Surviving against the odds: exploring the clinical and radiological features of iniencephaly compatible with life. Illustrative case

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BACKGROUND Iniencephaly is a rare neural tube defect (NTD) characterized by deformities in the occiput and inion, along with rachischisis in the cervical and thoracic spine, resulting in the head appearing in retroflexion.

OBSERVATIONS This report details the case of a female newborn who underwent surgery for an encephalocele. She survived up to 6 months, exhibiting good overall health, although she displayed physical abnormalities, including facial deformity, a short neck, and minor spasms in all limbs. Both cardiovascular and abdominal assessments remained stable, and imaging revealed defects in the occipital bone, a large cephalocele, and spinal dysraphism.

LESSONS Although iniencephaly is generally incompatible with life, a few cases have been reported otherwise. Our patient, one of these notable exceptions, remains alive at 6 months old, possibly due to the lack of major vascular deformities. However, she does exhibit significant psychomotor retardation.

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KEYWORDS iniencephaly; neural tube defect; encephalocele

Iniencephaly is a rare congenital anomaly that affects the neural tube. This condition is defined by abnormalities in the occiput and inion, accompanied by rachischisis in the cervical and thoracic vertebrae, with the patient’s head often positioned in retroflexion. The occurrence rate of iniencephaly is remarkably low, estimated to be between 0.1 and 10 cases per 10,000 births. Unfortunately, the overall prognosis is generally poor. It can also be associated with a variety of conditions, including anencephaly, encephalocele, hydrocephalus, cyclopia, mandible absence, cleft lip and palate, cardiovascular diseases, diaphragmatic hernia, renal abnormalities, disproportionate arm growth compared to the legs, clubfoot, and gastrointestinal atresia.1–3 Eighty-two genes have been identified as potentially influencing the development of this disorder.4

Illustrative Case

A 1-day-old female was brought to our Neurosurgery Department after being born via caesarean delivery at 38 weeks’ gestation. At birth, she was in generally good condition, displaying spontaneous breathing with brief cyanosis that required temporary oxygen support but showing no significant respiratory distress. However, physical abnormalities were observed, such as swelling resembling an encephalocele in the occipital region, facial deformity, a markedly short neck, retrognathia, and stretched skin in the area around the mandible (Fig. 1). The infant also experienced minor spasms in all four limbs and had bilateral clubfeet. A protruding rib cage was noticeable.

A cardiovascular check showed stable circulation and no pathological murmurs. An abdominal ultrasound displayed normal kidney, gallbladder, and spleen functions. The cardiac echocardiogram revealed slight left-sided enlargement but with proper heart activity, indicating mild coronary insufficiency. Mild tricuspid inefficiency was noticed, and the pulmonary flow was normal. The hematological report also fell within the standard range.

ABBREVIATIONS CSF = cerebrospinal fluid; KFS = Klippel-Feil syndrome; MRI = magnetic resonance imaging; NTD = neural tube defect.

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The patient had successful surgery to correct her occipital deformity, which involved significant meningeal involvement. The surgery involved removal of the excess meninges and elimination of a small amount of abnormal neurological tissue connected to the cerebellum through cauterization. The dura was easily reconstructed, and the surplus skin was also removed. A 3-month postoperative check-up displayed superb outcomes with no new deformities. However, the patient still had severe psychomotor retardation.

Magnetic resonance imaging (MRI) scans (Figs. 2–4) showed a defect in the occipital bone at the foramen magnum, linked with a large cephalocele. Inside the cephalocele, there was a very small, underdeveloped cerebellum and a considerable amount of cerebrospinal fluid (CSF) encased by the meninges, measuring approximately 3.8 × 5.1 × 5.3 cm. The skull seemed proportionally large in relation to the spine, and the head was retroflexed. The posterior pituitary gland was larger than typical for the infant’s age, measuring 5 mm, and exhibited a normal pronounced increase in signal intensity on T1-weighted images. There were small foci in the right occipital lobe that displayed heightened signal intensity on T1-weighted images and assumed a gyriform shape on sagittal images. The myelination process seemed normal for the patient’s age, whereas the rest of the cerebrum appeared normal with no signs of space-occupying lesions or other irregularities. The occipital horns of the lateral ventricles were slightly enlarged, and the CSF spaces were normally unremarkable for the age group. C1 to C3 had substantial evidence of spina bifida (spine dysraphism), and minor spina bifida was possibly present at C6. Lordosis was apparent at the cervicothoracic vertebrae, whereas the upper part of the spinal cord appeared normal. The lungs appeared small on sagittal and coronal images.

**Patient Informed Consent**

The necessary patient informed consent was obtained in this study.

**Discussion**

**Observations**

Iniencephaly, a rare neural tube defect (NTD), has an estimated occurrence of 0.1 case in 10,000 pregnancies. The recurrence risk varies from 1% to 5%.5,6 Most of these uncommon cases are considered incompatible with life.3,7 with very few instances viewed as compatible with life.6–11 Our study presents a unique case in which the infant survived up to 6 months of age.

Iniencephaly is characterized by abnormal joining of the occiput and cervical spine, causing severe retroflexion of the head.5 The root cause of this condition is still unclear, but possible contributing factors include environmental influences like socioeconomic conditions, a lack of folic acid, obesity, and certain medications. There may also be a genetic component to iniencephaly, with conditions such as monosomy X, trisomy 13, trisomy 18, and chromosomal abnormalities linked to the NTDs associated with this condition.4,5 It is important to note that researchers have identified 82 human genes potentially linked to this genetic risk.4,5,12

Iniencephaly, a type of birth defect, begins to appear early in pregnancy before the 24th day of gestation, which is when the cephalic neural fold closes. The paraver tebral mesoderm, a layer of embryonic cells, differentiates, forming ventral masses that contribute to the creation of vertebral bodies, pedicles, and cranial homologs. Similarly, the dorsal masses contribute to the formation of neural arches and cranial vault bones. In cases of iniencephaly, one or both of these cell masses could be affected.3,5

The process of neural tube closure, which involves numerous initiation sites, has been suggested to occur at the midcervical level, between the prosencephalon and mesencephalon, at the stomodeum, at the caudal end of rhombencephalon, and at the caudal end of the neural tube. Failures in closures at positions 2, 4, and 1 could lead to the development of craniorachischisis.13,14

Iniencephaly is linked to a variety of systemic abnormalities.1,2,15–17 These include brain conditions such as holoprosencephaly, anencephaly, encephalocele, hydrocephalus, and Dandy-Walker malformations. Spinal issues may feature spina bifida or pronounced spinal retroflexion. Cardiac abnormalities such as septal defects can occur alongside gastrointestinal issues like omphalocele, diaphragmatic hernia, and intestinal or anal atresia. Other potential abnormalities include pulmonary hypoplasia, cleft palate, adrenal or thymic hypoplasia, and bronchogenic cysts. Genital malformations may be present, along with conditions like horse-shoe kidney and accessory spleen. There may also be differences in limb length, with overgrowth of the upper limbs in comparison to the

![FIG. 1. A: The typical morphological aspect of the face. B: The presence of an occipital encephalocoele.](image1)

![FIG. 2. Sagittal T2-weighted (A) and T1-weighted (B) MRI revealing a relatively large skull in relation to the spine, accompanied by retroflexion of the head. An occipital bone defect at the foramen magnum is evident, associated with a large cephalocele containing a very small hypoplastic cerebellum and a significant amount of CSF exhibiting increased signal intensity on T2-weighted imaging, all surrounded by meninges. Notably, there is enlargement of the posterior lobe of the pituitary gland, consistent with the infant’s age group, showing a normal marked increase in signal intensity on T1-weighted imaging. These features are indicative of iniencephaly apertus.](image2)
lower limbs, exaggerated spinal retroflexion, and Dandy-Walker malformations.

Clinical diagnosis depends on the absence of the occiput and noticeable neck retroflexion, which require differentiation from Klippel-Feil syndrome (KFS). Although KFS stems from abnormal segmentation or formation of cervical somites during embryological development, it does not involve occipital retroversion.4,6 It is critical to differentiate between iniencephaly apertus and anencephaly.18 Anencephaly is characterized by a partial or complete lack of the cranial vault, resulting in a head bent backward, left uncovered by skin, with normal neck (cervical) vertebrae. In contrast, iniencephaly results in a head bent backward and covered by skin but with abnormal neck vertebrae.

In the present study, we discuss a case involving a newborn diagnosed with iniencephaly at birth, who later had surgery for an encephalocele in our department. The infant had a minor systemic abnormality that presumably contributed to their survival. During our observation period, the child showed significant developmental concerns. At 6 months old, the child was still alive but exhibited significant psychomotor retardation.

Lessons

Iniencephaly is a rare NTD that often leads to obstructed labor and needs meticulous obstetric management. There is a recurrence risk of 1% to 5% in future pregnancies, underscoring the importance of preconception counseling. This paper discusses an unusual case to
illustrate that noncomplicated iniencephaly is not invariably fatal. Timely prenatal diagnosis is vital for the planning of postnatal surgical procedures. Accurate use of MRI and ultrasonography is crucial to determine the necessity of pregnancy termination. The combined use of ultrasound to detect cardiac malformations and MRI to detect brain and spine abnormalities allows thorough evaluation of intricate neural tube malformations. All the diagnoses align closely with actual pathology.

References


Disclosures

The authors report no conflict of interest concerning the materials or methods used in this study or the findings specified in this paper.

Author Contributions

Conception and design: H Kadri, Shehadeh Agha, Bakleh. Acquisition of data: T Kadri. Analysis and interpretation of data: Dughly, H Kadri. Drafting the article: H Kadri, Dughly. Critically revising the article: H Kadri. Reviewed submitted version of manuscript: H Kadri, Shehadeh Agha. Approved the final version of the manuscript on behalf of all authors: H Kadri. Administrative/technical/material support: Mackieh. Study supervision: Abouharb.

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