Iniencephaly: Early Prenatal Diagnosis and Postdelivery Findings

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Iniencephaly is a rare malformation characterized by occipital bone defect,cervical dysraphism and retroflexion of fetal head.Because of its lethal prognosis termination of pregnancy is recommended when this condition is diagnosed before viability.Early prenatally diagnosed cases are rare because careful and early ultrasonograhic evaluation is necessary.In this case report we present an iniencephaly early diagnosed by sonography,in which therapeutic abortus was induced and examined after delivery.

Key Words: Iniencephaly, Prenatal diagnosis, Prenatal ultrasonography

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Introduction

Iniencephaly was first reported by Saint-Hilaire.[1] and is a rare, fatal neural tube defect including occipital bone defect,rachischisis of the cervical and thoracic spine and extreme retroflexion of the head. Incidence varies from 1/1000 to 1/100000.[2] Failure to recognize iniencephaly when it is a part of a more complex malformations may explain a lower incidence in some reports.In etiology, maternal use of some antibiotics, antihistamines, antitumor agents, smoking, alcohol use and syphilis are thought to be associated. We present a case of iniencephaly, diagnosed by early prenatal ultrasonography and discussed with postdelivery morphologic findings.

Case Report

Our patient was 24 years old with her first pragnancy, A 11 week old female fetus was observed by transvaginal ultrasonography.Prenatally there were no exposure to any teratogenic agents and no consanguineous family history. Transvaginal ultrasonographic examination of the fetus revealed occipital bone defect, abnormal vertebral structure and retroflexion of the head. The crown-rump length was compatible with the gestational age and we also observed encephalocele and contracted thorax of the fetus (Figure 1). The diagnosis was made as a result of these findings and discussed with the parents. Pregnany was terminated at 12 weeks gestation by labor induction. Postabortus morphologic findings were concordant with prenatal sonographic findings. The crown-rump length was 12 cm.The head was retroflexed with short neck and there was a large occipital bone defect in the cranium. Low-set ears and depressed nose were observed (Figure 2). There were no any other abnormalities in the visceral organs. We confirmd our diagnosis with these findings.

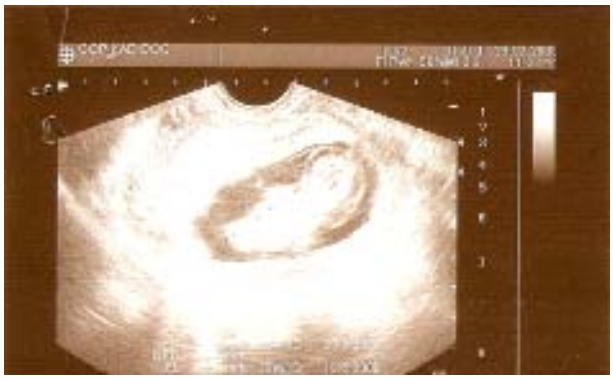

Figure 1: Ultrasonographic examination of the fetus revealed occipital bone defect,abnormal vertebral structure and retroflexion of the head.The crown-rump length was compatible with the gestational age

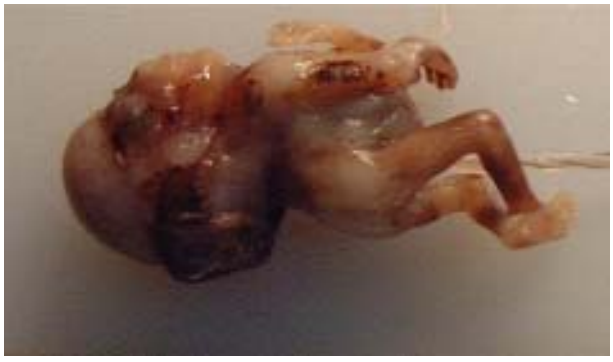

Figure 2: The crown-rump length was 12cm.The head was retroflexed witk short neck and was a large occipital bone defect in the cranium.Low-set ears and depressed nose were observed

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Discussion

Iniencephaly is a neural tube defect. The developmental defects of neural tube and vertebras were consisted just after the 26 and 30th days of conception. The recurrence incidence of iniencephaly is less than 1%.This rate may be higher in families with a history of neural tube defects.[3] The dilatation and rupture of previously closed neural tubes and any error at the time of embryogenesis were accepted as two hypothesis about iniencephaly.[4] In etiology,maternal use of some antibiotics,antihistamines,antitumor agents,smoking, alcohol use and syphilis are thought to be associated. In our patient none of them were present but low parity and low socioeconomic status were present as risk factors. Most cases are sporadic and probably secondary to polygenic inheritance.[3]

The prenatal diagnosis of iniencephaly can be confidently made by ultrasound during the early second trimester as it has been demonstrated by several authors. The main diagnostic features include dorsoflexion of the head and short and abnormal spinal structure. Ultrasound evaluation should include a sagital section through the spine for accurate evaluation of vertebral anomalies with close observation of the occiput and the foramen magnum.[5] In 1988,Shoham et al. described the sonographic diagnostic criteries as follows; the absence of parietal bones, cerebral tissue and normal cervical structure, rachischisis and myelomeningocele.[6]

Polihidramnios and multipl other anomalies are associated in approximately 84% of cases. These anomalies are, hydrocephalus, anencephaly, cyclopia, cleft lip/palate, single umbilical artery and congenital heart diseases.[7] In our case, none of them were present.

As it has been stated by several authors, prenatal diagnosis of iniencephaly can be made by ultrasound from the second trimester onwards. Marton et al. first diagnosed iniencephaly in the first trimester of pregnancy at 10 weeks gestation.[8] Cuillier et al. reported the earliest prenatal diagnosis of iniencephalus at 9 weeks gestation by transvaginal sonography.[9] There is no any other reported cases in the first trimester other than these two cases.

Prenatal diagnosis of iniencephaly should include a differential diagnosis of anencephaly, cervical myelomeningocele and Klippel-Feill syndrome. Because of the latality of the anomaly, termination of the pregnancy is commonplace in countries where elective abortion is legal. Although, fetuses with neural tube defects have associated chromosomal abnormalities, we were unable to confirm such association. Detailed sonographic evaluation must be recommended for the next pregnancies.

İniensefali: Erken Gebelikte Tanı ve Doğum Sonrası Bulgular

İniensefali, oksipital kemik defekti, servikal disrafizm ve fetal başın aşırı retrofleksiyonu ile karakterize nadir görülen bir malformasyondur. Prognozunun kötü olmasından dolayı tanı konulduğunda fetal viabilite kazanılmadan gebeliğin sonlandırılması düşünülmelidir. Dikkatli ve erken ultrasonografik değerlendirme gerektirdiğinden erken gebelik döneminde tanı zor olabilmektedir. Bu olgu sunumunda 13. gebelik haftasında tanı koyduğumuz hastayı ve doğum sonrası bulguları tartıştık.

Anahtar Kelimeler: İniensefali, Prenatal tanı, Prenatal ultrasonografi

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