A Case of Prenatally Diagnosed Bilateral Lateral Facial Cleft (Tessier No. 7) with Multiple Anomalies Diagnosed by 3D Ultrasonography

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Fetal lateral cleft lips are rare congenital deformities characterized by macrostomia with lateral facial muscular diastasis and usually associated with other anomalies, especially periauricular skin tags, bony abnormalities of the maxilla and zygoma, temporalis and ear deformities. We report a case of bilateral lateral facial cleft with single nostril and multiple anomalies observed through prenatal two dimensional and three dimensional ultrasonography examinations.

Key Words: Prenatal bilateral lateral cleft lip, Tessier No 7, Three-dimensional ultrasonography

Fetal cleft lips are fairly common facial malformations which can be detected by prenatal sono screening. Cleft lip can range from small vermilion notches to larger defects which extend to the nostrils and alveolar ridge of the maxilla. The worldwide incidence of cleft lips is 1 in 700 births and usually occurs between the middle and the lateral third of the superior lip. Whereas The lateral facial clefts (Tessier No 7) are rare type results from failure of the maxillary and mandibular portions of the first brachial arch to unite and accounts for 5.5% of all clefts with incidence being reported at about 1 : 3,000 to 1 : 5,642 births with generally normal karyotype. Unilateral is predominant compared to bilateral, seen in approximately 90% of cases. While the size of clefts is varied, fissures are seen from slight widening of the mouth to those extending back to the ear. In addition, hypoplastic mastication muscles, tongue, absence of facial nerve function and facial bone abnormalities, and ear deformities are commonly associated.

Although fetal cleft lips can be detected by two-dimensional ultrasonography (2D US), it is easy to miss small size clefts. Three-dimensional ultrasonography (3D US) may provide additional benefits by allowing visualization of these anomalies in a complementary manner. Here, we report a case of bilateral lateral facial clefts with single nostril and the details of accompanied anatomical deformities detected by US within a 27-week gestational patient.

Case report

A 29-year-old primigravida was referred to our hospital at 27 weeks for further evaluation because of an abnormal fetal face. She underwent intrauterine insemination (IUI) for this pregnancy. Quad-test and nuchal translucency showed unre-
marked results without family history for congenital anomalies.
The fetus was an appropriate size for date with polyhydramnios,
reaching an amniotic fluid index 26.8 cm (>97 percentile) by
2D US. The stomach was well noticed and, 2D US revealed
the presence of bilateral fissure of the fetal lip and a single
nostril (Fig. 1). Moreover, bilateral lateral ventriculomegaly
with third ventricular dilatation, muscular type of ventricular
septal defect (VSD), and periauricular skin tags were observed
(Fig. 2). The 3D US examination more accurately showed
macrostomia with bilateral extension to the masseter area,
bilateral abnormal eyelid swelling, right side nostril absence
and preauricular skin tags (Fig. 3).

At 38 weeks plus 6 days’ gestation, the woman delivered
a girl by cesarean section due to progress failure at a birth
weight of 2,612 g with height and head circumference within
normal range. The baby’s apgar scores were 7 at 1 min and
8 at 5 min. On examination, bilateral lip clefts extended to
the masseter area without cleft palate and swelling abnormal
eyelid were shown, the clefts were lined with skin externally
and buccal mucosa internally. The baby’s appearance was not
different as that observed by prenatal US (Fig. 3). Physical
examination on the face indicated single nostril with right
obstructed nostril and multiple bilateral preauricular skin tags.
Orbital area showed palpebral fissure anomaly with eyelid

Fig. 1. 2D ultrasonographic image of the fatal face. (A) Bilateral lateral cleft lips (arrow) (B) Suspicious single nostril of left side (arrow).

Fig. 2. 2D ultrasonographic image of the heart and brain. (A) Bilateral mild lateral ventriculomegaly (transverticular plane). (B) Muscular ventricular septal defect demonstrated by color doppler (arrow).
Fig. 3. Prenatal 3D ultrasonographic images & The newborn face (A) 3D ultrasonography demonstrated fetal bilateral lateral clefts and swelling abnormal eyelid with left single nostril. (B) Multiple preauricular skin tags. (C) Postnatal baby’s appearance is similar with prenatal 3D US images.

coloboma and congenital corneal opacity.

The karyotype of the newborn was a normal 46, XX. Postnatal neck computed tomography (CT) scan showed right maxillary hypoplasia, a low-lying skull base and widening interorbital distance. Brain US revealed absence of septum pellucidum with mild dilatation of both lateral ventricle posterior horns, but there was no evidence of hydrocephalus or craniosynostosis. In addition, muscular type of ventricular septal defect and type I trachea esophageal fistula were diagnosed. After a tracheoesophageal fistulectomy on the postoperative third day, the baby was referred to the other institution for further management.

Discussion

The Tessier’s classification of facial and cranial clefts was reported in 1976. There are 14 clefts based on specific axes along the face and cranium with interruption in relation to the orbit. Tessier number 0-7 clefts are facial clefts and number 9-14 clefts are cranial clefts. The lateral facial clefts include the numbers 6,7,8. The Tessier No 7 cleft is defined as the maxillary cleft in the molar region and between the maxillary tuberosity and the pterygoid process, absence of the zygomatic arch, soft tissue deformities including macrostomia, preauricular tags or microtia, temporalis abnormality, and absence of preauricular hair. 4
Generally, the macrostomia associated with this deformity should be repaired in infancy. Speech and airway management is required during childhood, bone grafting of the maxillar is appropriate at the late childhood if necessary. Additionally, if orthognathic surgery is required, it should be done after completion of growth.5

Up to now, severe Tessier No.7 cleft cases with other anomalies have been reported. They take different methods of postnatal surgical reconstruction and its prognosis depends on combined anomalies especially bony deformities of the mandible and maxillary.6,8

In our case, the fetus had type I trachea esophageal fistula and ventriculomegaly, single nostril, bilateral abnormal eyelids that were not reported previously. Thus, there could be various unexpected deformities. Although additional evaluations may be important after delivery, it is also necessary to observe prenatal US more carefully.

In this case, US revealed almost all abnormalities except type I trachea esophageal fistula and maxillary hypoplasia, which were diagnosed after delivery. We have excluded type I trachea esophageal fistula for the reason of the polyhydramnios because prenatal US allowed viewing of the stomach with appropriate size. 2D US revealed bilateral lateral lip clefts with single nostril, cardiac and brain abnormalities. 3D US was helpful in identifying the extension of the bilateral clefts and demonstrated bilateral multiple periauricular skin tags that were not clearly found through the 2D US.

3D US possesses many benefits helpful to identify fetal faces by offering rendering images and better diagnostic accuracy of face malformations. It can reveal cleft palate, micrognathia and other anatomies not detectable by 2D US due to position and location.5 Combination of 2D and 3D US could be used to clarify the type and severity of facial cleft in addition to other defects.

References

국문요약

태아 횡입술감립증은 드문 산전 기형이다. 특징적으로 큰입증이 관찰되며 다른 기형과 동반되는 경우가 대부분이다. 특히 귀 앞의 피부연성섬유종(periauricular skin tag), 상악골이나 광대의 기형, 측두근이나 귀의 기형이 동반될 수 있다. 2차원과 3차원 초음파(two dimensional and three dimensional ultrasonography)를 통해 횡입술감립증을 진단하였으며 이와 함께 동반되어 산전 진찰된 기형의 증례를 보고한다.

중심 단어: 양측 횡입술감립증, Teisser7번 감립증, 입체초음파