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A Patient with Cleft Lip Combined with Branchial Cleft Cyst : A Rare Case Report

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ABSTRACT

새열낭종을 동반한 구순열 환아의 증례보고

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전북대학교 치의학전문대학원 구강악안면외과학교실 전북대학교 음성과학연구소

구순구개열은 두경부에서 가장 흔히 발생하는 선천적 기형 중 하나로 554명 중 1명의 빈도로 나타나며 인종에 따라 다양하다. 구순구개열 환아들은 다른 선천적 기형을 동반하여 나타나는 경우가 흔하며, 그 빈도는 문헌에 따라 다르지만 1.5～63.4%로 나타난다. 새열낭종은 두번째 인두강의 폐쇄부전으로 나타나는 선천적 결손으로, 주로 흉쇄유돌근 전방에 나타난다. 구순구개열 환자에 있어 새열낭종을 동반하는 경우는 매우 드물다.

전북대학교 구강악안면외과학 교실에서는 새열낭종을 동반한 구순열 환아 1례를 경험하였다. 환아는 우측 불완전 편측성 구순열로 내원하여, 구순성형술과 함께 우측 목에 존재하던 새열낭종에 대한 제거 수술을 시행하였다. 환아는 출생 당시부터 심실중격결손 및 동맥관개존증 등의 선천적 심장질환 및 갑상선 기능저하증을 가지고 있던 환아로 다양한 선천적인 결손을 동반한 본 환아의 증례를 문헌고찰과 함께 보고하는 바이다.

Key words: 구순구개열, 새열낭종, 선천적 기형

I. INTRODUCTION

The development of the human face is a dynamic process that starts with the formation of facial prominences surrounding the primitive mouth. These structures involve the frontonasal prominence, two maxillary prominences, and two mandibular prominences. Cleft of the lip and palate are the most frequent congenital malformation of the head and neck[1]. These malformations are due to a partial or complete lack of fusion of those facial prominences. Most cases of cleft lip and palate are multifactorial. The incidence of cleft lip and palate is quoted a tup to 1 per 554 live births in south Korea[2]. The incidence varies among different racial groups and is highest among Asians[1].

Patients with cleft lip and palate are frequently associated with congenital malformations. The prevalence of associated anomalies with cleft lip and palate and the types of associated malformations vary between different studies.

Here we report the case of a patient with cleft lip associated with branchial cleft cyst which is also an anomaly often appears on the lateral aspect of the neck, a rare case.

II. CLINICAL REPORT

The female infant, born in October, 2009, was the third child of a 28-year old woman and her 31-year-old husband. Pregnancy was uneventful, no toxic, infectious, or traumatic incidents were reported. Delivery was at 39 weeks' gestation, through cesarean. Birthweight was 3,180 g (50-75 percentile), length 48.7 cm (50-75 percentile), and head circumference 33 cm (25-50 percentile). Cleft lip, congenital heart diseases and hypothyroidism were noted at birth (Figure 1). Echocardiogram showed ventricular septal defect (VSD) with moderate sized and patent ductus arteriosus (PDA) with moderate shunt. Thyroid ultrasound and scan showed a part of left lobe and isthmus of thyroid without ride lobe which comprise of hypothyroidism due to hypoplasia of thyroid. Result of visual evoked potential and auditory evoked potential were normal. Chromosome analysis was also normal.

The patient came to our department with a unilateral incomplete cleft lip on right side (Figure 2). While clinical examination, we found a 'pin-point' fistula on the right side of the anterior neck with serous discharge (Figure 3). We provisionally diagnosed as a branchial cleft cyst and decided to do the surgical management while undergoing primary cheiloplasty. Neither fistula track, nor cystic lesion was observed during the examination by neck ultrasound.

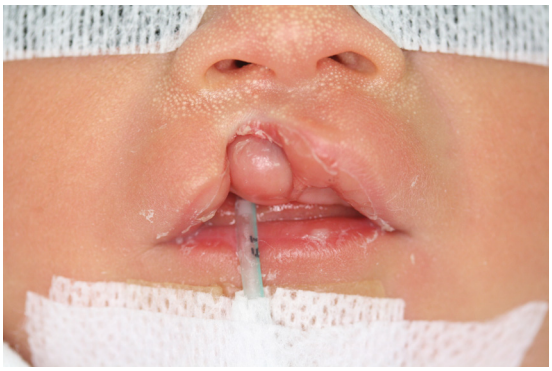

Figure 1. A patient of unilateral incomplete cleft lip on right side at birth.

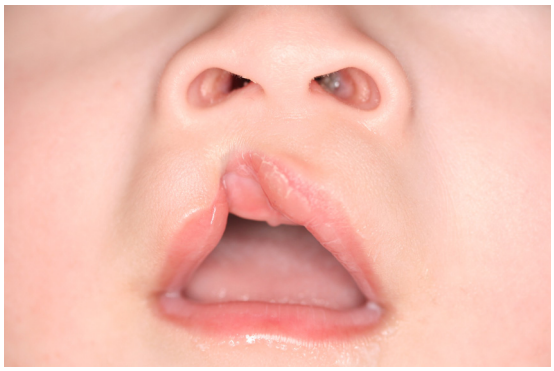

Figure 2. The patient at 3 months of age.

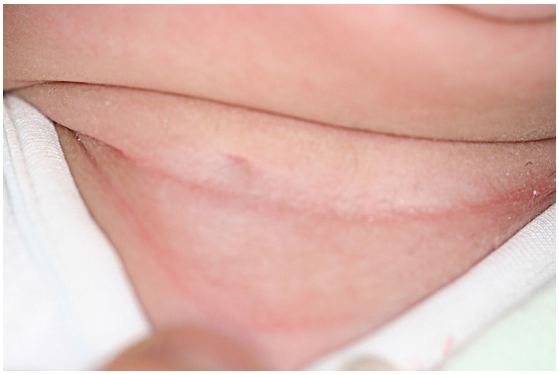

Figure 3. A 'pin-point' fistula on the right side of the anterior neck with serous discharge.

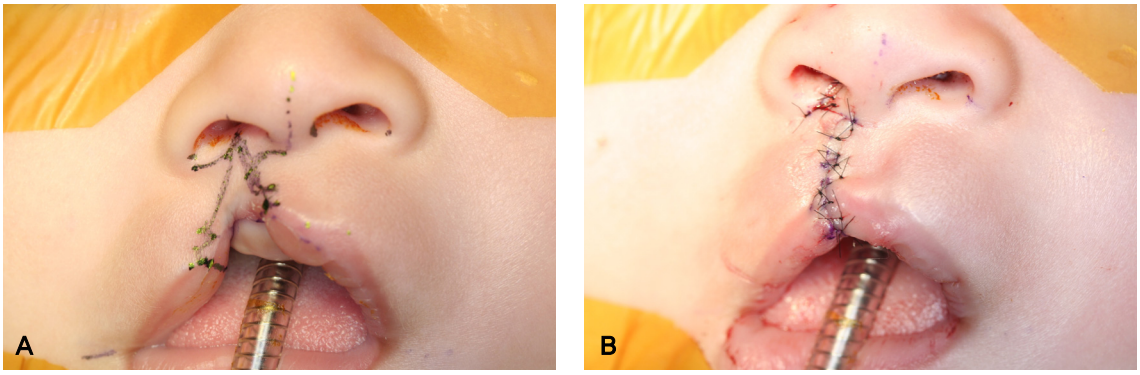

Figure 4. A patient of unilateral incomplete cleft lip on right side treated by the Millard rotation advancement technique with triangular flap, A : Design of triangular flap, B : Immediate post-operative view shows reconstructed symmetric Cupid's bow.

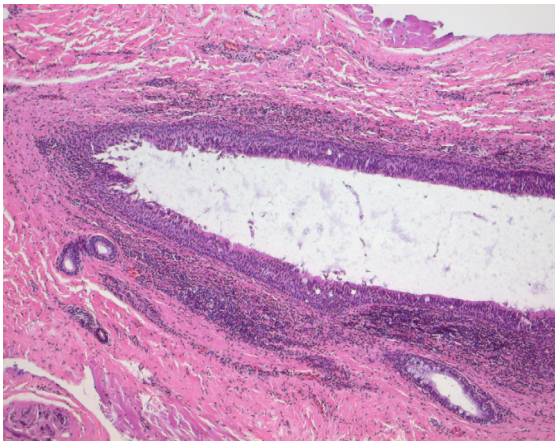

Figure 5. Microscopically, the cyst is lined with respiratory (ciliated columnar) epithelium. Lymphoid tissue is present outside the epithelial lining (×100).

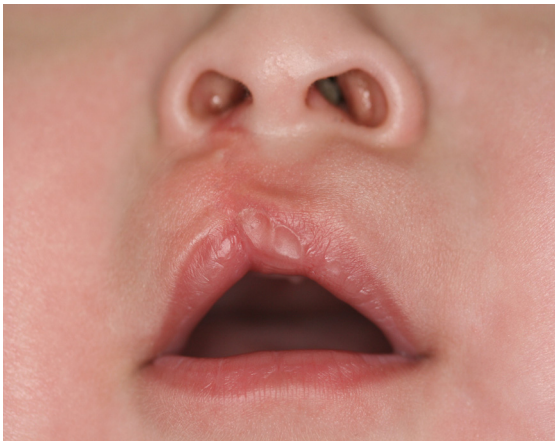

Figure 6. A 2 months after the operation. It shows reconstructed symmetric Cupid's bow and continuity on red vermillion border.

The operation was performed on January in 2010, at 3 months of age, under general anesthesia. The bodyweight was 6.2kg. The Millard rotation advancement technique with triangular flap was used in repair of the unilateral incomplete cleft lip (Figure 4A, B). Complete surgical excision was done on branchial cleft cyst. Final diagnosis was branchial cleft cyst with $1.3 \times 0.3 \times 0.2$cm sized, according to the biopsy result (Figure 5). Stitch out has done after 1 week of operation. We found a reconstructed symmetric Cupid's bow and continuity on red vermillion border after the operation (Figure 6).

III. DISCUSSION

Cleft lip and cleft palate are common defects that result in abnormal facial appearance. Such defects are due to a partial or complete lack of fusion of the maxillary prominence with medial nasal prominence on one or both side.

Patients with cleft lip and palate are frequently associated with other congenital malformations. The prevalence of associated anomalies with cleft lip and palate and the types of associated malformations vary between different studies, ranging from 6% to 63.5%[3]. Wyszynski et al, attributed this wide variation to methodologic factors. These authors suggested that this wide variation is due to 1) differences in case definition and inclusion/exclusion criteria, 2) knowledge and technology available to produce syndrome delineation, 3) variability of clinical expression of associated anomalies, 4) how long after birth cases are examined, 5) selection of patients, sources of ascertainment, and sample size, and 6) true population differences and changes in frequency over time[4].

There are some syndromes that occur with cleft lip and palate. Pierre Robin sequence, Treacher-collins syndrome, Hallermann syndrome, Goldenhar syndrome, Crouzon syndrome, and Cornelia de Lange are the syndromes which can cause cleft lip and palate. In this report, the patient had no evidence of relations with these syndromes as investigations which have done at birth.

Several organ systems might be affected in association with cleft lip and palate, however, there are variations in the literature regarding which organ system is most commonly affected[3]. The most common associated malformations were those affecting cardiovascular malformation, followed by the skeletal system, central nervous system and eye system[1-6]. Cleft lip and palate with branchial cleft cyst is very rare.

Branchial cleft cyst is believed to develop as a result of incomplete obliteration of the cervical sinus or from epithelial rests of the branchial clefts due to failure of the second pharyngeal arch growing caudally over the third and fourth arches. It occur primarily on the lateral side of the neck along the anterior border of the sternocleidomastoid (SCM) muscle at the level of the carotid bifurcation, but may also be found in or around the parotid gland and external ear.

Most (95%) of the branchial cleft cyst arise from the second cleft[7]. The incidence of spontaneous defects of the branchial cleft is approximately 1 per 4000 live births.

IV. CONCLUSION

The overall prevalence of malformations nearly one in five patients, this emphasizes the need for thorough investigation of all the patients with clefts. A routine screening for other malformations, especially cardiac, skeletal, and central nervous system defects may need to be considered in infants with clefts[1]. Also concerning about an unusual symptom found in patient with cleft lip and palate and more evaluation should be done likewise we found branchial cleft cyst during clinical examination.

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