

## Mandibular expansion in a Congenital aglossia patient

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## 국문초록

선천적 무설증은 매우 드물게 보고되는 증상이다. 무설증을 가진 환자는 구개 이상, 하순 결손, 하악 측절치 결손 등을 포함한 하악골의 발육 부전을 나타낸다. 무설증의 원인은 밝혀지지 않았으나 유전적 요인이나 발생 4주시기에 기형유발물질의 영향으로 추정되고 있다. 무설증의 치료는 혀를 재건해주는 외과적 술식과 하악골의 성장을 돕는 교정적 술식, 하악골의 길이를 늘리는 distraction osteogenesis 등이 있으며 무설증의 정도에 따라 달라진다. 본 증례는 선천적 무설증과 situs inversus(장기의 좌우가 바뀐 상태)를 가진 6세 여아에서 무설증으로 인한 하악골의 횡적 수축을 악정형 장치를 이용하여 치료하였으며 향후 2차 교정과 외과적 수술을 계획하기에 양호한 결과를 얻을 수 있었다.

**주요어** : 선천적 무설증, 하악골 발육 부전, situs inversus

## I. Introduction

*Aglossia* means an absence of the entire tongue<sup>1)</sup>. There are few reports on congenital aglossia (congenitally complete absence of the tongue) in the literature, and most reports were of partial aglossia or microglossia cases<sup>2-4)</sup>. Oral manifestations of an aglossia include micrognathia, high arched or cleft palate, defects of the lower lip, an absence of lateral incisors<sup>4)</sup>. The important influence of the tongue malformation on postnatal tooth alignment and jaw development is relatively well known. Embryologically, microglossia can be attributed to a growth failure of the lateral lingual swelling and the tuberculum impar. At 4th week of the fetal life, the anterior portion of the tongue begins to develop from three structures of the 1st branchial arch, the two lateral lingual swelling

and a median swelling, the tuberculum impar. The posterior part of the tongue is derived from the second and third branchial arches<sup>3)</sup>. However, the etiology of aglossia is unknown.

The treatment of aglossia includes the surgical rehabilitation of the tongue tip to some extent, orthopedic expansion of the mandible to guide mandibular growth, and mandibular expansion by distraction osteogenesis<sup>1,2)</sup>.

Some reported cases have been associated with oromandibular-limb hypogenesis syndromes. Patients with aglossia/hypoglossia can have coexisting anomalies, such as cleft palate, intraoral bands, and situs inversus(i.e. reversal of visceral organs). In the present report, the orthopedic management of mandibular deficiency in a congenital aglossia with situs inversus patient is discussed.

## II. Case report

A 6 year old female was observed to have a congenital absence of the tongue(aglossia) without hypodactylia syndrome(Fig.1, 2). She visited the

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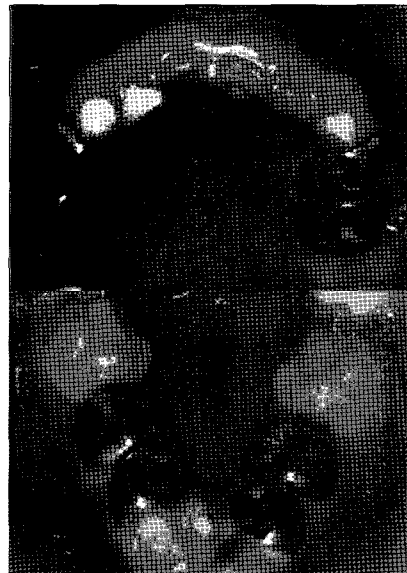
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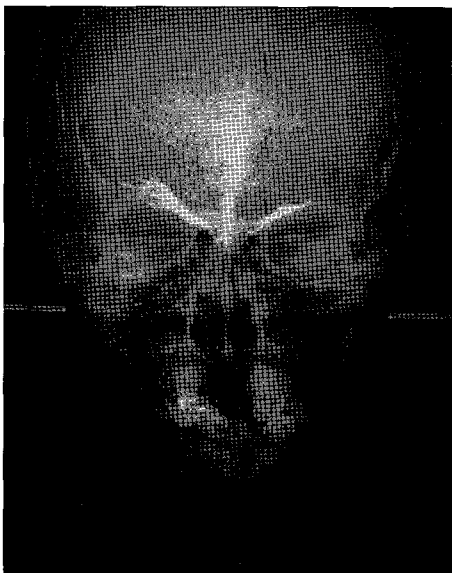
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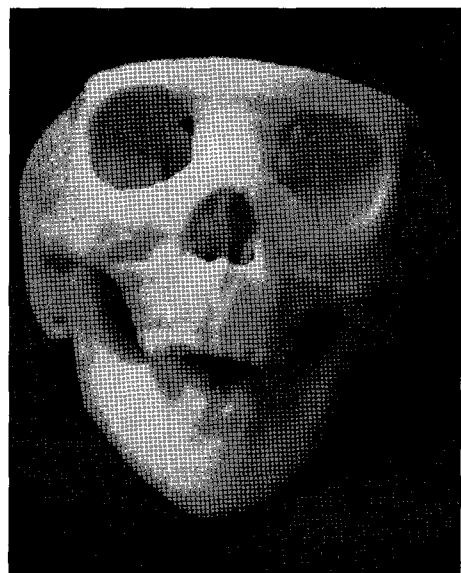
**Fig. 1.** Initial profile.



**Fig. 2.** Initial maxillary and mandibular arch. Note the constricted mandibular arch.



**Fig. 3.** PA view radiogram shows constricted lower alveolar process.



**Fig. 4.** 3D starch model. Note the constricted lower alveolar arch.

Department of Pediatric Dentistry, Seoul National University Dental hospital with a chief complaint of a mandibular retrusion. On the oral examination, the entire tongue was absent so that the floor of mouth was directly seen. The patient was born in a normal delivery and was diagnosed with situs inversus with dextrocardia when she was 12 days-old. There was no history of illness, trauma, or maternal drug use during pregnancy, and the familial history showed nothing

significance. The patient could pronounce the vowels relatively well, but the consonants were not clear, because they require the use of the tongue tip.

In order to test her taste sensation capacity, 5% cane sugar(sweet), 10% sodium chloride(salty), 0.1% quinine(bitter), and 1% acetic acid(sour) were prepared and applied with cotton swabs to the various regions of the mucous membrane of the oral cavity. The results indicated that she could identify the various

tastes in the sublingual caruncle and fauces. There was no signs or symptoms of any abnormalities on her both TMJs, and her hearing capacity was normal.

After a clinical examination of the patient, facial photos, panoramic view, lateral cephalometric, posterior-anterior(Fig. 3), and 3D-CT were taken. In addition, a study model and a 3D starch skull model(Fig. 4) were made. By using these materials, an orthodontic diagnosis and treatment plan was established. The mandibular alveolar process was severely constricted and one mandibular lateral incisor was

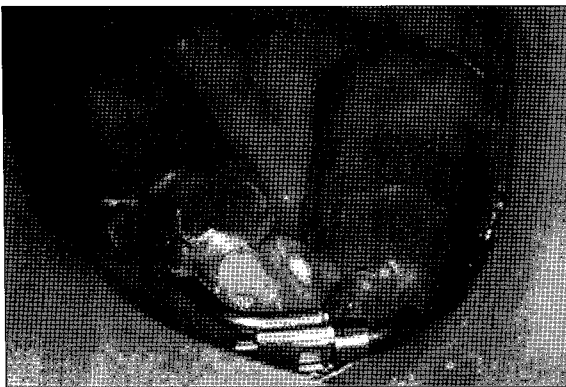
congenitally missing.

Combined orthopedic and surgical treatment was planned. Initially, a transverse expansion of the lower arch was planned by a bonded hyrax(Dentaurum, Germany) screw(Fig. 5) to correct the telescopic bite and gain space for the mandibular incisors. After some retention period, surgical intervention was planned as a second phase of treatment.

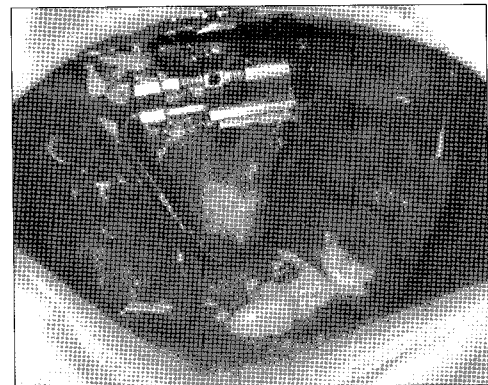
Upon delivery of the appliance, the guardian of the patient was educated to turn the screw once every 3 days. After 11 months of screw activation, the primary intercanine width had increased by 5.7mm, the intermolar width had increased by 8.7mm(Table 1, Fig. 6-1, 2). The telescopic bite was corrected and space for the mandibular incisors was gained. Retention was obtained by a Hawley type retainer which will be applied until the complete exfoliation of primary dentition. The retention period was 13 months, without any relapse of the intercanine width.

**Table 1.** Expansion amount of the lower arch

|                           | Before expansion | After expansion | Amount of expansion |
|---------------------------|------------------|-----------------|---------------------|
| Primary intercanine width | 8.4mm            | 14.1mm          | +5.7mm              |
| Intermolar width          | 21.5mm           | 30.2mm          | +8.7mm              |



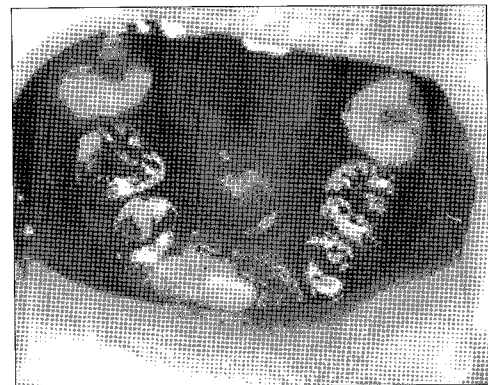
**Fig. 5-1.** Appliance design.



**Fig. 5-2.** Appliance design after 3 months.



**Fig. 6-1.** Before screw activation.



**Fig. 6-2.** After 11 months.

### III. Discussion

Cases of congenital aglossia are very rare in the literature<sup>2-6)</sup>, and orthodontic treatment cases are particularly unusual.

Although the etiology of congenital aglossia is unclear, both genetic and teratogenic mechanisms have been proposed. Amor and Craig proposed that sporadic genetic mutations and external teratogenic insults before the 4th embryonic week might be the cause<sup>3)</sup>.

Aglossia cases with limb anomalies have been reported which are known as oromandibular-limb hypogenesis syndromes (aglossia-adactylia syndrome, Hanhart's syndrome)<sup>3,4)</sup>. These syndromes feature associated limb anomalies, such as hypodactylia (an absence of digits) and hypomelia (hypoplasia of part or all of the limb). No limb anomalies were observed in the present case.

Situs inversus means the position of the heart chambers as well as the visceral organs such as the liver and spleen are reversed. It is a rare condition that occurs in only about one in every 8000-25000 birth. Dextrocardia with situs inversus is a rare heart condition characterized by an abnormal positioning of the heart. In this condition, the tip of the heart (apex) is positioned on the right side of the chest. However, most affected individuals can live a normal life without any associated symptoms or disability<sup>7)</sup>.

Orthodontic and orthopedic treatment of an aglossia patient depends on the nature and severity of the condition. Goto et al reported an orthodontic management case of an aglossia patient which was retained for 6 years<sup>6)</sup>. They recommended a long-term retention because the occlusion would be less stable in the aglossia condition.

In the present case, the primary intercanine width and intermolar width was increased by a bonded

hyrax screw, and the increased space was well retained after 13 month. Occlusal harmony was obtained and space for the permanent incisors was gained. The taste sensation, TMJ condition, and speech did not change after the orthopedic treatment. As Goto et al suggested, long-term retention would be needed until second phase orthodontic and surgical treatment is conducted.

### IV. Summary

In a congenital aglossia patient, constricted mandibular alveolar process could be expanded by an orthopedic treatment. The long-term retention results and second phase treatment is required.

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Abstract

MANDIBULAR EXPANSION IN A CONGENITAL AGLOSSIA PATIENT

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Congenital aglossia is a very rare condition. The oral manifestations of an aglossia include micrognathia, high arched or cleft palate, defects of the lower lip, an absence of lateral incisors and a mandibular growth deficiency. Although the etiology of congenital aglossia is unclear, both genetic and teratogenic mechanisms have been proposed. Treatment of aglossia patients depends on the nature and severity of the condition which includes surgical rehabilitation of the tongue tip to some extent, orthopedic expansion of the mandible to guide mandibular growth, and mandibular expansion by a distraction osteogenesis. In the present case, a 6 year old female aglossia patient with situs inversus was treated. A bonded hyrax screw was used to increase her mandibular primary intercanine width and intermolar width. A second phase orthodontic and surgical treatment will be possible after some retention phase.

**Key words** : Congenital aglossia, Mandibular growth, Situs inversus