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Case Report

Iniencephaly with Anencephaly: Case Report

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Abstract: Iniencephaly is an extremely rare neural tube defect characterized by the triad of fixed retroflexion of the head, variable degrees of cervical lordosis and dysraphism, and an occipital bone defect involving the foramen magnum. Incidence ranges from 0.1 to 10 in 10,000. Although there have been cases of iniencephaly reported as an isolated anomaly, it very rarely occurs alone. We report here a rare case of Iniencephaly in association with anencephaly.
Keywords: Iniencephaly, Anencephaly, Neural tube defect.

INTRODUCTION

Iniencephaly is an extremely rare neural tube defect characterized by the triad of fixed retroflexion of the head, variable degrees of cervical lordosis and dysraphism, and an occipital bone defect involving the foramen magnum [1]. Incidence ranges from 0.1 to 10 in 10,000 [2], more common in female babies. The word Inien is derived from the Greek word "Inion" which means nape of the neck [3].

Lewis classified iniencephaly into two groups on the basis of the presence or absence of encephalocele, - iniencephaly apertus and iniencephaly clauses [3].

Although there have been cases of iniencephaly reported as an isolated anomaly, it very rarely occurs alone [3]. Associated malformation involving CNS includes anencephaly, encephalocele, microcephaly, hydrocephaly, holoprosencephaly, posterior fossa defects and spinal defects [5-7] while non-CNS anomalies frequently associated include diaphragmatic hernia, omphalocele, thoracic cage deformities, genitourinary malformations, cyclopia, cleft lips and palate,club foot, congenital heart disease, imperforated anus and single umbilical artery [6, 7].

CASE REPORT

A 22 year old woman, primigravida was referred at 18 weeks of gestation for obstetrics ultrasound examination. On examination fetal cardiac activity was present with normal placenta, amniotic fluid volume and 3 vessel cords. There was absence of cranial vault, cerebral hemispheres and diencephalic structures. Facial structures and orbits were present. Findings were consistent with '*acrania-anencephaly sequence*' (Fig. 1). Neck was absent with persistent retroflexion of the head and rachischisis of the thoracic spine, findings wereconsistent with '*Iniencephaly*' (Fig. 2). Because antenatal findings were incompatible with life patient was counseled for termination of pregnancy.

Abortus confirmed findings of anencephaly i.e. absence of skull vault with exposed, disorganized brain tissue. There was absent neck with mandibular skin touching the chest, retroflexion of head what has been described as 'star gaze posture' in literature; with rachischisis of thoracic spine (Fig. 3).There was no abnormality of appendicular skeleton and anterior abdominal wall was intact. Radiograph of abortus (A.P. and Lateral views) confirmed absence of cervical spine and rachischisis of dorsal vertebrae (Fig. 2).

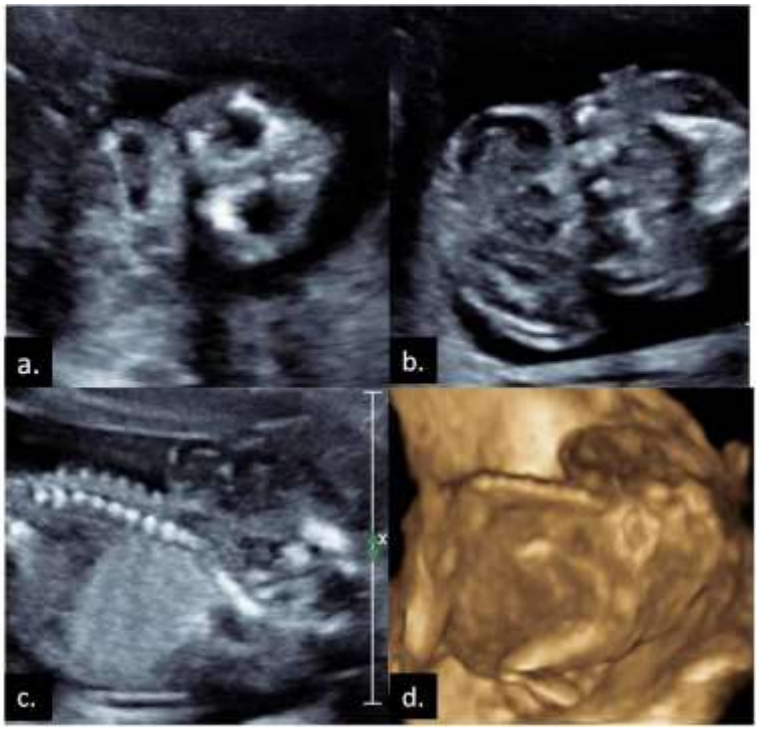

Fig.1: a and b (axial sections) c and d (sagittal sections) showing normal appearing orbits and face but with absence of calvarium and disorganized brain tissue with persistent retroflexion of head.

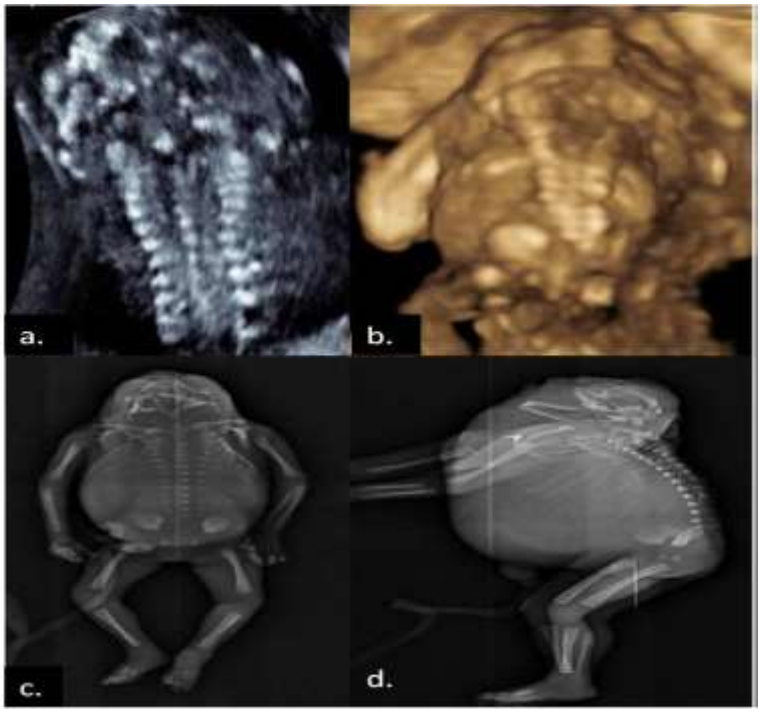

Fig. 2: a (Coronal section spine) b (3 D Image) c and d (Radiograph of abortus AP and Lat. views) showing absence of cervical vertebrae and rachischisis of upper thoracic vertebrae.

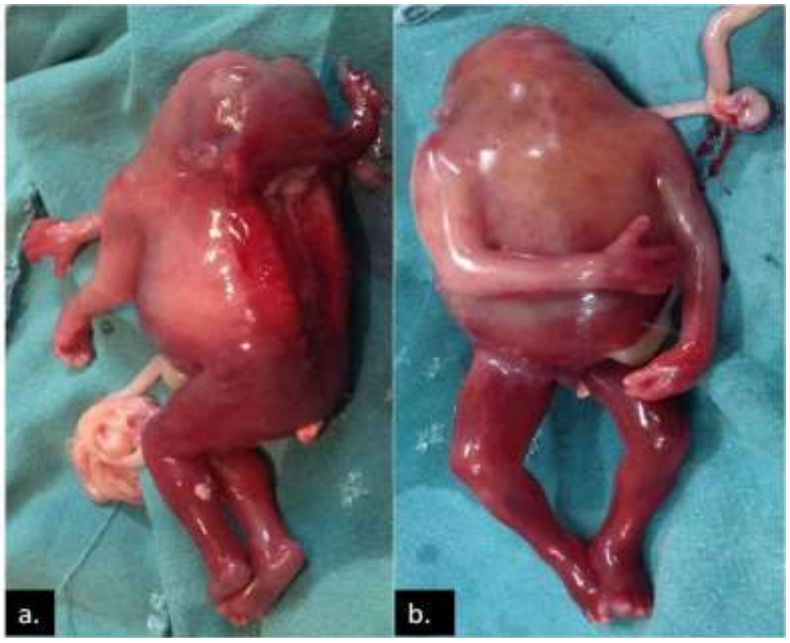

Fig. 3: a and b showing post abortion 18 week fetus showing features of anencephaly with absent neck and rachischisis of dorsal spine. Retroflexion of foetal head with mandibular skin touching chest.

DISCUSSION

Iniencephaly is an uncommon neural tube defect (NTD) [8]. The first description of iniencephaly is attributed to Saint-Hilare in 1836 [4]. The etiology is still unclear and no causative agent has been defined. Most cases of iniencephaly are sporadic and are probably secondary to multifocal inheritance [9]. Chromosomal abnormalities including trisomy 18, trisomy 13, and monosomy X have been associated with this disorder [10]. Environmental causes include poor socioeconomic conditions, low parity, and lack of folic acid supplementation, obesity and drugs including sulphonamide, tetracycline, antihistamines, and antitumor agents are shown to have increased risk [11]. Congenital syphilis and maternal use of teratogenic chemicals (streptonigrin, triparanol and vincoblastine), and antibiotics (sulfonamide, tetracyclin), antihistamines, cigarette smoking, alcohol use, and substance abuse were reported [12-14].

The important features for diagnosis of iniencephaly include occipital bone deficit leading to enlarged foramen magnum, irregular fusion of malformed vertebrae, incomplete closure of vertebral arches and bodies, retroflexion of the cervical spine, upward turned face with chin continuous with chest because of the absence of neck [2, 3].

Iniencephaly apertus should be differentiated from anencephaly with retroflexion of spine. Iniencephaly clausus should be differentiated from Klippel–Fiel syndrome (KFS) and cervical meningomyelocele [10].

Iniencephaly is usually fatal but rare cases with long term survival had also been reported [15]. It depends on the severity of associated anoamalies and the degree of retroflexion [4].

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