AN INIENCEPHALY CLAUSUS AS A RARE FORM OF COMPLEX NEURAL TUBE DEFECT: A CASE REPORT FROM RURAL ETHIOPIA

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ABSTRACT

BACKGROUND: Iniencephaly is a rare and severe form of neural tube defect characterized by a combination of malformations of the skull and spine, resulting in the head being severely retroflexed (bent backward) on the neck. Its prenatal diagnosis may be suspected during the first-trimester scan and can be confirmed from the early second trimester onwards. Unfortunately, most fetuses are stillborn, and others die after a few hours of life.

CASE SUMMARY: A thirty-year-old, Para II woman with one previous abortion from Western Ethiopia was referred to Wollega University Comprehensive Specialized Hospital with a diagnosis of second-trimester pregnancy and severe anemia secondary to recurrent malaria attacks. The ultrasound examination revealed findings suggestive of iniencephaly, and the patient was then counseled on termination of the pregnancy. After stabilizing the patient with a blood transfusion, the termination was carried out using 200 mg of mifepristone administered orally, followed by sublingual misoprostol. On the third day of the termination, the patient was discharged with advice on preconception care and the need for taking folic acid before and during her next pregnancy.

CONCLUSION: Prenatal diagnosis and management of such rare fetal anomalies pose significant challenges, especially in resource-limited settings where expertise in prenatal scanning is scarce. Consequently, it is advisable to identify women with risk factors for neural tube defects and to refer them for expert scanning by experienced sonographers or maternal-fetal medicine specialists.

KEYWORDS: Fetal anomaly, iniencephaly, neural tube defect, prenatal care, anatomic scanning

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INTRODUCTION

Iniencephaly is a rare neural tube defect characterized by a severe malformation of the skull and spine, specifically the occipital bone and cervical vertebrae. The defining feature is a marked retroflexion (backward bending) of the head, often appearing as if the face is looking upwards, with the back of the head fused to the upper spine[1, 2, 3]. The condition is divided into two types based on whether an encephalocele is present: iniencephaly apertus, which includes a sac-like protrusion of brain and membrane tissue through an opening in the skull, and iniencephaly clausus, which does not involve an encephalocele but still presents with a spinal defect[2, 3, 4].

Environmental factors, including poor socioeconomic conditions, insufficient folic acid supplementation, and the use of certain medications, as well as maternal diabetes and obesity, are all recognized as potential risk factors for the development of iniencephaly. While the exact cause remains unknown, it is likely a combination of these and other genetic factors. Some studies have also linked chromosomal abnormalities, such as trisomy 18, trisomy 13, and monosomy X, to the disorder[3, 4, 5].

Iniencephaly can be detected during pregnancy through ultrasound[5, 6]. It is almost always fatal, with most affected fetuses miscarrying or being stillborn[4,6]. There is no specific treatment for iniencephaly, but some cases with less severe malformations may survive with surgical intervention for spinal deformities[6]. In this report, the authors present a rare form of neural tube defect, iniencephaly, which was diagnosed at a tertiary hospital but missed at a primary hospital. This case highlights the importance of focused and early anatomic scanning by expert sonographers or maternal-fetal medicine specialists.

CASE PRESENTATION

Our case was a 30-year-old Para II Abortion I woman from Western Ethiopia who did not recall her last menstrual period (LMP) and was amenorrheic for the past five months. She was referred to Wollega University Comprehensive Specialized Hospital (WUCSH) with a diagnosis of second-trimester pregnancy complicated with severe anemia secondary to malaria. She received antenatal care at the primary hospital, where she was given iron tablets and tetanus and diphtheria (Td) vaccine. However, she did not receive folic acid supplementation during or before this pregnancy. The patient denied any history of smoking, khat chewing, or alcohol consumption. She did not take any other drugs during the current pregnancy. Furthermore, she had no known chronic medical conditions such as hypertension, diabetes mellitus, tuberculosis, or heart disease. Her past reproductive history included one abortion, two vaginal deliveries that resulted in one stillbirth, and one live neonate. There were no reported fetal anomalies in these pregnancies.

The patient was acutely sick-looking. Her vital signs were blood pressure 100/60 mmHg, pulse rate 80 beats per minute, respiratory rate 20 breaths per minute, and temperature 36°C. She had pale conjunctivae. The lymphoglandular, chest, and cardiovascular systems were normal. Abdominal examination showed a 26-week-sized uterus, no contractions felt, and the fetal heartbeat was 160 per minute. The genitourinary system showed no costovertebral angle tenderness; the cervix was closed, and the pelvis was adequate. She had palmar pallor and was oriented to time, person, and place. Reflexes were normal, with no neurologic deficits.

Blood group, random blood sugar (RBS), peripheral morphology, stool examination, liver function test, renal function test, VDRL, HBsAg, complete blood count (CBC), echocardiography, and electrocardiogram (ECG) (Table 1) were done.

Table 1: Laboratory investigations of woman managed for iniencephaly at Wollega University Comprehensive Specialized Hospital, Western Ethiopia, 2025

Laboratory tests	Results
CBC count	WBC count= 7,100 cells/µL; RBC count= 2.65 million cells/µL; *Hematocrit= 18.4%; Platelet count= 168,000 cells/µl; MCV=88.4 fL; MCH=31 pg
Liver function test	ALT=32 U/L; AST=37 U/L
Renal function test	creatinine=0.5 mg/dl
Urinalysis	Non-revealing
Stool exam	No ova or parasite seen
HBsAg	Non-reactive
RBS	140 mg/dl
Blood group	O+
Obstetric ultrasound	Positive FHB, AGA of 24 weeks, fetal thorax directly connected to head, no measurable fetal neck, fundal placenta, and DVP of amniotic fluid=8.3 cm
ECG	Normal finding
Echocardiography	Normal finding

AGA: Aggregate gestational age;
ALT: Alanine aminotransferase;
AST: aspartate aminotransferase;
CBC: Complete blood count;
DVP: Deepest vertical pocket;
ECG: electrocardiogram;
FHB: Fetal heart beat;
HBsAg: Hepatitis B surface antigen;
RBC: Red blood cell;
RBS: Random blood sugar;
WBC: white blood cell;
MCV: Mean Corpuscular Volume;
MCH: Mean Corpuscular Hemoglobin

*Hematocrit at discharge=28.1%

The obstetric ultrasound examination showed a positive fetal heartbeat, a fundal placenta, and the deepest vertical pocket of amniotic fluid at 5 cm. The fetal head directly connected to the thorax, with no clearly visible fetal neck, raising a high index of suspicion of a severe fetal anomaly, most likely iniencephaly (Figure 3). No structural anomalies were observed in the other body parts.

With the final diagnosis of second-trimester pregnancy complicated with severe anemia secondary to repeated attacks of malaria and severe fetal anomaly, the patient was admitted. In the ward, she was transfused with three units of whole blood. Following the stabilization of the patient, the managing team provided the family with advice and counseling regarding the seriousness of the fetal anomaly and the implications for pregnancy termination. Then, an informed decision was made to terminate the pregnancy. She was given 200 mg of mifepristone orally. Then, after 24 hours of mifepristone, 400 mcg of misoprostol was given sublingually. Eight hours later, she expelled a 600 g female abortus that had a hyperextended neck and short cervical spine but no encephalocele or other gross anomalies (Figure 1 and Figure 2). Autopsy and MRI examinations were not done. On the third postpartum day, the patient was discharged from the hospital with advice on preconception care and to take 4 mg of folic acid starting from three months prior to the next planned pregnancy.

Figure 1: Dorsal view of the iniencephalic fetus showing the absence of encephalocele (iniencephaly clausus), Wollega University Comprehensive Specialized Hospital, Western Ethiopia, 2025

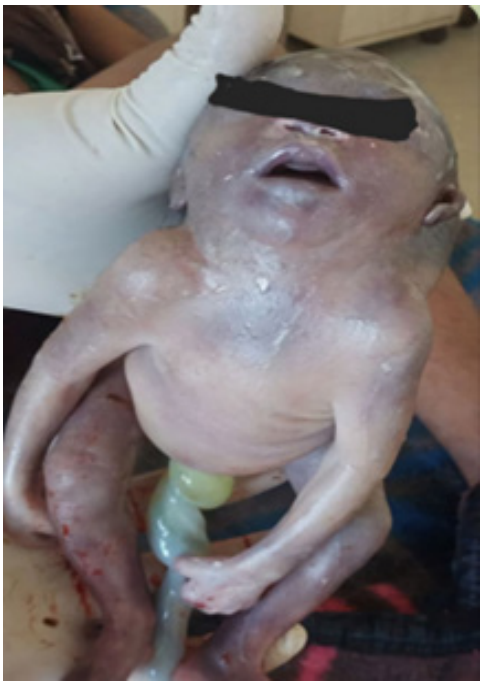

Figure 2: Antral view of the iniencephalic fetus showing hyperextension of the neck and continuation of mandibular skin to the chest wall, Wollega University Comprehensive Specialized Hospital, Western Ethiopia, 2025

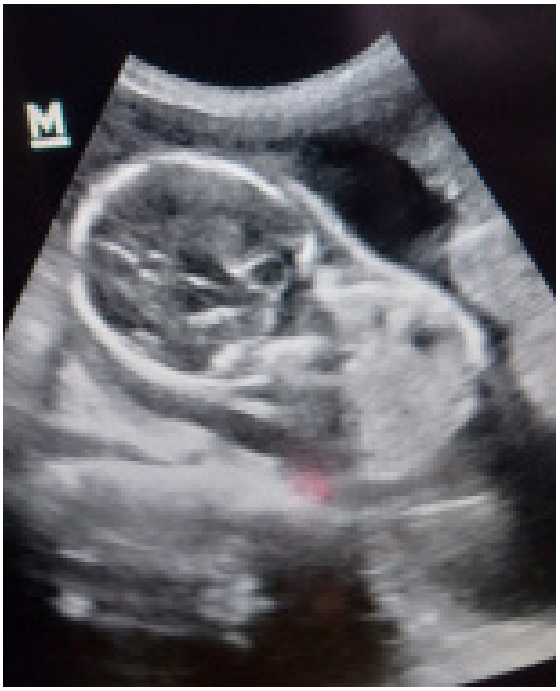

Figure 3: The direct contact between the foetal head and thorax, Wollega University Comprehensive Specialized Hospital, Western Ethiopia, 2025

DISCUSSION

The terminology "iniencephaly" is derived from the Greek words "inion" (meaning neck) and "encephalos" (meaning brain). It describes the attachment of the posteriormost part of the occipital bone to the back, resulting in the absence of the neck and retroflexion of the head[4, 7, 8]. Based on the presence or absence of encephalocele, there are two groups of iniencephaly: iniencephaly apertus and iniencephaly clausus[4, 8]. The current case has no encephalocele and is thus called iniencephalus clausus.

Iniencephaly has three distinct features: occipital bone defect, cervicothoracic vertebral body abnormality (part or all of the vertebral body is missing or fused), and arch fusion abnormalities. These anomalies cause shortening of the cervical spine and hyperextension of the cervicothoracic spine, causing the mandibular skin to continue with the chest wall[4, 5, 8, 9].

Iniencephaly is a rare form of neural tube defect. The incidence ranges from 0.1 to 10 in 10,000 pregnancies, and it is seen more frequently in females. It has a recurrence risk of 1–5% in subsequent pregnancies[9]. This case is also female iniencephaly.

The exact cause and pathogenesis of iniencephaly are not fully understood, but both genetics and the environment may play a role. Environmental factors that increase the risk of iniencephaly include poor socioeconomic status, low parity, lack of folic acid supplementation, diabetes, obesity, and certain drugs like sulfonamides, tetracycline, antihistamines, and cytostatic agents[4, 7, 9, 10]. In this case, there was no folic acid supplementation, and the woman was from a low socioeconomic status.

Chromosomal abnormalities such as trisomy 18, trisomy 13, and monosomy X have been detected accompanying this anomaly[5]. Because of a lack of facility, genetic tests were not done in the current case. Pregnancy at an extreme maternal age and self or family history of neural tube defects are also risk factors[4, 5].

Iniencephaly can be diagnosed during antenatal follow-up by careful ultrasound scanning. The fetus typically shows a "star-gazing" appearance on ultrasound examination[8, 10]. The fetus's head is hyperextended and retroflexed, facing upward as if looking at the stars[11]. A detailed examination of the spine and cranial bones is possible with magnetic resonance imaging (MRI)[8]. Furthermore, concomitant polyhydramnios may be seen on ultrasound imaging in fetuses with iniencephalus clausus[4]. In this case, prenatal diagnosis was missed at the primary (referring) hospital. However, the sonographers at the treating hospital successfully made the diagnosis, which can be attributed to their expertise and the quality of the ultrasound machine they used.

Iniencephaly, despite being diagnosed prenatally, is invariably fatal during the neonatal period. However, a limited number of case reports indicate instances of increased longevity. Consequently, termination of pregnancy is typically considered the preferred course of action in such circumstances[4,8,10]. In the present case, the pregnancy was terminated following a comprehensive discussion with the woman about its prognosis.

Iniencephaly has a recurrence rate of 1 to 5%[2]. Therefore, it is crucial to focus on prevention. Women should receive counseling regarding folic acid supplementation prior to future pregnancies. Taking a daily supplement of at least 400 mcg of folic acid before and during early pregnancy can significantly reduce the risk of neural tube defects, including iniencephaly. Pregnant women should avoid certain medications, including antiepileptic drugs, diuretics, antihistamines, and sulfa drugs, as these have been linked to an increased risk of neural tube defects[1, 2, 4].

CONCLUSION

Prenatal diagnosis and management of iniencephaly pose significant challenges, especially in resource-limited settings where expertise in prenatal scanning is scarce. Consequently, it is advisable to identify women with risk factors for neural tube defects and refer them for focused early ultrasound scans to facilitate the diagnosis of such severe fatal anomalies.

Abbreviations:
AGA: Aggregate gestational age;
ALT: Alanine aminotransferase;
AST: aspartate aminotransferase;
CBC: Complete blood count;
DVP: Deepest vertical pocket;
ECG: electrocardiogram;
FHB: Fetal heart beat;
HBsAg: Hepatitis B surface antigen;
RBC: Red blood cell;
RBS: Random blood sugar;
WBC: white blood cell

DECLARATIONS

Consent for publication: Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

Availability of data and materials: The datasets used during the current study are available from the corresponding author on reasonable request.

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