Complete and isolated congenital aglossia: case report and treatment of sequelae using rapid prototyping models

Frederico Salles, DDS,[a] Marcos Anchieta, DDS,[b] Patrícia Costa Bezerra, MS,[c] Maria Lúcia G.M. Torres, MS,[d] Elizabeth Queiroz, PhD,[e] and Jorge Faber, PhD,[f] Brasília and Rio de Janeiro, Brazil
%UNIVERSIDADE CATÓLICA DE BRASÍLIA, UNIVERSIDADE DE BRASÍLIA, AND UNIVERSIDADE FEDERAL DO RIO DE JANEIRO

Aglossia is a rare anomaly caused by failed embryogenesis of the lateral lingual swellings and tuberculum impar from the fourth to eighth gestational weeks. Most cases of aglossia and hypoglossia reported in the literature were associated with limb deformities, cleft palate, deafness, situs inversus, and several syndromes, such as Moebius, Pierre Robin, and Hanhart. This report describes the case of a 14-year-old girl with complete aglossia. As the tongue plays an important role in facial growth, this patient had dentofacial deformities that affected the mandible in particular. She also had severe malocclusion and agenesis of permanent mandibular incisors. Thyroid dysfunction, recently associated with aglossia, was not observed. The use of rapid prototyping models of the jaws as an aid to osteogenic distraction of the mandibular symphysis is also described. **(Oral Surg Oral Med Oral Pathol Oral Radiol Endod 2008;105:e41-e47)**

Aglossia is an extremely rare condition caused by failed embryogenesis of the lateral lingual swellings and tuberculum impar from the fourth to eighth gestational weeks.[1-4] Most cases of aglossia and hypoglossia reported in the literature were associated with limb deformities, cleft palate, deafness, situs inversus, and several syndromes, such as Moebius, Pierre Robin, and Hanhart. As the tongue plays an important role in facial growth, dentofacial deformities, particularly affecting the mandible, are usually observed. Thyroid dysfunction was also recently associated with aglossia.[5]

The term aglossia refers to the congenital absence of the entire tongue, whereas microglossia and hypoglossia refer to abnormal smallness of the tongue. However, both terms have been used to describe the same entity in the literature, which complicates a review of cases of true aglossia. The first case of hypoglossia was reported in 1718 by de Jussieu,[6] who described "how a girl born without the tongue performed functions which depended on this organ." The earliest report on the association of tongue anomalies and limb malformations is ascribed to Kettner[7] in 1907, but his patient also had glossopalatine ankylosis.[8] It was Rosenthal,[9] in 1932, who first described a true case of aglossia-adactylia, with cleft of the lower lip, absence of the maxillary incisors, and only a small rudimentary tongue. The case reported by Rosenthal was the fifth in the 2 centuries since de Jussieu's report. Since then, most reports of tongue anomalies have described its co-occurrence with other malformations, particularly adactylia and hypodactyly in hands or feet, or in both.[10-26] Glossopalatine ankylosis; situs inversus[2,4]; deafness[27,28]; and Moebius, Hanhart,[29,30] and Pierre Robin syndromes have also been reported in association with aglossia. In 1971, Hall[8] classified these entities into 5 different groups, all under the generic term Syndromes of Oromandibular and Limb Hypogenesis (Table I).

This report describes a case of aglossia (Table I, Type I-B) and consequent dentofacial deformities without any other comorbidity except for agenesis of the 4 mandibular incisors, both in primary and permanent dentitions. It also describes the use of rapid prototyping models as an aid to planning osteogenic distraction of the mandibular symphysis.

[a]Private practice; former Visiting Professor at School of Medicine, Universidade de Brasília, Brasília, Brazil; former Head of the Maxillofacial Surgery Department, Hospital do Aparelho Locomotor–Sarah, Brasília, Brazil.
[b]Private practice.
[c]Professor, School of Nutrition, Universidade Católica de Brasília, Brasília, Brazil.
[d]Professor and Associate Researcher, Universidade de Brasília, Brasília, Brazil.
[e]Professor, Institute of Psychology, Universidade de Brasília, Brasília, Brazil.
[f]Private practice.
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Table I. Hall's Classification: Syndromes of Oromandibular and Limb Hypogenesis[8]

Type I
A) Hypoglossia
B) Aglossia
Type II
A) Hypoglossia-Hypodactyly
B) Hypoglossia-Hypomelia (Peromelia)
C) Hypoglossia-Hypodactylomelia
Type III
A) Glossopalatine Ankylosis (Ankyloglossum Superius Syndrome)
B) With Hypoglossia
C) With Hypoglossia-Hypodactyly
D) With Hypoglossia-Hypomelia
E) With Hypoglossia-Hypodactylomelia
Type IV
A) Intraoral Bands and Fusion
B) With Hypoglossia
C) With Hypoglossia-Hypodactyly
D) With Hypoglossia-Hypomelia
E) With Hypoglossia-Hypodactylomelia
Type V
A) Hanhart Syndrome
B) Charlie M. Syndrome
C) Pierre Robin Syndrome
D) Moebius Syndrome
E) Amniotic Band Syndrome

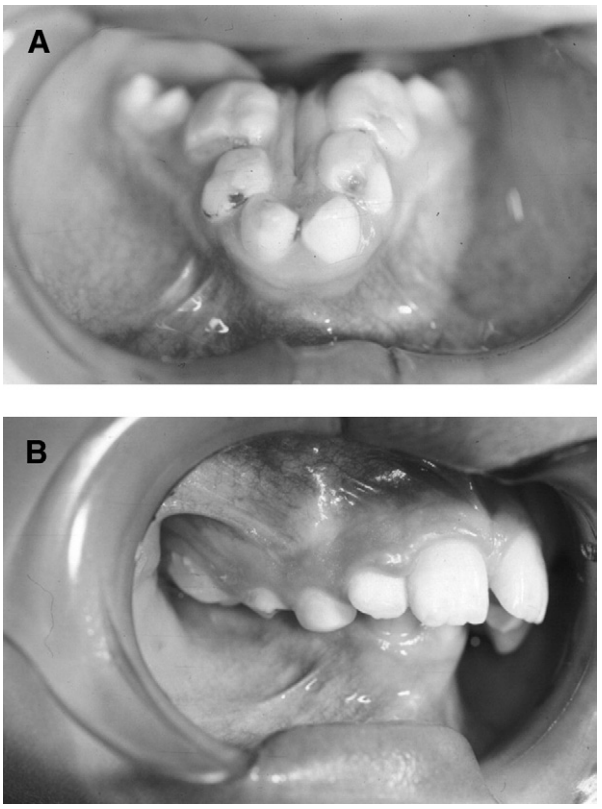

Fig. 1. **A,** Patient's mandible at 5 years of age. **B,** Maxilla obscures mandible completely.

CASE REPORT

Medical history

A 14-year-old white girl with aglossia was seen in our private practice. She was the first born of a white 31-year-old Brazilian woman, gravida 2, para 2, abortions zero, blood type A, Rh positive. Her mother's serologic tests were all negative. No trauma, ingestion of drugs, or exposure to toxic substances or radiation occurred during pregnancy. The girl was born to young and healthy nonconsanguineous parents after normal and spontaneous cephalic delivery at term. Weight, body measurements, and head circumference were normal. She cried at birth, and the obstetrician did not notice any abnormality. Her psychomotor development was within normal parameters: she walked at 1 year and 6 months of age, spoke at 2 years, and entered preschool at 4. At present, she is in her sophomore year in a regular high school program. Today she is 1.20 m tall. Her sister, born 8 years later, is healthy. Her family members are all short; the tallest person in the family is her father, whose height is 1.58 m.

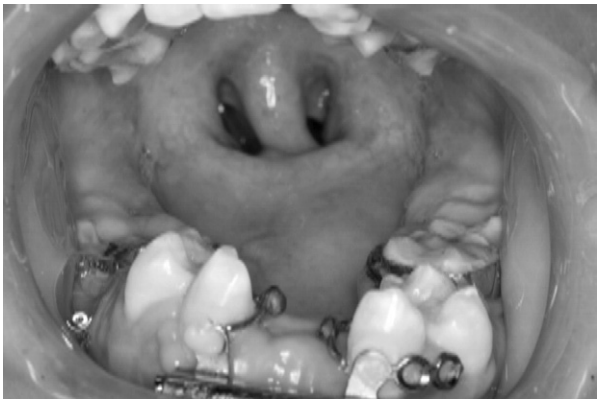

Fig. 2. Total absence of tongue and hypertrophic uvula.

Immediately after birth, the girl had difficulties suckling and swallowing. Therefore, she was fed via nasogastric tube and kept in the hospital to learn how to suckle. After 25 days, no progress was observed, and she was discharged from the hospital. At home, her mother enlarged the opening of the nipple of a nursing bottle and introduced milk directly into the infant's throat. Later she was misdiagnosed with ankyloglossia and hospitalized for an operation, which, fortunately, was not performed.

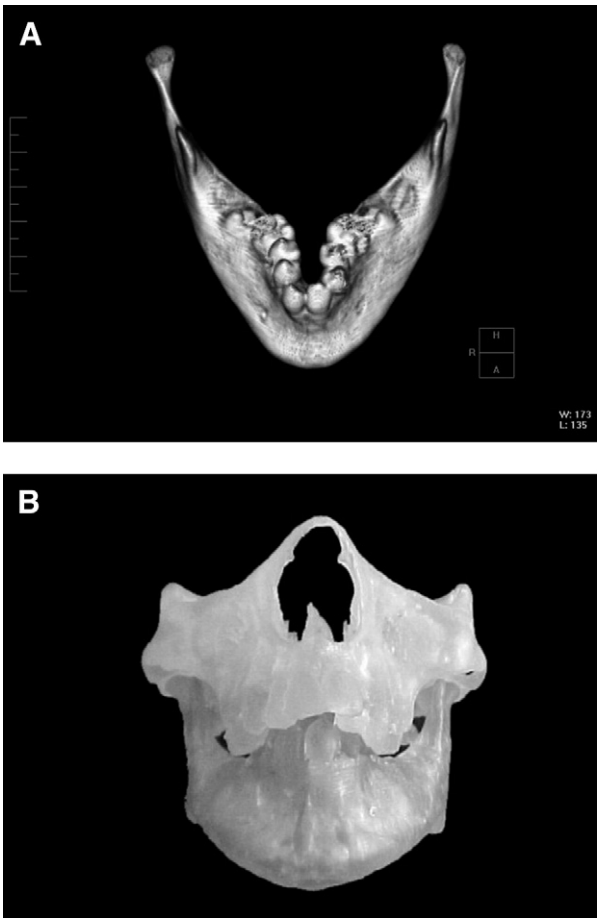

Fig. 3. **A,** Virtual image of mandible reconstructed from CT scans. **B,** Rapid prototyping model in acrylic resin.

Clinical findings

The patient's chief complaint was that it was impossible to chew foods because the lower teeth touched and hurt the palatal mucosa (Fig. 1, *A* and *B*). Although she could answer questions easily, a definite speech impediment was noticed. Her uvula was hypertrophic, and the tongue was completely absent with no rudimentary structure resembling it (Fig. 2). The alveolar arches had not developed transversally, and the mandible had not grown in the anterior direction, probably because of lack of muscular stimulus between the alveolar arches. A number of dentofacial deformities were observed: drastic reduction of space between mandibular bodies (Fig. 3, *A*), bilateral buccal crossbite, excessive overjet, deep overbite, and high palatal vault. Maxillary teeth were normal in form and number and had no caries. Mandibular incisors were absent. She had no abnormalities in the head, ears, eyes, nose, or any other part of the body (Fig. 4). The absence of the tongue was compensated by the fact that the floor of the mouth was smooth and its posterior portion could be elevated to contact the palate, which allowed her to develop speech and swallowing functions.

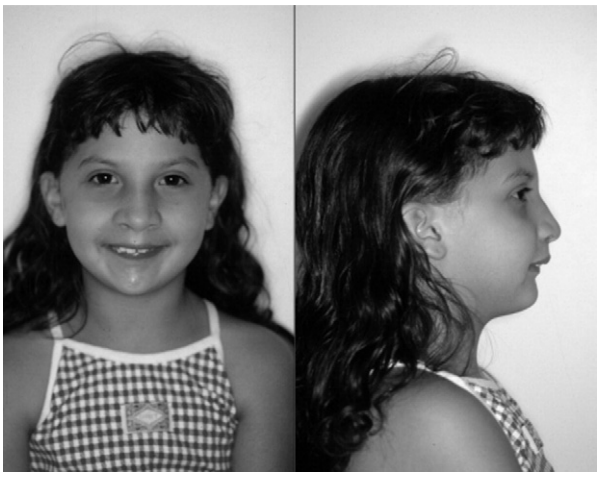

Fig. 4. PA and lateral photo of patient at 5 years of age.

Findings of complementary tests

Panoramic radiographs revealed the presence of all permanent teeth except the 4 mandibular incisors (Fig. 5, *A*). Results of blood tests, urinalysis, thyroid sonography, scintigraphy, karyotype, brain computed tomography (CT), radiographs of hands and feet (Fig. 5, *B* and *C*), and audiometry were normal. Spiral CT scans of the maxilla and mandible, at 1-mm 2-dimensional slices and 0.5-mm intervals, were used to produce a rapid prototyping model according to the protocol described by ARTIS Prototipagem.[31] The model was fabricated in acrylic resin using the PolyJet technology (Fig. 3, *B*), which was chosen because of the transparency and accurate reproduction of anatomic details that it provides, and because it is possible to use burs and surgical saws on the model.

Feeding characteristics and taste perception

The patient's eating pattern was analyzed according to Duarte and Castellani[32] and the National Center for Health Statistics.[33] Her body mass index (BMI) was in the 50th percentile; the weight/age ratio, between the 5th and 10th percentiles; and the height/age ratio, below the 5th percentile. No signs of micronutrient deficits were found. Her low height was not attributed to chronic undernourishment due to possible deficient nutrition in the first years of life because the average height of her parents, 4 grandparents, 3 aunts, and 1 sister was 1.55 m.

Most authors do not provide details of taste testing and only report on whether their patients perceive tastes or not.[34] Eskew and Shepard,[35] however, tested their

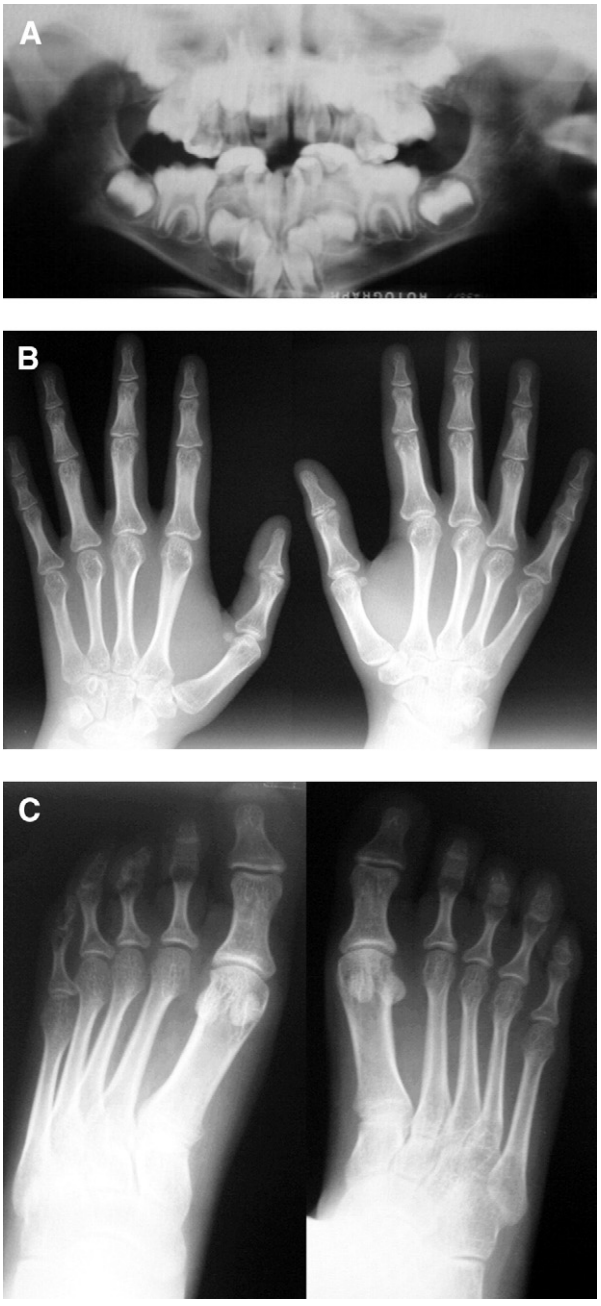

Fig. 5. **A-C,** Panoramic radiograph and plain radiograph of hands and feet.

patient's taste perception by applying sucrose, sodium chloride, sulfuric acid, and quinine solutions to the oral mucosa with a brush, and concluded that only the soft palate mucosa was capable of identifying different tastes. Therefore, our patient's taste perception was evaluated with threshold tests recommended by Teixeira et al.[34] Briefly, 6 increasing concentrations of acid, bitter, sweet, and salty solutions were used to determine the minimally detectable concentration thresholds and the recognition of the specific taste for each test solution. Solutions were prepared with citric acid, caffeine, sucrose, and sodium chloride. At taste changes, the patient washed her mouth with distilled water.

The patient recognized the acid taste from the lowest concentration (0.015 g/L); the bitter taste was identified as a new taste at the third concentration (0.012 g/L), and classified as sweet up to the last concentration; the sweet taste was detected as a new taste at the fifth concentration (8 g/L), and identified as bitter at the last concentration (16 g/L); the salty taste was detected as a new taste at the fourth concentration (0.75 g/L), and identified as salty from the fifth concentration on (1.5 g/L). These results suggest a greater sensitivity to acid and salty tastes and an impaired identification of sweet and bitter tastes, which she inverted. However, this did not interfere in her eating choices, and she ate her family's usual diet.

Treatment

Few studies have discussed possible treatments of aglossia sequelae. Boraz et al.[1] and Sarmiento et al.[36] suggested the use of palatal expanders for mandibular expansion. The results obtained by the first authors were not successful; the other authors reported their case before the expander was applied. Boraz et al.[1] used a palatal expander after vertical symphyseal osteotomy, but their patient lost the ability to talk and swallow that had been developed during facial growth. We concluded that palatal expanders block the elevation of the hypertrophic floor of the mouth and make swallowing and speech difficult.

Aglossia sequelae involve several conditions that cannot be treated with a single procedure. To treat the anatomic, functional, psychological, nutritional, and esthetic problems, we chose a multidisciplinary approach with the participation of professionals in the areas of nutrition, psychology, speech and hearing, general dentistry, orthodontics, maxillofacial surgery, and implantology. Our patient received routine psychological, nutritional, orthodontic, and speech and hearing care. Her clinical treatment consisted, initially, of symphyseal osteogenic distraction using a buccal intraoral distractor[37,38] (Fig. 6). A 30-mm symphyseal expansion was performed in three 10-mm stages to keep the vector of the distractor inside the mouth. The rapid prototyping model was used to build a customized distractor and to determine the correct position of the distraction vector (Fig. 3, *B,* and 6, *A*). The distractor was attached to the cortical bone, canines, and first premolars after the separation of the mandibular bodies by means of vertical symphyseal osteotomy in the midline (Fig. 6, *B*).

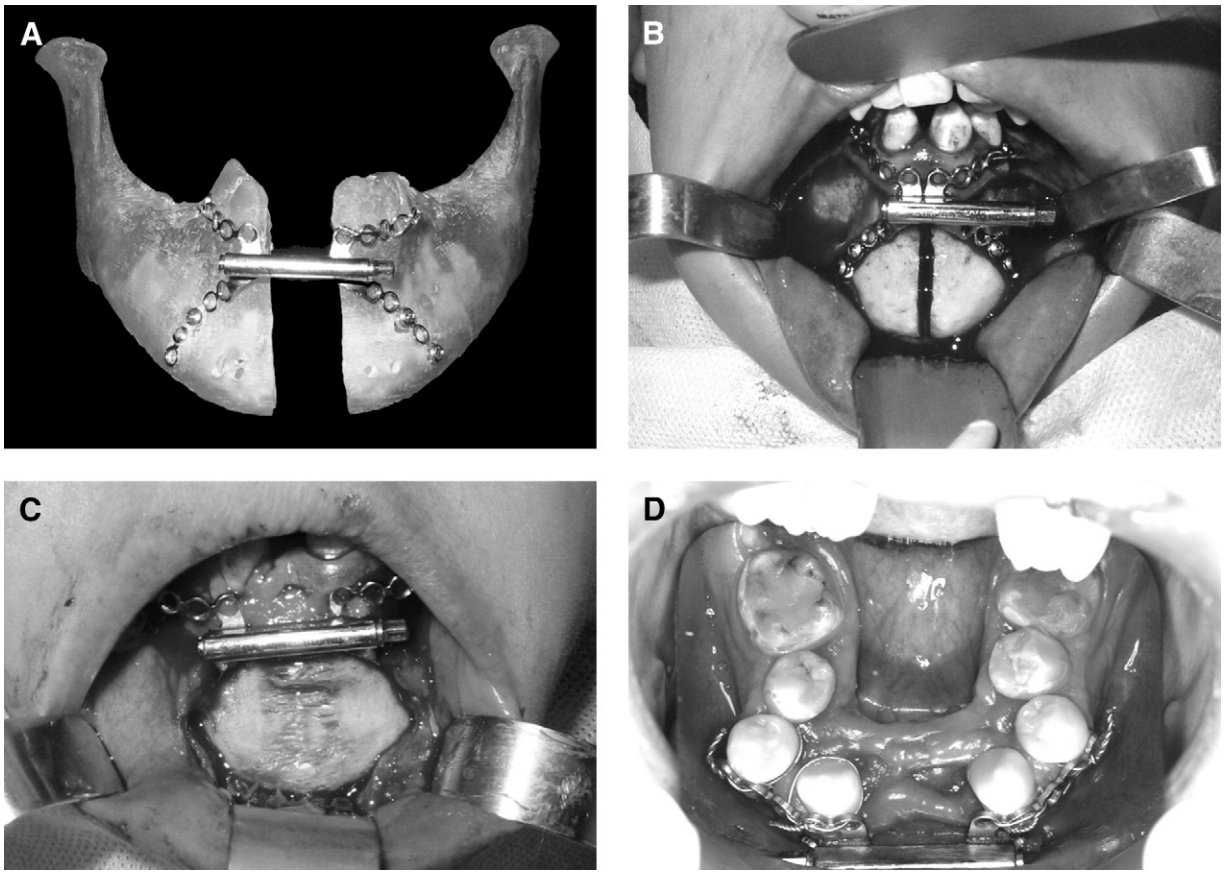

Fig. 6. **A,** Adaptation and test of distractor to rapid prototyping model. **B,** Transfer of distractor to mandible after symphyseal osteotomy. **C,** New bone formation following distraction vector. **D,** Partial result of symphyseal distraction.

DISCUSSION

Complete aglossia is a condition that is practically incompatible with life. Because nourishment is not possible without the suckling reflex, these patients seldom live longer than 3 days.[28] Many cases may have gone unreported because infants did not survive until a diagnosis was made. Maternal instinct has been responsible for a few of those who survived. The Portuguese mother in de Jussieu's report[6] pressed her infant's lips against her nipple and expressed milk intermittently. In the cases reported by Fulford et al.,[39] as well as in our case, the mother enlarged the opening of the nipple of a nursing bottle and placed milk directly into the infant's throat. This may explain why not more than 10 cases of complete aglossia have been reported in 3 centuries. Maternal participation goes beyond helping for immediate survival. The development of children with congenital illnesses depends on the capacity of their families to adhere to treatment programs and to help their social, school, and vocational inclusion. In this respect, the involvement of a multidisciplinary team committed to full rehabilitation objectives is a facilitating resource to achieve good quality of life for patients and their families.

Besides the fundamental functions of speech, mastication, and swallowing, the tongue also has an important role in the growth of jaws, particularly of the mandible, and in the prevention of malocclusion. Moreover, it affects facial esthetics and, therefore, aglossia impairs psychological, sexual, and social development.

Our patient's enhanced taste perception, rarely reported in the literature reviewed, was a surprising finding in this case. De Jussieu[6] reported that the taste perception of the adolescent that he evaluated in Lisbon was similar to that found in healthy people. He also reported a marked enlargement of the uvula, an abnormality found in most cases reported so far. This vicarious hypertrophy of the uvula partially blocks the throat's opening and leads part of the air into the nasal cavity, which makes the articulation of nasalized sounds possible.

Speech, found in all cases of aglossia, is precarious and made possible by the gradual hypertrophy of the muscles of the floor of the mouth, particularly the

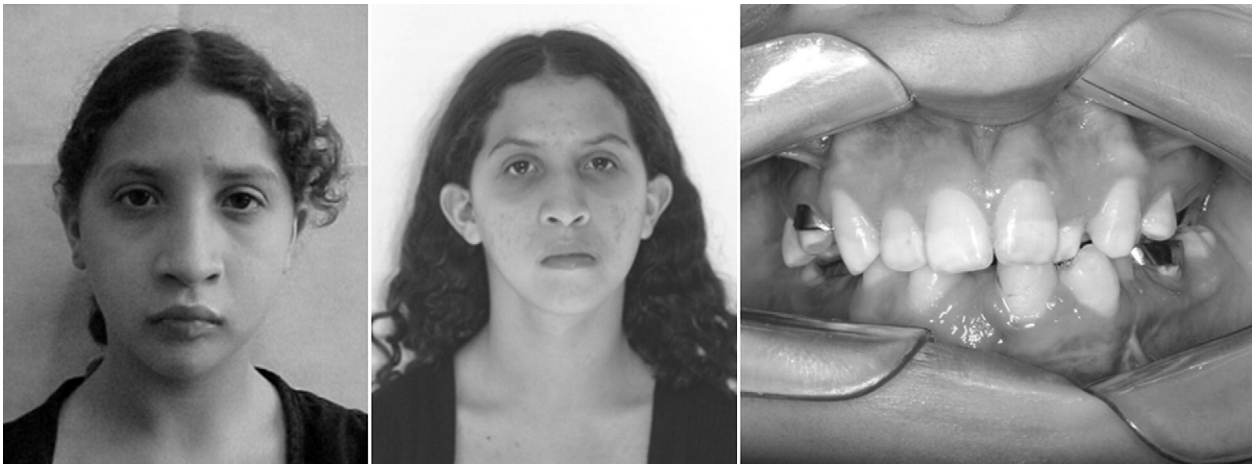

Fig. 7. **A,** Patient's facial features at 13 years of age, before symphyseal distraction. **B,** Patient's facial features at 14 years of age, after symphyseal distraction. **C,** Current maxilla-to-mandible relationship; patient at 14 years and 8 months of age.

mylohyoid muscle, and by the stimulation of swallowing since the first days of life. As a result, the floor of the mouth can be elevated and touch the palate. The Chinese person studied by Eskew and Shepard[35] could speak Chinese and English.

Our patient had a neutral voice quality and moderate impairment of nasal resonance. She also had severe articulation problems, with more marked distortions in the articulation of the phonemes /t/, /d/, /n/, /s/, /z/, /š/, /ž/, and omission of the phonemes /l/ and /r/. She tried to make the lower lip touch the maxillary incisors to articulate linguodental phonemes, but had marked difficulties because of lack of muscle tone, which made her speech sound imprecise and slurred. Speech and hearing therapy improved this pattern, and after 10 months of exercises, the plosive phonemes /t/ and /d/ and the nasal phoneme /n/ became clearer and better articulated. The fricative phonemes /s/ and /z/ also became clearer, but distortions in the articulation of the phonemes /š/ and /ž/ remained.

CONCLUSIONS

Facial sequelae of aglossia compromise facial esthetics and severely affect psychological well-being. We agree with the concept formulated by Tallmadge in the question written for the epilogue of the book edited by Dingman and Natvig[40]: "Is there any part of the body dearer to a person than their own face?"(p.367) The treatment of these sequelae is possible and may improve quality of life, as could be confirmed even before treatment completion in the case reported here (Fig. 6, *C* and *D*, and 7). We believe that such results would have been impossible without the participation of a multidisciplinary team. Transparent rapid prototyping models built in acrylic resin are excellent aids to surgery because they provide preoperative simulation, reduce operation time and risks, and help build patient confidence.

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Reprint requests:

Frederico Salles, MD
SMDB Conj. 12 Bloco F
1° andar. CEP 71.680-120
Brasília, DF. Brazil
salles.frederico@gmail.com