J. Fetal Med. (March 2018) 5:23–26
https://doi.org/10.1007/s40556-017-0149-8

ORIGINAL ARTICLE

First Trimester Diagnosis of Iniencephaly Confirmed by Postnatal Autopsy

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Received: 27 November 2017 / Accepted: 1 December 2017 / Published online: 4 January 2018

Abstract Iniencephaly (Greek: "inoin" meaning nape of neck) is an extremely rare and uniformly lethal form of neural tube defect. We report a case diagnosed in the first trimester using two-dimensional and three-dimensional ultrasound. The diagnosis was confirmed on postnatal autopsy. This case highlights the importance of diagnosing structural abnormalities in the first trimester itself.

Keywords Iniencephaly · First trimester · Ultrasound · Fetal autopsy · Neural tube defect

Introduction

Iniencephaly (Greek "inoin" nape of neck) is an extremely rare form of neural tube defect (NTD) and is mostly fatal. The typical triad described in literature for its diagnosis includes fixed retroflexion of the head, variable degrees of cervical lordosis and dysraphism, and an occipital bone defect involving the foramen magnum [1, 2]. We report a case of Iniencephaly diagnosed in the first trimester of pregnancy with 2D and 3D ultrasound.

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Case Report

A 31-year-old G3P1 was referred to our fetal medicine department at 13 weeks gestation with suspected spinal deformity on ultrasound done in an outside facility. She had a 5-year-old healthy daughter. An earlier pregnancy was terminated in the second trimester due to encephalocele diagnosed at 19 weeks on anomaly scan. This pregnancy was a spontaneous conception; however, she started folic acid only after 6 weeks. The couple was nonconsanguineous and had no other significant family history. She was not diabetic. Also, she had no history of exposure to any teratogenic agent or substance abuse.

Ultrasound was done using E8 scanner (GE Healthcare, Milwaukee, WI) equipped with a convex 4–8 MHz abdominal transducer and a 6–12 MHz transvaginal probe. Two-dimensional ultrasound showed a single live fetus with CRL of 41 mm which was below the 3rd centile for gestation. The fetal head was continuously fixed in hyperextension which was confirmed on 3D render imaging (Fig. 1). There was no discernible neck region and the cervical and thoracic vertebrae showed 'rachischisis' (Greek 'rhachis'—spine and 'schisis'—split) though the overlying skin appeared to be intact. There was no encephalocele. The fetal trunk was small and bulky with exaggerated lordosis and the upper limbs seemed to be relatively long and placed in extension. The lower limbs appeared normal. The nuchal translucency was increased; there was no tricuspid regurgitation and Doppler flow was normal in the ductus venosus. In addition, there was a small exomphalos. Color Doppler showed a single umbilical artery. Fetal brain appeared normal. Mediastinal shift of the heart was noted along with non-visualization of fetal stomach suggestive of diaphragmatic hernia. The eyes appeared prominent, however limited views of the face

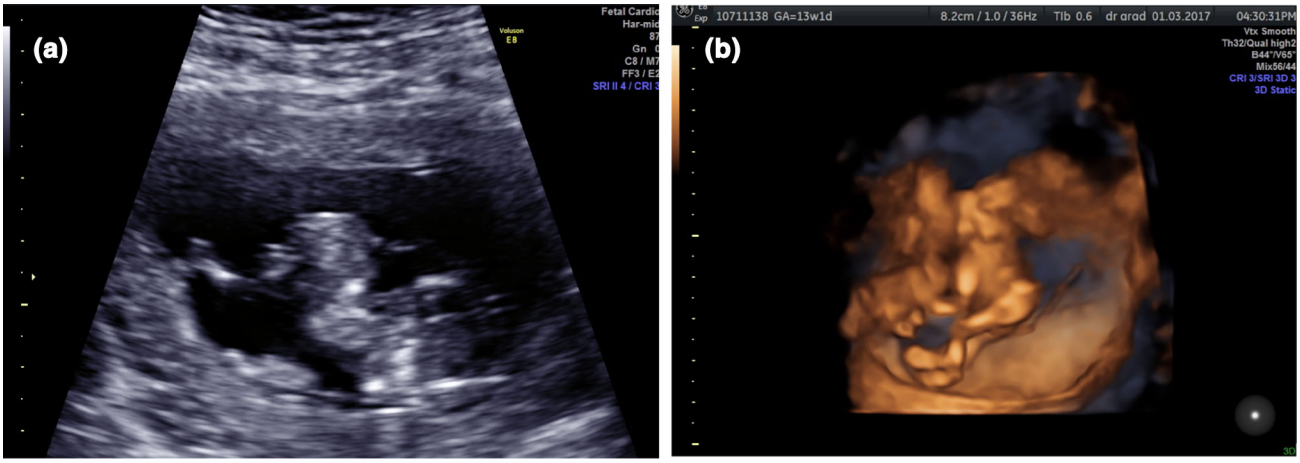

Fig. 1 **a** 2D USG image of fetus in prone position with head in hyperextension and no distinct neck region. **b** 3D rendered image of the same fetus

could be obtained owing to the fetal prone position. The placenta and amniotic fluid appeared normal. A provisional diagnosis of Iniencephaly clausus was made and the couple was counseled regarding the uniformly poor prognosis. The couple opted for termination of pregnancy, which was performed with vaginal misoprostol. A male fetus weighing 20 g with a CRL of 38 mm was evaluated postnatally. Gross examination showed hyperextension at the level of cervicodorsal region and a short trunk with lordosis due to an abnormally shaped vertebral column (Fig. 2). Infantogram was not informative due to poor mineralization; however, it did demonstrate an abnormal cervicothoracic region (Fig. 3). The occipital bone was absent.

Fetal autopsy confirmed exomphalos containing gut loops and spleen. In addition, there was choanal atresia, imperforate anus and single umbilical artery. There was a left sided diaphragmatic hernia, with gut loops present in the posterior aspect of left hemithorax. The heart was pushed to the right side and the lungs were relatively large in size owing to choanal atresia. Owing to the small size of heart, detailed anatomical study of the heart was not possible. The other organs appeared normal. The couple was counseled regarding the recurrence risk of 2–5% and that this risk can be reduced to nearly half by periconceptional folic acid intake [3].

Discussion

Iniencephaly was first reported by Saint Hilaire in 1836 [4]. It is characterized by a variable defect of the occipital bone, resulting in a large foramen magnum, partial or total absence of the cervical and thoracic vertebra accompanied by incomplete closure of arcs and/or vertebral bodies,

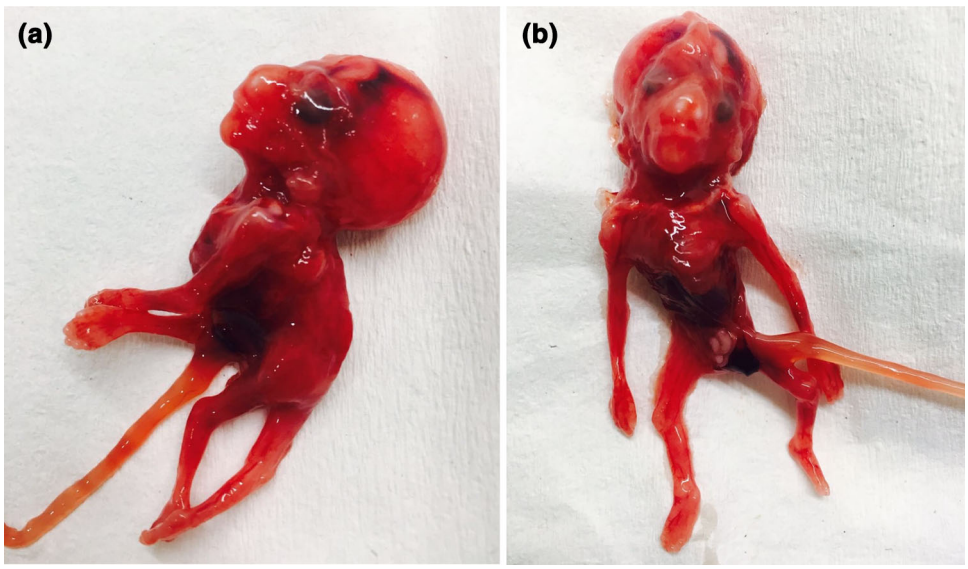

Fig. 2 **a** Postnatal image of the fetus with head in hyperextension, short trunk and lordosis, **b** image showing no discernible neck region and omphalocele

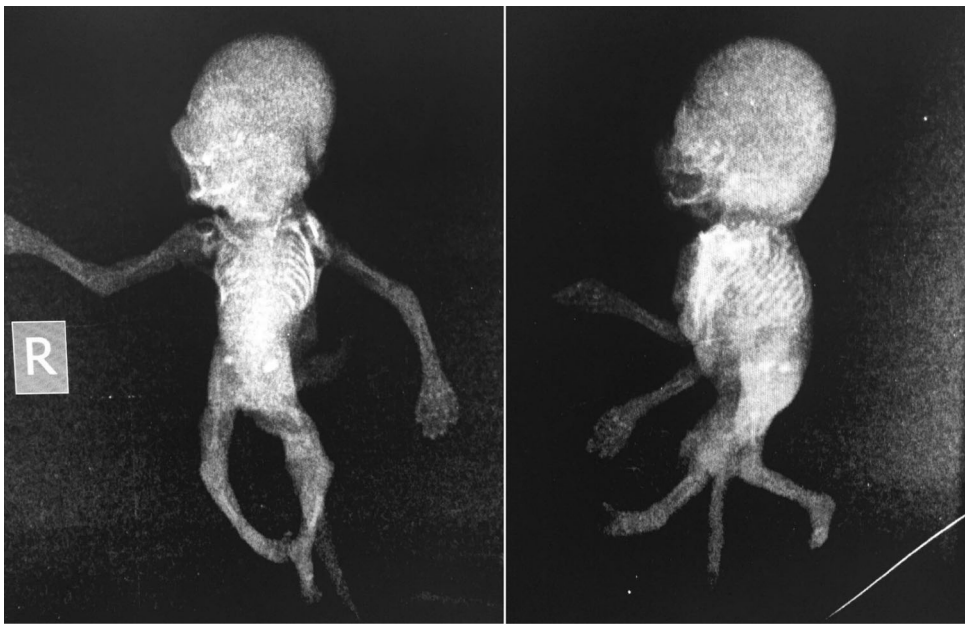

Fig. 3 Infantogram (AP and lateral views) non visualisation of ossification centres in cervical region suggestive of raschischisis

marked lordosis, hyperextension of the cervico-thoracic spine causing the face to deflect upwards (extreme retroflexion or 'stargazing') [5]. Lewis classified it into apertus and clausus based on the presence or absence of a cephalocele respectively [6]. There is a wide variation in the reported incidence ranging from 1 in 900 to 1 in 65,000 births, being more prevalent in regions where anencephaly is common [1]. There is a female preponderance (male: female = 0.28) [7].

Being part of the spectrum of neural tube defects, poor socioeconomic status and lack of folic acid supplementation is known to have a causal association [8]. In our case, this was the second consecutive neural tube defect, and the patient, started folic acid supplementation only after 6 weeks gestation.

Although there have been more than 200 cases of Iniencephaly reported in English literature [9, 10], the diagnosis of most of them was made in the second trimester of pregnancy. With the paradigm shift of inversion of pyramid of antenatal care [11], first trimester screening is not limited to aneuploidies. Recent literature also shows an increased interest in diagnosing neural tube defects in early pregnancy in order to discern the possible etiology and unravel specific recurrence rates [12]. In our case, diagnosis was made in the first trimester, which serves as an example that fortifies the importance of the first trimester scan. Differential diagnosis of persistent hyperextension of the fetal head include fetal neck masses, congenital torticollis secondary to uterine malformations or leiomyomata, congenital neuromuscular disorders, and tight nuchal cords which are rather easily distinguished on ultrasound [13]. Further, early diagnosis of a lethal anomaly is beneficial in the context that patients can take an early decision for discontinuation of pregnancy, thereby alleviating the physical and psychological implications of second trimester termination of pregnancy.

Although diagnosis can be established on 2D USG, 3D imaging provides better spatial orientation. Clearer images of the fetal malformation on 3D USG make fetal MRI optional [14]. Also, rendering methods improve visualization for the patient, helping them understand the abnormality better and aids counseling.

Iniencephaly is known to be associated with other anomalies (75%) which include omphalocele (20%), clubfoot (19%), encephalocele (14%), single umbilical artery (14%), congenital diaphragmatic hernia (12.7%), facial clefts (9.5%), duodenal atresia (6.4%), renal agenesis or dysgenesis (6.4%), myelomeningocele (6.4%); ventriculomegaly or hydrocephalus (6.4%), genital abnormalities (3.2%), cystic hygroma (3.2%), heart defects (3.2%), holoprosencephaly (1.6%), imperforate anus (1.6%), lissencephaly (1.6%) and syringomyelia (1.6%) [8]. Our fetus, based on the postnatal evaluation, was confirmed to have omphalocele, congenital diaphragmatic hernia, single umbilical artery, imperforate anus and chaonal atresia. Thus, a detailed postnatal examination is imperative not only to confirm antenatal diagnosis, but also to reveal findings, which cannot be detected by USG early in gestation e.g. imperforate anus. Like other neural tube defects, Iniencephaly also carries the risk of recurrence in subsequent pregnancies (1–5%), which can be reduced by periconceptional folic acid starting at least 3 months prior to conception.

In conclusion, Iniencephaly is a part of the spectrum of neural tube defects. Since the outcome is invariably lethal, early diagnosis can help parents make early decision regarding termination. The clinical ease as well as psychological benefits of early termination makes diagnosis of lethal abnormalities in the first trimester desirable. We present this case for its rarity, early diagnosis of Iniencephaly and associated abnormalities confirmed on postnatal correlation.

Compliance with Ethical Standards

Conflict of interest The authors declare that they have no conflict of interest.

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