Case
Report

Prenatal Diagnosis of Iniencephaly: Clues and Pitfalls

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Iniencephaly is a congenital malformation consisting of occipital bone defect with enlargement of the foramen magnum, cervical dysraphism and fixed retroflection of the head due to spinal deformities. A 38-years-old woman presented to Etlik Zubeyde Hanim Maternity Hospital perinatology clinic at 18 week of gestation. Ultrasonography revealed a large occipital encephalocele and short cervicothoracic spine. Termination of pregnancy was recommended with the diagnosis of iniencephaly but the family refused it because of religious concerns. She was lost in follow-up but at 38 weeks she presented to our department with regular uterine contractions. Ultrasonography revealed extreme retroflexion of the head, a very short cervicothoracic spine, an encephalocele and marked polyhydramnios. Due to severe retroflexion of the head, a caudal sweep motion of the ultrasound probe gave an impression of a posteriorly placed bladder. Normal chin and neck relation was lacking. A cesarean section was decided due to marked flexion of the spine. A female fetus was delivered weighing 2790 g, with Apgar scores of 1 at 1 min and 1 at 5 min. Iniencephaly is a lethal abnormality which may be diagnosed prenatally with striking ultrasonographic features.

Key Words: Iniencephaly, Cervical dysraphism, Encephalocele

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Introduction

Iniencephaly is a congenital malformation consisting of occipital bone defect with enlargement of the foramen magnum, cervical dysraphism and fixed retroflection of the head due to spinal deformities. This rare abnormality is usually lethal, however surviving fetuses have been reported.[1] Iniencephaly is classified into two groups by Lewis;[2] cases with or without encephalocele are classified as iniencephaly apertus and iniencephaly clausus, respectively. Here we would like to report an iniencephaly apertus case which was diagnosed prenatally but delivered at term due to parents will.

Case Report

A 38-years-old woman presented to Etlik Zubeyde Hanım Maternity Hospital perinatology clinic at 18 week of gestation. This was her 3rd pregnancy and had one healthy son. There was first degree consanguinity. She did not receive routine prenatal care; she did not get a first or second trimester Down syndrome screening test. Her past medical history was otherwise unremarkable. At 18 weeks' gestation, a sonographic examination revealed a suspicion for fetal spine anomaly and she was referred to our center.

This was a female fetus and biometric measurements were appropriate for gestational age. Ultrasonography revealed a large occipital encephalocele and short cervicothoracic spine. No additional anomaly was present. Termination of pregnancy was recommended with the diagnosis of iniencephaly but the family refused it because of religious concerns. She was lost in follow-up but at 38 weeks she presented to our department with regular uterine contractions. Two-dimensional (2D), 3-dimensional (3D) and 4-dimensional (4D) ultrasonography were used for examination. Ultrasonography revealed extreme retroflexion of the head causing upward-turned face so-called star-gazing fetus, a very short cervicothoracic spine, an encephalocele and marked polyhydramnios (Figure 1). The posterior part of the skull was wider than the anterior part (Figure 2) and due to retroflexion of the head; femoral heads were visible in the biparietal diameter (BPD) plan. Because of an almost 180 degrees retroflexion of the head, a caudal sweep motion of the ultrasound probe from the BPD plan revealed an image which gives an impression of a posteriorly placed bladder (Figure 2). Performing 3D and 4D ultrasonography were challenging due to complexity of the malformation combined with an engaged term fetus. However, lack of normal chin and neck relation was striking. The heart anatomy was normal with four chambers and normally oriented great vessels. Pelvic ex-

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amination revealed 4 cm cervical dilation and face presentation. A cesarean section was decided due to marked flexion of the spine. In the operation, after Pfannenstiel incision, a low transverse uterine incision was made but was modified to a T-shaped incision because of roundness of the fetus. A female fetus was delivered weighing 2790 g, with Apgar scores of 1 at 1 min and 1 at 5 min (Figure 3). The fetus remained alive only for 10 minutes after delivery. Autopsy was not accepted by the family. X-ray imaging of the fetus showed marked hyperextension and distortion of the vertebra (Figure 4).

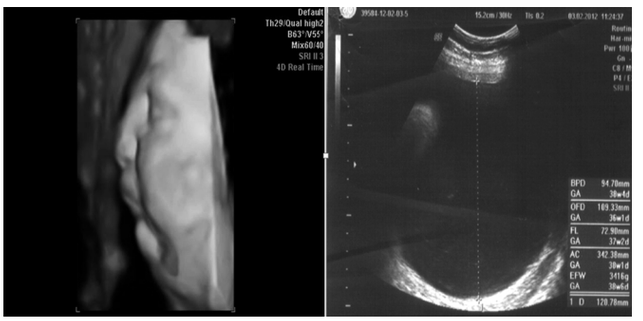

Figure 1: 3D image of the fetal head (left) and marked polihidramnios.

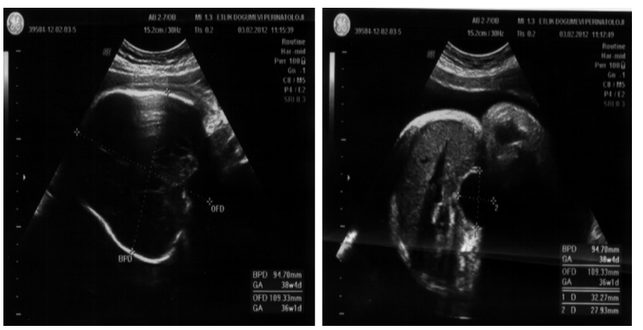

Figure 2: The posterior part of the skull was wider than the anterior part (left) and because of an almost 180 degrees retroflexion of the head, a caudal sweep motion of the ultrasound probe from the BPD plan revealed an image which gives an impression of a posteriorly placed bladder (right)..

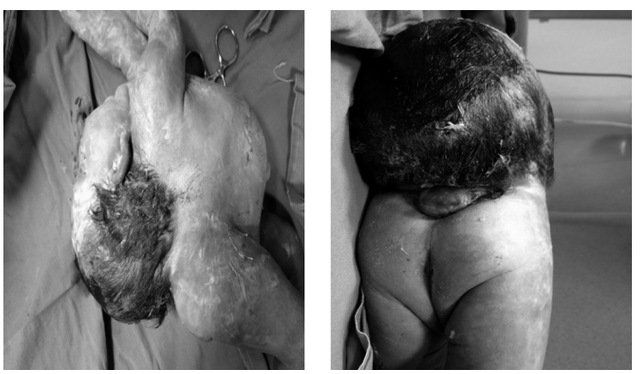

Figure 3: Extreme retroflexion of the head causing upward-turned face so-called star-gazing fetus (left) and encephalocele (right)

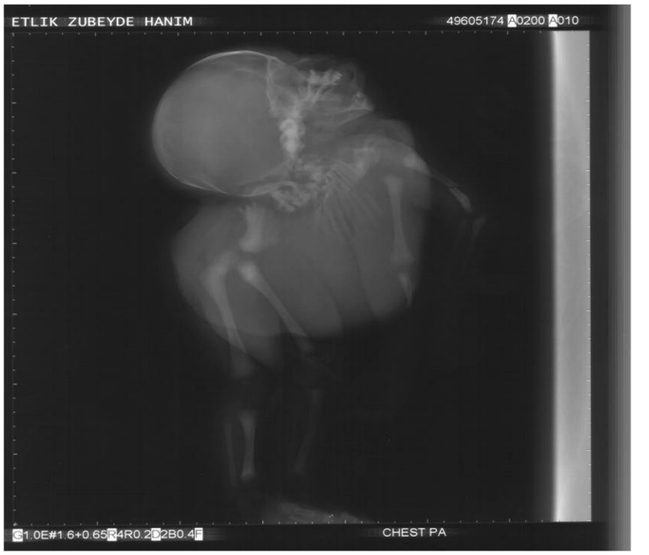

Figure 3: Extreme retroflexion of the head causing upward-turned face so-called star-gazing fetus (left) and encephalocele (right)

Discussion

Iniencephaly is an almost lethal congenital abnormality with an incidence of 0, 1-10:10000.[2] There is a female tendency; females are nine times more affected than males.[3] The pathogenesis of iniencephaly is not known. The differential diagnosis of iniencephaly should include Klippel-Feil syndrome, Jarcho-Levin syndrome, cervical myelomeningocele and lymphangioma.[4] Iniencephaly may be isolated or may be associated with other anomalies such as encephalocele, meningomyelocele, cyclopia, anencephaly, holoprosencephaly, Dandy-Walker malformation, hydrocephalus, absence of mandible, cleft lip/palate, congenital diaphragmatic hernia, cardiac defects, omphalocele, polycystic kidneys, hydronephrosis, gastrointestinal atresia, single umbilical artery, caudal regression sequence, arthrogryposis, and club foot.[4] In our case, encephalocele was present.

Prenatal diagnosis is possible in the first or early second trimester.[5] Ultrasonographic features of this syndrome are occipital bone defects, exaggerated lordosis, rachischisis, reduction of number of cervical and thoracic vertebrae, extreme retroflexion of the head and a short spine.[6] At term, extreme retroflexion and short spinal column may challenge the ultrasonographer for orientation. Additional clues for iniencephaly are lack of normal chin and neck relation and impression of a posteriorly placed bladder by the caudal sweep motion of the ultrasound probe due to severe retroflexion of the head.

Termination of pregnancy is usually offered due to ominous prognosis but in our case, the family declined this option and a term iniencephalic fetus was delivered. Term delivery of

this fetus is especially difficult due to extreme retroflexion. Thus, a cesarean section may be chosen and a classical vertical incision may be more appropriate in cases with iniencephaly.

İniensefalinin Prenatal Tanısı: İpuçları ve Zorluklar

İniensefali, oksipital kemik defekti ve buna eşlik eden foramen magnum genişlemesi, servikal disrafizm ve spinal deformitelere başın fikse retrofleksiyonu ile karakterize konjenital malformasyondur. 38 yaşındaki bayan hasta Etlik Zübeyde Hanım Doğumevi Perinatoloji Kliniği'ne gebeliğinin 18. haftasında başvurmuştur. Yapılan ultrasonografide geniş bir oksipital ensefalosel ve kısa servikotorasik omurga saptandı. Hastaya iniensefali ön tanısı ile gebelik terminasyonu önerildi fakat dini gerekçeler ile hasta tarafından reddedildi. Gebelik takibine gelmeyen hasta gebeliğinin 38. haftasında ağrı şikayeti ile acile başvurdu. Yapılan ultrasonografide başın ileri derecede retroflekse olduğu, çok kısa servikotorasik omurganın, ensefalosel ve polihidramnios bulunduğu tespit edildi. Başın aşırı retrofleksiyonu nedeniyle, ultrason probunun kaudal hareketi ile posterior yerleşmiş imajı veren mesane izlendi. Normal çene ve boyun ilişkisi kaybolmuştu. Omurganın belirgin fleksiyonu ve yüz geliş nedeni ile sezaryen doğuma karar verildi. 2790 gram kız bebek 5. dakika Apgar skoru 1 ile doğurtuldu. İniensefali, belirgin ultrasonografik özellikleri ile prenatal dönemde belirlenebilen lethal bir anomalidir. Bu vaka takdiminde term iniensefali vakası sunulmaktadır.

Anahtar Kelimeler: İniensefali, Ensefalosel, Servikal disrafizm

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