and the mother did not attend any prenatal consultation regarding alcohol use or abuse during pregnancy. Cesarean delivery at term (40 weeks) produced a 2800-g, 40-cm newborn with Apgar scores of 7 at 1 minute and 9 at 5 minutes. The father was 32 years old and mother 29 years at birth and the patient was their third daughter. The first attempt at breastfeeding revealed the tongue abnormality. The patient was initially fed via a nasogastric tube, but she rejected this after 2 months and subsequently had difficulty adjusting to bottle feeding. Her motor and speech development was slightly delayed: speaking at 12 months, and walking at 18 months. In the first year of life, her weight was below normative values because of difficulty swallowing. She had pneumonia and problems breathing throughout her childhood.

On physical examination, she exhibited mild middle finger hypomelia. The oral cavity was separated from the oropharynx by a thin membrane (persistent buccopharyngeal membrane) with a central orifice that permitted feeding and phonation. On intraoral examination she exhibited severe maxillary and mandibular hypoplasia with micrognathia, excessive horizontal overjet,

Table I. Hall classification

Type1
A) Hypoglossia
B) Aglossia
Type 2
A) Hypoglossia—hypodactylia
B) Hypoglossia—hypomelia
C) Hypoglossia—hypodactylomelia
Type 3
A) Glossopalatine ankylosis
B) With hypoglossia
C) With hypoglossia—hypodactylia
D) With hypoglossia—hypomelia
E) With Hypoglossia—hypodactylomelia
Type 4
A) Intraoral band and fusion
B) With hypoglossia
C) With hypoglossia—hypodactylia
D) With hypoglossia—hypomelia
E) With Hypoglossia—hypodactylomelia
Type 5
A) The Hanhart syndrome
B) Charlie M syndrome
C) Pierre Robin syndrome
D) Moebius syndrome
E) Amniotic band syndrome

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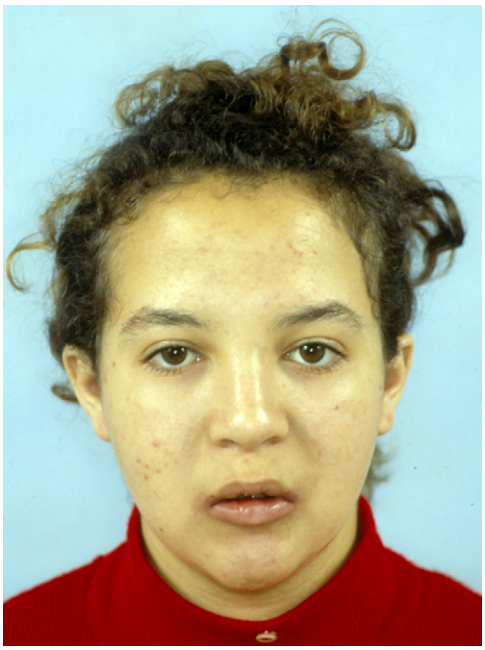

Fig. 1. Frontal face photograph.

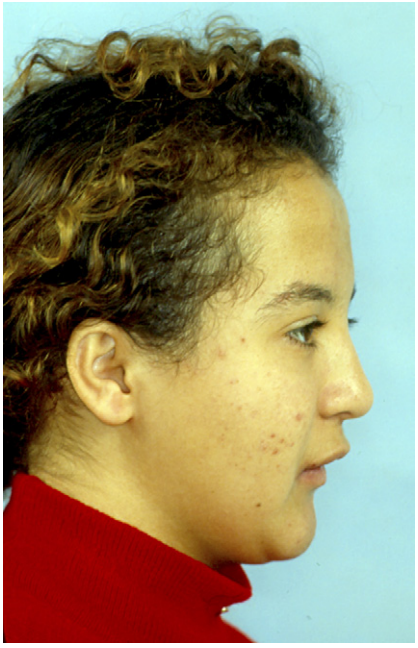

Fig. 2. Lateral face photograph.

severe tooth crowding, bilateral posterior crossbite, and agenesis of 5 permanent teeth (1 incisor, both canines and second mandibular molars). The soft palate had a submucous cleft and a bifid uvula and the anterior two-thirds of the tongue was underdeveloped and limited to the floor of the mouth with inability to protrude or move laterally. There was a persistent buccopharyngeal membrane that joined the soft palate to the inferior

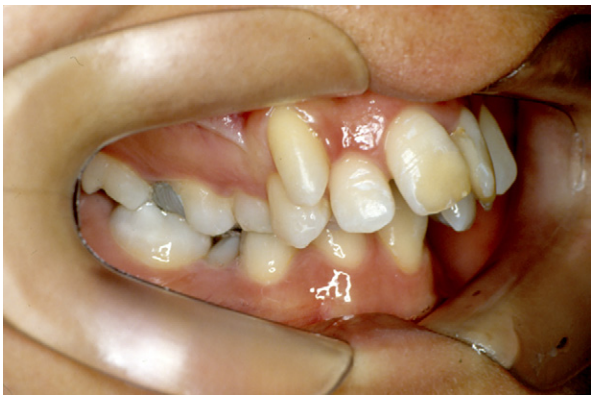

Fig. 3. Right intrabucal photograph.

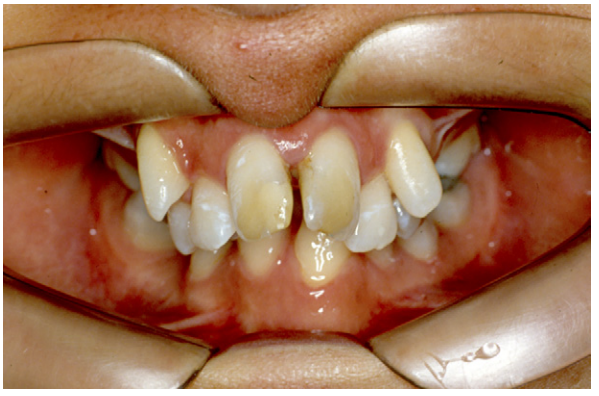

Fig. 4. Frontal intrabucal photograph.

anterior two-thirds of the tongue laterally and superiorly, partially isolating the oral and the pharyngeal cavities (Figures 3-8).

The patient phonated and ate through a tear-drop–shaped orifice in the center of the bucopharyngeal membrane. The orifice was visible only during speech when the bifid uvula and part of the pharyngeal cavity could be observed through it. In addition to hypoglossia, the patient exhibited glossopalatine ankylosis and hypertrophy of the muscles of the floor of the mouth. She breathed through her nose and was able to swallow, though abnormally. The temporomandibular joint functioned normally, but the upper lip was hypotonic and the lower lip everted. Abnormal speech was characterized by omission of linguoalveolar and linguopalatal phonemes and an altered voice quality. Neurological and psychomotor assessments were normal.

Cephalometric teleradiography disclosed a convex bone profile, a correct relationship between apical bases, with mandibular and maxillary retrusion, and hyperdivergent facial pattern; the radiographs disclosed an incorrect anterior interdental relation of the upper incisors, vestibular inclination of the maxillary incisors, and lingual inclination of the mandibular incisors (Figures 9 and 10).