DOI: 10.7860/IJARS/2020/44889:2570

Case Series

Radiology Section

A Case Series of Iniencephaly- An Uncommon Congenital Anomaly

SENDHIL COUMARY ARUMUGAM[1], PAVITHRA BALAKRISHNAN[2], NITHYA MANI[3], SWATHY SRINIVASAN[4]

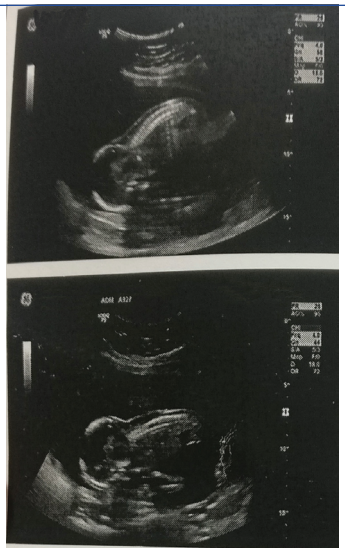
ABSTRACT

Iniencephaly is a complex and rare neural tube defect. It is characterised by the fusion of the cervical and cervicothoracic vertebrae associated with acute retroflexion of the head, a very short neck, marked lordosis of the cervical spine and thereby an upturned face. Anatomically there is a large foramen magnum and brain tissue herniates through the enlarged foramen magnum. Its incidence is about 1:1000-1:2000 births with female preponderance. Most of Iniencephalic babies are still born or die soon after birth. Here, two cases of Iniencephaly at different gestational ages were presented because of the rarity of this condition these cases have been reported.

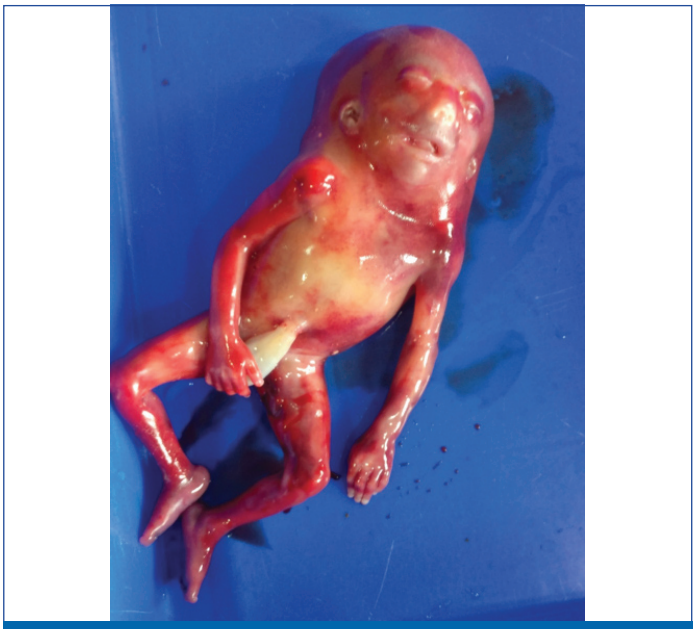
Keywords: Foramen magnum, Neural tube defect, Spinal dysraphism

CASE REPORT 1

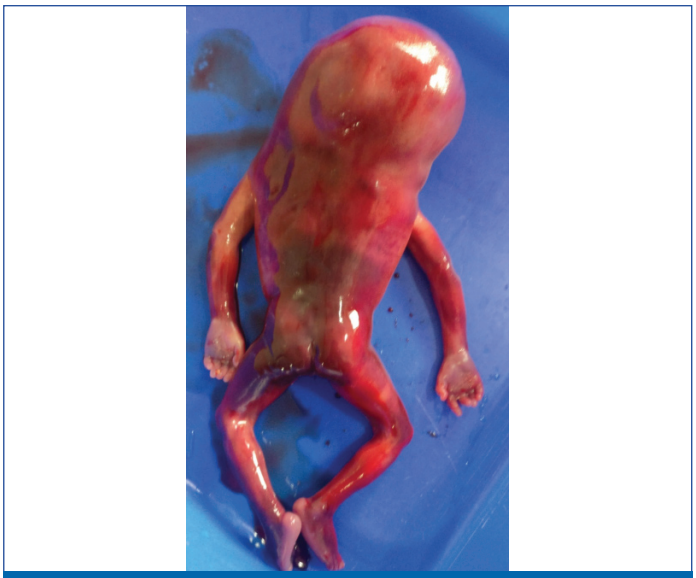
A 23-year-old primigravida presented to the OPD for routine antenatal booking at 16 weeks. Her previous antenatal visits were with a nearby health centre. There was no first trimester screening. All her antenatal blood investigations were within normal limits. An anomaly scan was carried out at 19 weeks. It showed a single live intrauterine fetus with gestational age corresponding to 19 weeks 1 day with subcutaneous oedema of the scalp and neck with an abnormal nuchal fold thickness of 8.5 mm. The fetal calvarium was normal but cerebellum was not visualised. There was abnormal retroflexion of neck with abnormal shortened spine with absent cervical spine, dilated lateral ventricles, abnormal posterior cranial fossa and a small thorax. Dorsal and lumbar spine were normal. No obvious spinal dysraphism was noted. All the features represented Iniencephaly [Table/Fig-1]. A second opinion from another fetal medicine specialist was sought and the diagnosis was confirmed. Patient was counselled regarding the poor prognosis and so she went ahead with medical termination of pregnancy. A dead female fetus of weight 300 g with fixed retroflexion of the head, without a neck, and with low-set ears was found. There was no lumbar meningocele [Table/Fig-2,3]. Since, the family did not accept autopsy, further evaluation for the presence of additional anomalies was impossible.



[Table/Fig-1]: Ultrasonography done using a GE C1-6 with 1-5 Mhz convex probe, showing the sagittal section of the fetus with abnormal retroflexion of neck with shortened spine (abnormal cervical spine) with increased nuchal fold thickening.



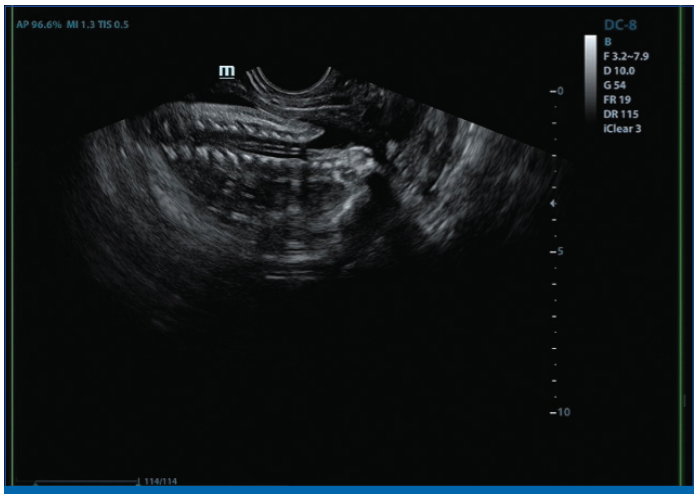
[Table/Fig-2]: Ventral view of the fetus showing cervical shortening with hyperextension of the neck and continuation of mandibular skin to the chest wall.



[Table/Fig-3]: Dorsal view of the fetus showing absence encephalocoele (iniencephaly clausus).

CASE REPORT 2

A 22-year-old second gravida with previous normal pregnancy outcome (G2P1L1) presented for her first antenatal visit. She did not had any previous visits to any of the hospitals. Therefore, she had not got any investigations done. After a basic clinical examination, as the uterine fundal height of the pregnancy did not correspond to the pregnancy dates she was stating she was asked to do an obstetric ultrasound. This ultrasound revealed a 34-weeks fetus with iniencephaly [Table/Fig-4]. A multidisciplinary team was formed comprising of neonatologist, obstetrician, anesthesiologist and a geneticist. The patient was counselled regarding the disease, the prognosis of the baby, need for resuscitation and the need for a caesarean if necessary. As termination of pregnancy at 34-weeks would not be legal, patient was counselled to continue pregnancy to term. Patient did not return back for further follow-up.



[Table/Fig-4]: Ultrasonography was done using Mindray Dc8 machine GE with a transvaginal probe 11-3Mhz showing the sagittal section of the cervical spine. The cervical vertebrae show spinal rachischisis - a feature of iniencephaly.

DISCUSSION

The Greek word "inion" means back of the neck. Here in iniencephaly, inion (external occipital protuberance) joins with the back, leading to retroflexion of the head and absence of the neck. Persistence of the embryonic cervical lordosis at the third week, leads to failure of closure of the neural tube. Thereby, abnormal development of the rostral portion of the notochord and somites of the cervico occipital region leads to development of iniencephaly. Iniencephaly is a rare neural tube defect with three distinctive features of occipital bone defect, cervicothoracic verterbral fusion (complete or partial) and abnormal arch fusion [1].

Iniencephaly leads to a very short neck with hyperextension causing the mandibular skin to be in continum with the skin over the chest wall. Saint Hillary described it first and hypothesised that the arrest of the embryo in physiological retroflexion caused this anomaly. Lewis classified them as iniencephaly apertus and iniencephaly clausus based on the presence or absence of encephalocele, respectively [2,3]. Both the cases presented here did not have an encephalocoele. The extreme extension of the head, which may be fixed, thus giving the characteristic appearance of star gazing position.

Other anomalies that lead to congenital retroflexion of spine are anencephaly and Klippel-Feil Syndrome (KFS). Cervical vertebrae are abnormal and retroflexed in iniencephaly while they are almost normal in anencephaly. Anencephaly shows partial or complete absence of neurocranium and no skin coverage of the retroflexed head while the retroflexed head is completely covered with skin in iniencephaly [4]. The KFS represents abnormal fusion of two or more cervical vertebrae leading to shortening of neck resulting in facial asymmetry. Patients with KFS can be polysyndromic and it is compatible with life [5].

Iniencephaly can be associated with neural and non neural anomalies. Commonly associated neural anomalies are anencephaly, holoprosencephaly, microcephaly, cyclopia, cerebellar cysts etc. Non neural anomalies include facial dysmorphism, chest deformity and pulmonary hypoplasia. Anomalies of the gastrointestinal, urinary, cardiovascular and urogenital systems are also seen frequently with iniencephaly. Both the cases presented here did not have other associated anomalies on ultrasound; however, an autopsy would have helped reveal other underlying minor anomalies.

Association of iniencephaly with other anomalies increases the fatality rate. The presence of a single anomaly necessitates a thorough examination of the other systems as well. Also, as open neural tube defects carry the risk of recurrence in subsequent pregnancies (1-5%), the patient should be counselled and encouraged to take folic acid supplementation at least three months prior to conception.

Early prenatal diagnosis of iniencephaly is possible with ultrasound as early as 14 weeks of pregnancy. Prenatal diagnosis of iniencephaly by ultrasonography is relatively easy due to severe anomalies of axial anatomy and vertebrae and typical fetal position [3]. Early diagnosis helps in counselling and decision making by parents.

The exact aetiology of iniencephaly is not clear, genetic, low socioeconomic status, low parity, folic acid and environmental factors have been implicated. In both the cases there was a lack of folic acid supplementation periconceptionally, which may have contributed to its development. It is invariably lethal in the neonatal period. However, there a few case reports showing increased longevity [6].

REFERENCES

[1] Romero R, Jeanty P, Pilu G, Ghidini A, Hobbins JC. "Iniencephaly" Prenatal Diagnosis of Congenital Anomalies, Appleton & Lange, Norwalk, Conn, USA 1988 Pp. 65-66.
[2] Kulkarni PR, Rao RV, Alur MB, Joshi SK. Iniencephaly clausus: A case report with review of literature. J Pediatr Neurosci. 2011;6:121-23.
[3] Pungavkar SA, Sainani NI, Karnik AS, Mohanty PH, Lawande MA, Patkar DP, et al. Antenatal diagnosis of Iniencephaly: Sonographic and MR correlation: A Case Report Korean J Radiol. 2007;8(4):351-55.
[4] Chandrupatla M, Swargam N. Anencepahaly: A case report. Int J Sci Stud. 2014;2(7):255-57.
[5] Agarwal AK, Mohit Goel, Jeetendra Bajpai, Sourav Shukla, Nikhil Sachdeva. Klippel Feil Syndrome: a rare case report. J Orthop Case Rep. 2014 Jul- Sep; 4(3): 53-55.
[6] Bakhti S, Tighilt N, Khoudir W, Djennas M. Iniencephaly with Long Survival: A Rare Condition. IrJNS. 2016;2 (3): 19-21.

PARTICULARS OF CONTRIBUTORS:
1. Professor, Department of Obstetrics and Gynaecology, Mahatma Gandhi Medical College and RI, Pondicherry, India.
2. Postgraduate, Department of Obstetrics and Gynaecology, Mahatma Gandhi Medical College and RI, Pondicherry, India.
3. Consultant, Department of Fetal Medicine, Nithya Scans, Senthil Speciality Hospital, Pondicherry, India.
4. Assistant Professor, Department of Obstetrics and Gynaecology, Mahatma Gandhi Medical College and RI, Pondicherry, India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:
Sendhil Coumary Arumugam,
9, Thillai Mestri Street, Pondicherry, India.
E-mail: sendhilcoumary1975@yahoo.in

PLAGIARISM CHECKING METHODS: [Jain H et al.]
- Plagiarism X-checker: May 05, 2020
- Manual Googling: May 21, 2020
- iThenticate Software: Sep 01, 2020 (21%)

ETYMOLOGY: Author Origin

AUTHOR DECLARATION:
- Financial or Other Competing Interests: None
- Was informed consent obtained from the subjects involved in the study? Yes
- For any images presented appropriate consent has been obtained from the subjects. Yes

Date of Submission: **May 04, 2020**
Date of Peer Review: **Jun 01, 2020**
Date of Acceptance: **Jul 24, 2020**
Date of Publishing: **Oct 01, 2020**