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Iniencephaly: Prenatal diagnosis and management—Case report

Tewodros Getahun Asfaw, Birhanu Kebede Dina, Yared Tesfaye Wube

ABSTRACT

Introduction: Iniencephaly is a rare and complex neural tube defect characterized by significant deformities in the cervical and thoracic spine, resulting in an extremely retroflexed head. Although environmental and genetic factors have been proposed as potential causes, the precise etiology and pathogenesis are not well understood. Prenatal diagnosis of iniencephaly through ultrasound is rare, but it can often be confirmed with a second-trimester ultrasound. Due to its almost universally fatal prognosis, pregnancy termination is often considered when this condition is diagnosed before the fetus reaches age of viability.

Case Report: We present two cases of prenatal iniencephaly diagnosis, which, to the best of our knowledge, represent the second documented instances of iniencephaly diagnosis in Ethiopia, where the condition was first identified during Fetal Anatomic Scanning.

Conclusion: Iniencephaly is a severe and devastating condition that demands early prenatal diagnosis and counseling. Timely anatomic scanning enables an accurate diagnosis and provides families with the essential information to make informed decisions. In this case, early diagnosis facilitated appropriate management, including pregnancy termination, and was followed by a smooth post-expulsion period.

Keywords: Ethiopia, Iniencephaly, Neural tube defect, Prenatal diagnosis

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INTRODUCTION

Iniencephaly is a rare and complex neural tube defect characterized by significant deformities in the cervical and thoracic spine, resulting in an extremely retroflexed head. The condition also causes the absence of a visible neck, with the skin extending continuously from the back of the scalp to the skin on the back, and from the face to the chest [1]. The neck is absent due to the partial or complete absence of the occipital squama, along with defects, abnormal fusion, and splitting of the cervical and thoracic vertebrae, as well as issues with the closure of the vertebral arches. The lordosis in the cervicothoracic region causes the face to tilt upwards (stargazing head), while the trunk is typically deformed and shortened [2–4].

CASE SERIES

Case 1

This is a 24-year-old primigravid woman who was referred from a nearby health center at a gestational age of 23 weeks with a suspicion of nuchal edema for further investigation at Abebech Gobena Referral Hospital in Addis Ababa, Ethiopia. All her baseline investigations were normal, and she had no known medical conditions or drug use.

Upon fetal anatomic scanning at the Maternal Fetal Medicine unit, a singleton viable intrauterine pregnancy was identified with a fixed, hyperextended neck and a

continuous scalp extending to the skin of the thorax, as shown in Figure 1A–C. Additionally, neck edema likely secondary to lymphatic obstruction was observed (as shown in Figure 2). Unilateral clubfoot was observed on the right lower limb, while the left lower limb appeared normal (as shown in Figure 3, right and left lower limbs, respectively).

The final diagnosis of iniencephaly was made, and the prognosis was discussed with the family. They opted pregnancy termination. Mifepristone and misoprostol were administered, and the mother expelled a 500 g male fresh stillborn (Figure 4). The post-expulsion period was uneventful. The mother was counseled on the risk of recurrence and advised to seek early preconceptional care. She was then discharged. A postmortem X-ray was not performed due to the unavailability of X-ray services at the hospital.

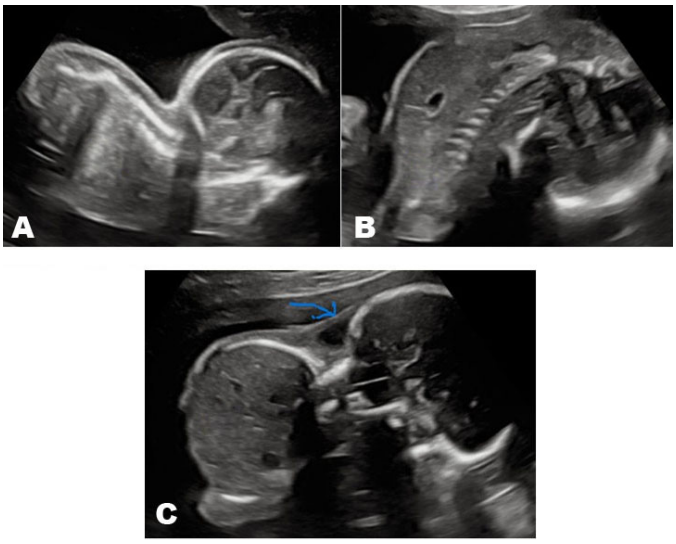

Figure 1: A 23-week fetus with a hyperextended neck (A and B) and a continuous scalp extending to the skin of the thorax (C, arrow).

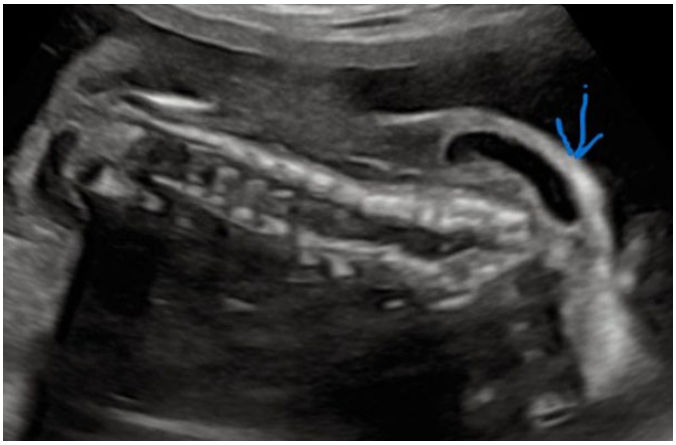

Figure 2: Neck edema (blue arrow), likely secondary to lymphatic obstruction.

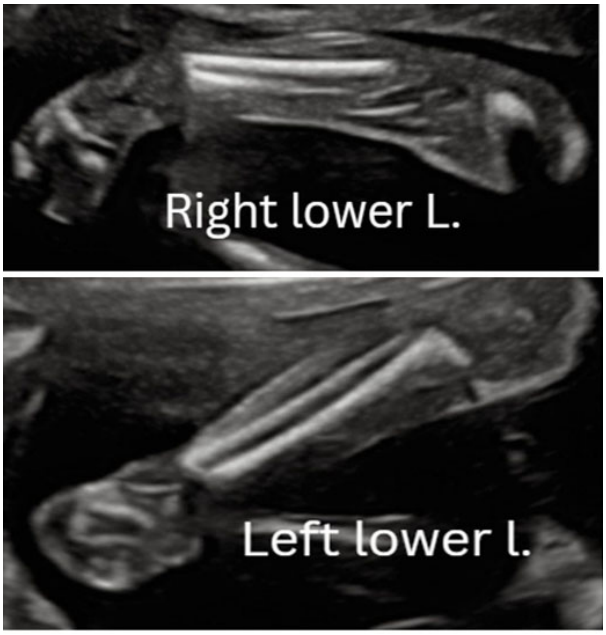

Figure 3: A 23-week fetus with unilateral clubfoot, showing the right lower limb with clubfoot and the left lower limb without clubfoot.

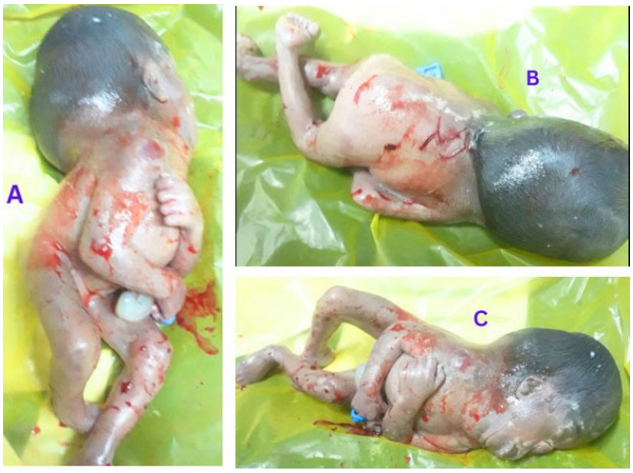

Figure 4: Photograph of an iniencephaly stillborn after termination of pregnancy at 23 + 6 weeks, with lateral view (A), posterior view (B), and anterolateral view (C).

Case 2

A 24-year-old primigravid woman, with an unknown last menstrual period, had her gestational age calculated at eight weeks via crown-rump length (CRL). She was first referred to Abebech Gobena Referral Hospital at 8 weeks and 4 days, where a vanishing twin was noted. Her baseline investigations were uneventful. She continued follow-up care at the same hospital and was subsequently referred to the Maternal Fetal Medicine unit for a fetal anatomic scan at 20 weeks gestational age.

During the anatomic scan, the vanishing co-twin was not visible, and a singleton viable intrauterine fetus was identified. The fetus exhibited a fixed hyperextended neck with a stargazing facial appearance, along with a

continuous scalp extending to the skin of the back. The axial view of the neck revealed bilateral neck edema, likely caused by lymphatic obstruction (Figure 5).

Additionally, the spine appeared unusually short with vertebral splaying, particularly in the cervicothoracic region, although it was covered by overlying skin as shown in Figure 6A and B respectively.

The final diagnosis was iniencephaly. After counseling on the prognosis, the mother opted for pregnancy termination. Following the initiation of mifepristone and misoprostol, she delivered a 450 g freshly died male stillborn (Figure 7). The post-expulsion period was smooth, with no complications. The autopsy was not performed because the family chose not to pursue further investigation of the stillborn baby due to cultural and religious beliefs.

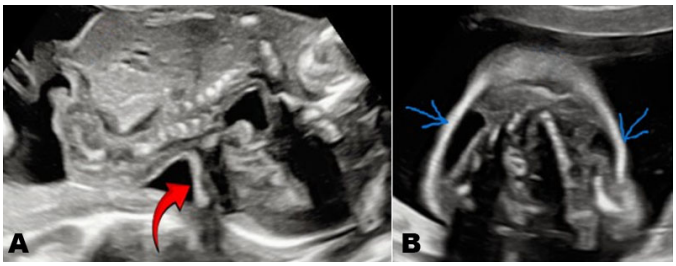

Figure 5: A 20-week fetus with a hyperextended neck (A), along with a continuous scalp extending to the skin of the back (red arrow). The axial view of the neck (B) shows bilateral neck edema (arrow), likely due to lymphatic obstruction.

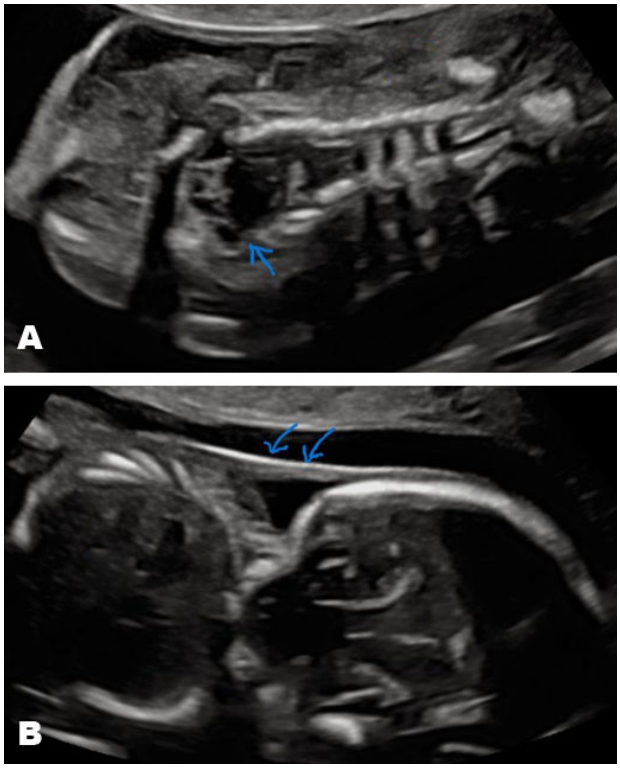

Figure 6: Grossly shortened spine with vertebral splaying (arrow), particularly in the cervicothoracic region (A). A continuous scalp (double arrow) extending to the skin of the back as shown in (B).

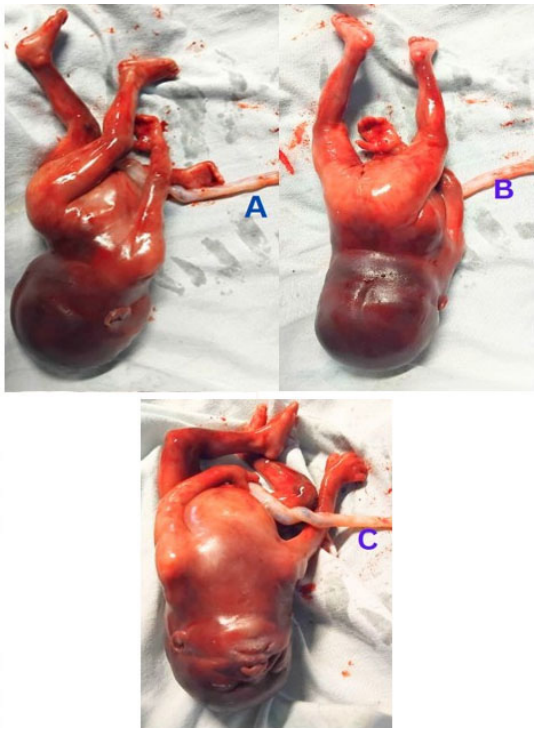

Figure 7: Photograph of an iniencephaly stillborn after termination of pregnancy at 20 + 4 weeks, with lateral view (A), posterior view (B), and anterior view (C).

DISCUSSION

Iniencephaly is an abnormality in cervical vertebrae associated with cervicothoracic spinal retroflexion and neural tube defect [5]. The incidence of iniencephaly is approximately 1 in 100,000 to 1 in 1000 births, with a notable female predominance (90%) observed [6–10]. This contrasts with our case, as both of the stillborns were male.

Two types of iniencephaly are recognized based on the presence or absence of encephalocele: iniencephalus apertus, which involves the presence of encephalocele, and iniencephalus clausus, where encephalocele is absent [11, 12]. Iniencephaly can be diagnosed prenatally through ultrasound starting in the early second trimester. The diagnosis is typically straightforward due to the characteristic star-gazing appearance of the fetus [6].

The sonographic features of iniencephaly include: (1) A defect in the occipital bone, resulting in an enlarged foramen magnum; (2) Partial or complete absence of the cervical and thoracic vertebrae, with irregular fusion of the remaining vertebrae and incomplete closure of the vertebral arches and/or bodies; (3) A significantly shortened spinal column due to marked lordosis and hyperextension of the malformed cervicothoracic spine; and (4) An upward-turned face, with the mandible's skin directly connected to the chest, caused by the absence of a neck [7, 13, 14]. Polyhydramnios is commonly associated with iniencephaly due to impaired fetal swallowing [10].

Iniencephaly is thought to occur due to the arrest of the embryo in physiological retroflexion during the third week of gestation or from a failure in the normal forward bending process that typically happens in the fourth week. This results in a fixed retroflexion of the head and severe lordosis of the cervicothoracic spine [15]. While there is no definitive evidence for the etiologic factors, it appears that nutritional, environmental, and genetic factors have been suggested as potential causes [16, 17].

It has been observed that in geographic areas with a higher prevalence of anencephaly, there is also an increased prevalence of iniencephaly [5]. The majority of cases of iniencephaly are sporadic and likely related to polygenic inheritance [18]. However, it has also been reported as a result of congenital syphilis, maternal diabetes, and exposure to teratogenic substances such as triparanol, streptonigrin, and vincaleukoblastine [18]. Additionally, chromosomal abnormalities, including trisomy 18, trisomy 13, and monosomy X, have also been associated with this anomaly [19].

Although most patients are either stillborn or pass away within a few hours after birth, only a few survivors have been documented in the literature to date. However, the prognosis for infants with iniencephaly remains extremely poor [16, 17, 20]. Due to its nearly universally fatal prognosis, termination of pregnancy is common when this condition is diagnosed before the fetus reaches the age of viability [3, 18].

Given that iniencephaly has a recurrence rate of 1% to 5%, the Centers for Disease Control and Prevention (CDC), Institute of Medicine, all recommend that women who have previously had a pregnancy affected by neural tube defects take 4000 mcg of folic acid daily, starting at least one month before pregnancy and continuing through the first three months of pregnancy [17, 18, 21].

CONCLUSION

Iniencephaly is a severe and devastating condition that demands early prenatal diagnosis and counseling. Timely anatomic scanning enables an accurate diagnosis and provides families with the essential information to make informed decisions. In this case, early diagnosis facilitated appropriate management, including pregnancy termination, and was followed by a smooth post-expulsion period.

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Author Contributions

Tewodros Getahun Asfaw – Conception of the work, Design of the work, Acquisition of data, Analysis of data, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Birhanu Kebede Dina – Conception of the work, Design of the work, Acquisition of data, Analysis of data, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Yared Tesfaye Wube – Conception of the work, Design of the work, Analysis of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

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The corresponding author is the guarantor of submission.

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Conflict of Interest

Authors declare no conflict of interest.

Data Availability

All relevant data are within the paper and its Supporting Information files.

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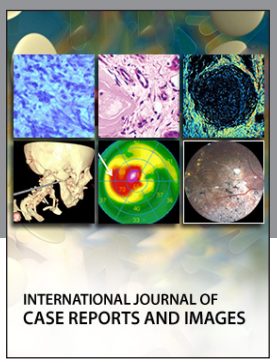

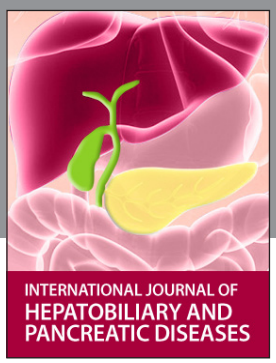

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