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CASE REPORT

Pediatric/Craniofacial

Long-term Prognosis of Mandibular Distraction in 3 Cases of Hypoglossia-Hypodactyly Syndrome without Extremity Anomalies

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Summary: Hypoglossia-hypodactyly, or aglossia-adactylia syndrome with or without limb anomalies, is an extremely rare congenital condition. It is characterized by a narrow, V-shaped mandibular dental arch with micrognathia, and is typically challenging to treat. We have previously reported 3 patients with hypoglossia-hypodactyly syndrome without limb anomalies who were treated with transverse distraction osteogenesis at the mandibular symphysis during childhood. In this report, we present the long-term prognoses of these 3 cases, until 18 years of age. Of the 3 total cases, sufficient and stable results in occlusion, speech, mastication, and facial appearance were obtained in 2 cases with subsequent orthodontic treatments. Similar results were not observed in the remaining case when continuous orthodontic treatments were not performed. Based on these results, we propose that the transverse distraction osteogenesis procedure at the mandibular symphysis during childhood could be a reliable method to correct the V-shaped mandibular arch of hypoglossia-hypodactyly syndrome, when subsequent orthodontic treatments are provided during the patient's growing period. *(Plast Reconstr Surg Glob Open 2019;7:e2211; doi: 10.1097/GOX.0000000000002211; Published online 3 May 2019.)*

Hypoglossia-hypodactyly, or aglossia-adactylia syndrome with or without limb anomalies, is an extremely rare congenital condition that is characterized by a narrow V-shaped mandibular dental arch with micrognathia, due to aplasia of the tongue.[1–6] There are as few as 40 cases reported in the global body of literature.[7,8] Intelligence is normal in most of the cases[9]; patients have problems with speech, swallowing, mastication, and facial appearance, due to jaw deformity. Treatment for the jaw deformities is challenging, and there are few reports on successful surgical intervention. We have previously reported 3 cases of hypoglossia-hypodactyly syndrome without limb anomalies in which treatment was performed with the transverse distraction osteogenesis procedure at the mandibular symphysis during childhood.[10] The reports include descriptions of functional improvement in speech, swallowing, and mastication just after the treatment.

In this report, we presented long-term prognoses of 3 hypoglossia-hypodactyly cases until 18 years of age, and estimated the application of this treatment (Table 1) as a follow-up report.

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CASE REPORTS

Case 1

The patient, a 9-year-old female, presented to our clinic with the chief complaints of tongue and mandibular deformities.[10] The patient was born via normal delivery at full-term gestation, and there was no family history of congenital anomalies. At the age of 9, the patient underwent a distraction osteogenesis procedure of 21 mm in length at the mandibular symphysis, and her mandibular arch was successfully widened.[10] She received subsequent orthodontic treatments until adolescence. After the active orthodontic treatments, we provided her a removable orthodontic retainer. No additional orthognathic surgery was needed. Her occlusion and facial appearance were still well-maintained at the age of 18 (Fig. 1). Her masticatory function and articulation were adequate for her daily life, although she was diagnosed by our speech therapist with a slight articulation error due to hypoglossia.

Case 2

The patient, a 9-year-old female, presented to our clinic with the chief complaints of tongue and mandibular

Table 1. Treatment and Long-term Prognoses in 3 Cases of Hypoglossia-Hypodactyly Syndrome

	Age at DO	Length of DO	Disturbance for OT	Additional OGS	Occlusal Prognosis
Case 1	9	21	–	–	Good
Case 2	9	19	AAS	–	Poor
Case 3	12	21	–	+	Good

AAS, atlantoaxial subluxation; DO, distraction osteogenesis; OT, orthodontic treatments; OGS, orthognathic surgeries; +/-, with/without.

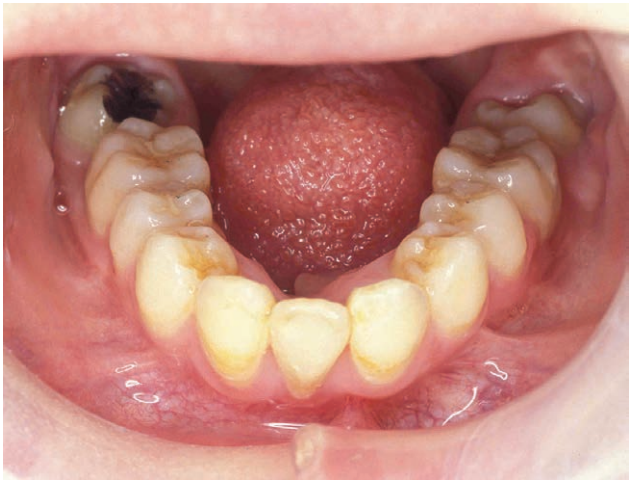

Fig. 1. The corrected mandibular arch in case 1 at the age of 18.

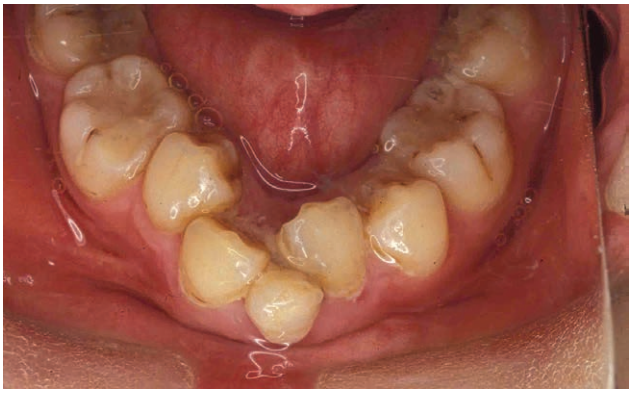

Fig. 2. The remaining V-shaped deformity of the mandibular arch in case 2 at the age of 18.

deformities.[10] At the time of presentation, her parents were healthy and nonconsanguineous. The patient's medical history included a normal birth at full-term gestation. At the age of 9, the patient underwent a distraction osteogenesis procedure of 19 mm in length at the mandibular symphysis, and her mandibular arch was successfully widened.[10] However, she could not receive subsequent orthodontic treatments due to her atlantoaxial subluxation. Although atlantoaxial arthrodesis was performed when the patient was 14, the limited extension of her neck interfered with adequate orthodontic treatments. As a result, she had a remaining telescopic bite and V-shaped mandibular arch (Fig. 2); therefore, she had problems with diet and articulation at the age 18.

Case 3

The patient, a 12-year-old female, presented to our clinic with the chief complaints of tongue and mandibular deformities (Fig. 3A). Her medical history included a normal full-term delivery to healthy parents. At the age of 12, the patient underwent the distraction osteogenesis procedure of 21 mm length at the mandibular symphysis.[10] Subsequent orthodontic treatments were performed to arrange the mandibular teeth and to maintain the arch. She demonstrated remarkable maxillary protrusion and retrognathia at age 19, thus additional orthognathic surgeries were performed, such as anterior maxillary segmentation to correct the teeth axis and a mandibular advancement of 10 mm with a conventional bilateral sagittal split of the ramus. After the active orthodontic treatments, we provided her a removable orthodontic retainer. Finally, satisfactory facial profile and occlusion were achieved at the age of 21 (Fig. 3B). She had no problems with diet and speech in her daily life even though a slight articulation error due to hypoglossia remained.

CONCLUSIONS

We followed and reviewed 3 cases of hypoglossia-hypodactyly syndrome without extremity anomalies, which had been reported previously.[10] Those patients who were able to receive subsequent orthodontic treatments had sufficient and stable results with regard to occlusion, speech, mastication, and facial appearance. Based on these results, we propose that the transverse distraction osteogenesis

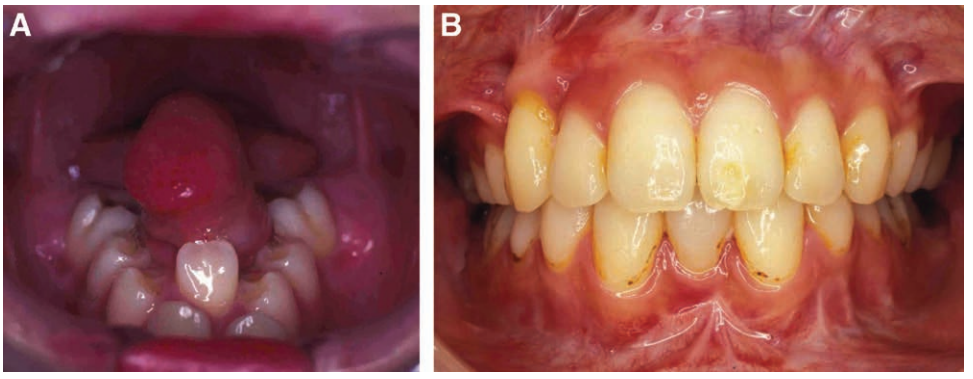

Fig. 3. Oral findings in case 3. A, The V-shaped mandibular arch before distraction. B, The corrected mandibular arch and occlusion at the age of 21 after the additional orthognathic surgeries.

procedure at the mandibular symphysis during childhood could be a reliable treatment to correct the V-shaped mandibular arch associated with hypoglossia-hypodactyly syndrome, when subsequent orthodontic treatments are provided during the patient's growing period.

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