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INIENCEPHALY A RARE NEURAL TUBE DEFECT WITH REVIEW OF LITERATURE.

Radiodiagnosis

Surekha B* MD, DNB, Assistant Professor, Department of Radiodiagnosis, PES Institute of Medical Science and Research, Kuppam, Andhra Pradesh, India. *Corresponding Author

Naveen D DMRD, DNB, Assistant Professor, Department of Radiodiagnosis, PES Institute of Medical Science and Research, Kuppam, Andhra Pradesh, India.

ABSTRACT

Iniencephaly is rare neural tube defect. Multifactorial etiologies have been proposed. Two main types have been described apertus and clausus.

KEYWORDS

Iniencephaly, neural tube defect

INTRODUCTION:

Iniencephaly is an uncommon and fatal neural tube defect involving the occiput and inion combined with rachischisis of the cervical and thoracic spine with retro flexion of the head.[1]

Iniencephaly, from the ancient Greek "inion" meaning "nape of the neck", had its first report by Saint-Hilaire in 1836.[2]

Etiology:

The exact etiology and pathogenesis is not known, both genetic and environmental causes have been implicated.

Saint-Hilaire[2] first described this entity as being caused by arrest of the embryo in physiological retroflexion during the third week of gestation or by failure of normal forward bending during the fourth week.

Chromosomal abnormalities including trisomy 18, trisomy 13, and monosomy X have been associated with this disorder.[3]Environmental factors like poor socioeconomic conditions, low parity, and lack of folic acid supplementation, maternal syphilis obesity and drugs including sulphonamide, tetracycline, clomiphene citrate ,antihistamines, and antitumor agents a have been implicated as etiological factors, but there is no definite evidence.[4,5]

Previous reports have mentioned about risk of NTDs in mothers having hyperhomocysteinemia. Folic acid lowers the raised homocystein levels and reduces the risk of NTDs.

Two main groups were classified by Lewis:[1]

- Iniencephaly apertus which has an encephalocele.
- Iniencephaly clausus which has a spinal defect but no cephalocele.

Prevalence:

Reports vary from 0.1-10:10,000 .[6]

In India, incidence is reported to be 1 per 65,000 deliveries with the Female: Male ratio of 4.5:1.[7]

Iniencephaly is seen in families with a history of neural tube defects and the recurrence risk is 5%.[7]

Pathogenesis

Iniencephaly is a developmental error occurring in early pregnancy prior to closure of cephalic neural folds at 24 days gestational age. It is characterized by arrest as well as imperfect development of base of skull (portion of neural tube) and vertebral column (cervicothoracic region mainly).[8] According to some author iniencephaly is not considered to be an open defect because usually the sac containing CSF contains neural tissue and is covered by an intact dermis.[9]

At the early stage of embryo, paravertebral sclerotome (mesoderm) differentiates into two parts.[8]

1] A ventral mass (forms the vertebral bodies, pedicles and their cranial homologous)
2] A dorsal mass (forms neural arches and vault bones of the cranium).

In iniencephaly, one or both of these masses are hypoplastic or ill developed.

In Simple Iniencephaly, the defect found in occipitocervical region indicates site 1 and probably site 4 closure defects.

In iniencephaly with anencephaly where vault and facial bones as well as lower vertebrae are involved, in addition to occipitocervical region, favours all 5 sites closure defect hypothesis.[8]

Site 1 - Mid cervical.
Site 2 - Between prosencephalon and mesencephalon.
Site 3 - At stomodeum.
Site 4 - Caudal end of rhombencephalon.
Site 5 - Most caudal end of neural tube.

Exaggerated spinal retroflexion is due to absence of neural arches.
On microscopic examination of the brain, several anomalies have been detected "including microencephaly, polymicrogyria, heterotopic glial tissue in the leptomeninges, atresia of the ventricular system, marked disorganization of the brain stem, vermian agenesis, large cerebellar cyst, and disorganization of the spinal cord tissue".[10]

Diagnostic criteria:[11,12]

- A variable deficit of the occipital bones resulting in an enlarged foramen magnum
- Partial or total absence of cervical and thoracic vertebrae with an irregular fusion of those present, accompanied by incomplete closure of the vertebral arches and/or bodies
- Significant shortening of the spinal column due to marked lordosis and hyperextension of the malformed cervical-thoracic spine
- Upward turned face and mandibular skin directly continuous with that of the chest due to the lack of neck.

Most cases diagnosed prenatally have presented with high alpha-fetoprotein and/or polyhydramnios.

The diagnosis is made on the extreme dorsal flexion of the head, the abnormally short and deformed cervical and thoracic spine and the overall shortening of the fetus.[10,13] The retroflexion of the head and the spinal disorganization is visible on medial-sagittal scans of the spinal column.[14]

DISCUSSION:

We have detected 3 cases in our department. All of them came for NT scan with no previous scans done.

Primigravida with age group of 20 – 23 years.

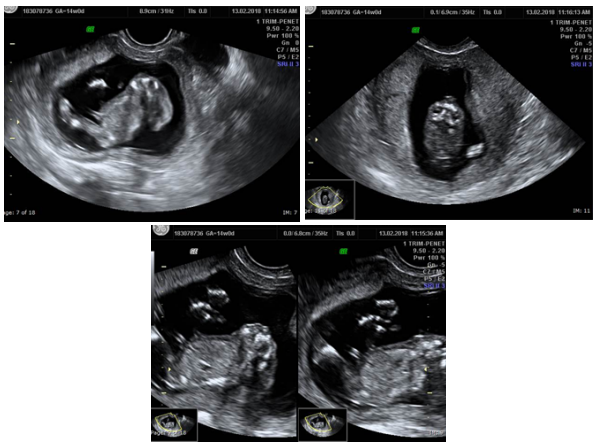
CASE1:

20-year-old primigravida with consanguineous marriage was referred to our department for NT scan. No previous ultrasound done. No history of folic acid supplementation.

Ultrasound findings (Fig - 1):

Single live intrauterine gestation corresponding to 13 weeks

Absence of skull vault with brain tissue free floating in amniotic fluid, exaggerated cervico thoracic lordosis, head in fixed retro flexion, Short neck, upturned face, multilevel segmentation abnormalities of the spine and short and stubby fingers

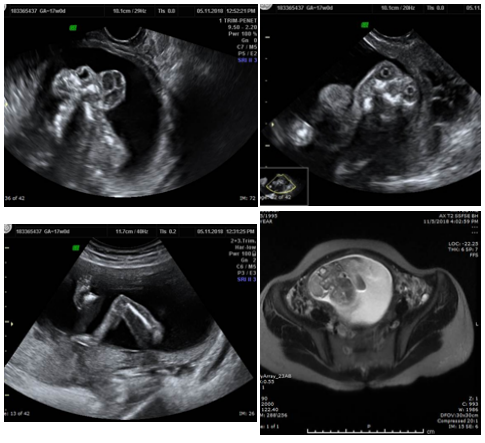


CASE2:
23year old female was referred to our department for NT scan. No previous ultrasound done.

Ultrasound findings (Fig - 2):
Single live intrauterine gestation corresponding to 15 weeks

There was near total absence of cranial vault with deformed brain seen free floating in the amniotic fluid.

Fixed retroflexion of the neck with upturned face. Hypotelorism seen. Kyphoscoliotic deformity of the spine with segmentation anomalies seen at multiple levels. Left club foot.

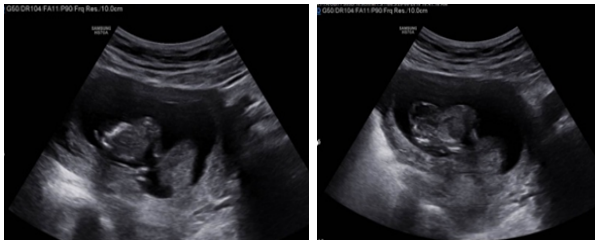


CASE3:
20year old female was referred to our department for NT scan. No previous ultrasound done.

Ultrasound findings (Fig - 3):
Single live intrauterine Gestation corresponding to 13 weeks 4 days

Absence of cranial vault with brain tissue seen free floating in the amniotic fluid. Spinal elements not well developed with exaggerated kyphotic deformity. Short neck with fixed retroflexion of head and upturned face.

Protuberant abdomen.



Associated anomalies
Several other anomalies have been associated with iniencephaly. These include anencephaly, cephalocele, holoprosencephaly, agnathia-microstomia-synmelia, spina bifida, omphalocele, gastroschisis, diaphragmatic hernia or agenesis, pulmonary hypoplasia or hyperplasia, cardiac malformations, renal anomalies, overgrowth of the arms compared to the legs, genu recurvatum, arthrogryposis, club-foot and gastrointestinal atresia.[1,15,16,18]

Differential diagnosis:
To make the diagnosis of iniencephaly, differential diagnosis must be made with cervical hyperextension, nuchal or cervical tumors such as teratomas, goiter, lymphangiomas, anencephaly, cervical myelomeningocele, encephalocele, Klippel-Feil syndrome and Jarcho-Levin syndrome.[19,6]

Klippel-Feil syndrome is caused by the unsuccessful segmentation of cervical vertebrae during early fetal development; is characterized by a short neck, low hairline implantation, and limited cervical movements. It can be associated with deafness, congenital heart defects, neurocognitive disorders, cleft palate, rib defects, Sprengel anomaly, dermoid cysts, scoliosis and kidney abnormalities. The involved gene (s) in this syndrome has not yet been determined, possibly located at 5q11.2, 5q35.1, 8p21.1, and 17q23.[19] The differentiation between iniencephaly clausus and Klippel-Feil syndrome is difficult and controversial. Some authors feel that Klippel-Feil syndrome may be the mildest form of iniencephaly.[20]

The distinction between iniencephaly apertus and anencephaly with spinal retroflexion relates to the time of onset.[15] Anencephaly arises prior to the closure of the cephalic neural folds at 24 days gestation[18]. Iniencephaly, on the other hand, arises after the cephalic neural tube has closed.[15]

The other important differential diagnosis is the spondylocostal dysostosis or Jarcho-Levin syndrome, which is an autosomal recessive disorder syndrome, is characterized by a short trunk, head in opisthotonus, short neck, a barrel chest, vertebrae minted or block, spina bifida and rib abnormalities.

Prognosis
Iniencephaly apertus is always fatal in the neonatal period.[21] Four cases of long-term survival of very mild Iniencephalus clausus have been reported,[22,23] although in these cases, the deformity was minimal and they should probably have been classified as Klippel-Feil syndrome.

Prevention
All women of reproductive age should consume at least 0.4 mg (400 mcg) of folic acid daily to prevent neural tube defects. For women who have previously had a fetus affected with acrania, the Centers for Disease Control and Prevention (CDC), recommends increasing the intake of folic acid to 4 mg (4000 mcg) per day beginning at least one month prior to conception.

REFERENCES

1. Lewis HL: Iniencephalus. Am J Obstet 35; 11-53, 1897.
2. Saint-Hilaire IG: Iniencephalus. In Histoire des Anomalies de l"Organisation. Paris J.B. Bailliere. (ed). Vol.2, Paris, 1836 pp 308-310.
3. Chen CP. Prenatal diagnosis of iniencephaly. Taiwan J Obstet Gynecol. 2007;46:199–207. [PubMed: 17962097]
4. Nyberg DA, Mahomy BS, Pretorius DH: Diagnostic ultrasound of fetal anomalies-Text and atlas. Littleton MA Year Book Medical Publishers, Inc p146 1990.
5. Semi T, Mehmet U, Oya P, Husamettin U, Cem C, Firat E. Iniencephaly: Prenatal diagnosis with postmortem findings. J Obstet Gybocol. 2007;33:566–9.
6. Romero R, Pilu G, Jeanty P et al: Prenatal diagnosis of congenital anomalies. Norwalk, CT: Appleton and Lange, 1988 pp. 64-67.
7. Jayant K, Mehta A, Sanghvi LD. A study of congenital malformations in Mumbai. J Obstet & Gynaecol India 1961;11:280-294
8. Congenital malformations associated with iniencephaly Bhagirath Kandhare1, Ashutosh Chitnis2, Divya Bansal3, Bhavin Patel4
9. Williams F, Griffiths PD. Spinal neural tube defects on in utero MRI. Clin Radiol. 2013;68:e715–22. [PubMed: 24034547].
10. Morocz I, Molnar P, Toth Z, et al: Diagnostic criteria of iniencephaly based on the authors" own cases and the review of the literature. Morphol Igazsagugyi Orv Sz 25:58-65, 1985.
11. Cimmino CV, Painter JW: Iniencephaly. Radiology 79:942-944, 1962.
12. Bose S, Makhani JS, Thaker SV: Iniencephaly. Indian J Med Sci 18:590-594, 1964.
13. Hrgovic Z, Panitz HG, Kurjak A, et al: Contribution to the recognition of iniencephaly on the basis of a new case. J Perinat Med 17:375-9, 1989.
14. Morocz I, Szeifert T, Molnar P, et al. Prenatal diagnosis and pathoanatomy of iniencephaly. Clin Genet 30:81-86, 1986.
15. Lemire, RJ, Beckwith, B, Shepard TH: Iniencephaly and anencephaly with spinal retroflexion, a comparative study of eight human specimens. Teratology 6:27-36, 1972.
16. Osborne DF: Iniencephalus. Can Med Assoc J 59:474-475, 1948.
18. Schram JHN, Krenning RA, Olthof R, et al. A case of iniencephaly. Ned Tijdschr Geneeskd 124: 1108-1111, 1980.

19. J Pediatr Neurosci. 2015 Apr-Jun; 10(2): 181–184.doi: 10.4103/1817-1745.159211PMCID: PMC4489071Iniencephaly: Case ReportHernando R. Alvis-Miranda, Doris A. Bula-Anichiarico, Willem G. Calderón-Miranda,1 and Luis R. Moscote-Salazar.
20. Gilmour JR: The essential identity of Klippel-Feil syndrome and iniencephaly. J Pathol 53:117-131, 1941.
21. Nishimura H, Okamoto N: Iniencephaly. In PJ Vinken & GW Bruyn (eds) Handbook of Clinical Neurology. Vol 30, New York: North-Holland Biochemical Press, 1977 pp. 257-268.
22. Sherk HH, Shut L, Chung S: Iniencephalic deformity of the cervical spine with Klippel-Feil anomalies and congenital evaluation of the scapula. J Bone Joint Surg 56-A:1254, 1974.
23. Katz VL, Aylsworth AS, Albright SG: Iniencephaly is not uniformly fatal. Prenat Diagn 9:595-599, 1989.