Antenatal Diagnosis of Iniencephaly: Sonographic and MR Correlation: A Case Report

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Iniencephaly is an uncommon and fatal neural tube defect involving the occiput and inion, this occurs together with rachischisis of the cervical and thoracic spine, and retroflexion of the head. We report the ultrasound (US) and magnetic resonance (MR) imaging findings of a case of iniencephaly with clubfeet and arthrogryposis. The diagnosis of iniencephaly is easy to make on ultrasound due to the typical star-gazing fetus. However, the details of the fetal brain and spinal cord may not be adequately delineated on US. We found MR imaging to be superior for depicting central nervous system abnormalities. MR imaging has evolved as an imaging modality and it is complementary to fetal US, yet US remains the screening modality of choice.

Index terms:
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Although ultrasound (US) is the imaging modality of choice for conducting antenatal evaluation of fetal anomalies, limitations are sometimes encountered. Magnetic resonance (MR) imaging has evolved as a useful adjunct because of its lack of radiation, non-operator dependence and a better inherent soft tissue contrast. It can be used to complement the US examination for evaluating the fetal central nervous system (CNS). Iniencephaly is a complex neural tube defect and it can be easily diagnosed on US due to the typical fetal position known as the "star-gazing appearance". The US findings of this anomaly are well described in the medical literature; however, there are not many articles describing the MR findings, and the evaluation of the intracranial and intraspinal abnormalities also remains inadequate. We found MR to be superior for delineating these anomalies compared to US.

CASE REPORT

A 25-year-old Indian woman was referred to our hospital for an antenatal anomaly scan at 28 weeks of pregnancy. She was pregnant from a non-consanguineous marriage. A previous scan done at the 26th week of this pregnancy had revealed a diamniotic, dichorionic twin pregnancy with gross polyhydramnios in one of the sacs. There was no history of radiation exposure or any drug intake during this pregnancy. On examination, the uterus was enlarged, which corresponded to about 32 weeks of gestation.

Transabdominal gray-scale US was performed on a HDI 5000 US scanner (ATL-Philips, Netherlands) using a broadband 2 –5 MHz curved-array transducer. It revealed a dichorionic, diamniotic twin pregnancy. Fetus I was normal. Fetus II had increased head parameters for its age. The neck was short and hyperextended. The face was upturned, corresponding to the description of a star-gazing fetus (Fig. 1). The

lateral and third ventricles were normal in size, but there was enlargement of the fourth ventricle (Fig. 2). No cephalocele or meningocele was detected. The craniocervical junction appeared deformed with fusion of the occiput with the upper cervical region (Fig. 3). This fetus did not demonstrate any limb activity during the examination, which lasted for 30 minutes, suggesting akinesia. There was hyperextension at the knees, with flexion of the arms and clenching of the fists. Equinovarus deformity of both feet was detected (Fig. 4), and polyhydramnios was also noted. The possibility of iniencephaly with arthrogryposis and fetal akinesia was raised.

Fetal MR was performed on a 1.5 Tesla MR scanner (Signa, GE Echospeed, Milwaukee, WI) for further evaluation after obtaining an informed consent. Single-shot fast spin-echo sequences (ssFSE; TR/TE- 1392/95) and a body coil were used. MR examination revealed two fetuses. The twins were separated by a thin hypointense membrane, which was difficult to visualize. Fetus I was normal, fetus II had a brachycephalic skull and hyperextended short neck. For fetus II, the supratentorial brain parenchyma was normal. The lateral and third ventricles were normal in size and shape, except for mild unilateral colpocephaly (Fig. 5). The fourth ventricle was enlarged and it communicated with the cisterna magna (Figs. 6, 7). The brainstem from the level of the pons was split into two symmetrical halves. This split extended caudally beyond the cervicomedullary junction into the upper portion of the cervical cord (Figs. 6, 7). The spinal canal in this region was widened (Fig. 8); however, there was no bony spur. Distally, the spinal cord appeared normal in shape and thickness (Fig. 8). No split could be identified in the vertebral column. The knees were extended. Both feet demonstrated talipes equinovarus deformity (Fig. 4). Two placentae were identified; one was anterior, while the other was posterior and fundal. Both the umbilical cords had three vessels. The cervix was short and effaced.

The patient experienced preterm labor pains at 30 weeks with rupture of membranes, resulting in the spontaneous vaginal delivery of a 1300 gm (fetus I) fetus and a 750 gm (fetus II) fetus; both were females. Fetus II was an iniencephalic fetus that had a short hyper-extended neck, retroflexed head, micrognathia, hyperextension at knees and bilateral clubfeet. These characteristics confirmed the antenatal imaging findings. Fetus II remained alive for a few hours after delivery. An autopsy to confirm the

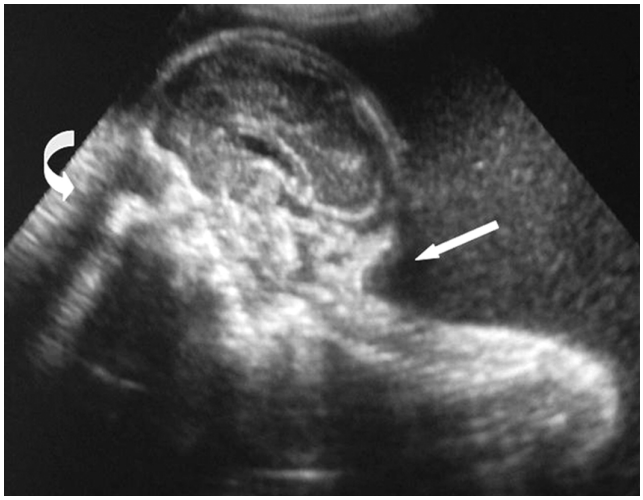

Fig. 1. Ultrasonography reveals the short fetal neck with a hyperextended cervical spine (straight arrow) and upturned star-gazing face (curved arrow).

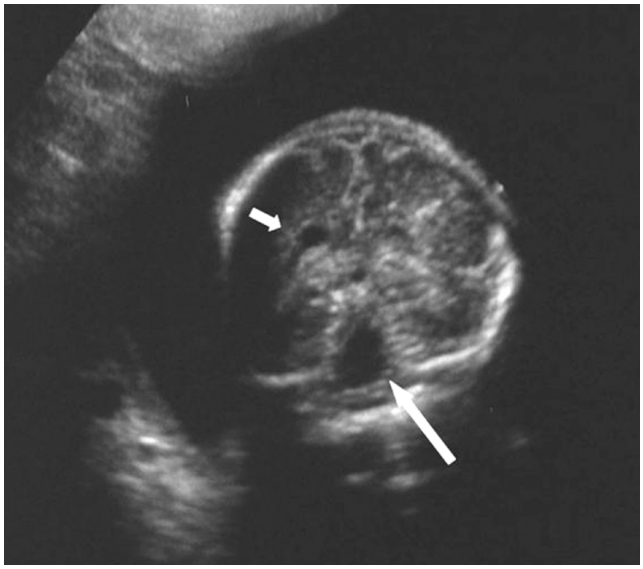

Fig. 2. Ultrasonography shows the enlarged fourth ventricle (long white arrow) with normal lateral ventricles (small white arrow).

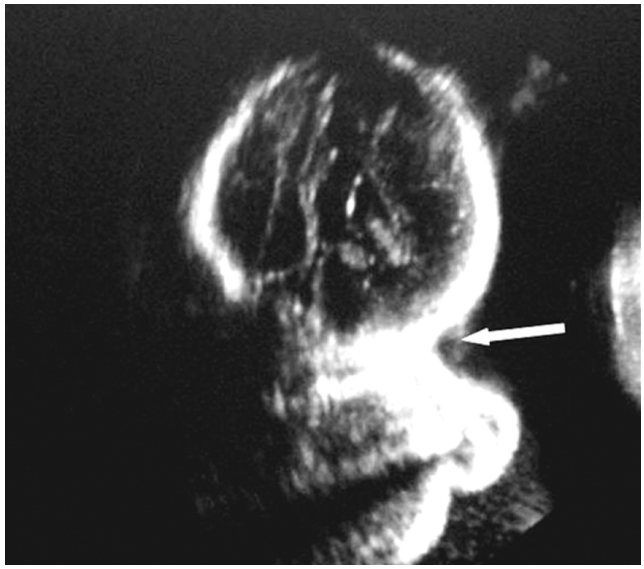

Fig. 3. Fusion of the occiput with upper cervical region (white arrow) is demonstrated on ultrasonography.

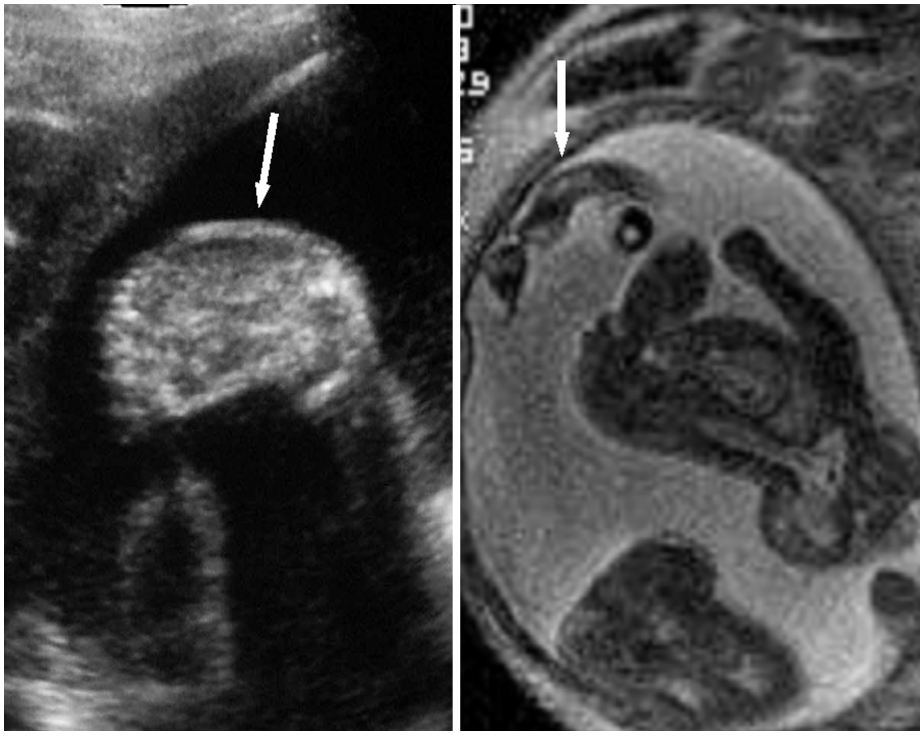

Fig. 4. Club foot is visualized on US (**A**) and MR imaging (**B**) (white arrows).

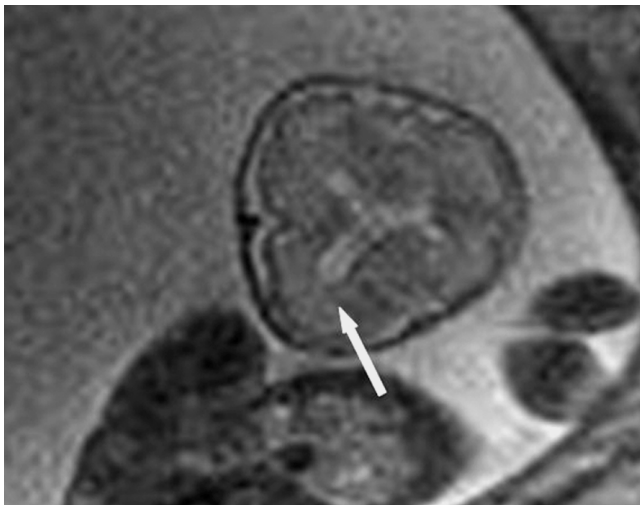

Fig. 5. MR image reveals the normal supratentorial brain of fetus II, except for mild unilateral colpocephaly (white arrow).

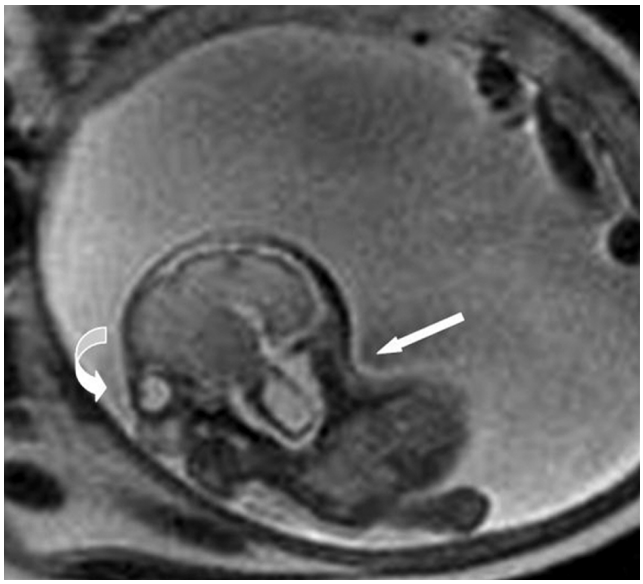

Fig. 6. MR image reveals the short fetal neck, hyper-extended cervical spine (straight arrow) and upturned face (curved arrow) of fetus II.

findings could not be performed in accordance with the wishes of the parents.

DISCUSSION

Iniencephaly is a rare neural defect involving the occiput and inion combined with rachischisis of the cervical and thoracic spine and retroflexion of the head (1). Saint-Hilaire (2) first described this entity as being caused by arrest of the embryo in physiological retroflexion during the third week of gestation or by failure of normal forward bending during the fourth week. Environmental factors like maternal syphilis and drugs such as clomiphene citrate and sedatives have been implicated as etiological factors, but there is no definite evidence (3). There was no such medical history for our patient. The overall incidence of iniencephaly is 0.1 to 10 per 10,000 (3). In India, the

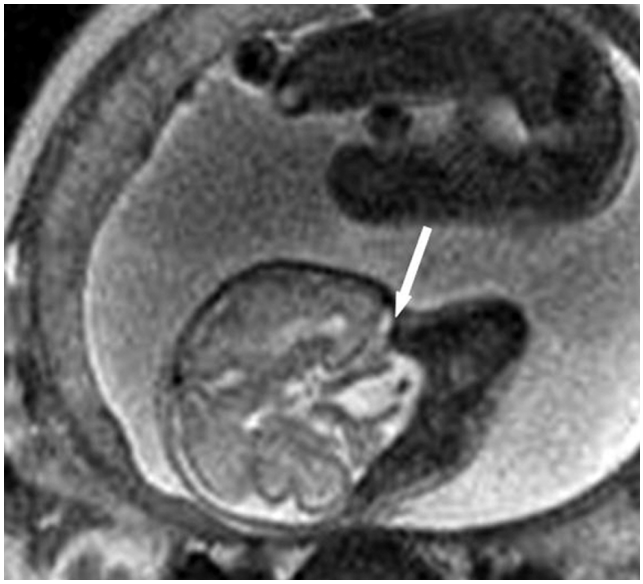

Fig. 7. MR image reveals the enlarged 4th ventricle of fetus II, which communicates with the cisterna magna (straight arrow) with a split brainstem extending to cervico-medullary junction.

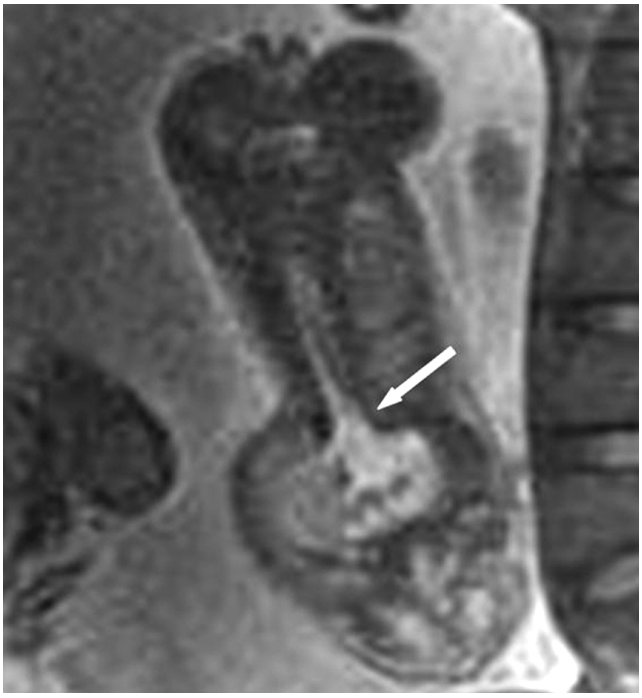

Fig. 8. MR image reveals a widened upper spinal canal (straight arrow) with a normal distal spine.

incidence is reported to be 1 per 65,000 deliveries (4). A female preponderance (90%) has been noted (4) as was seen in our case. Iniencephaly is seen in families with a history of neural tube defects and the recurrence risk is 5% (4). There was no contributory family history in our case. Children with iniencephaly are almost always still born, but live born fetuses have been reported (4), as in our case. Lewis (1) classified iniencephaly in two main groups: 1) iniencephaly apertus, which is associated with encephalocele, and 2) iniencephaly clausus, which is associated with a spinal defect, but no cephalocele. The main features of iniencephaly include: 1) a variable deficit of the occipital bones that results in an enlarged foramen magnum, 2) partial or total absence of the cervical and thoracic vertebrae with an irregular fusion of those vertebrae that are present, and this is accompanied by incomplete closure of the vertebral arches and bodies, 3) hyper-extension of the malformed cervico-thoracic spine with significant shortening of the spinal column due to marked lordosis, and 4) an upward-turned face and the mandibular skin being directly continuous with that of the chest owing to the short neck (3). The fetus in our case had all the above-mentioned features. It had no swelling in the cervical region and so our case is the second group type. Iniencephaly may be associated with other malformations like arthrogryposis and clubfeet (5), as was seen in our case. Other anomalies include anencephaly, holoprosencephaly, spina bifida, absence of mandible, cleft palate, low set ears, pulmonary hypoplasia or hyperplasia, cardiac malformations, diaphragmatic hernia or agenesis, omphalocele, gastroschisis, gastrointestinal atresia, single umbilical artery, renal anomalies, overgrowth of the arm compared to the legs, genu recurvatum, and hydramnios (5). Cyclopia as an association with iniencephaly has also been reported (6). Arthrogryposis is a combination of neurologic, muscular and connective tissue disorders that lead to limitations of joint mobility, contractures and rigidity. The US features of arthrogryposis include fetal immobility, fixed extremities and especially the arms, clenched hands, hyperextension at the knee and a talipes equinovarus deformity (5). Our fetus demonstrated arthrogryposis on US, this can be difficult to assess on MR alone. However, the constancy of the fetal position over several sequences may provide a clue to the akinetic condition. US is the modality of choice for prenatal screening and it provides cost-effective, real-time fetal images. However, diagnostic difficulties occasionally arise owing to maternal body habitus, oligohydramnios or complex fetal anomalies (7). MR imaging is the next best alternative, as it does not expose the patient to ionizing radiation. The evolution of faster imaging with half-Fourier techniques (ultrafast MR imaging) has virtually eliminated any artifacts due to fetal motion. It is relatively better than US in cases of maternal obesity and oligohydroamnios. The CNS and other anatomic structures of a fetus are well visualized on MR beginning from the early second trimester (6), so the US findings can be confirmed in most cases and additional information can be acquired about the precise diagnosis,

severity and location of the anomaly. Thus, further clinical management can be guided with regard to terminating pregnancy or providing adequate postnatal care (7). It is important to make the decision to proceed with fetal MR on a case-by-case basis in close consultation with the referring obstetrician. In some instances, fetal MR in the late third trimester may be of sufficient quality to replace or at least delay immediate postnatal MR, thereby eliminating the need for administering neonatal sedation (7). We found ultrafast MR to be a useful adjunct to US for confirming the diagnosis of iniencephaly. MR delineated the intracranial and intraspinal anomalies to a much greater extent that did US. Arthrogryposis could only be suspected on MR, and US was the superior modality to diagnose this. The diagnosis of clubfeet could be made using both modalities.

In conclusion, prenatal ultrafast MR is superior to US for delineating the abnormalities involving the brainstem, cervical cord and cervico-dorsal spine in a fetus with iniencephaly. It can also be used to image other CNS anomalies for conducting meticulous evaluation, which may influence the clinical management.

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