

Iniencephaly

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Introduction

Iniencephaly is a rare, usually lethal neural tube defect characterised by occipital bone abnormality, spinal dysraphism, fixed retroflexion of head and severe lordosis of cervico-thoracic spine¹. We report a case of iniencephaly associated with multiple congenital malformations.



Figure 1: Absent right thumb

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Case report

A live born female weighing 1850g was delivered at term to a 24-year-old primigravida, married to a non-consanguineous 28 year old partner. Baby cried weakly at birth and had an Apgar score of 7 at one minute. On examination, the baby had a posteriorly retracted head, facial dysmorphism, short neck, cleft palate and low set ears. There was sutural diastasis, retracted head, facial dysmorphism, short neck, cleft palate and low set ears. There was sutural diastasis, small thoracic cage and spina bifida aperta at the level of T5-T7. Right upper limb was hypoplastic with absence of the right pollux (Figure 1).

There was camptodactyly in the phalanges of the right upper limb. The left upper limb was normal. Right sided talipes was present. X-ray of the spine revealed fusion of cervical and upper thoracic vertebrae, scoliosis and a hemivertebra (Figure 2).



Figure 2: X-ray of spine. Hemivertebra shown by arrow

Ultrasonography of the abdomen revealed absence of the right kidney. Ultrasonography of the brain revealed presence of hydrocephalus and confirmed the encephalocele. Magnetic resonance imaging (MRI) of the brain revealed an occipital encephalocele with ventriculomegaly (Figure 3).

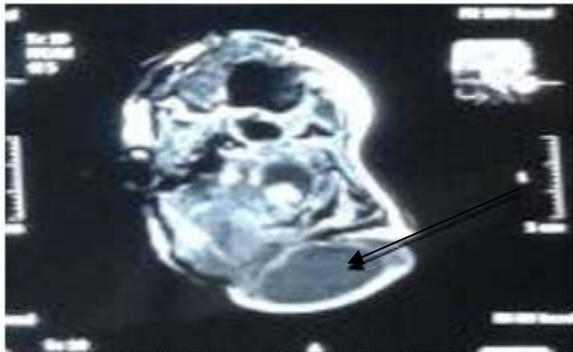


Figure 3: MRI of the brain. Encephalocele is shown by arrow

The baby was managed with supportive care for the first 2 days. Tube feeding was initiated with expressed breast milk; empirical antibiotics were started as patient was taken up for surgical removal of the encephalocele on day 3 of life. The surgery was uneventful; patient was taken off oxygen on day 4. The postoperative course was not significant and the baby was discharged at 15 days of life on nutritional supplements and spoon feeding. She was alive and well till the age of 2 month on follow up.

At 75 days postnatal age, the infant presented with acute respiratory difficulty and high grade fever. On admission the baby was tachypnoeic, with severe intercostal retractions. Chest radiograph showed bilateral bronchopneumonia. In spite of treatment with appropriate antibiotics and supportive care, the baby succumbed to this infection after two days of hospitalisation.

From the clinical features, a diagnosis of iniencephaly was made in this case. There was no family history of neural tube defects, drug ingestion or any illness during pregnancy. There is no history of folic acid supplementation in the first trimester.

Discussion

There are 2 types of iniencephaly. The severe group is iniencephaly apertus, involving the development of an encephalocele. In the other group, iniencephaly clausus, the encephalocele is absent². Iniencephaly has an incidence rate estimated at 0.1 to 10 in 10,000 deliveries. For unknown reasons, this disease seems to occur mostly in newborn females (about 90%)³.

Iniencephaly is found to be associated with other malformations such as anencephaly, encephalocele, hydrocephalus, agnathia, microstomia, cleft lip and palate, diaphragmatic hernia, pulmonary hypoplasia, cardiovascular disorders, renal abnormalities,

gastrointestinal atresia, genitourinary malformations, talipes, and overgrowth of arms as compared to the legs⁴. The most accurate method of diagnosis in prenatal screening is through real-time fetal images, although MRI and sonography are the most commonly used technique since there is no exposure to ionizing radiation⁵.

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