Antenatal Diagnosis of Cleft Lip and Palate by 2-D and 3-D Ultrasound, Case Report and Review of the Literature

YARIK DAMAK VE DUDAĞIN 2 VE 3 BOYUTLU ULTRASONOGRAFİ İLE ANTENATAL TANISI, OLGU SUNUMU VE LİTERATÜR DERLEMESİ

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Abstract

Cleft lip and cleft palate (CL/CP) are among the most common craniofacial birth defects. Although, early diagnosis of CL/CP is possible by prenatal ultrasound, the majority are recognized in the delivery room. The use of ultrasound for the prenatal diagnosis of cleft lip and palate has aided considerably in the early diagnosis of orofacial clefting. As the reliability and validity of ultrasound diagnosis increase, the impact and consequences of early diagnosis should be taken into consideration as well. In this article we present a patient with CL/CP. An extensive review of the literature considering the prenatal diagnosis by ultrasound both two and three-dimensional is also included.

Key Words: Prenatal diagnosis, prenatal ultrasonography, cleft lip, cleft palate

Özet

Yarık damak ve dudak yüz bölgesinde en sık görülen doğumsal kusurlardandır. Doğum öncesi erken tanı ultrasonografi ile mümkün olsa da çoğu hastaya tanı doğum odasında konmaktadır. Yarık damak ve dudak tanısının doğum öncesi konmasında ultrasonografinin kullanılması orofasial yarıklaşmanın erken tanısının konmasına hayli yardımcı olmaktadır. Ultrason tanısının güvenilirliği ve geçerliliği arttıkça, erken tanının etkisi ve sonuçları da göz önüne alınmalıdır. Bu yazıda yarık damak/dudaklı bir hasta sunulmuştur. Aynı zamanda, hem 2 hem de 3 boyutlu ultrasonografi ile prenatal tanı konusunda ayrıntılı bir literatür taraması yapılmıştır.

Anahtar Kelimeler: Prenatal tanı, prenatal ultrasonografi, yarık dudak, yarık damak

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Bilateral CL/CP appears as a defect on both sides of the upper lip extending to the ipsilateral nostril. Orofacial clefting is among the most common congenital abnormalities with rising incidence.[1] Cleft lip with or without an associated cleft palate is encountered in about 1/750 births in the European population. Isolated cleft palate is less common than cleft lip and palate.[2] Major medical and psychological problems may ensue following this diagnosis, for both the child and the family.

Isolated CL/CP is inherited as a complex trait (multifactorial inheritance), and demonstrates strong familial aggregation with a significant genetic component.[3] There is no evidence for classic Mendelian inheritance attributable to any single gene, although a number of genes or loci have been implicated, including transforming growth factor α and a region on chromosome 6p23-24.[4] Maternal teratogens including phenytoin, sodium valproate and methotrexate may play role in the etiology of CL/CP as well.[5]

Herein we present a case of CL/CP in a fetus diagnosed at the 25th gestational week by prenatal USG. Three-dimensional (3-D) ultrasound was a

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helpful diagnostic tool which allowed us to image the defect clearly. An extensive review of the literature about prenatal diagnosis of CL/CP by classical 2-D USG as well as 3-D USG are also included in the discussion section.

Case Report

An 18 year-old woman, gravida 1, para 0, attended our hospital at the 25th week of gestation for routine antenatal control. She had no complaints about her pregnancy. Her first visit was at 19th week of her gestation in an outside antenatal clinic and her antenatal follow-up was uneventful.

Her systemic examination was normal. The laboratory results were also normal except a mild iron deficiency anemia. Zinc and folic acid concentrations were studied as well and they were within normal ranges. During the conversation with the family, a scar on the philtrium of the father was noticed. Upon questioning, the father indicated that he had a cleft lip anomaly which was treated successfully by surgery in his infanthood.

Keeping this in mind, a two-dimensional ultrasonography scan was performed with special emphasis on the facial features of the fetus. The detailed ultrasound examination revealed a cleft lip anomaly with no associated organ or extremity anomalies. No hydramnios was detected (Figure 1). Fallowing this 2-D USG examination a 3-D/4-D (3 dimensional / 4 dimensional) scan was performed and bilateral cleft lip and palate anomalies were definitely demonstrated after rotating the planar images into standard anatomic positions (Figure 2). Both 2-D and 3-D/4-D USG examinations were performed using Voluson 730 (Kretztechnik GE).

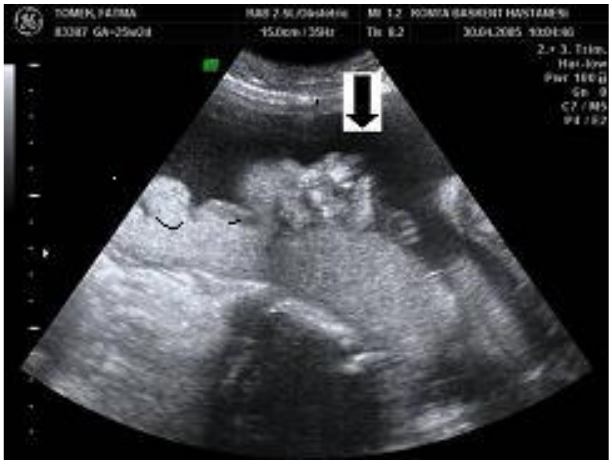

Figure 1. Coronal image of the lower face demonstrates bilateral clefts in the upper lip.

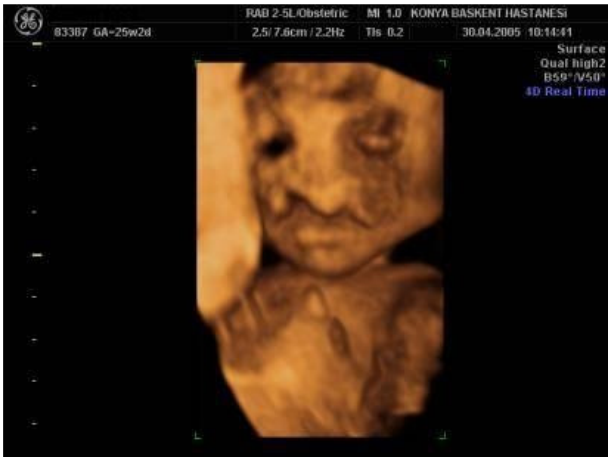

Figure 2. The coronal plane illustrating fetal cleft palate by 3-D USG.

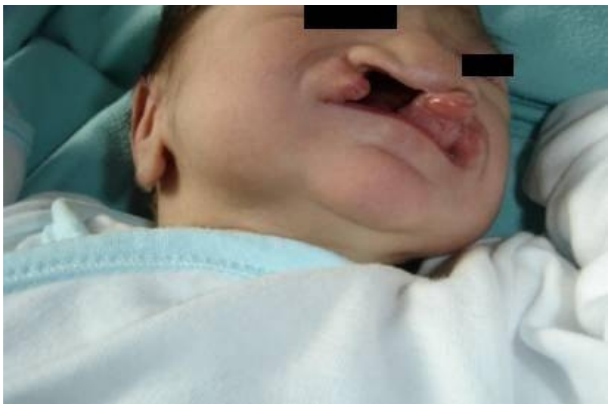

Figure 3. The picture of the newborn confirms presence of bilateral cleft lip and cleft palate.

The family was informed about the outcomes and therapeutic options. Cordocentesis was also offered to the family and performed after obtaining a written informed consent. Chromosomal analysis revealed a normal 46 XY karyotype. At the 40th week of gestation, a 3190 g male fetus with bilateral cleft lip and palate was delivered by cesarean section due to the presence of late decelerations in labor (Figure 3). The early postnatal period was uneventful and the patient was referred to the pedi-

atric and plastic surgery clinics for nutritional rehabilitation and corrective surgery.

Discussion

Although the midline structures of fetal face are completely fused by the 7th week of gestation, visualization of the mandible and maxilla are not clear until the 10th week. Therefore CL/CP cannot be reliably diagnosed until the soft tissues of the fetal face become distinct, which is at the 13-14th weeks, by transabdominal sonography and a little earlier by transvaginal sonography. Prenatal diagnosis of cleft lip by ultrasound is possible as early as 12 weeks. Karyotype determination should be offered in these cases.[6]

A detailed ultrasound exam is mandatory to rule out associated syndromes. Early diagnosis will allow the parents and doctors to discuss and decide about the outcome of the pregnancy and consider possible termination if associated anomalies and syndromes are present. The incidence of structural abnormalities and syndromes associated with cleft lip and palate range between 21% and 38%.[7,8] Over 300 syndromes have been described in association with facial clefts. If the defect lies in the midline, presence of neural tube defects should be considered as well especially holoprosencephaly. Some associated syndromes are Van der Woude syndrome (2%), amnion band sequence, arthrogryposis, chromosomal anomalies especially trisomy 13, holoprosencephaly, Meckel-Gruber syndrome, MURCS association, Nager syndrome, Pierre-Rubin sequence, Roberts syndrome, short rib-polydactly syndrome.[9]

The success of ultrasonographic diagnosis varies widely from series to series. Stoll et al reported a 5.3% detection rate of facial clefts between 1979 and 1988 and 26.5% during the years 1989-1998.[6] Berge et al mentioned in his series of 70 cases that none of the fetuses with isolated cleft lip had additional anomalies. He reported that associated malformations were more frequent in cases with bilateral clefts than in those with unilateral clefts.[10]

Today high resolution 2-D scanning remains the cornerstone of prenatal diagnosis. But with conventional 2-D ultrasound, detection and comprehension of imaging features of cleft lip and palate are difficult. 4-D scanning, by allowing rotation of fetal images and evaluation of the tooth bearing alveolar ridge contour and anterior tooth socket alignment, which were defective in the present case, provides a better anatomic detail and recognizable imaging features.

The ultrasound examination in our case was performed using Voluson 730 (Kretztechnik, GE) which has 2-D, 3-D, 4-D and Doppler facilities. The fetal face was visualized by obtaining a 2-D profile first and defective lip was detected. To rule out whether an associated cleft palate was present or not, 3-D/4-D images were obtained. Coronal plane was chosen to obtain an optimal surface image. Care should be taken to obtain a true coronal plane which is the most critical technical aspect of the study. The three-dimensional ultrasound technique helps us to rotate the frontal facial image 180º along the vertical axis so that the palate, nasal cavity and orbits can be examined one at a time.

Johnson et al, showed that the rate of diagnosis of cleft lip and palate increased from 48% to 76% when three-dimensional ultrasound was employed.[11] Chen et al, claimed that by the use of 3-D sonography the accuracy of the diagnosis reached 100%.[12] On the contrary, Ghi et al, found no additional advantages of 3-D over 2-D considering the diagnosis.[13]

In our opinion, a good high resolution 2-D ultrasound is quite successful at diagnosing abnormalities but inclusion of 3-D and 4-D ultrasound imaging in the examination protocol allows easier and more rapid screening and better comprehension of the anomaly by the examiner and the family as well.

Because of the possible association with other anomalies, a careful fetal survey including fetal echocardiography should be performed when a facial cleft is identified. Due to inadequate swallowing reflex, hydroamnios may develop which justifies frequent antenatal checks including sonograms.

Accurate and early diagnosis of CL/CP is important, and in our case this has allowed informed

parental choice regarding the continuation of the affected pregnancy, better postnatal parental adjustment and early planning of surgical procedures to correct the malformation.

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