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Case Report

Early diagnosis of iniencephaly: a case report with review of literature

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ABSTRACT

Iniencephaly is a rare and fatal neural tube defect characterized by retroflexion of fetal head with absent neck due to spinal defects. Herein, we describe a case of iniencephaly with a brief review of literature.

Keywords: Anencephaly, Iniencephaly

INTRODUCTION

Iniencephaly is a rare and fatal neural tube defect characterized by retroflexion of fetal head with absent neck due to spinal defects. Often seen in association with other systemic abnormalities. Herein, we are describing a case of iniencephaly with a brief review of literature.

CASE REPORT

A 25 years $G_2P_1L_1$ presented at 13 weeks of gestation. She had one normal child. There was no significant past history in self or family. She denied intake of folic acid supplementation.She had an ultrasound showing a single live intrauterine fetus with non-visualised fetal neck and cranial bones. Repeat Scan in our radiology department revealed short and rotated spine, head fixed in retro flexion, facial structure seen clearly and showed grotesque appearance (Figure 1). This was constant observation even after repeat scan after an hour and on next day, appearing as typical star gazed fetus. Based on fetal anomaly and patients desire, pregnancy was terminated (Figure 2 & 3). The patient and her family refused genetic counselling and autopsy.

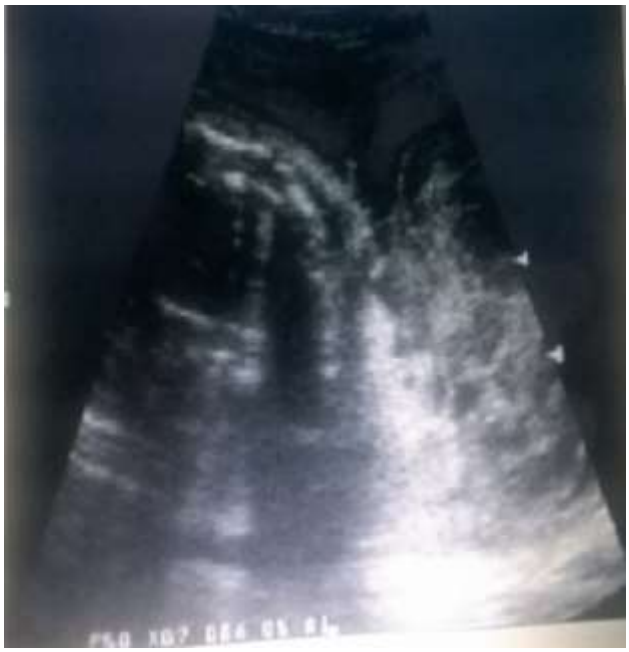

Figure 1: Ultrasound showing absent fetal neck with head fixed in retroflexion.

Figure 2: Fetus showing absent fetal neck with gross retroflexion of fetal head.

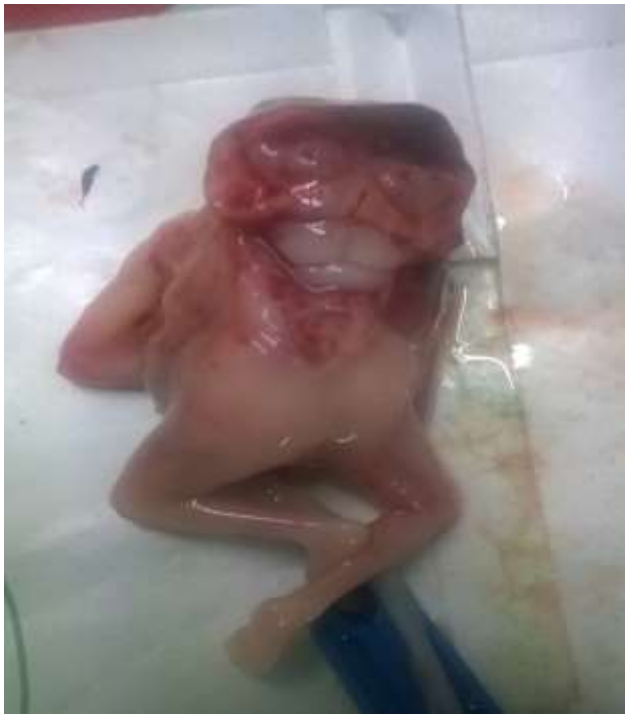

Figure 3: Posterior aspect showing occipital bone deficit with enlarged foramen magnum.

DISCUSSION

Iniencephaly is a rare and fatal neural tube defect with a reported incidence of 0.1-10/ 10,000 deliveries.[1] The word Inien is derived from the Greek word "Inion" which means nape of the neck. Lewis has classified Iniencephaly into iniencephaly apertus and iniencephaly clausus, according to the presence of encephalocele.[2]

Exact etiology remains unknown. Both environmental and genetic factors have been associated with iniencephaly. Environmental factors such as poor socioeconomic conditions, lack of folic acid supplementation, obesity and drugs (sulphonamides, tetracycline, antihistaminic) have been implicated.[2] Chromosomal abnormalities including trisomy, trisomy and Turner syndrome have been associated with this disorder.[3] Iniencephaly may be associated with other anomalies such as encephalocele, meningomyelocele, hydrocephalus, Dandy-Walker malformation, holoprosencephaly, omphalocele, congenital diaphragmatic hernia, hydronephrosis, polycystic kidneys, cardiac defects, caudal regression sequence, arthrogryposis, club foot, single umbilical artery, and gastrointestinal atresia.[4] In a survey of 24 cases of iniencephaly, the maternal serum alpha-fetoprotein (AFP) values ranged between 0.7 and 3.9 (median 2.0) MoM, while the amniotic fluid AFP values were between 0.9 and 2.7 (median 1.4). Moreover, they also noted that the tendency to develop polyhydramnios was very high (75%).[5]

The characteristic features include a short or absent neck with extreme retroflexion of the fetal head, occipital bone deficit with enlarged foramen magnum and malformed cervico-thoracic vertebrae. These features give a typical star gazed appearance to the fetus on ultrasound and detailed CNS and spine abnormalities may be known by MRI or CT.

Iniencephaly need to be differentiated from anencephaly, Klippel-Feil syndrome (KFS) and nuchal tumours. In anencephaly, there will be partial or complete absence of cranial vault, which is not covered by skin, and cervical vertebrae are normal whereas in iniencephaly, the head is retroflexed and covered by skin and cervical vertebrae is abnormal. Though fusion of cervical vertebrae and malformation may be seen in both KFS and iniencephaly, retroflexion of head is usually not seen in KFS.[6]

This condition can cause obstructed labour because of retroflexion of the fetal head in advanced gestational weeks and can recur in subsequent pregnancies.[7] Because of its invariable lethal prognosis, termination is justified when detected.

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