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CASE REPORT

Iniencephaly

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ABSTRACT

Iniencephaly is an uncommon form of neural tube defects which is characterized by retroflexion of the head and absence of neck as a consequence of defective closure of the vertebral body and arch. Multiple identified risk factors for its causation include environmental, genetic and drugs. We report a case of 38-year-old woman with prior history of still birth and abortions who presented at 35 weeks of gestation with lower abdominal pain and high blood pressure. Mother had consanguineous marriage. Her hypothyroidism was untreated in the first and second trimester. She delivered an iniencephalic baby girl via emergency c-section with multiple malformations at 38 weeks gestation secondary to fetal cardiac deceleration. Baby survived for less than 18 hours. In this case, proper antenatal care and follow up visits were needed along with postnatal genetic and pathological evaluation including assessment of risk factors. Appropriate management is important to prevent complications and recurrence in subsequent pregnancies.

Keywords: Hypothyroidism, Iniencephaly, Pregnancy

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Introduction

Iniencephaly is a rare form of neural tube defects (NTDs). It is manifested by the retroflexion of the head and absence of neck due to the abnormalities in vertebral body and arch closure. The exact mechanisms involved in its pathogenesis are not completely known; however, various environmental and genetic factors have been implicated in its causation.[1] Its incidence has been reported to be 0.1 to 10 per 10,000 births. The majority of the affected babies are females. Severe forms have a very short duration of survival however; milder forms are not usually fatal. [2] Here we report a case of iniencephaly born to a hypothyroid mother in the third trimester. In the present case report, modifiable risk factors for iniencephalic baby were obesity, socioeconomic status, folate intake, hypothyroidism, hypertension and consanguineous marriage whereas, female fetal sex and genetic factors may have played role as non-modifiable factors.

Case Report

A 38-year-old obese female G9 P6 A2 (none of the baby alive), admitted to Gynecology and Obstetrics Unit-I, Liaquat University Hospital, Hyderabad with 35 weeks of gestational amenorrhea, increased blood pressure and lower abdominal pain. She was a known case of hypothyroidism and hypertension for 9 years. She did not receive antenatal visits in her first trimester and did not take folic acid supplementations before and during pregnancy. Before pregnancy, she was non-compliant in taking medication for hypothyroidism and hypertension. Patient stopped taking thyroxine medication as she came to know about her pregnancy. However, in her third trimester, she was prescribed tablet thyroxine (dose: 200mcg/day) and tab methyldopa by a local doctor due to the hypothyroid and hypertensive profile,

respectively. No previous ultrasound scans and thyroid profile reports were available from the patient. She had a consanguineous marriage to her first cousin; however, no significant family history was found. She had history of late intrauterine fetal deaths (IUFD) in her first, second, sixth and eighth pregnancy and stillbirths in fifth and seventh pregnancy; however, no congenital anomaly was detected. In addition, she had history of two abortions in the third and fourth pregnancy. During her hospital stay, she was vigilantly monitored by cardiotocography, kick count and blood pressure monitoring. On the ultrasound, unstable lie, short and rotated spine with the retroflexed head was observed and CTG showed decelerations with loss of variability and bradycardia at 35th week. She was shifted to operation theatre for emergency lower segment caesarean section and alive baby girl was delivered with difficulty. The baby was in breech presentation and with the hyperextended neck for which uterine incision was extended. The baby was sent to the Paediatric ward for evaluation. The baby had low set ears, depressed nasal bridge, fixed retroflexed head and multiple cardiac and pulmonary abnormalities. Baby died after 18 hours of birth. Autopsy was refused by the parents, so further evaluation was not possible.

Discussion

Iniencephaly occurs before the closure of cephalic neural folds and characterized by the defects in the occipital bone, abnormalities of cervicothoracic vertebrae and retroflexion of the head. Its risk factors include low socioeconomic conditions, obesity, poor folic acid intake prior to conception and during pregnancy, drugs (such as antihistamines, sulphonamides and tetracyclines), methylenetetrahydrofolate reductase gene (MTHFR) C677T mutation, hyperhomocysteinemia and chromosomal defects such as monosomy X, trisomy 13 and 18.[3,4] It is associated with multiple anomalies, including anencephaly, hydrocephalus, cardiac septal defects, cleft lip and palate, pulmonary hypoplasia, genital malformations and skeletal abnormalities.[5] In the present case; obesity, poor socioeconomic conditions and female fetal sex, were important risk factors. Moreover, low levels of folate have been linked to increased homocysteine levels in a hypothyroid patient further increasing risk.[6] As patient did not take folic acid before and during pregnancy, the risk may have further increased. Patient had a marriage with her first cousin. The consanguineous marriages have been reported as a significant risk factor for neural tube defects in the Pakistani population.[7] Furthermore, hypothyroidism itself is associated with increased risks of abortion, prematurity, IUFD, intrauterine fetal growth restrictions (IUGR), congenital anomalies in the fetus, impaired brain development of the fetus, increased maternal and fetal morbidity and mortality[8]; suggesting risk factors for poor obstetrical outcomes in the present case. Although chronic hypertension is considered as a risk factor for congenital malformations in the fetus[9], its role in iniencephaly causation has not been discussed earlier. The ultrasound imaging in the second trimester is sufficient to diagnose neural tube defects; nonetheless, it requires competent ultrasonologist and proper equipment for detailed anatomical evaluation. Prenatal magnetic resonance imaging (MRI) may also provide an important tool for detailed neurological examination of the fetus, especially associated with neural tube defects.[10] It is therefore advised to have antenatal and follow up visits along with postnatal genetic and pathological examination. Furthermore, proper evaluation of risk factors and relevant management is important to prevent complications and recurrence in subsequent pregnancies.[11]

Conclusion

Iniencephaly is one of the less common and complex neural tube defects that requires proper obstetrical management. Patients with previous history should be counselled and recommended to take folic acid supplementations.

Figure I and II: An iniencephalic baby presented with retroflexion of head, low set ears and depressed nasal bridge.

Consent: An informed consent from the patient to report the study and for publishing photograph was taken.

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