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Case Report

INIENCEPHALY AND ASSOCIATED ANOMALIES IN A 13 WEEK FETUS

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ABSTRACT

Iniencephaly is a rare neural tube defect (1 in 65,000 births in India). It involves defect of occiput and inions combined with rachischisis and retroflexion of head. We present a rare case of inencephaly diagnosed on ante-natal ultrasound in a 13 weeks fetus and associated with cystic hygroma and single umbilical artery. The fetal cervico-thoracic spine was retroflexed, occipital area was soft and brain tissue and spinal cord was visible externally in cervical region. The side of neck showed subcutaneous edema and two vessels (1 umbilical vein and 1 umbilical artery) were present in the umbilical cord. On x-ray examination spinous processes of all vertebrae showed presence of ossification centers except in the cervical region.

KEY WORDS: Iniencephaly, Rachischisis, Cystic hygroma.

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INTRODUCTION

Iniencephaly is a rare neural tube defect which is incompatible with life. The incidence of iniencephaly is reported to be 1 in 65,000 births in India and is more common in female fetuses [1].

However, iniencephaly has rarely been reported pre-natally and in association with cystic hygroma and single umbilical artery. We report a case of iniencephaly diagnosed on ante-natal ultrasound in a 13 weeks fetus and associated with cystic hygroma and single umbilical artery.

CASE REPORT

A rural 27 year old, gravid 2, para 1 presented with fetus with 13.2 weeks of gestation. She had previous history of a child born with intra-uterine growth retardation (IUGR) but no known congenital anomaly who died 10 days after birth due to sepsis. Rest of maternal and family history was insignificant.

The ultra-sound results of present pregnancy showed a single live fetus of 11-12 weeks of gestation with features of iniencephaly, cystic hygroma and single umbilical artery. Keeping in view the poor prognosis of the fetus, the parents decided to undergo medical termination of pregnancy (MTP).

A physical examination and x-ray study was done of the 13 week fetus and the findings were correlated with pre-natal sonographic findings. On external examination of the fetus, the cervico-thoracic spine was retroflexed.

The occipital area was soft, flattened and brain tissue and spinal cord was visible externally in cervical region [Figure 1]. Below the level of cervical region, no defect was observed and vertebral column was covered by normal skin. The skin overlying the side of neck was loose and soft indicative of subcutaneous edema. The umbilical cord showed the presence of 2 vessels (1 umbilical vein and 1 umbilical artery). The x-ray showed presence of ossification centers in spinous processes of all vertebrae except in the cervical region [Figure 2].

Fig. 1: Photographs showing retroflexion and rechischisis of cervical spine.

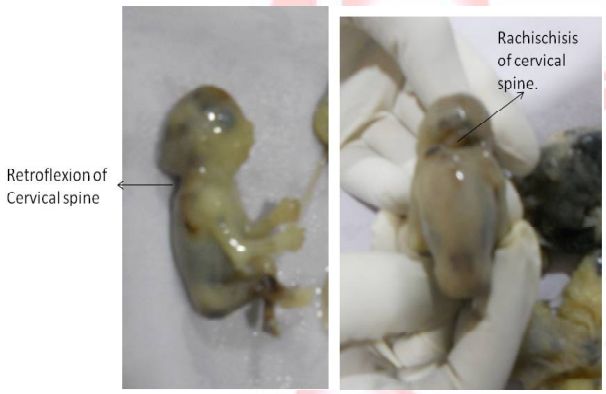
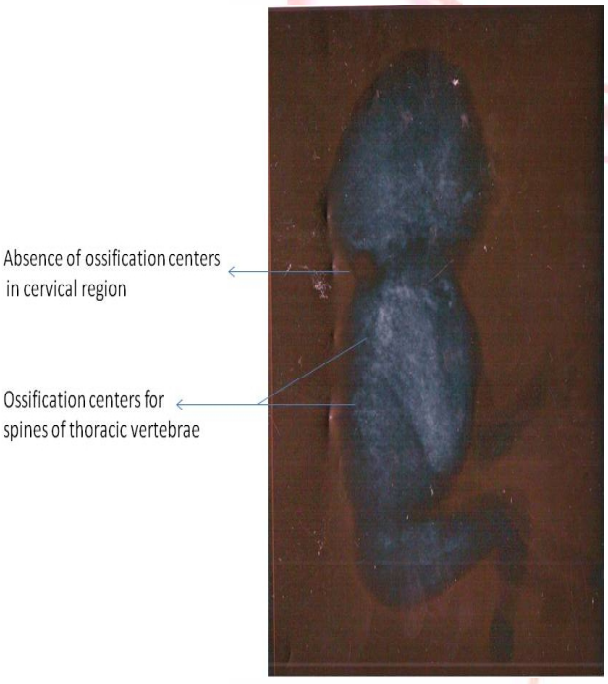


Fig. 2: Radiograph showing ossification centers.



DISCUSSION

The term inien is derived from the Greek word inion which means nape of the neck. In iniencephaly the posterior most part of occipital bone fuses with the back leading to absence of the neck and retroflexion of the head. The diagnosis of iniencephaly rests on following criteria described by Ballantyne i) imperfect formation of base of skull, mainly in region of occiput and inions. ii) retro-flexion of the spine forcing the head of the foetus to look upwards iii) rachischisis of cervical and thoracic spine [2].

It is believed that the failure of development of both paravertebral sclerotomes of the ventral and dorsal masses is the primary defect responsible for this malformation [3]. Other possibilities include arrest of the embryo in physiologic retroflexion in 3rd week of gestation, rupture of neural tube, disturbance of the paraxial mesoderm.

The exact etiology and pathogenesis is not known, both genetic and environmental causes have been implicated. Chromosomal abnormalities including trisomy 18, trisomy 13 and monosomy X are associated with this disorder [4]. Environmental causes like poor socio-economic conditions, low parity, lack of folic acid supplementation, obesity and drugs including sulphonamides, tetracyclines, antihistamines and antitumor agents are known to have increased risk [5].

Iniencephaly is associated with various congenital anomalies like cardio-vascular disorders, diaphragmatic hernias and gastrointestinal malformations. In the present case it is associated with cystic hygroma and single umbilical artery. Cystic hygroma is a condition with marked lymphoedema and lymphatic dilatation occurring particularly in cranial region. It results from delayed communication of jugular lymphatic sacs with jugular veins or due to jugular lymphatic obstruction. Cases of cystic hygroma have been reported in association with anencephaly but not with iniencephaly [6]. Fetuses with single umbilical artery have a high prevalence of associated anomalies involving various organ systems including nervous, musculoskeletal, cardio-vascular, gastro-intestinal and genitor-urinary system. A previous study reports a case of iniencephaly with single umbilical artery [7].

Iniencephaly has rarely been reported prenatally. Antenatally it can be diagnosed by ultrasound or MRI. The fetus shows typical star-gazing appearance on USG and detailed CNS and spine abnormalities may be known on MRI [8]. It needs

to be differentiated from anencephaly with retroflexion of spine. In anencephaly the skin over retroflexed head is absent and cervical vertebrae are usually normal, whereas, in inience-phaly the skin overlying head is normal and cervical vertebrae are abnormal. Other differential diagnoses include Klippel-Feil Syndrome, congenital torticollis and nuchal tumors [9].

CONCLUSION

Iniencephaly is a very rare congenital anomaly. It may be associated with various anomalies like cystic hygroma and single umbilical artery. With the advances in modern imaging techniques, this condition can be diagnosed ante-natally in the early second trimester.

Conflicts of Interests: None

REFERENCES

[1]. Jayant K, Mehta A, Sanghvi LD. A study of congenital malformations in Mumbai. J Obstet & Gynaecol India. 1961;11:280-294.

[2]. Ballantyne JW. The embryo. In: Manual of Antenatal Pathology and Hygiene. Edinburgh: W Green and Sons, 1904: 272-285.

[3]. Ramakrishnan S, Sharma D, Ramakrishnan V, Parihar PS, Sharma S, Kanther DK. Iniencephaly with Cyclops (a case report). J Postgrad Med. 1991;37:51-2.

[4]. Chen CP. Prenatal diagnosis of iniencephaly. Taiwan J Obstet Gynecol. 2007; 46:199-207.

[5]. Tugrul S, Uludoğan M, Pekin O, Uslu H, Çelik C, Ersan F. Iniencephaly: Prenatal diagnosis with postmortem findings. J Obstet Gynaecol Res. 2007; 33: 566–569.

[6]. Sharma S, Singh M. Anencephaly with cystic hygroma: Antenatal detection. J K Science. 2000; 2(2):107-9.

[7]. Kulkarni PR, Rao RV, Alur MB, Joshi SK. Iniencephaly clauses: A case report with review of literature. J Pediatr Neurosci. 2011; 6(2): 121-123.

[8]. Gadodia A, Gupta P, Sharma R, Kumar S, Gupta G. Antenatal sonography and MRI of iniencephaly apertus and clausus. Fetal Diagn ther. 2010;27(3):178-80.

[9]. Hemal U, Solanki RS, Varsheney A, Baliga S. Prenatal diagnosis of iniencephaly on ultrasound. Indian J Radiol Imaging. 2004;14(3):265-6.

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