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■ Case report ■

Macroglossia secondary to lymphangioma of the deep neck space: Report of two cases

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= Abstract =

Lymphangioma is a rare, benign, and hamartomatous tumor of the lymphatic vessels that shows a marked predilection for the head and neck region. When this tumor occurs on the tongue or mouth floor or in the deep neck space, blockage of the efferent lymphatic vessels can result in secondary macroglossia. We report here two patients who showed unusual macroglossia from birth. Initially, there was no noticeable cervical or mandibular swelling. However, mandibular swellings were noted during follow-up examinations, which led to MRI scans on the two infant patients at 5 months and 5 weeks of age, respectively. Subsequently, both patients were diagnosed with lymphangioma or lymphangiohemangioma in the deep neck space. (Korean J Pediatr 2010;53:97-102)

Key Words : Macroglossia, Lymphangioma, Cervical

Introduction

Macroglossia, or enlarged tongue, is a component of numerous syndromes, many caused by inherited metabolic anomalies in which the increase in tongue size is a manifestation of visceromegaly related to lysosomal storage diseases, such as Hurler syndrome, Hunter syndrome, and Maroteaux-Lamy syndrome. Other macroglossia-associated disorders include BeckwithWiedemann syndrome, neurofibromatosis type 1, hemangiomatosis associated with Sturge-Weber syndrome, and congenital lymphangioma (cystic hygroma)[1]. Macroglossia can be classified as either congenital/primary or secondary. Primary macroglossia is due to over-development of the musculature[2], while the secondary form may result from a tumor of the tongue (such as diffuse lymphangioma or hemangioma), neurofibromatosis, or, occasionally, blockage of the efferent lymphatic vessels, as in cases of malignant neoplasm of the tongue[3].

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Lymphangiomas and/or lymphangiohemangiomas are benign, relatively rare tumors characterized by a proliferation of lymphatic and/or blood vessels[4, 5]. When they appear within the neck, lymphangiomas are commonly located in the anterior or posterior cervical triangles, though submandibular or submental involvement is also frequent[6, 7]. In these two unusual macroglossia case we observed, macroglossia was the initial manifestation of lymphangioma or lymphangiohemangioma in the deep neck space.

Case report

Case 1

A seven-day-old female infant was admitted to our hospital with a chief complaint of feeding difficulty associated with macroglossia. There was no specific antenatal or family history. She had been delivered vaginally after 38 weeks' gestation with a birth weight of 3,370 g (50-75th percentile), length of 50.9 cm (50-75th percentile), and head circumference of 34.5 cm (50-75th percentile). She showed no abnormal laboratory findings, including CBC and blood glucose. Physical examination revealed insufficient mouth closure with a large, normalcolored tongue (Fig. 1A), which protruded anteriorly over the lower alveolar

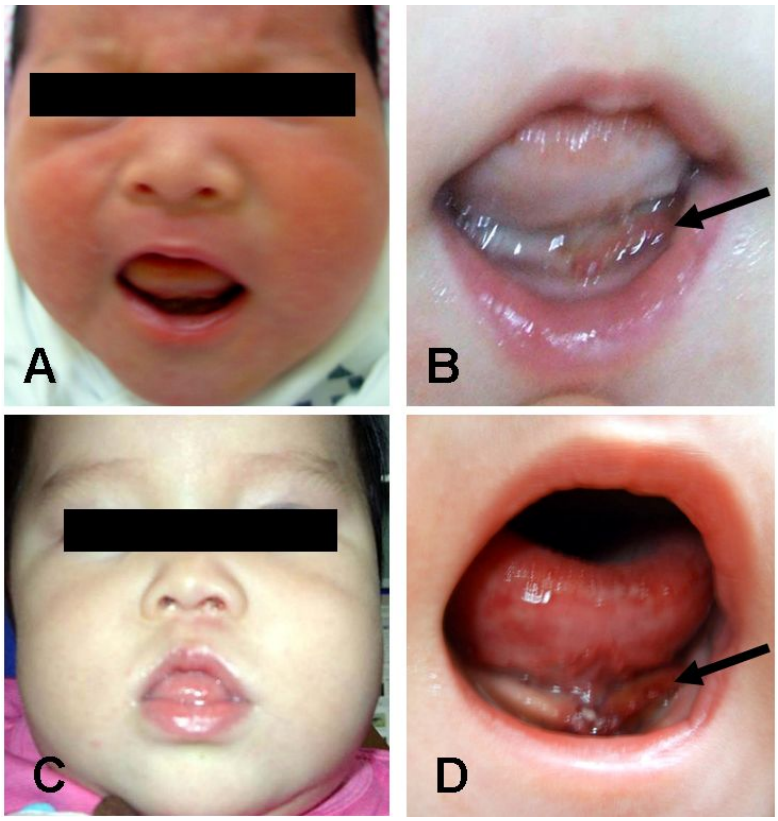

Fig. 1. Gross features of case 1. (A) At birth, insufficient mouth closure with a large, normal-colored tongue was apparent. (B) At 5 months of age, a red-beef-colored, small protruding lesion below the left side of the tongue base was noted (black arrow). (C, D) At 8 months, the tongue had increased in size and changed to a more reddish color, with a red-beef-colored lesion at the tongue base (black arrow).

ridge and reached superiorly to the hard palate. In spite of the macroglossia, the airway was not obstructed. Cranial ultrasonography, a metabolic screening test (including thyroid function), and a hearing test all were normal. The patient was discharged with improved feeding.

Upon follow-up examination at 1 month of age, the patient's growth percentiles and development were within normal ranges. We suspected Beckwith-Wiedemann syndrome because of the insufficient mouth closure with macroglossia. However, results of methylation tests of the H19, IGF2, and LIT1 genes on 11P15.5 (for the evaluation of Beckwith-Wiedemann syndrome), a chromosomal study, and a follow-up thyroid function test were all normal.

Upon follow-up at 5 months of age, the patient's macroglossia was still normalcolored, but we noted a redbeef-colored, small, protruding lesion below the left side of tongue base, with left mandibular swelling (Fig. 1B). A neck MRI showed macroglossia and a multiseptated cystic lesion involving both mandibular deep neck spaces, manifesting as an infiltrative signal intensity pattern in the left mandibular area suggesting lymphangioma or lymphangiohemangioma (Fig. 2A, B).

At 8 months of age, the patient's tongue had increased in size and changed to more reddish color (Fig. 1C). Also, the red-beef-colored lesion in the base of the tongue had enlarged, showing intermittent bleeding with trauma (Fig. 1D). At 11 months of age, her physical status, except for macroglossia with mandibular swelling, growth, and developmental status were normal. A follow-up MRI showed no specific change during this interval (Fig. 2C, D).

Case 2

A four-day-old male infant was brought to the outpatient department due to the presence of macroglossia since birth. There was no specific antenatal or family history. He had been delivered vaginally after 39 weeks'

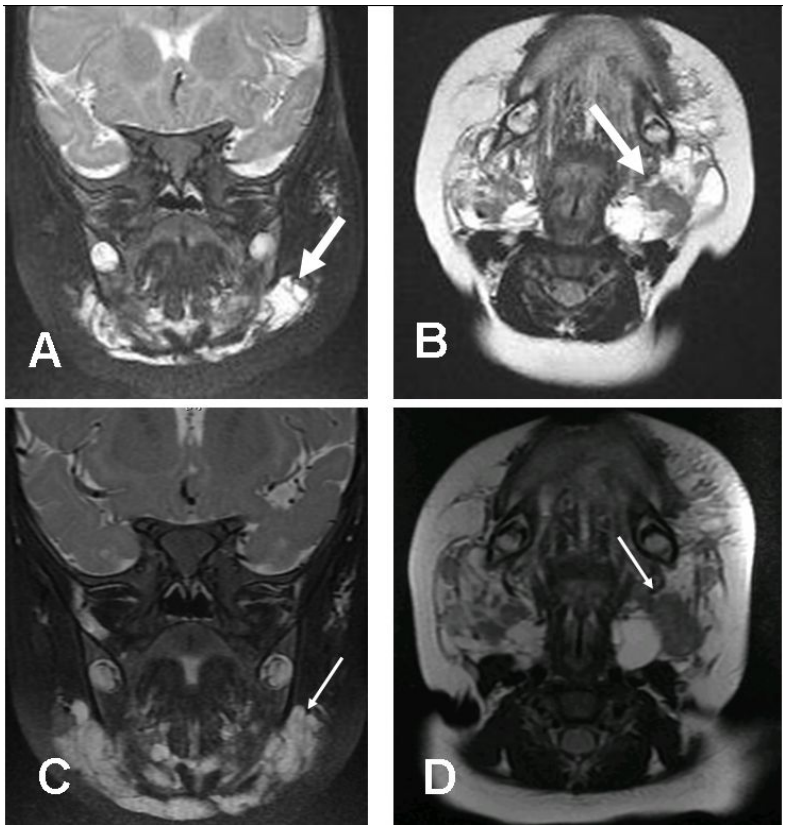

Fig. 2. Neck magnetic resonance imaging (MRI; T2-weighted) of case 1. (A, B) At 5 months of age, macroglossia and multiseptated cystic lesions involving both mandibular deep neck spaces and manifesting as an infiltrative signal intensity pattern in the left mandibular area (suggesting lymphangioma or lymphangiohemangiomas) were noted (white wide arrow). (C, D) By 11 months of age, there had been no specific interval change (white narrow arrow).

gestation with a birth weight of 3,300 g (25-50th percentile), length of 52.1 cm (50-75th percentile), and head circumference of 34.6 cm (50-75th percentile). Upon physical examination, his general appearance was healthy and nonspecific except for the tongue, which was a notably dark, violet color, without vesicles or pebble-like lesions (Fig. 3A, B). He had been fed on breast milk without any feeding difficulties. He did not seem to have pain or discomfort when his tongue was compressed or touched. A neck ultrasound at 8 days of age showed no definite abnormal findings in the mandibular or tongue region.

Upon follow-up examination at 5 weeks of age, his growth and developmental status were normal, however, the tongue had increased in size, with no change in color. Swelling of the left mandibular area was also noted; however, there was no pain or tenderness. A facial MRI showed a poorly defined, infiltrating lesion, largely cystic internally with a subtle enhancement involving the left face and deep neck space (parapharyngeal, parotid and submandibular), suggesting lymphangioma or lymphangiohemangioma (Fig. 4A, B).

At 6 months of age, the patient's tongue had become normal in color and in size, and left mandibular swelling was reduced (Fig. 3C). A small, painless cystic lesion suggesting a dermoid cyst was also noted on the midline suprasternal area (Fig. 3D). Follow-up MRI showed a dermoid or epidermoid cyst in the anterior lower neck, without the remarkable change indicative of lymphangioma or lymphangiohemangioma (Fig. 4C, D), in spite of the improvement in the tongue lesion and the left mandibular swelling.

Discussion

Lymphangiomas are congenital malformations of lymphatic vessels filled with a clear protein-rich fluid con-

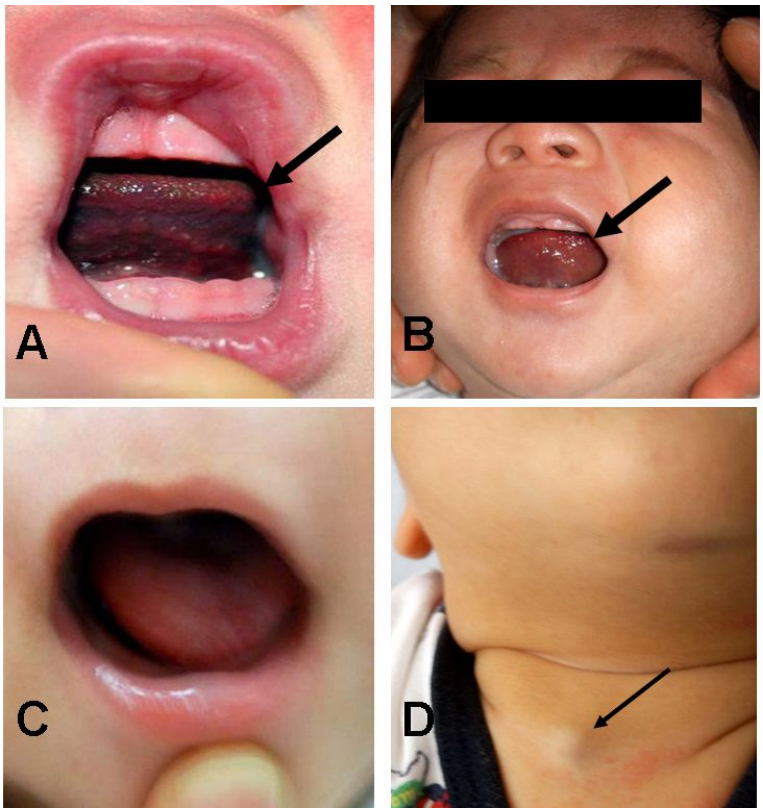

Fig. 3. Gross features of case 2. (A, B) On postnatal day 4, a large, dark, violet-colored tongue was noted (black wide arrow). (C) At 6 months, the tongue had become nearly normal in color and size. (D) A small, painless, cystic lesion, suggesting a dermoid cyst (black narrow arrow), was noted on the midline suprasternal area.

taining few lymph cells. It can also occur in association with hemangioma. Depending on the size of the vessels, this lesion may be classified as capillary lymphangioma (lymphangioma simplex), cavernous lymphangioma, or cystic lymphangioma (cystic hygroma). Among these, the cavernous type is the most common[8]. The prevalent sites are the head and neck region[8, 9], however, lymphangioma rarely affects the oral cavity. Antenatal diagnosis is sometimes possible through ultrasonography[10].

Lymphangioma is the most common cause of secondary macroglossia in infancy. The tumor may be localized in a small area of the tongue or floor of the mouth, or it may diffusely infiltrate these areas[6]. If the tumor is located in a deeper area, it may present as a submucosal mass[11]. Lingual lymphangioma presents specific therapeutic problems because of the almost exclusively microcystic character of the lesions and the marked functional problems they can cause, such as obstruction, bleeding, edema, pain, feeding or breathing difficulty, lingual extrusion, dental issues, and jaw deformities[11].

Our two cases were not initially diagnosed as lymphangioma or lymphangiohemangioma because the patients showed only macroglossia without mass lesions in their necks or mandibular areas. However, later follow up examinations revealed the patient's mandibular swellings, which led us to perform MRIs. Based on the MRI results, the diagnosis of lymphangioma or lymphangiohemangioma was confirmed at 5 months of age in case 1 and 5 weeks of age in case 2.

Lymphangioma may often be misdiagnosed as a number of oral lesions including hemangioma, teratoma, lingual thyroid, dermoid cyst, thyroglossal duct cyst, heterotopic gastric mucosal cyst, and granular cell tumor[12]. It may increase in size, producing macroglossia, which causes interference with swallowing and speech and respiratory difficulties, if left untreated[13]. Lymphangioma lesions are not tender or painful in the initial stage. However, inflammation from trauma or infection causes excessive lym-

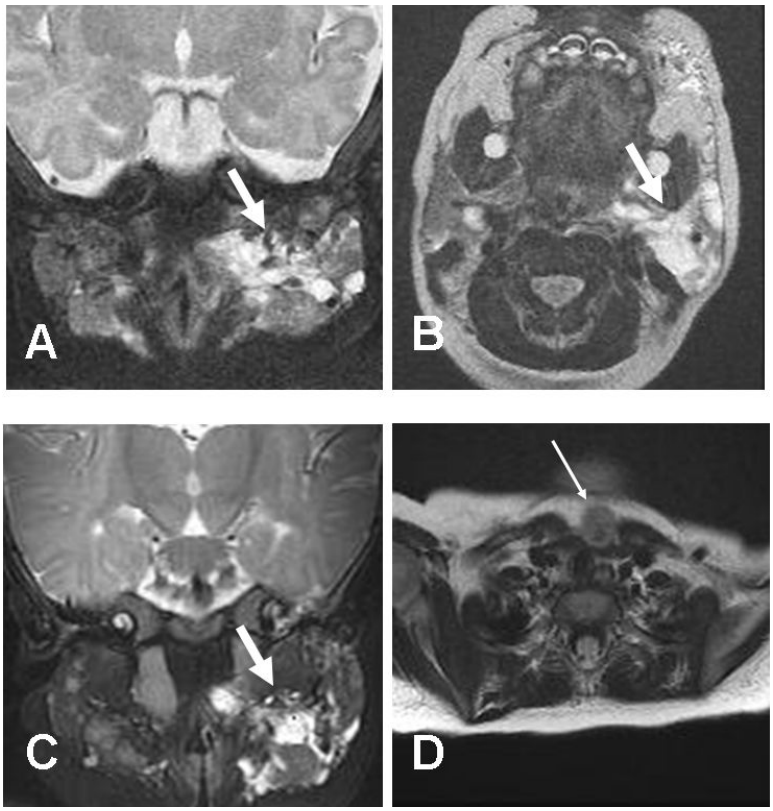

Fig. 4. Facial magnetic resonance imaging (MRI; T2-weighted) of case 2. (A, B) At 5 weeks of age, a poorly defined infiltrating lesion (and subtle enhancement involving the left face and deep neck space (parapharyngeal, parotid, and submandibular), suggesting lymphangioma or lymphangiohemangioma (white wide arrow), was noted. (C, D) By 6 months of age, there had been no remarkable changes except the appearance of a dermoid or epidermoid cyst (white narrow arrow) at the anterior lower neck.

phatic tissue formation, with accompanying severe pain, and may lead to swallowing difficulties and airway obstruction[5].

In lymphangioma, unlike hemangioma, spontaneous regression in patients with symptomatic obstruction is rare[14]. If there is any symptomatic airway obstruction due to lymphangioma, surgery of the upper aerodigestive structures is mainstay of treatment[15, 16]. Hartl et al.[17] surveyed the postoperative outcome of 18 cases of pediatric lymphangiomas with dyspnea from encroachment on the tongue base, the parapharyngeal space, and/or the larynx. They concluded that involvement of the upper airway seems to be the determining prognostic factor in extensive lymphangiomas. Patients with dyspnea due to external compression on the airway responded well to surgery. However, in patients with intrinsic involvement, aggressive surgical treatment did not seem to significantly improve the prognosis. The less aggressive, symptomatic therapy may be an alternative treatment, to avoid mutilating surgery in patients with intrinsic involvement of the airway.

If this tumor occurs in the tongue and/or the floor of the mouth, complete eradication of the tumor is difficult because of its infiltrative nature. Padwa et al.[18] advocated conservative surgery with repeated partial resection, while Neville et al.[8] did not recommend surgery for non-enlarging lymphangioma of the tongue because of the difficulty in removing the tumor and its high recurrence rate.

For macroglossia, repeated partial glossectomy has the approval of many researchers[13]. For obstructive sleep apnea syndrome, bi-level positive pressure ventilation at home can be useful as an alternative to tracheotomy in some patients presenting with moderate airway obstruction from parapharyngeal or oropharyngeal lymphangioma. Sclerosing agents and hemolytic streptococcal preparation have had no notable effect on residual cervical lesions[17]. Recently, surface radiofrequency ablation was suggested for

improving functional impairment, such as impairment of the bite and hindrance due to large volume vesicles and bleeding, and, in particular, for reducing the bleeding[19].

In our two cases, macroglossia-associated feeding difficulty was noted with only one case (case 1) for a short time. Up to the present time, the patients' growth and development are within normal ranges, and no respiratory or speech problems have been noted. For these reasons, we observe the patients regularly without any intervention. The follow-up MRIs showed no remarkable changes in either case, although the size and color of the tongue with left mandibular swelling had improved in case 2.

In conclusion, if an infant shows a tongue of unusual size or color, without congenital hypothyroidism or any associated syndromes, lymphangioma or lymphangiohemangioma in the deep neck space should be suspected, and neck and/or facial MRIs should be performed to confirm, in spite of the absence of neck swelling. Proper diagnosis and management, according to a risk-benefit evaluation, may help improve the patient's quality of life.

한 글 요 약

심 경부 림프관종에 의한 거설증 2례

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림프관종은 드물게 발생하는 림프혈관의 양성 과오종으로, 주로 두경부에 호발하는데, 혀나 구저부 또는 심 경부에 발생할 경우 원심성 림프관 흐름의 방해로 이차적 거설증이 발생할 수 있다. 저자들은 출생 시에는 경부 또는 하악 부종 없이 거설증만 보였던 두 명의 환아에서 각각 생후 5개월과 5주에 하악 부종이 보여 MRI로 심 경부 림프관종을 확인한 2례를 경험하였기에 보고하는 바이다.

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