

Iniencephaly: neuroradiological and surgical features

Case report and review of the literature

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✓ Iniencephaly is a rare congenital anomaly characterized by spina bifida of the cervical vertebrae, fixed retroflexion of the head on the cervical spine, and occipital bone defect. There are only five reports of surviving patients with iniencephaly. The authors report the case of a newborn who presented with iniencephaly and an encephalocele that were surgically treated in our service. Neurological examination of the patient yielded normal results except for a moderate psychomotor retardation. The neuroradiological and surgical findings of the case suggested that the trigger of the anomaly was the occipital bone defect and rachischisis of the posterior vertebral arches.

KEY WORDS • iniencephaly • congenital abnormality • neural tube defect

THE term “iniencephalus,” originating from the Greek word “inion,” is used to describe an anomaly that involves both the brain and the neck.⁷ Iniencephalus has three cardinal characteristics: 1) occipital bone defect; 2) spina bifida of the cervical vertebrae; and 3) fixed retroflexion of the head on the cervical spine.⁹ There are only five reported cases of surviving patients with iniencephaly; the remaining cases were identified from autopsy findings.^{3,4,6,9}

The present report includes the complete neuroradiological workup and surgical findings in a case of iniencephaly that had 3 years of follow-up review.

Case Report

History. The mother of the patient was a 26-year-old woman who was pregnant for the first time and had visited the obstetrical–gynecological clinic for prenatal care only once at the 8th month of gestation. An ultrasound examination performed at that time revealed a huge occipitocervical mass in the fetus that was diagnosed as an occipital encephalocele. The woman had no history of substance abuse, and there was no significant individual or family medical history.

Examination. The male infant was delivered at 34 weeks of gestation by Cesarean section; he had an Apgar score of 7. The infant weighed 3360 g and was 51 cm in length. He had a short, broad neck, and his head was fixed in the retroflexion position. An occipital mass over the cranio-cervical junction was covered by intact skin with hemangiomas (Fig. 1 *upper*). The occipital bone defect was palpable at the borders of the occipital encephalocele.

The transillumination test was negative. No motor deficit was detected. The anterior fontanel was normal. Magnetic resonance (MR) imaging was performed when the patient was 5 days of age; the image revealed the absence of the posterior arches of the upper cervical vertebrae and an occipital bone defect. An encephalocele protruded from the occipital defect and widely open cervical spina bifida. The configuration of the cerebellum, vermis, occipital lobes, brainstem, and fourth ventricle was highly disturbed (Fig. 2 *left*). No associated congenital anomaly of other organ systems was found on systemic evaluation.

The infant was discharged home 6 days after birth. He had periodic medical examinations and displayed good physical development at 13 months of age. Magnetic resonance imaging was performed at that time and showed accentuated deformities at the brainstem with an important increase in the pontomamillary distance when compared with previous MR images. Areas of cerebrospinal fluid (CSF) accumulation appeared in the encephalocele (Fig. 2 *right*). Our impression was that there was a continuous and progressive posterior displacement of the brain through the large opening at the craniocervical junction. A three-dimensional computerized tomography (CT) scan demonstrated defective development of the endochondral portion of the occipital bone and rachischisis of the cervical vertebrae (Fig. 3). The remainder of the calvaria was normally developed and the calvarial sutures were partially synostosed. On cerebral angiography, cerebellar and occipital vessels were found in the encephalocele, and the transverse sinuses were located at the occipital bone border (Fig. 4). Although the brainstem auditory evoked responses were normal, it was decided that the infant should



FIG. 1. *Upper:* Preoperative photograph of the patient. *Lower:* Latest available photograph of the patient obtained when he was 3 years of age.

undergo surgery to avoid additional posterior displacement of the brainstem and to reduce the size of the encephalocele.

Operation. At the first stage of the operation, to create a space for intracranial growth of the brain, we established a “floating forehead” and bilateral parasagittal linear craniectomies. Two weeks later, we proceeded with the repair of the encephalocele. With the patient in the prone posi-

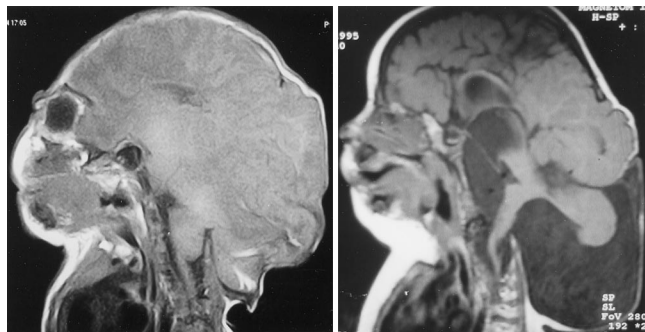


FIG. 2. *Left:* Midsagittal T₁-weighted MR image obtained when the patient was 5 days old showing the occipitocervical encephalocele with a significant deformation of posterior fossa structures protruding into the encephalocele. The medulla oblongata and cerebellar vermis are in the cervical canal. *Right:* Magnetic resonance image of the same area shown at the *left* obtained when the patient was 13 months of age. Further displacement of the brainstem posteriorly and partial filling of the cyst by CSF can be seen.



FIG. 3. Three-dimensional CT scan revealing rachischisis of the cervical vertebrae and defective development of the endochondral portion of the occipital bone.

tion, we made a median vertical skin incision along the entire encephalocele and performed dissection until the encephalocele was entirely exposed. The dura was intact but densely adherent to the subgalea. The transverse sinuses were located under the occipital bone border with the encephalocele as seen on cerebral angiography. The dura was opened the same way as the skin incision. A significant amount of CSF was discharged. All neural tissues were covered by arachnoid. We observed that the normal configuration of the cerebellar hemispheres, vermis, and the fourth ventricle was extremely distorted and the occipital lobes were extended into the encephalocele. The floor of the fourth ventricle and the proximal cervical spinal cord were easily seen with minimal upward retraction of the cerebellar hemispheres (Fig. 5). All neural tissues were apparently functional, and we performed a reduction duraplasty by folding over the dural edge as one would form an envelope. The encephalocele mass was thus reduced, and a solid posterior support, formed by the double



FIG. 4. Cerebral angiogram demonstrating cerebellar and occipital vessels coursing within the encephalocele.

Iniencephaly

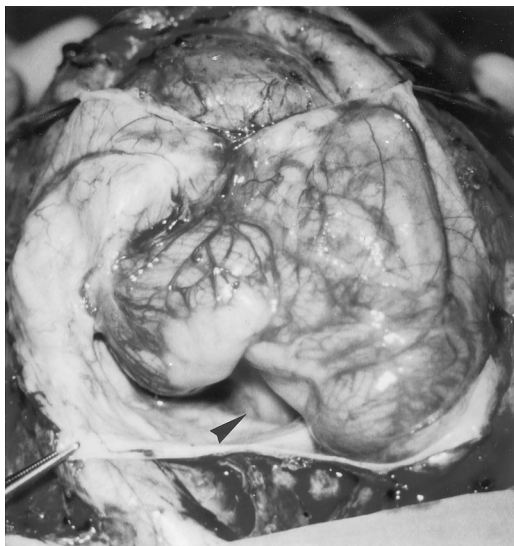


FIG. 5. Intraoperative photograph demonstrating the disturbed configuration of the cerebellar hemispheres, vermis, and fourth ventricle. The occipital lobes lie partially in the encephalocele. The floor of the fourth ventricle and the proximal spinal cord are seen deep in the operative view (*arrowhead*).



FIG. 6. Magnetic resonance image obtained 14 months postoperatively revealing no further displacement of the brainstem. The encephalocele is markedly reduced in size, and there is no CSF accumulation.

layers of dura, was created. The skin was also reduced in size.

Postoperative Course. Postoperatively, the patient suffered from severe respiratory distress and a third nerve paralysis on the right side. He was maintained on a ventilator for 10 days. A control CT scan demonstrated enlargement of the lateral and third ventricles, and a ventriculoperitoneal (VP) shunt was placed. Several days after shunt placement, we removed the ventilatory support and the third nerve paralysis improved gradually. Fourteen months postoperatively, no further increase in pontomamillary distance was found on control MR imaging (Fig. 6). The size of the encephalocele was markedly decreased and there was no CSF accumulation. The results of control brainstem auditory evoked potentials and visual evoked potentials were normal. The child, now 3 years of age, still holds his head in the retroflexion posture but has a smaller occipital encephalocele and markedly improved neck motion, probably because of the smaller encephalocele (Fig. 1 *lower*). His neurodevelopmental status is moderately retarded. He is able to stand up and walk with minimal assistance, and he can also speak several words.

Discussion

Review of the Literature

As in other cases of neural tube defect, there is no identified cause of iniencephaly. In five cases of iniencephaly with anencephaly reported by Rodriguez, et al.,⁸ the only common denominator was a Hispanic mother aged in her 20s or early 30s. Katz, et al.,⁴ classified cases according to those with an encephalocele (*apertus*) and those without (*clausus*). Sherk and associates⁹ categorized those with a large encephalocele protruding from a widely open for-

men magnum and cervical spina bifida in the most severe forms, such as in our case. Associated anomalies of the cerebrum include anencephaly, hydrocephaly, cyclopia, microcephaly, polymicrogyria, holoprosencephaly, and atresia of the ventricular system.¹ Although we did not detect evidence in our case, iniencephaly may be associated with anomalies of other organ systems such as the urinary tract, cardiovascular, skeletal, and gastrointestinal systems.² Undoubtedly, it is the severity of associated anomalies that contributes to the compatibility with life or to living standards. One of the patients described by Sherk and associates and the patient discussed by Munden, et al.,⁶ were both 17-year-old boys who had no intellectual problems. Table 1 provides a summary of the clinical data of reported surviving patients with iniencephaly.

Developmental Pathogenesis

The developmental pathogenesis of the various axial dysraphic disorders is unresolved. Among the many theories that have been proposed, the majority have been used to try to explain the pathogenesis from the standpoint of primary neural anomaly.¹⁰ Marin-Padilla and Marin-Padilla⁵ suggested a primary mesodermal deficiency as the cause of all neural developmental abnormalities. In our case, we infer from the surgical and neuroradiological findings that the formation of neural structures as well as vascular arborization were complete. The trigger for the anomaly was only the occipital bone defect and rachischisis of the posterior vertebral arches. The remainder of the anomaly was a simple herniation of neural tissue through this posterior bone opening that occurred during the gestational period. The morphological appearance of iniencephaly may resemble a large occipital encephalocele. The huge occipital bone defect and rachischisis of the cervical vertebrae are the main features that distinguish iniencephaly from a large occipital encephalocele.

TABLE 1
*Clinical profiles of five surviving patients with iniencephaly**

Authors & Year	Age, Sex	Neurological Exam	Radiological Findings	Associated Abnormalities
Sherk, et al., 1974	17 yrs, M	normal	enlarged foramen magnum, cervical rachischisis, cerebellar tonsils & medulla extending into cervical canal on myelography, bilat scapular elevation, fusion defects of VBs	Klippel-Feil syndrome, bilat Sprengel's deformity
	16 mos, F	normal	enlarged foramen magnum, cervical rachischisis, bilat scapular elevation, fusion defects of VBs	Klippel-Feil syndrome, Sprengel's deformity
	3 yrs, F	lt CN VI & VII palsy, mild bilat conductive hearing loss	cervical rachischisis, extremely elongated fourth ventricle extending down into thoracic spine on myelography, bilat scapular elevation, fusion defects of VBs	Klippel-Feil syndrome, Sprengel's deformity
Katz, et al., 1989 & Gartman, et al., 1991	21 mos, M	normal	cervical rachischisis between C-1 & T-4, cerebellum extending into cervical canal & agenesis of corpus callosum on MR imaging	pyloric stenosis, agenesis of corpus callosum
Munden, et al., 1993	17 yrs, M	normal	complete fusion of cervical & upper thoracic VBs, enlarged foramen magnum, posterior defects of C1-3, fusion of posterior arches of lower cervical & upper thoracic spines, bilat scapular elevation	Klippel-Feil syndrome, Sprengel's deformity
present study	3 yrs, M	normal	cervical rachischisis, occipital defect, bilat scapular elevation, cerebellum, medulla & occipital lobes protruding into occipital encephalocele on MR imaging	Sprengel's deformity

* CN = cranial nerve; VBs = vertebral bodies.

Surgical Treatment for Iniencephaly

Although the posterior extracranial herniation of the brain did not alter the normal function of the brainstem, the progressive increase in the pontomamillary distance was the main indication for surgery. Among the previously reported cases, we have found only one case that was treated surgically.^{3,4} The authors of that case report believed that many thickened fibrous bands connecting the occiput to the posterior aspect of the cervical spine contributed to the retroflexion deformity. They suggested that these bands represented fibrosis and underdevelopment of the trapezius muscle. They improved the retroflexion deformity by untethering the fibrous bands.³ That case differs from our own by the absence of occipital bone defect and encephalocele. Also, we did not observe intraoperatively any fibrosis that contributed to the retroflexion deformity. We think that the primary event that caused the characteristic retroflexion deformity was the disturbed configuration of the craniocervical junction. The progressive extracranial growth of the posterior fossa component into the huge encephalocele probably favored the retroflexion deformity. Our initial surgical strategy was to create space for intracranial growth of the brain by the "floating forehead" and parasagittal craniectomies; at the second stage of the operation, we reduced the size of the encephalocele by reduction duraplasty. Double layers of dura were formed to provide a solid posterior support so that further herniation of the brain could be avoided. The cause of the respiratory stress and the third nerve palsy was undoubtedly mechanical compression of the brainstem as a result of the reduction duraplasty. We think that the VP shunt placement contributed significantly to the lack of CSF refilling within the encephalocele. As seen in the postoperative MR studies, an important reduction in encephalocele size was provided and the pontomamillary distance remained stable.

Conclusions

The benefits our patient received from three consecu-

tive surgical procedures were a considerable reduction in the size of the encephalocele and stabilization of the brain herniation through the occipitocervical defect. It is not known whether the same benefits could be achieved if treatment consisted simply of early VP shunting. It is difficult to predict what problems this patient will develop in the future. This is a rare example of a surviving patient with iniencephaly.

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