INIENCEPHALY: A RARE NEURAL TUBE DEFECT◆

(İniensefali: Nadir Bir Nöral Tüp Defekti)

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Summary

Background: Iniencephaly is a rare craniocervical deformity characterized by marked, fixed retroflexion of the head and a short, immobile neck. We report a case of iniencephaly diagnosed prenatally by ultrasound examination.

Case presentation: A 20-year-old gravida 1 woman was first seen in our antenatal clinic at 24 weeks' pregnancy. On ultrasound examination a fixed retroflexion of the head, severe microcephaly, anencephaly, meningocele, deformed spine with cervical dysraphism, and omphalocele were found. She delivered a 440 g, 24 weeks- old female fetus. Postmortem examination confirmed the diagnosis of iniencephaly.

Discussion: The ultrasonic diagnosis of iniencephaly should be based on the finding of extreme retroflexion of the head accompanied by an abnormally short and deformed spine. Early diagnosis and termination of pregnancy reduces the maternal risks. The mother should be recommended folic acid supplementation for future pregnancies.

Key words: Iniencephaly, neural tube defect, prenatal ultrasonography.

Özet

Giriş: *İniensefali, başın fikse ve belirgin retrofleksiyonu, ayrıca kısa ve hareketsiz ense ile karakterize nadir bir kranioservikal deformitedir. Biz bu vaka sunumunda prenatal dönemde ultrasonografi ile tanı koyduğumuz iniensefali vakasını bildirdik.*

Vaka Sunumu: *İlk gebeliğin 24. haftasında gebe polikliniğine başvuran hastanın yapılan ultrasonografi muayenesinde fetal başın fikse enseye yapışık olması, şiddetli mikrosefali, anensefali, meningosel, servikal açıklıkla beraber deforme olmuş omurga ve omfalosel saptandı. Bu bulgularla 440 g ağırlığında kız bebek doğurtuldu. Doğum sonrası yapılan otopside iniensefali tanısı doğrulandı.*

Tartışma: *İniensefalinin ultrasonografik olarak tanısında temel olarak oldukça kısa ve deforme olmuş omurga ile birlikte başın ileri derecede retrofleksiyonu mutlaka bulunmalıdır. Erken tanı ve gebelik terminasyonu maternal riskleri azaltmaktadır. Sonraki gebeliklerinden önce folik asit takviyesi önerilmelidir.*

Anahtar kelimeler: *İniensefali, nöral tüp defekti, prenatal ultrasonografi*

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BACKGROUND

Iniencephaly is a lethal and rare neural tube defect characterized by the triad of occipital bone defect cervical dysraphism, fixed retroflexion of the fetal head and severe lordosis of the cervicothoracic spine [(1)]. Iniencephaly is a rare neural tube defect. Early diagnosis and termination of pregnancy reduces maternal risks. The prognosis is extremely poor, as most affected newborns die within a few hours. A case with iniencephaly is diagnosed by prenatal ultrasonography performed at Haseki Research and Training Hospital.

CASE PRESENTATION

A 24 -weeks old female fetus, the first sib of the family. The prenatal history was unremarkable. Sonography performed at 24 weeks gestation showed that the amniotic fluid volume was normal, and that fetal cardiac activity was present. The femur length of the fetus was compatible with 24 weeks of gestation. In ultrasonographic examination, an occipital bone defect, marked retroflexion of the fetal head with a short neck, anencephaly, meningocele and omphalocele were noted (Figure 1). A therapeutic abortion was induced. The vaginal delivery resulted in a stillborn infant. Necropsy revealed 440 g, 24 weeks- old female fetus. The crown-heel length was 25 cm. The cranium was anencephalic with iniencephaly: the head was retroflexed with a short neck and rachischisis extending from cervical thoracic region were observed. The face was upturned due to the neck; the skin of the mandible was joined to the breast. An omphalocele sac including the liver and intestines and left sided diaphragmatic hernia were noted. In postmortem examination, a retro placental hematoma was also demonstrated. Club foot was present on both sides (Figure 2). The maternal serologic tests for syphilis were negative. The mother didn't use folic asid before and during the prenatal period.

Figure 1. Anencephaly, meningocele and omphalocele (ultrasonography)

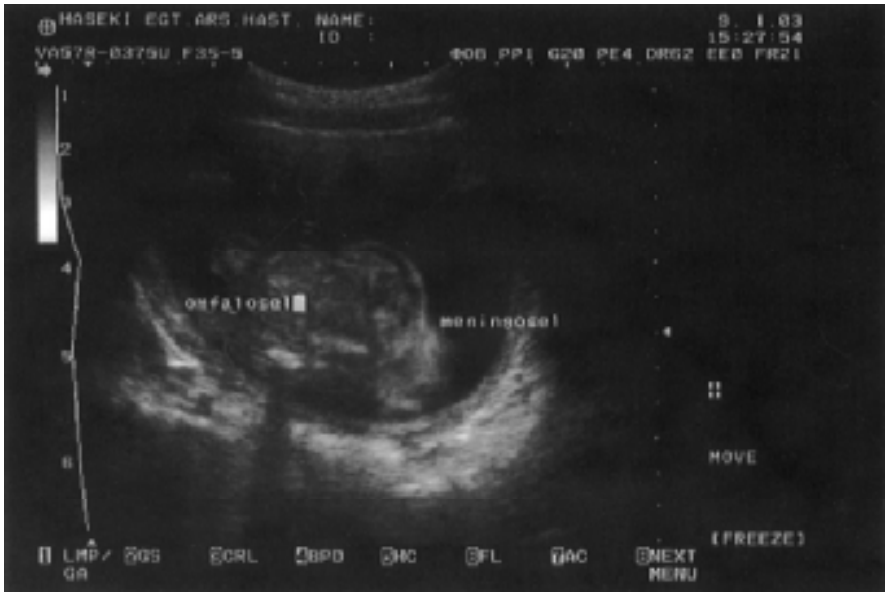
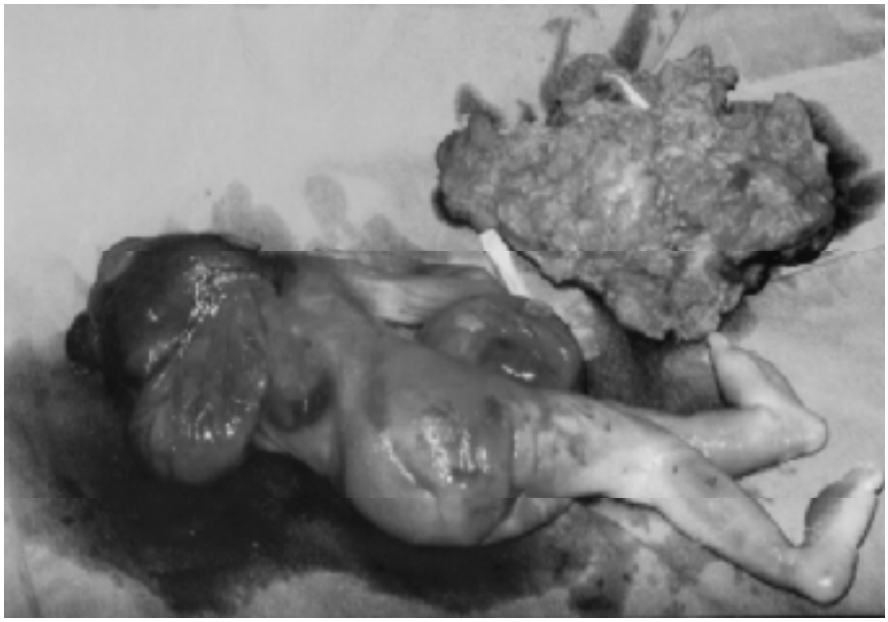


Figure 2. Iniencephaly, anencephaly, rachischisis (cervical and thoracic vertebrae), omphalocele, club-foot, retroplacental hematoma (after delivery).



DISCUSSION

The first description of iniencephaly is attributed to Saint-Hilarie in 1836 [2]. Almost 200 cases have been reported in the literature to date. The incidence varies from 1 in 1000 to less than 1 in 100000 deliveries [3,4]. The developmental pathogenesis of this condition is not known, although it has been described as a consequence of congenital syphilis and, in animal models, associated to the use of teratogenic chemicals such as triparanol, streptonigrin and vincaleukoblastine [4]. By this case, none of the signs related congenital syphilis were detected. Antibiotics, especially sulfonamides and certain antihistamines were thought to be associated with neural tube defects [5]. Most cases, however, are sporadic a probably secondary to polygenic inheritance [6]. Folic acid deficiency may be etiologic factor in neural tube defects. Ultrasound evaluation should include a sagittal section through the spine for accurate evaluation of vertebral anomalies with close observation of the occipital bone and foramen magnum. Associated anomalies involving the CNS include cerebral defects such as anencephaly, encephalocele, microcephaly, hydrocephaly, holoprosencephaly, posterior fossa defects and spinal defects such as the characteristic cervical dysraphism and fixed cervical hyperlordosis [7,8]. The most frequent accompanying malformation is spina bifida in 74% of cases, followed by diaphragmatic hernia 37%; small adrenal glands 37%, and club-foot 32%, hypoplastic lungs, single umbilical artery and omphalocele were found in 26% of the cases. Cardiovascular defects, genitourinary malformations, cyclopia, cleft lip and palate, imperforated anus and club-foot are also associated anomalies [8]. A case with chorangiosis of the placenta has been reported [9]. The literature has only documented six survivors with iniencephaly [10,11, and 12]. Mild iniencephaly must be differentiated from severe Klippel-Feil anomaly if surgical release of the retroflexed head is planned. The survivor with omphalocele underwent three surgical interventions, but this infant had a normal neurological examination up to the third year of age.

During delivery the fetus may cause obstructed labor and maternal trauma. Early diagnosis and termination of pregnancy reduces the maternal risks. There is a recurrence risk of iniencephaly with an incidence of less than 1% and this rate may be higher in families with a history of neural tube defect. The

mother was recommended folic acid supplementation for future pregnancies. We recommend supplementation of the diet of women, beginning three months prior to an anticipated pregnancy.

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