Congenital Aglossia with Situs Inversus Totalis
- A Case Report -

Hypoglossia or aglossia is an uncommon anomaly, either of which may occur as an isolated finding or in association with other deformations, especially limb anomalies. Their genetic background is uncertain, and drug induced teratogen has not been clearly identified. We experienced a case of congenital aglossia with situs inversus in a female infant aged twelve days. Her initial complaints at admission were feeding difficulty and weight loss. In a review of literature, the association with situs inversus is very rare and only three cases have been reported until now. *(JKMS 1997; 12 : 55~7)*

Key Words : *Congenital aglossia, Situs inversus*

Gi-Young Jang, M.D., Kwang-Chul Lee, M.D., Ji-Tae Choung, M.D., Chang-Sung Son, M.D., Young-Chang Tockgo, M.D.

Department of Pediatrics, College of Medicine, Korea University, Seoul, Korea

Received : June 18, 1996
Accepted : November 20, 1996

Address for correspondence
Chang-Sung Son, Department of Pediatrics, Korea University Hospital, #126-1, Anam-Dong 5Ka, Sung Buk-Ku, Seoul, Korea
Tel : (02) 920-5090, Fax : (02) 922-7476

INTRODUCTION

Total or partial absence of the tongue associated with abnormalities of the limbs is rare. In addition to the tongue defects, oral manifestations include micrognathia, high arched or cleft palate, defects in the lower lip, and absence of lateral incisors. Abnormalities of the limbs may be mild to severe, ranging from defects of a single digit to complete peromelia. Most reported cases of aglossia have been associated with limb abnormalities. The first reported case of aglossia with situs inversus appears in the literture in 1925 (1). Thereafter, very rare cases have been reported until 1990 (2, 3).

Herein we present a case of congenital aglossia with situs inversus in a female infant aged twelve days, and make a brief review of the literature.

CASE REPORT

A twelve day-old female infant was admitted due to feeding difficulty and weight loss. The infant was born at term, and weighed 2,600 grams. The delivery was normal. There were no histories of illness, trauma, or maternal drug use during pregnancy. There was no family history of congenital anomalies.

On physical examination, the head was normal in size (circumference 33cm), and eyes and ears were normal. The absence of the tongue and underdeveloped mandible were observed (Fig. 1). The upper and lower alveolar ridges were palpated. There were no limb abnormalities. A chest X-ray showed dextrocardia associated with inversion of the abdominal organs (Fig. 2). On electrocardiogram, the inversion of P-wave, QRS complex and T-wave in lead-I and the reversal of QRS complex and T-wave in lead-II and aVR were noted (Fig. 3). On abdominal sonograms, the liver was located in the left side, the spleen was located in the right side and the positions of aorta and inferior vena cava were switched (Fig. 4). On echocardiogram, there were no cardiac malformations except mirror image dextrocardia. The karyotype revealed a normal female chromosome constitution (46, XX). The infant was supported by tube feeding during neonatal period. During 2 years- follow-up the child was doing well except for

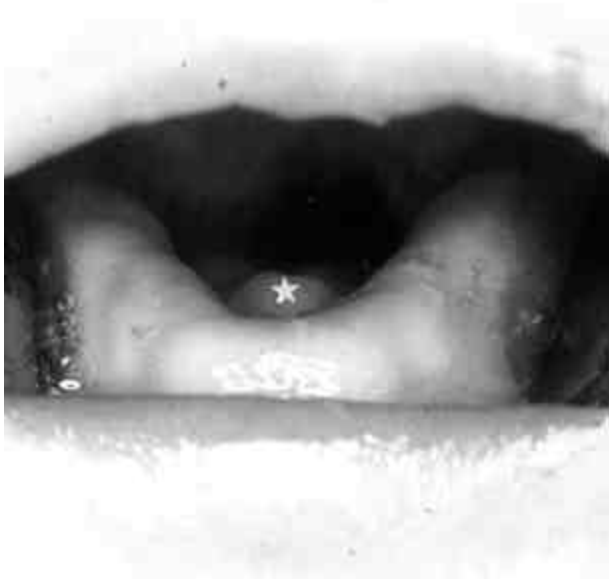

Fig. 1. Photograph showing the absence of the tongue(★).

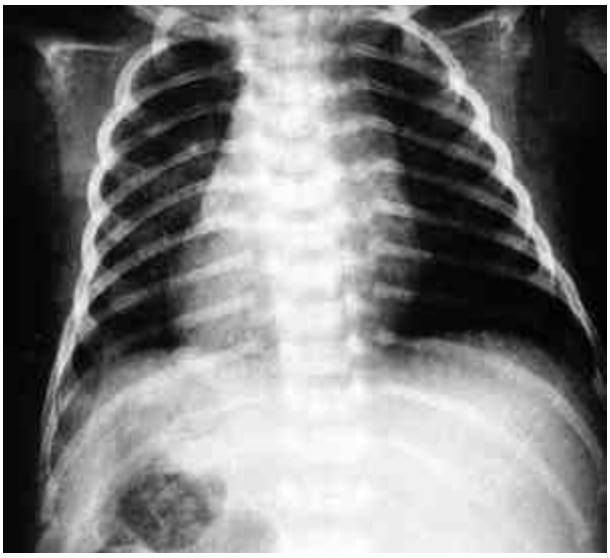

Fig. 2. Chest X-ray showing right deviation of the cardiac apex and stomach air shadow to the right upper abdomen.

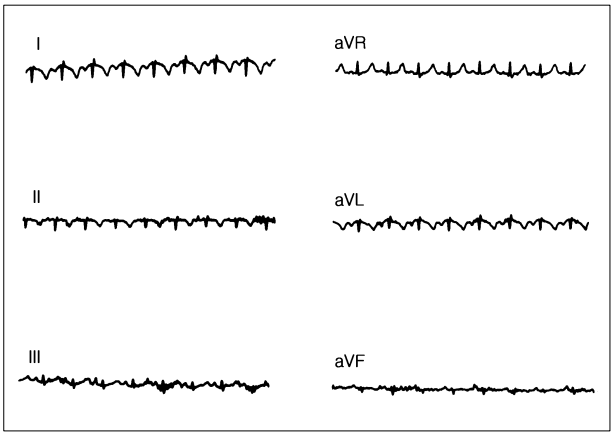

Fig. 3. Electrocardiogram showing the inversion of P-wave, QRS complex and T-wave in lead-I and reversal of QRS complex and T-wave in lead-II and aVR(1/2 voltage).

mild physical underdevelopment and phonation disturbance.

DISCUSSION

Aglossia is a part of the familial malformation syndrome in which asymmetric reduction defects of the limbs are associated with oromandibular anomalies. These anomalies, sometimes referred to a "family" of syndromes, are now called "oromandibular limb hypogenesis syndromes". In 1971, Hall (4) classified oromandibular limb hypogenesis syndrome to 5 major types (Table 1). Their genetic origin is uncertain, and no drug-induced teratogen has been clearly identified (5, 6).

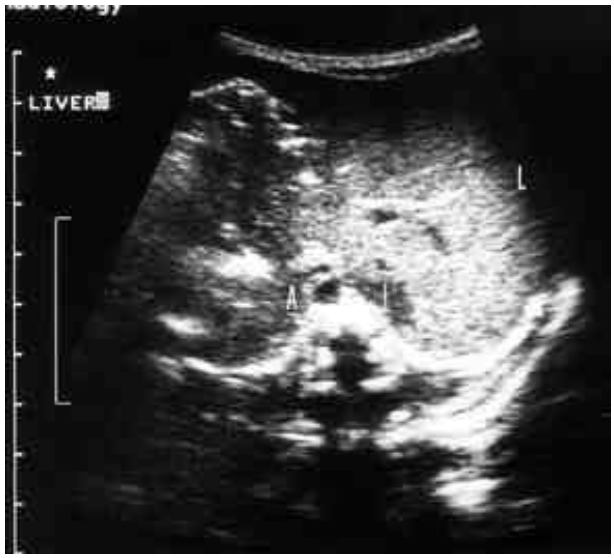

Fig. 4. In the abdominal ultrasound scan, the liver(L) is located in the left side and the positions of aorta(A) and inferior vena cava(I) are switched.

Embryologically, microglossia can be attributed to a growth failure of the lateral lingual swelling and the tuberculum impar. At fourth week of the fetal life, the anterior portion of the tongue starts to develop from three structures of the first 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 (7). Although a number of theories have been proposed, the etiology of aglossia is largely unknown (8).

However, in the case of situs inversus, several studies have identified its origin, especially in the genetic aspect. Laterality is randomly determined and directed by a three-step process in which the molecular laterality is converted to the cellular laterality (9). The iv/iv mouse has a 50% chance of developing situs inversus totalis and cardiac defects (10). The mapping of the autosomal recessive conditon of the situs inversus in mouse (the iv locus) on chromosome 12 suggested the existence of a human counterpart gene. This gene could be on human chromosome 14q32 by homology (11). Yokoyama et al.(12) reported a situs inversus mutation associated with inv gene. According to their reports, the recessive mutation was identified in a family of transgenic mice that resulted in a reversal of left-right polarity (situs inversus) in 100 percent of the homozygous transgenic mice tested. This insertional mutation identifies a gene that controls embryonic turning and visceral left-right polarity.

In this case, the karyotype revealed a normal female chromosome constitution(46, XX). Type I oromandibular limb hypogenesis syndrome includes lingual hypogenesis

Table 1. Oromandibular Limb Hypogenesis Syndrome. (Hall's classification)

Type I :
A. Hypoglossia
B. Aglossia
Type II :
A. Hypoglossia-hypodactylia
B. Hypoglossia-hypomelia
C. Hypoglossia-hypodactylomelia
Type III :
A. Glossopalatine ankylosis(ankyloglossum superius syndome)
B. With hypoglossia
C. With hypoglossia-hypodactylia
D. With hypoglossia-hypomelia
E. With hypoglossia-hypodactylomelia
Type IV :
A. Intraoral bands and fusion
B. With hypoglossia
C. With hypoglossia-hypodactylia
D. With hypoglossia-hypomelia
E. With hypoglossia-hypodactylomelia
Type V :
A. The Hanhart syndrome
B. Charlie M syndrome
C. Pierre Robin syndrome
D. Möbius syndrome
E. Amniotic band syndrome

without associated limb abnormalities. So, we consider this case as a type I oromandibular limb hypogenesis syndrome by Hall's classification.

REFERENCES

1. Watkin HG. *Congenital absence of the tongue. Int J Orthod 1925 ; 11 : 941-3.*
2. Dunham ME, Austin TL. *Congenital aglossia and situs inversus. Int J Pediatr Otorhinolaryngol 1990 ; 19 : 163-8.*
3. Hussels IE. *Microglossia, hypodontia, micrognathia, situs inversus. Birth Defects 1971 ; 7 : 282.*
4. Hall BD. *Aglossia-adactylia. Birth defects 1971 ; 7 : 233-6.*
5. Chicarilli ZN, Polayes IM. *Oromandibular limb hypogenesis syndrome. Plast Reconstr Surg 1985 ; 76 : 13-24.*
6. Nevin NC, Kernohan DC, Ross AM. *Ankyloglossum superius sydrome. Oral Surg 1980 ; 50 : 254-6.*
7. Johnson GF, Robinow M. *Aglossia-adactylia. Pediatrics 1978 ; 128 : 127-32.*
8. Lustmann J, Lurie R, Struthers P, Garwood A. *The hypoglossia-hypodactylia syndrome. Oral Surg 1981 ; 51 : 403-8.*
9. Brown NA, Wolpert L. *The development of handedness in left/right asymmetry. Development 1990 ; 109 : 1-9.*
10. Layton WM Jr. *The biology of asymmetry and the development of the cardiac loop. In : Ferrans VJ, Rosenquist GC, Weinstein C, eds. Cardiac morphogenesis. New York : Elsevier, 1985 : 134-40.*
11. Brueckner M, D'Eustachio P, Horwich AL. *Linkage mapping of a mouse gene, iv, that controls left-right asymmetry of the heart and viscera. Proc Natl Acad Sci USA 1989 ; 86 : 5035-8.*
12. Yokoyama T, Copeland NG, Jenkins NA, Montgomery CA, Elder FFB, Overbeek PA. *Reversal of left-right asymmetry : A situs inversus mutation. Science 1993 ; 260 : 679-82.*