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ANTENATAL DIAGNOSIS OF A RARE NEURAL TUBE DISORDER – INIENCEPHALY APERTUS

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Abstract:
Iniencephaly, a rare neural tube defect, was initially described by Saint-Hilaire in 1836. Its occurrence varies from 0.1 to 10 in 10,000 deliveries, with a higher frequency observed in females. Common characteristics of iniencephaly include cervical bifida, occipital bone defect, and retroflexion of the head on the cervical spine. Here, we present a case involving a 24-year-old primigravida with a history of consanguinity. At 14 weeks gestation, she was found to have a malformed fetus, diagnosed as iniencephaly apertus through detailed ultrasonographic anatomy scan.

Key Words: Iniencephaly, apertus, neural tube defect, congenital malformation, antenatal

Introduction

Iniencephaly is one of the uncommon neural tube defects (NTD) that was first reported by Saint-Hilaire in 1836.1 The word inion is a Greek word that means the occiput or the nape of the neck.2 Its incidence ranges from 0.1 to 10 in 10,000 being more common in female babies.3 Three common features that characterize this disorder are: spina bifida at the cervical region, defect in the occipital bone and retroflexion of the head at the cervical spine.4 There are two main types of Iniencephaly: Iniencephaly apertus and Iniencephaly clauses. Iniencephaly apertus is the most severe form involving the development of an encephalocele. In Iniencephaly clauses there is absence of encephalocele.5 Other disorders associated are cardiovascular disorders, diaphragmatic hernias, and gastrointestinal malformations.6 Iniencephaly is not a genetic disease; however, the exact etiology for its occurrence is still unknown. Women with lower socioeconomic status and poor nutrition especially folic acid deficiency and hyperhomocysteinemia have shown to be at higher risk.7 Herein,

we present a rare case of iniencephaly apertus accidentally diagnosed at 14 weeks gestation in apparently healthy pregnant woman.

Case Report:

A 24 years old primigravida patient presented at 14 weeks of gestation who was referred to our clinic for suspicious anomaly. She was currently married for 2 years from her cousin that resembles a history of strong consanguinity. She had no history of diabetes or hypertension. She had not taken folic acid during the pregnancy. She had no family history of any congenital anomalies.

Ultrasound evaluation reported a single fetus with average biometry of 14 weeks gestation. The fetus showed occipital bone defect with small encephalocele (Figure 1), bilateral mild ventriculomegaly (Figure 2), absent cervical vertebra with star gazing appearance (Figure 3a and 3b) and associated left diaphragmatic hernia (Figure 4). Mild subcutaneous edema with few anechoic areas noted in neck region (Figure 5). Diagnosis of iniencephaly apertus was made based on ultrasonographic findings.

Discussion:

Closure of the neural tube mostly happen around 28th day of pregnancy and failure of its closure may lead to a defect graded from a small meningocele to life threatening conditions.8 Iniencephaly nearly represents about 1% of all fetal abnormalities, with an incidence rate ranging between 0.1 and 10 in 10,000 deliveries.9 The important features of iniencephaly are abnormalities of the occipital bones, leading to a larger foramen magnum, absence of cervico-thoracic regions of the vertebral column associated with an abnormal fusion of the present vertebrae and defective fusion of the vertebral arches and bodies, shortening of the spinal column as a result of marked lordosis and hyperextension of the malformed spine, upward-directed face and star-gazing appearance.10 The most common associated anomalies are spina bifida (74%), diaphragmatic hernia (37%), adrenal hypoplasia (37%) and club foot (32%).11 Prenatal diagnosis of iniencephaly is feasible by either ultrasonography or MRI. Iniencephaly has a very poor prognosis and both types are incompatible with life. There are only six cases reported in the literature of iniencephaly with relatively long-term survival.12 In cases associated with minor malformations, cervical deformities could be improved by neurosurgical intervention.5

Conclusion:

In conclusion, iniencephaly is an extremely rare NTD with a lethal outcome. Prenatal diagnosis by ultrasonography is relatively easy due to presence of multiple severe anomalies.

Conflict of Interest:-

No conflict of interest.

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Figures:-

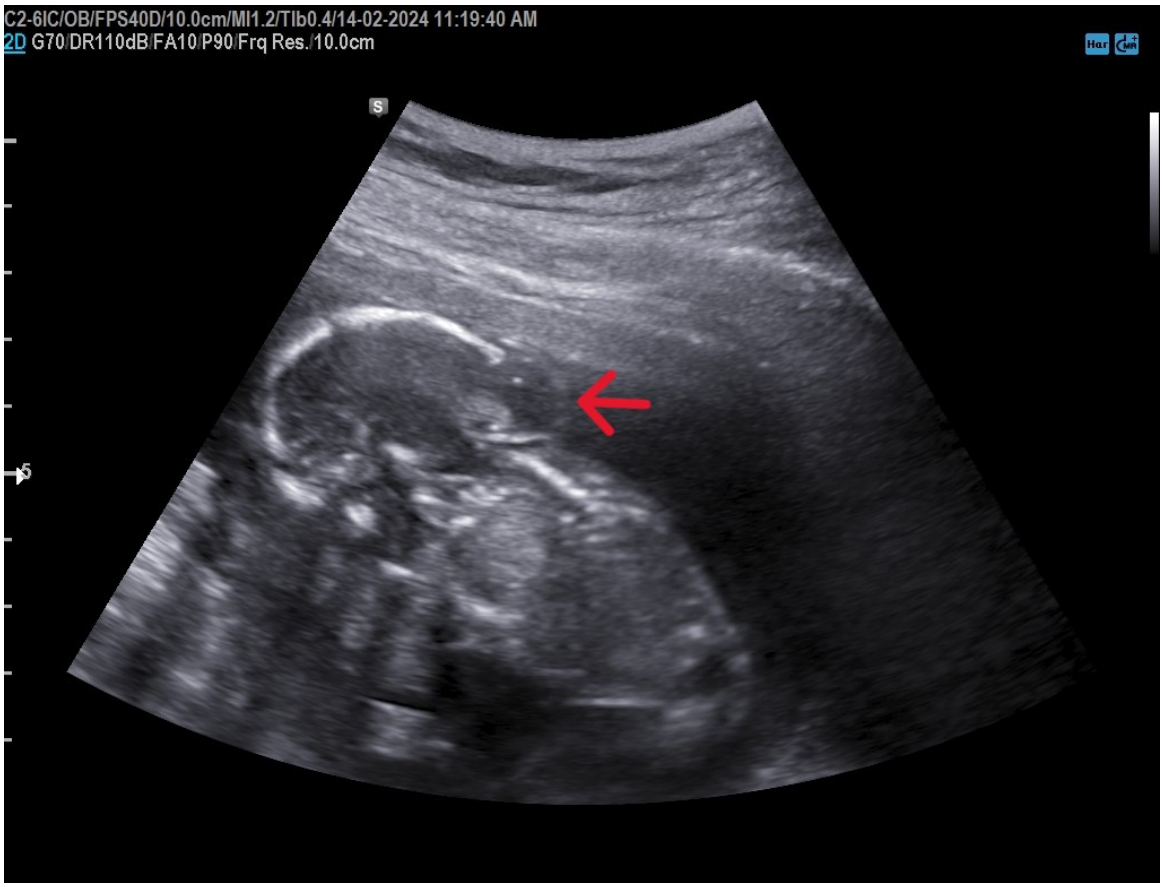


Figure 1: Defect in the occipital bone noted with small encephalocele (Red arrow).

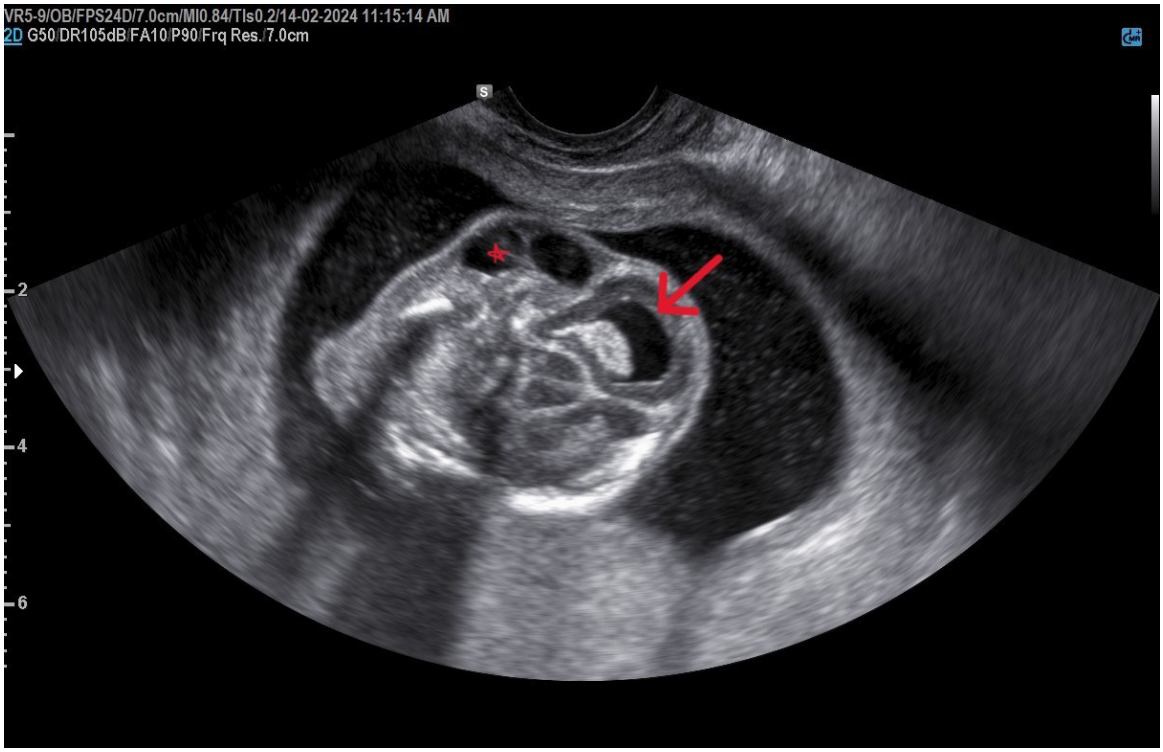
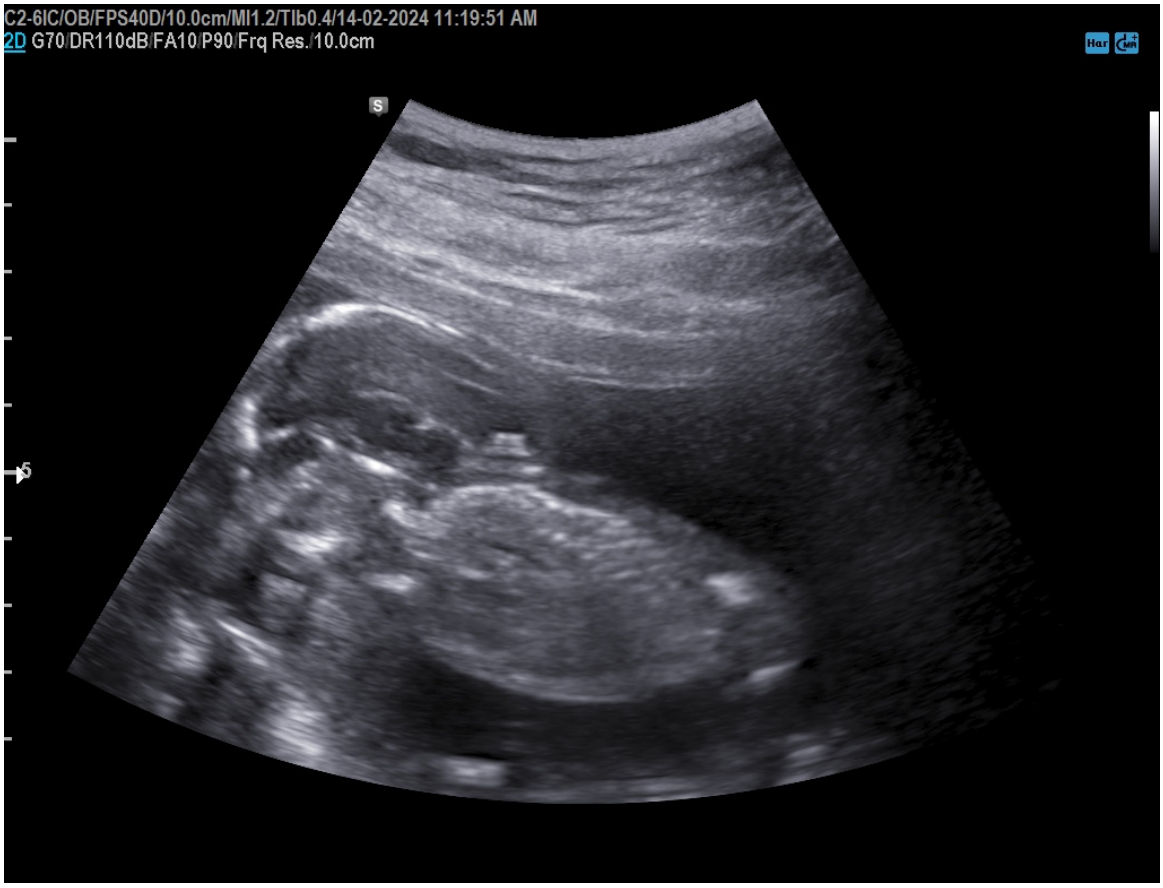


Figure 2: Dilated ventricles (Red arrow) and subcutaneous edema with anechoic areas noted in neck region (*).



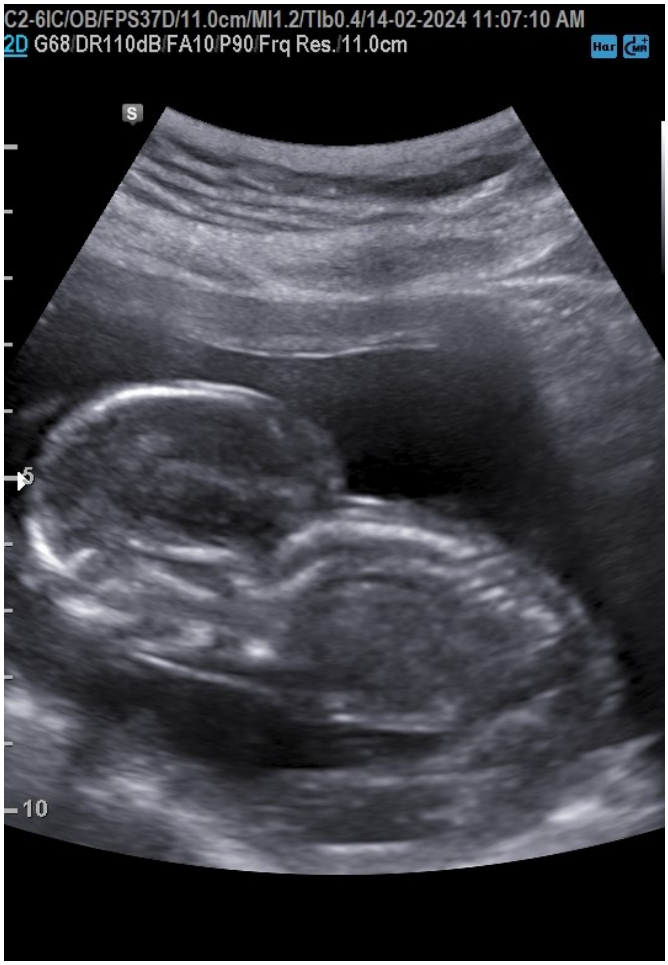


Figure 3A and 3B:

Absent of cervical vertebra noted with head in hyperextension (Star gazing appearance).

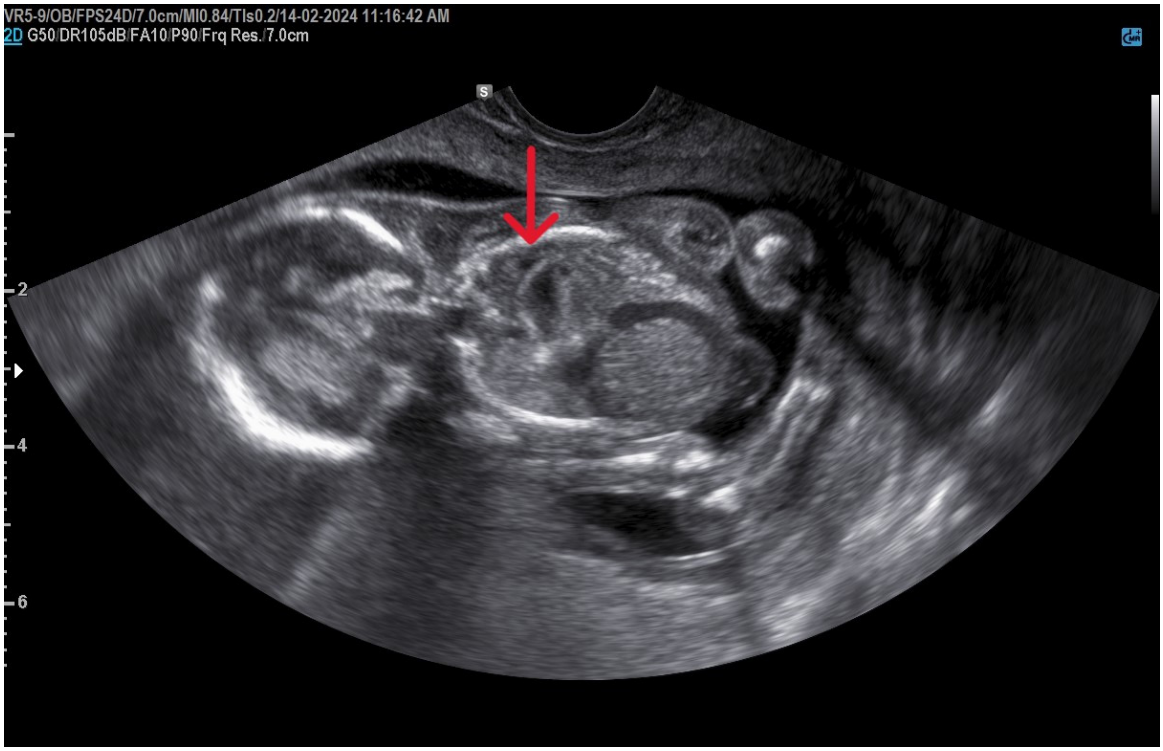


Figure 4: Left diaphragmatic hernia noted with herniation of stomach (Red arrow).

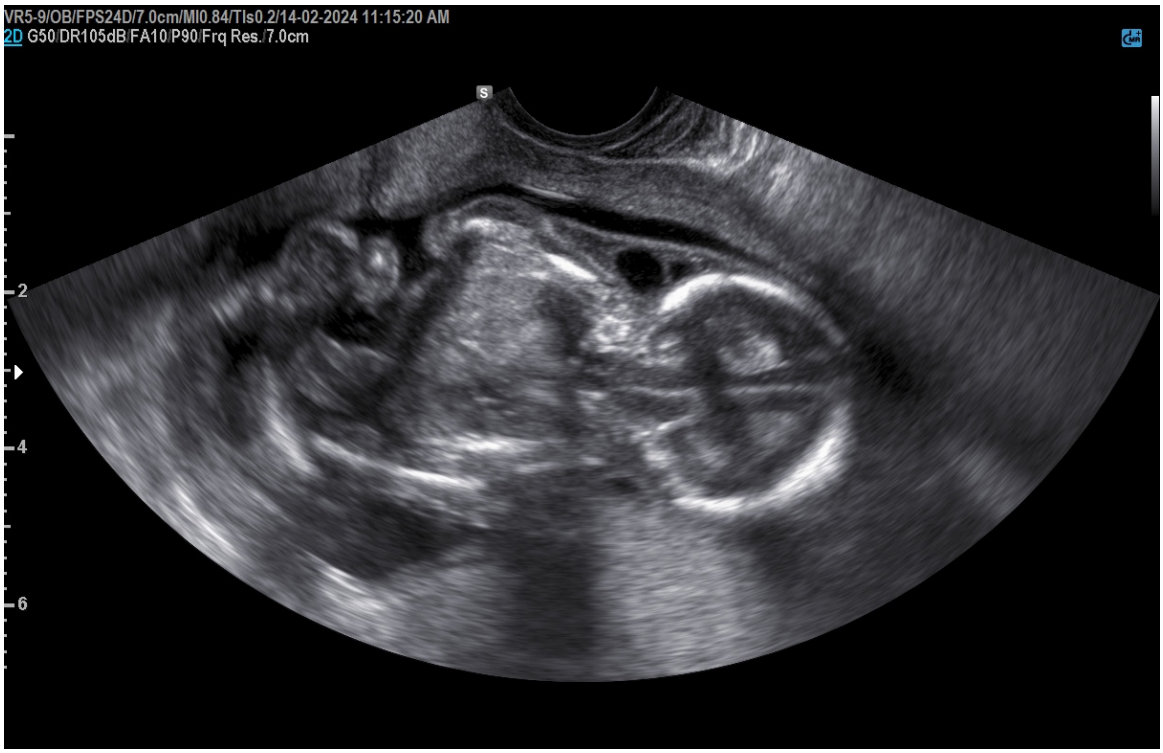


Figure 5: Subcutaneous edema noted with anechoic areas in it in neck region.