Iniencephaly: A Rare Anomaly

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Abstract

Following case report is an attempt to ascertain that the fetus obtained and evaluated presents with iniencephaly, which is one of the rare variants of neural tube defects. Most of the cases of iniencephaly are reported as isolated incidences. However, some researchers have reported its occurrence in cluster, in a short span of time in a specific population. This case report elaborates various gross anomalies in a fetus which differentiates it from other neural tube defects. The study was performed by examining the external features as well as the radiograph of the abortus. The study also elaborates in brief the different versions of neural tube anomalies which are sometimes concomitantly observed in the same fetus. Prenatal or postnatal diagnoses of iniencephaly with or without associated anomalies are suggestive of chromosomal abnormalities and in such cases cytogenetic investigations are highly recommended.

Keywords
- neural tube defects
- anencephaly
- cervical rachischisis
- prenatal diagnosis

Introduction

Congenital malformation (CMF) is a physical, metabolic, or anatomical defect which is apparent before birth, at birth, or detected during the first year of life.1 CMF is among the most distressing diseases that affect mankind and some of them are more bizarre and unique than others. Periodic reporting of rare variants of congenital anomalies in a given geographic location and in a specific community might help the clinicians improve their diagnosis and avoid forthcoming catastrophes. Improved knowledge of the unexpected fetal losses will lead to better couples counseling and this will eventually help prevent recurrences. Early diagnosis of life-threatening CMFs can also pave the way for surgical correction in certain anomalies (by doing fetal surgery). Also, it might help prevent further congenital anomalies in subsequent conceptions. Besides, in an academic institute, such research and feedback will be useful in maintaining higher standards of teaching and clinical practice.

Even though the conventional methods of antepartum ultrasonography and biochemical analysis might suffice to diagnose the anomalies, they are surely not enough to completely elaborate the structural anomalies. The definition of neural tube defects (NTDs) includes variety of defects, such as anencephaly, cranial rachischisis, iniencephaly, meningocele, meningo(myelo)cele, and encephalocele.2 Anencephaly was the first fetal malformation diagnosed prenatally by Camp-belle et al, using transabdominal ultrasound.3 However, iniencephaly (“inion” means the nape of the neck) was first reported by Saint-Hilaire in 1836.4 The incidence rate of iniencephaly ranges from 0.1 to 10 in 10,000 in general population all over and has been found most often in female fetuses.5 Whereas, in India, the incidence has been reported to be 1 per 65,000 deliveries.6 Iniencephaly is almost always associated with numerous other anomalies such as anencephaly, encephalocele, meningo(myelo)cele, hydrocephalus, Dandy–Walker malformation, hydronephrosis, polycystic kidneys, cardiac defects, caudal regression syndrome, clubfoot, etc.7 And because of this, it often goes unnoticed or unreported in many cases giving a false impression of its incidence rate in general population.

Extreme forms of NTDs (such as anencephaly, encephalocele, cervical rachischisis, and iniencephaly) are incompatible with life. Among these, iniencephaly is a very rare NTD which is characterized by the triad of retroflexion of the head, cervical lordosis, and an occipital bone defect.8

Case Report

The following study was conducted in the Department of Anatomy, R.C.S.M. Government Medical College, Kolhapur,
Maharashtra, India. A stillborn fetus was obtained from the Department of Obstetrics and Gynecology. The patient had poor antenatal care and almost no follow-up or prenatal investigations. Because antenatal findings on ultrasonography were found to be incompatible with life, the patient was counseled for termination of pregnancy. The pregnancy was medically terminated citing severe anomalies. A brief maternal history regarding any disease, intake of drugs, socioeconomic status, paternal consanguinity, etc., was collected. But these factors were found to be insignificant and futile in the present study. The fetus was clinically evaluated by external examination as well as by radiography. All the findings were documented and photographed.

The external examination of the fetus revealed the following:

The foot length and crown rump length of the fetus under study was 5.5 and 15.5 cm, respectively. The gestational age of the aborted fetus was estimated as 20 weeks approximately. The placenta and umbilical cord were found but showed no gross anomalies. On examining the fetus properly, the cranial vault, cerebral hemispheres, and diencephalic structures were absent. Facial structures and orbits were present but poorly developed. Orbits showed large protruding eyes (►Fig. 1) and pinnae were poorly developed. There was prominent retroflexion of the head which has been described as “star gaze posture” (►Fig. 2) in the literature. The skin over the parietal and occipital region and over the nape of the neck was poorly developed and appeared dark and shriveled (►Fig. 3). However, there was no overt rachischisis of the thoracic or cervical spine externally. The skin over the rest of the spinal column showed no signs of abnormality on external examination. The neck was almost completely absent and skin below the mandible was continuous with that of the chest. There was no abnormality of appendicular skeleton in general. Both anterior abdominal wall and thoracic wall were intact and showed no abnormalities externally. All these findings were partly consistent with NTDs of cranial end, mostly suggestive of iniencephaly.

Examination of the radiograph (anteroposterior view) of the abortus showed the following defects:

The squamous part of the occipital bone was nearly completely absent resulting in an enlarged foramen magnum. The parietal, frontal bones were not seen and the temporal bones were partially formed. There was partial absence of cervical and thoracic vertebrae with no fusion of those present. This was accompanied by incomplete closure of the vertebral arches and their bodies. There was significant shortening of the spinal column in the cervical region due to hyperextension of the malformed cervicothoracic spine (►Fig. 4). The ribs in the upper thoracic region were partly formed and not fused, whereas those in the lower region were absent. The growth in the rest of the skeletal system was appropriate to the age and did not show any significant defects.

Since prenatal investigations were not available for evaluation, the above-mentioned findings could not be corroborated with that of the antenatal studies.
Iniencephaly: A Remarkable Anomaly

Chavan

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Discussion

NTDs are common CMFs resulting from complete or partial failure of the neural tube to close during the phase of development. NTDs can be classified as open type, if neural tube is exposed or covered only by membrane, and closed type, if the defect is closed by the skin. However, the closed types are rarely seen.

The prevalence of NTD varies greatly from region to region in the same country and in populations of different countries. The highest incidence of NTD has been reported from Ireland and Wales (6.38–10.92 per 1,000 births). The prevalence of NTD from different parts of India has been reported to vary from 0.5 to 11 per 1,000 births.

Iniencephaly is one of the types of NTDs which presents with characteristic features which include a short or absent neck with extreme retroflexion of the head, occipital bone defect with enlarged foramen magnum, and malformed cervicothoracic vertebrae (i.e., craniospinal rachischisis). It is a developmental error occurring in early pregnancy prior to closure of cephalic end of the neural tube at 24 days of gestational age. The defective neural arches reach directly into the cervical part of the spinal canal, causing the formation of a common cavity between most of the spinal cord and the brain. The spine is severely distorted and it is significantly shortened due to marked lordosis.

Even though both genetic and environmental factors have been implicated; however, the exact etiology of this condition remains unresolved. Environmental factors such as poor socioeconomic conditions, teratogenic effects of drugs (especially antibiotics and antiepileptic drugs), and lack of folic acid supplementation have been implicated. Genetic factors mostly associated with iniencephaly include chromosomal abnormalities such as trisomy 13, trisomy 18, or Turner syndrome. Most of iniencephalic babies are stillborn or die soon after birth.

Some of the variants of NTDs are concomitantly seen in the same fetus. Based on these facts, iniencephaly has been classified into two groups, namely, iniencephaly apertus when it is associated with encephalocele and iniencephaly clausus when it is not associated with encephalocele. Iniencephaly apertus should be differentiated from anencephaly with retroflexion of the spine. Iniencephaly clausus should be differentiated from Klippel–Fiel syndrome (KFS) and cervical meningomyelocele. Although fusion of cervical vertebrae and malformation may be present in both, retroflexion of the head is not seen in KFS and is classically seen in the cases of iniencephaly. It is important to differentiate these two conditions because KFS is not lethal and can be surgically corrected. Other entities that can be listed in differential diagnosis are teratoma, lymphangioma, and Jarcho–Levin syndrome. This condition can cause obstructed labor because of retroflexion of the fetal head in advanced gestational weeks and can also recur in subsequent pregnancies.

The fetus in our case had most of the above-mentioned features. It had no swelling in the cervical region and so our case can be classified as iniencephaly clausus. Besides these, no other signs of other NTDs were seen in the...
aborted. Iniencephaly may be associated with other external deformities like arthrogryposis and clubfeet, but no such defects were seen in the present study.

Prenatal diagnosis can be performed by various methods of which ultrasonography remains the investigation of choice in the 1st trimester. However, magnetic resonance imaging provides a reliable tool to evaluate the fetus and outline the characteristics of fetal disorders in utero, thus allowing proper classification of the NTD. Detailed study of the scan of the fetus can reveal associated systemic anomalies. Assessment of serum biochemical markers for NTD such as α-fetoprotein levels in the maternal blood remains the gold standard for diagnosis, as it is for other types of NTDs.

After the diagnosis of such rare variants of NTDs in the prenatal period, it is advisable that the physicians should obtain detailed information of any other disorder detected in the family. There is a risk of recurrence of iniencephaly, especially among families with a history of NTD. Proper counseling of the couple with the help of a geneticist and psychologist is recommended. The expectant parents should be cautioned regarding anticipated complications.

Conclusions

Although not a novel work, the above-mentioned case has been reported for its rarity and uniqueness, as it has rarely been reported from this region. Most of our findings are consistent with the available literature on the NTDs and iniencephaly. Most of the NTDs are diagnosed by ultrasound examination and biochemical analysis, but fetal autopsy remains a neglected method of studying major CMFs in unviable fetuses, which usually reveals other structural anomalies. Failure to recognize iniencephaly in many cases explains the low incidence of reporting. And this error can be corrected by regular reporting of major anomalies. Surveillance and monitoring of congenital anomalies is important for reporting patterns of CMF. Termination in early pregnancy should be recommended to patients with iniencephaly fetus. Establishment of effective preventive measures by identifying major anomalies will reduce the socioeconomic burden of the patients to a great extent.

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Conflict of Interest

None declared.

References