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Case Report

Iniencephaly apertus: a rare case with early first-trimester diagnosis by ultrasonography and fetoscopic correlation

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ABSTRACT

Iniencephaly (IE) is a neural tube defect (NTD) of infrequent occurrence caused by developmental arrest of neural tube formation. It is usually complemented with other congenital malformations carrying lethal prognosis. Hysteroscopy(fetoscopy) can be used as an add-on tool for confirmation of lethal malformations such as iniencephaly, along with ultrasonography in very early first trimester pregnancy. Our patient was diagnosed on ultrasonography, with a fetus possibly carrying Iniencephaly apertus variant (with encephalocele) with cardiovascular and spinal cord malformations, at 8 weeks 5 days period of gestation. The ultrasound findings suggested IE, which were confirmed by fetoscopy before termination. The objective of this case report is to reinstate the importance of early ultrasound scan in first trimester of pregnancy to diagnose and intervene timely in case of relatively rare neural tube defects with grave outcome, also delineate the role of fetoscopy (hysteroscopy) as a diagnostic and therapeutic tool.

Keywords: First trimester ultrasonography, Fetoscopy, Iniencephaly, Neural-tube defect

INTRODUCTION

Congenital disorders, called malformations, are structural or functional anomalies occurring during intrauterine life. An estimated 3-6% of babies are born with a congenital disorder worldwide every year.[1] Neurological anomalies, including neural tube defects, form a significant bulk. Iniencephaly (IE) is a neural tube defect (NTD) of infrequent occurrence caused by developmental arrest of neural tube formation. It is usually complemented with other congenital malformations carrying lethal prognosis. Literature has only a few case reports of this rare anomaly. To the best of our knowledge, this seems to be the earliest suspected, confirmed (by hysteroscopy), and managed case of Iniencephaly.

CASE REPORT

A 24-year-old 2 nd gravida with previous full-term vaginal delivery presented to our obstetrics out-patient department (OPD) for her first antenatal check-up. She was 8 weeks 5 days by last menstrual period and had regular menstrual cycles. Pregnancy was confirmed by urine pregnancy test kit 15 days after missing the periods; no folic acid supplementation was started. It was a non-consanguineous marriage and her first child had no congenital abnormalities. Her past medical and surgical history were unremarkable. There was no significant personal or family history; and no history of any substance abuse or drug intake.

She belonged to low socio-economic status. She had complaints of nausea and vomiting of pregnancy. Her routine serum biochemistry and physical examination were within normal limits. So, we performed an antenatal Ultrasonography, which revealed a fetus of CRL 17.9 mm, corresponding to the period of gestation of 8 weeks and 2 days, with complex developmental anomalies involving the head, spine, heart, and body, as depicted in Figure 1.

There was a depression on the occipital surface of the fetal head, a small and irregular neck

with a hump on the back of the neck, along with a wide open cervical and upper thoracic spine. We performed fetoscopy (hysteroscopy) for confirmation of our findings before terminating the pregnancy. We could see encephalocele (cervical), wide open cervical and upper thoracic spine, short developing upper limb, wide open sternum with heart protruding out of the chest (ectopia cordis), and early physiological herniation (Figure 1).

Authors had already counselled the patient and husband regarding the presence of anomalies and grave fetal prognosis. They were willing for termination of pregnancy. Suction and evacuation were done for the same. The family refused for DNA storage or genetic evaluation of fetal tissues due to financial constraints.

DISCUSSION

Iniencephaly is derived from the Greek word 'inion', which means 'nape of the neck'. It is a complex lethal neural tube defect first described in 1836 by Saint-Hilaire.[2] It accounts for approximately 1% of all fetal disfigurements. Iniencephaly results from a failure of fusion in the cervical and upper thoracic segments of the upper spine. Three main features of Iniencephaly are, firstly, a defect in the occiput involving the foramen magnum; secondly, retroflexion of the entire spine forcing the fetus to look upward; lastly, open spinal defects of variable degree extending to the cervical and thoracic spine. The estimated incidence is about 0.1-10 per 10,000 live births, depicting the pretty rare nature of malformation.[3]

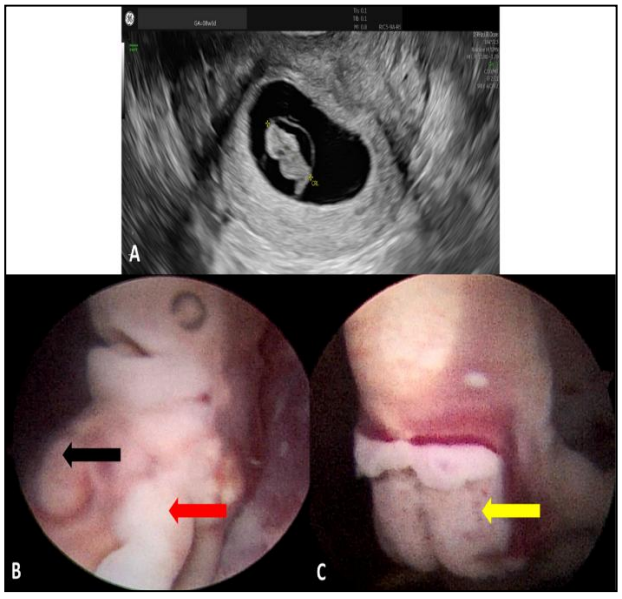

Figure 1: (A) Transvaginal ultrasound image of fetus. (B) Hysteroscopic view of fetus demonstrating ectopia cordis (black arrow) and short upper limb (red arrow). (C) Hysteroscopic view of fetus demonstrating wide open cervical and upper thoracic spine with encephalocele (cervical) (yellow arrow).

Complete neural tube closure occurs around 28 days after fertilization- anterior neuropore closes by 24 days and posterior by 27 days. Arrest of closure of neural groove before 24 days may result in anterior neuropore malformations ranging from grave anomalies like anencephaly, which are irreconcilable with life, to small meningoceles.[4] Non-closure of the anterior neuropore results in the persistence of the embryonic cervical retroflexion, which leads to the failure of the neural groove to close in the area of the cervical spine or the upper thorax, hence Iniencephaly.

Multifaceted etiology has been proposed for Iniencephaly, both genetic and environmental factors, though exact etiology is still poorly understood. Chromosomal anomalies such as trisomy 13, trisomy 18, and monosomy X have been associated.[5] Environmental factors such as low socio-economic status (SES), low parity, obesity, certain drugs (tetracycline, antihistamines, sulphonamides, anti tumor agents) and lack of folic acid supplementation during early pregnancy have shown increased risk.[6] As in our case, the patient belonged to low SES, had low parity, and did not have folic acid supplementation. Since the pathogenesis is poorly understood, averting the malformation can be troublesome.

Iniencephaly has been divaricated into two types by Lewis, based on the presence or absence of encephalocele. First is iniencephalus clausus (also called the closed type), and second is apertus (also called the open type). In the latter, an occipital cephalocele is seen protruding through the foramen magnum and occipital bone.[7]

Prenatal diagnosis can be easily made with the help of ultrasonography. In utero, MRI can also be used to properly characterize and classify fetal disorder.8 However, its high cost and non-availability at all health centres make ultrasonography the most favourable diagnostic tool. Sonographic findings of Iniencephaly include a large retroflexed head in the sagittal plane, giving the star gazing appearance.[9] The neck is not visualized separately, and the anterior chest wall is in continuation with the chin. An occipital encephalocele (protruding through the foramen magnum) can be seen in iniencephalus apertus. In transverse section, an open spinal defect can be seen.

Associated defects include anencephaly, microcephaly, hydrocephalus, open spina bifida, and facial clefts. Iniencephaly, especially the open type, is essentially a lethal anomaly. Most of the fetuses are either stillborn or die shortly after birth. Thus, termination should be offered as soon as the diagnosis is made.

Genetic analysis and fetal karyotype are also recommended once Iniencephaly is suspected. Autopsy should be performed in all cases after termination. Iniencephaly is incompatible with life, and its unknown etiology requires further experimental studies.

CONCLUSION

Neural tube defects, though uncommon, form a group of rather preventable congenital malformations requiring merely the supplementation of folic acid, hence warranting stringent patient care protocols. Fetal body distortion due to disproportionately large head with extreme retroflexion increases delivery related morbidity and mortality. Considering the advancements in technology, early diagnosis and termination is possible even before 9 weeks of gestation, as the fetus is incompatible with life. The skill and knowledge of the sonographer are equally crucial for diagnosis.

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