A Rare Case of Fetal Neural Tube Defect; Iniencephaly Clausus

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Abstract

Introduction Iniencephaly is an extremely rare type of neural tube defect characterized by the fusion of the cervical and cervicothoracic vertebrae. This condition results in acute retroflexion of the head, a short neck, significant lordosis of the cervical spine, and an upturned facial appearance. This condition typically results in poor fetal outcomes, with many cases ending in stillbirth or neonatal death.

Case summary Here, we present a case of iniencephaly diagnosed during intrapartum ultrasound in a 34-year-old gravida 5 woman referred from a health center to a primary hospital due to preterm premature rupture of membrane and labor. The fetus died intrapartum a few minutes before delivery.

Conclusion Iniencephaly remains a rare but critical condition that poses significant challenges for prenatal diagnosis and management. This case underscores the importance of early and accurate imaging in the detection of such severe anomalies, which can provide essential information for clinical decision-making and counseling.

Keywords
- iniencephaly clausus
- neural tube defect
- stillborn

Iniencephaly is a rare and lethal neural tube defect that was first described by Saint-Hilaire in 1836. This disorder is characterized by three main features: retroflection of the fetal head, occipital bone defect, and variable rachischisis.[1] Lewis discovered two types of iniencephaly in 1897: iniencephaly clausus, characterized by a spinal defect without encephalocele, and iniencephaly apertus, which shows both spinal defect and an encephalocele.[2–4] This condition can occur in 0.1 to 10 out of 10,000 cases, with females constituting more than 90% of the patients.[5–7] In this report, we describe a case of iniencephaly clausus that was initially suspected during intrapartum evaluation and subsequently confirmed postnatally.

Case Presentation

A 34-year-old gravida 5 para 4 (all alive) with a gestational age of $28^{6/7}$ weeks, according to a reliable last menstrual period, presented with vaginal fluid discharge lasting 8 hours, accompanied by lower abdominal and back pain for the past 2 hours. She had no prior antenatal care contact and was not supplemented with iron and folic acid. All her previous deliveries were normal, with no gross anomalies detected. On examination, she exhibited stable vital signs, pale conjunctiva, a 26-week-sized gravid uterus, positive cardiac activity, wet perineum, and a cervix dilated to 3 cm with effacement. Ultrasound evaluation revealed a single live intrauterine fetus with a short cervical spine, a retroflexed head, and no measurable fluid. After an 8-hour hospital stay, she delivered a 1.1 kg stillborn female fetus with a hyperextended neck and short cervical spine, but no other gross anomalies (►**Figs. 1** and **2**; ►**Video 1**). The family did not allow advanced examinations of the baby, such as an autopsy, due to religious reasons. Other investigations, such as MRI and X-rays, were not available at that hospital. The mother

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Fig. 1 Lateral view of the fetus showing cervical shortening with hyperextension of the neck and continuation of mandibular skin to the chest wall.

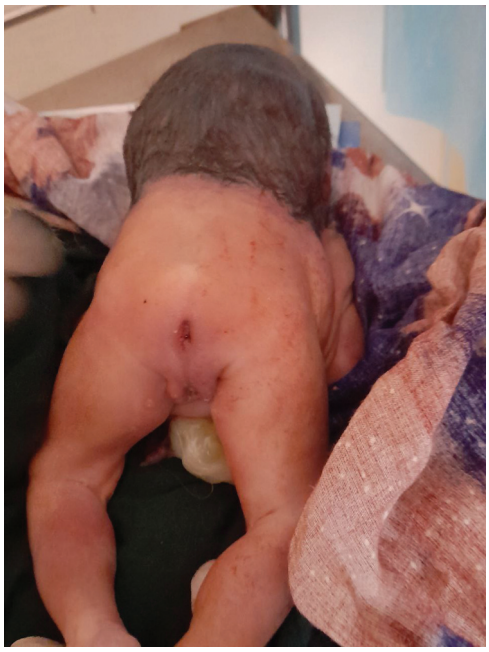

Fig. 2 Dorsal view of the fetus showing the absence of encephalocele (iniencephaly clausus).

was discharged in good condition after she was given folic acid and therapeutic iron.

Video 1

This video shows the immediate postdelivery examination video of a stillborn with iniencephaly calusus, demonstrating hallmark features such as a retroflexed head and spinal anomalies, with no associated encephalocele. Online content including video sequences viewable at: https://www.thieme-connect.com/products/ejournals/html/10.1055/a-2496-2417.

Discussion

The terminology "iniencephaly" is derived from the Greek word "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] Persistence of cervical lordosis in the third week of pregnancy causes the neural tube not to close. For this reason, abnormal development of the rostral portion of the notochord and somites of the cervical–occipital region may lead to the development of iniencephaly.[8] This condition is a rare neural tube defect with three distinct features: occipital bone defect, cervicothoracic vertebral body abnormality (part or all 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.[4,9,10]

The exact cause and pathogenesis of iniencephaly are not fully understood, but both genetics and environment may play a role. Environmental factors that increase the risk of iniencephaly are poor socioeconomic conditions, low parity, lack of folic acid supplementation, diabetes, obesity, and certain drugs. In this case, lower socioeconomic status and lack of folic acid supplementation during pregnancy are the identified risks for the disorder. Chromosomal abnormalities such as trisomy 18, trisomy 13, and monosomy X have been detected accompanying this anomaly.[10] Another possible risk is pregnancy early or late in a woman's life cycle.

Both iniencephaly and anencephaly can present with congenital retroflexion of the spine. However, it is important to distinguish iniencephaly clausus from Klippel–Feil syndrome and cervical meningomyelocele or teratoid masses.[3] Anencephaly is characterized by a complete or partial absence of the neurocranium, and in iniencephaly, the retroflexed head is completely covered with skin. Cervical spine abnormalities are common in iniencephaly, but almost always normal in anencephaly. In our case, retroflexion of the cervical spine was observed with spinal dysraphism, which was completely covered by skin.[3]

Regarding Klippel–Feil syndrome and iniencephaly, there are cervical segmental abnormalities in both, but hyperextension is not observed in Klippel–Feil syndrome.[9] Additionally, Klippel–Feil syndrome is not fatal and can be treated with surgery. The most common anomalies associated with iniencephaly include spina bifida (74%), diaphragmatic hernia (37%), adrenal hypoplasia (37%), clubfoot (32%), hypoplastic lung, single umbilical artery, omphalocele (26%), cardiovascular anomalies, genitourinary malformations, cyclopia, cleft lip and palate, and imperforated anus.[11,12]

Iniencephaly is always lethal in the neonatal period; however, a few case reports show increased longevity.[13] Prenatal diagnosis is possible with careful and early ultrasonographic examination. Magnetic resonance imaging can be used to evaluate those with equivocal fetal ultrasonographic findings.[14] Some of the ultrasound evidence for the diagnosis of iniencephaly includes a fusion of malformed vertebrae, incomplete closure of the vertebral arches and bodies, retroflexion of the cervical spine, hyperextended ("stargazing") position, and upward turned to face with a chin continuous with the chest.[9] Additionally, fetuses with iniencephaly clausus may present with concurrent polyhydramnios on ultrasound. Our case had some of these features, and hence iniencephaly was suspected prenatally and confirmed postnatally.

Prevention

Since iniencephaly has a recurrence rate of 1 to 5%, the women should be counseled for folic acid supplementation before future pregnancies. All women of reproductive age should be supplemented with 0.4 mg (400 µg) of folic acid daily to prevent neural tube defects. For a woman who has previously had a fetus affected with neural tube abnormalities, the Centers for Disease Control and Prevention (CDC) recommends increasing the intake of folic acid to 4 mg per day beginning at least 1 month before conception.

Conclusion

This case report highlights the importance of early and accurate prenatal diagnosis in managing rare and fatal congenital anomalies like iniencephaly clausus. The diagnosis was suspected based on characteristic ultrasonographic findings, which were later confirmed postnatally. Although iniencephaly is a rare neural tube defect with a poor prognosis, early identification can help in counseling the family regarding the condition and its outcome, and in preparing for the perinatal management of the mother. The report emphasizes the critical importance of folic acid supplementation in prenatal care to prevent neural tube defects, especially in women with a history of affected pregnancies, who should receive close monitoring and counseling to reduce the risk of recurrence.

Authors' Contributions

B.N.A., S.D.E., and D.M.H. were involved in the diagnosis, management, and follow-up of the case. B.N.A. was responsible for drafting the manuscript and revising it for submission. All authors have read and approved the manuscript and consented to its submission.

Data Availability

The data supporting the findings of this study are available from the corresponding author upon reasonable request.

Consent for Publication

The parents have provided consent for the publication of this case and the use of images for this purpose.

Conflict of Interest

None declared.

Acknowledgments

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