First Trimester Prenatal Diagnosis of Anencephaly

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Abstract

Background: Our main aim was to discuss a pregnant patient, whose fetus aided by first trimester ultrasonography had been diagnosed with anencephaly, and who later terminated her pregnancy.

Case: The patient was 34 years old, gravida 1, ultrasonographic imaging revealed the cranial surface to be mishapened, and instead of the normal oval shape it was lengthened and resembled a cap. An ultrasound scan performed at 11 weeks 2 days showed the characteristic form of anencephaly, and conclusively it was decided to terminate the pregnancy.

Conclusion: Anencephaly is a malformation that bears a great mortality rate. Although it is thought that the cranium can be visulized ultrsonographically after 11 weeks of gestation, and therefore cannot be scanned for before then, cephalic length, shape, signsof disorganised tissue and a scan specific to anencephaly may reveal cases of anencephaly at a higher rate.

Keywords: Anencephaly, ultrasonography, pregnancy termination.

Birinci trimesterde prenatal anensefali tanısı: olgu sunumu

Amaç: Birinci trimesterde yapılan ultrasonografik inceleme ile anensefali tanısı konan ve gebelik terminasyonu yapılan bir hastayı olgu sunumu olarak tartışmayı amaçladık.

Olgu: Otuzdört yaşında, 12 haftalık ilk gebeliği olan hastanın ultrasonografik incelemesinde, fetusun baş kısmının geliştiği bölgede düzensiz görünüm izlendi. Bu bölgenin oval şekli yerine, bir şapka gibi uzanarak üçgen biçimini aldığı izlendi. Gebeliğin 13 haftasında tekrar edilen sonografik incelemede başın karakteristik görüntüsüyle anensefali olduğu tespit edilip, terminasyonuna karar verildi .

Sonuç: Anensefali yaşamla bağdaşmayan bir malformasyondur. Bu nedenle ne kadar erken tanı konulursa hem hekim hem de aile yönünden tedavide büyük kolaylık söz konusudur. Kraniyumun gebeliğin 11 haftasından itibaren ultrasonografide vizüalize olması nedeniyle birinci trimesterde ultrasonla sefalik boy ve şekil bozuklukları, dezorganize görünüm gibi sonografik bulgular bize erken tanı için uyarıcı işaret olmakta ve erken tanı konulmasına imkan vermektedir.

Anahtar Sözcükler: Anensefali, birinci trimester, prenatal tanı.

Introduction

Following cardiac anomalies,neural tube defects (NTD) are the most common of congenital malformations (with a fequency of 3/1000 in our country). With the breakthrough and development of ultrasonography starting from the 1970s this entity is currently ultrasonographically scanned for during routine antenatal visits.[1] Anencephaly,a subtype of NTD,occurs when the cephalic end of the neural tube fails to close by the 6th week.Although anencephaly may be defined as the complete or partial absence of the fetal brain, it is also

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strongly believed that anencephaly starts out as exencephaly (meaning the defective development of the cranium) or acrania (the absence of the cranium) and that chronic exposure to amniotic fluid and certain mechanical traumas later transform the existing cerebral hemisferes into degenerated, disorganised tissue covered with an edematous and angiomatous memebrane.[2] Squamous cranial bones develope transmemebraneously and start to ossify by the 10th week. this may be detected ultrasonographically by the 11th week.[3] Anencephaly can generally be detected ultrasonographically in the 2nd and 3rd trimesters when signs like; absence of bones above the base of the head (especially absence of supraorbital bones); absence of both hemispheres; protruding eyes leading to the characteristic 'frog like' look is visualised.İt can be therefore said that ultrasonographiacal diagnosis of anencephaly in the second trimester may be based on the absence of both cerebral hemispheres and calvarium,while in the first trimester visualization of normally developing cerebral hemispheres,enclosed in a non calcifying structure is characteristic.[4,5] That anencephaly brings about lethal results, leads us to persue means of diagnosing and scanning for this malformation, allowing us to offer parents better counselling and if warrented an earlier right to terminate such pregnancies. In this review we aimed at reporting a case diagnosed with anencephaly at an early gestational week.

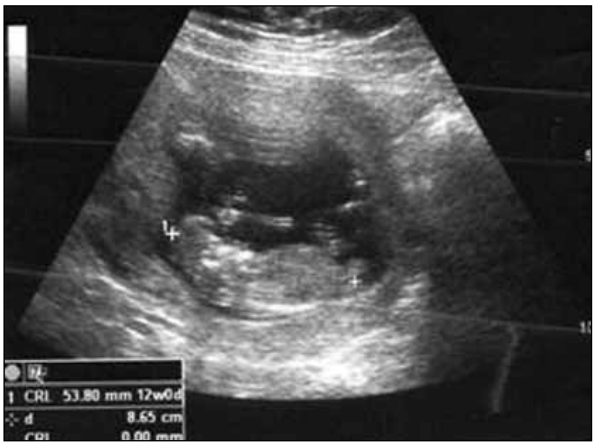

Figure 1. Anensefali düşünülen fetusun 12. gebelik haftasındaki ultrasonografik görünümü.

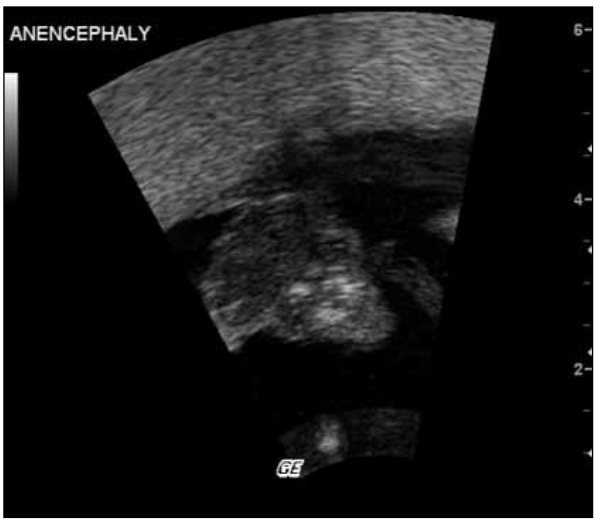

Figure 2. Anensefali düşünülen fetusun 13. gebelik haftasındaki ultrasonografik görünümü.

Case

A thirtyfour year old primipara with a pregnancy consistant with 12 weeks, applied to our day clinic for a routine antenatal visit. An ultrasonographical scan performed at this week showed the fetus as having an irregularly shaped cephalic end where a triangular caplike structure replace the expected normal oval shape. Additoinally the midline was indistinctive and both of the choroid plexi took up a disorganized appearance. When looked at in the saggittal profile the usually rounded forehead appeared flattened (Figure 1). The parents were informed of our suspicion of anencephaly and was told to return in two weeks time for confirmation. At 13 weeks the abnormal shape of the fetal head and its characteristic appearance prompted us to believe the fetus was anencephalic,and after necessary counselling on the topic, it was decided to terminate the pregnancy (Figure 2). Gross evaluation of the fetus showed that it lacked all cranial bones as well as brain tissue (Figure 2). The diagnosis of anen-

Figure 3. Termine edilen fetusun makroskopik görünümü.

cephaly was made macroscopically although hystopathological analysis of tissue samples taken from the base of the head detected the presence of disorganized glial tissue.

Discussion

Anencephaly, a form of NTD can currently be detected in the second trimester with increased levels of maternal serum alpha fetoprotein (MSAFP) or with an ultrasonographic scan in the 2nd or 3rd trimester showing absence of cranium above the orbita or the absence of the cerebral hemispheres. When suspected, amniyocenthesis may be performed, in which case an increased level of AFP and acetylcholinesterase may support the diagnosis.[6] According to M. Jaquier et al, 15% of anencephalic fetuses die in utero,and 67% of anencephalic fetuses born viable die in the first 24 hourrs where only a small percentage (3%) live up until 3-6 days.[7] The fact that this entity has such a high mortalite rate renders early scanning important, and if found necessary allows the parents the chance to terminate the pregnancy or to continue with a much more conscient state of mind. Due to the fact that the cranium begins to ossify at 10 weeks and that this becomes evident by the 11th week in the form of an ultrasonographical hyperecogenity, some believe early diagnosis of anencephaly via ultrasonographical scanning unsafe.[8] For example in a multicentered first trimester scan 26% of 31 anencephalic fetuses were not detected between 10-14 weeks.[9] This has prompted many others to find and utilize other characteristic markers that will tie in with its pathogenesis, besides the second and third trimester appearances of anencephaly. Accordingly, some have elaborated on detection of; acrania seen between 9-10 weeks, a small and irregular cephalic pole, cerebral lobes chronically exposed to amniotic fluid finally taking on the appearance of 'Mickey Mouse' in a sagittal plan, CRL retardation,[10] also 'cap' or horn like appearances of disorganised or deformed brain tissue left exposed to amniotic fluid. İn our case we detected a mis-shapened cephalus, but the cephalic pole was not small for gestational age. We believe that the distinctive cap like shape was due to the protrusion of cerebral tissue through the defective cranium. Becker et al reported a case where at 9 wks 3 days displayed a smaller and misshapened (loss of oval shape and protrusion of cephalic tissue) cephalic pole arousing suspicion of exencephaly.[11] Kennedy et al performed an ultrasound scan and was able to identify acrania by showing disorganized brain tissue at 10 weeks of gestation. Bronshtein and Ornoy identified a case of acrania at 9-11 wks,and later supported this by repeating their ultrasonogarphical scan at 12 wks, finally diagnosing the fetus with anencephaly at 14 wks. Sepulveda et al describes progressive decrease in the ratio of crown chin length (CCL) to crown rump length (CRL). This finding shows that in anencephalic fetuses, the fetal brain tissue gradually decreases in size. That CCL can be easily demonstrated and is unlikely to change according to fetal position makes the CCL measurement a potentially useful parameter.[13] Chatzipapas et al conducted a scan including 5388 fetuses and reported 6

cases of anencephaly (with an incidence of 1:1000). 5 of these 6 cases exhibited the 'Mickey Mouse' sign and CRL was smaller in comparison with uneffected fetuses. Follow up confirmed that all diagnosis of anencephaly was obtained in the first trimester.[10] Johnson et al scanned 55.237 fetuses between 10th-14th wks and detected 47 fetuses demonstrating signs of anencephaly (with incidence of 1/1175). In the first part of the study, 34.830 scans detected only 8 of the 31 cases of anencephaly, whereas in the second part of the study, specific search for signs of anencephaly yielded beter results where all cases of anencephaly were found between 10-14 weeks (16/16). In conclusion, anencephaly is a malformation showing high mortality. That the cranium is ultrasonographically visualized by the 11th week has given some authors reason to think that anencephaly cannot be successfully diagnosed in the first trimester. On the contrary, findings involving cephalic size and shape, and disorganized appearance,and specific scanning for anencephaly renders detection possible with high success rates. Standarised measurements are crucial for successful future first trimester scanning.

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