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Case Report

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The association of congenital hand reduction defect and uterine anomaly

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Abstract

Objective: Fetal hand anomalies can be overlooked in ultrasonographic examinations. In our report, we aim to present the case of hand reduction defect in a patient with uterine anomaly that we diagnosed during antenatal follow-up with the use of 2D and 3D ultrasound examinations.

Case: Genetic consultation was provided to a patient who had absence of hand in one extremity during the ultrasound control performed for triple test while nothing was detected in the ultrasound for double test screening. After the amniocentesis, the karyotype of fetus was found as normal (46 --). No other anomaly was observed in perinatology consultation, anomaly screening and fetal echocardiography. Remaining gestational period of the patient was free of problem and a single 3300 g fetus was delivered by cesarean section through breech presentation and the pictures of the hand were taken with the permission of the family.

Conclusion: Congenital hand reduction defects may occur together with uterine anomalies. Such anomalies can be diagnosed easily during ultrasonographic examinations in early weeks of gestation, and therefore they should be a part of early anomaly screening procedures.

Keywords: Congenital diagnosis, congenital hand deformities.

Özet: Konjenital el redüksiyon defekti ile uterus anomalisinin birlikteliği

Amaç: Fetal el anomalileri prenatal ultrasonografik incelemelerde gözden kaçabilmektedir. Bu yazımızda uterus anomalisi olan bir olguda; 2 boyutlu ve 3 boyutlu ultrason ile antenatal takipte tanısını koyduğumuz bir el redüksiyon defekti olgusunu sunmayı amaçladık.

Olgu: İkili test taramasında ultrasonda fark edilmeyen ancak üçlü test amacıyla yapılan ultrason kontrolünde bir ekstremitesinde el yokluğu tespit edilen hastaya genetik danışmanlık verildi. Hastaya amniosentez yapıldıktan sonra fetüsün karyotipi normal (46 --) olarak geldi. Perinatoloji konsültasyonu, anomali taraması ve fetal ekokardiyografide eşlik eden başka bir anomali tespit edilmedi. Hastanın geri kalan gebelik süreci sorunsuz geçti ve 3300 g, tekiz canlı makat ile prezante olan fetüs sezaryen operasyonu ile doğurtuldu ve ailenin izni ile elin fotoğrafları çekildi.

Sonuç: Konjenital el redüksiyon defektleri uterus anomalileri ile birliktelik gösterebilen durumlardır. Bu anomalilerin tanısı erken gebelik haftalarında yapılan ultrason incelemesinde rahatlıkla konabilir ve bu nedenle erken anomali taramalarının bir parçası olmalıdır.

Anahtar sözcükler: Konjenital tanı, doğumsal el deformiteleri.

Introduction

Congenital hand reduction defect is the stump end or full or partial absence of an extremity at the distal of a point. Transverse forming defect is also known as transverse reduction defect, transverse melia or transverse arrest.[1]

Congenital anomaly is seen about 1% of newborns, and 10% of such cases are upper extremity anomalies. Congenital hand reduction defect is seen one in every 20,000 births. In 50% of these cases, there is simple transverse reduction defect on forearm or hand, and no other concomitant anomaly. In other 50% of the cases,

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there is more than one reduction and 25% of such cases also have anomalies in other organs or in craniofacial structures.[2] Congenital hand reduction defect of upper extremity is usually an isolated anomaly.

Congenital hand anomalies may occur together with uterine anomalies. In our case, we aimed to discuss hand reduction defect that we diagnosed during antenatal follow-up with the use of 2D and 3D ultrasound examinations.

Case Report

In the obstetric examination of a 27-year-old patient who referred to our clinic for the complaint of delayed menstruation, gestation was observed through fetal cardiac activity which was intrauterine but located on cornual region close to right uterine. The patient who was pregnant for the first time had no kin marriage and specific finding in her family history. The patient was prescribed folic acid support and called for follow-up two weeks later in terms of cornual pregnancy. In her follow-ups, the localization of gestational sac was normal. Double screening test of the patient between 11 and 14 weeks of gestation were within normal ranges. The patient who started to take iron support visited the clinic for AFP screening at 17 weeks of gestation. In the USG examination, absence of hand in the extremity was found and genetic consultation was provided to the patient (**Figs. 1** and **2**). After the genetic amniocentesis, the karyotype of fetus was found as normal (46 --). No other anomaly was observed in perinatology consultation, anomaly screening and fetal echocardiography. Remaining gestational period of the patient was free of problem and a single 3300 g fetus was delivered by cesarean section through breech presentation and the pictures of the hand were taken with the permission of the family (**Fig. 3**). It was found during the operation that uterus was unicornuate, and the presence of non-communicating horn was detected on the left. Both tubes and ovaries were normal.

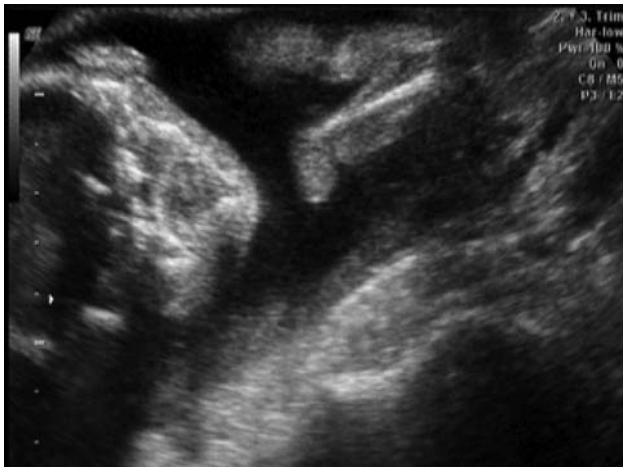

Fig. 1. 2D USG image for the absence of hand.

Discussion

The best time to evaluate fetal hands with ultrasound is between the end of first trimester and mid-period of sec-

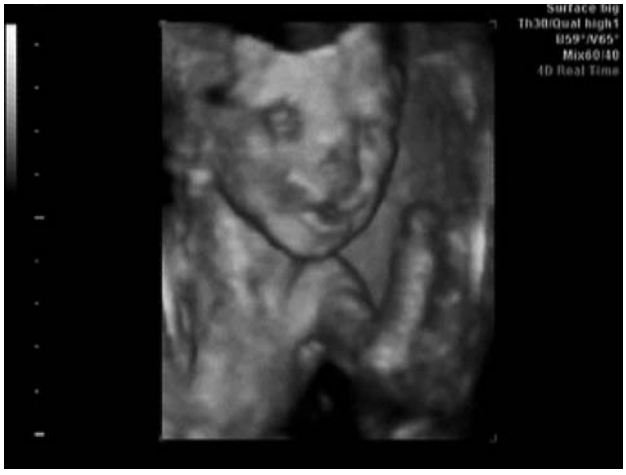

Fig. 2. 3D USG image for the absence of hand.

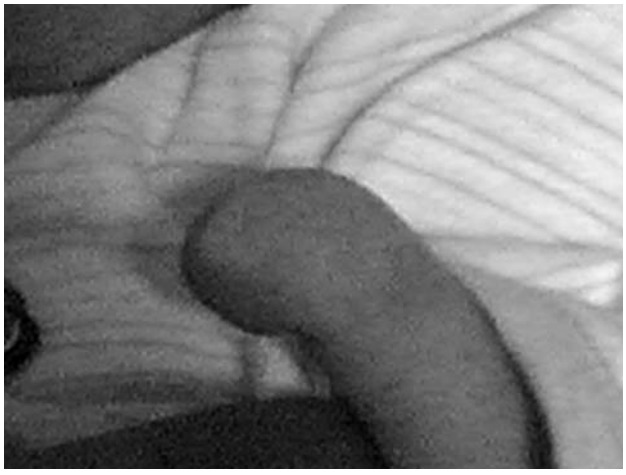

Fig. 3. Postnatal image for the absence of hand.

ond trimester. During this period, fetus moves frequently and hands are on open position more frequently than next periods. 3D USG is not necessary; however, it may be useful to define the morphology better.[3] We detected our case who referred for AFP test at 17 weeks of gestation when we observed the absence of the hand in the extremity. We applied 3D USG to the patient at the same session and confirmed the diagnosis.

Hand anomalies may be isolated and also may accompany to other skeletal and organ anomalies, aneuploidies, syndromes and bone dysplasias.[4,5] Therefore, if a hand anomaly is detected, the patient should be referred to a center where all fetal and cardiac anomalies can be investigated. We therefore carried out genetic screening by amniocentesis, and investigated other anomalies at further weeks by perinatology consultation and anomaly screening. We found no additional concomitant pathology.

Congenital transverse arrest, amniotic band syndrome and Adams-Oliver syndrome (an autosomal dominant inherited syndrome with aplasia cutis and asymmetric transverse extremity defects) are among the distinctive diagnosis of congenital distal hand reduction defect which is also a complication of early period chorion villus sampling.

Amniotic band syndrome is usually sporadic and generally causes asymmetric anomaly.[6] Amniotic band, which develops after embryologic development is completed, may cause many hand defects from hand reduction up to syndactyly.

Congenital transverse arrest is seen rarely although there are great differences among societies and countries in terms of its incidence rate. Genetics, environmental factors and teratogenic agents are shown as the reason in the etiology; however, actual reasons have not been clearly identified. Ninety-eight percent of congenital transverse arrest is unilateral.[7] It was reported that transverse arrest may be seen together with some congenital anomalies; and transverse arrest was found together with congenital band syndrome in one case.[1] Unicornuate uterine anomaly was found in the patient. However, before identifying this uterine anomaly, it was seen in the ultrasound during the first referral of the patient that gestational sac was located in a region close to cornual area. Uterine anomaly was confirmed by the cesarean section carried out due to breech presentation in the case. In the literature, it has been stated that there is a relationship between uterine anomalies and extremity defects. It has been reported that it may cause reduction defects and problems in extremity development associated with compression in uterine anomalies.[8]

Extremity processes can be seen with ultrasound at 8 weeks of gestation at the earliest. Femur and humerus can be seen as of 9 weeks of gestation, tibia/fibula and radius/ulna as of 10 weeks of gestation, fingers and toes as of 11 weeks of gestation, and all long bones can be seen fully as of 11 weeks of gestation.[9,10]

The aim in early anomaly screening is to diagnose minor or major anomalies before it is too late and to be able to get early management chance. Therefore, carefully checking the extremities of fetuses in the ultrasound examination to be performed for double test will help to identify this anomaly at early weeks.

Conclusion

Congenital hand reduction defects may occur together with uterine anomalies. Such anomalies can be diagnosed easily by carrying out ultrasonographic examinations in early weeks of gestation, and therefore they should be a part of early anomaly screening procedures.

Conflicts of Interest: No conflicts declared.

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