Deformity correction and long-term survival in an infant with iniencephaly

Case report

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The authors describe the case of a male infant who was diagnosed prenatally as having iniencephaly. Since birth, the child has grown, thrived, and undergone two successful operations to correct his cervical deformity. This case demonstrates that the iniencephaly defect is not uniformly fatal and that neurosurgical intervention may offer significant improvement in the cervical deformity.

Key Words: iniencephaly · congenital malformation · rachischisis · myocutaneous flap

Iniencephaly is an uncommon congenital malformation characterized by imperfect skull base formation, a variable degree of rachischisis, and retroflexion of the head on the cervical spine. The prognosis for such fetuses and infants has remained dismal with only three previously documented long-term survivors. Because of the high incidence of stillbirth and death in the neonatal period, many iniencephalic fetuses diagnosed in the prenatal period are terminated.

The iniencephaly abnormality involves a range of malformations from the more benign (as demonstrated by the patient presented here) to the severe, including associated anencephaly and open neural-tube defects. The increasing capacity for detailed prenatal diagnosis allows for a more accurate in utero description of the associated malformations. We describe the survival of an iniencephalic infant and the successful partial correction of his retroflexion deformity. We conclude that iniencephalic fetuses with less severe associated anomalies may be carried to term and may be restored to a more functional state than was previously thought possible, using surgical procedures similar to that utilized in this patient.

Case Report

This baby boy, now 21 months old, was delivered to a 35-year-old white woman, and was a member of a twin gestation. The mother had previously experienced two failed pregnancies; however, there was no family history for congenital anomalies or consanguinity.

Prenatal Course. At 17 weeks of gestation, an ultrasound examination demonstrated the other twin to be structurally normal, while our patient was noted to have a poorly defined malformation of the skull and cervical spine. Amniocentesis was performed on each of two apparently isolated amniotic sacs. Giemsa-banded chromosome analysis of cultured cells demonstrated the other fetus to be a cytogenetically normal female and our patient to be a cytogenetically normal male. Acetylcholinesterase determinations were negative on both amnion fluid samples. Radioimmunoassay for alpha-fetoprotein levels was 10.0 µg/ml for the female and 7.0 µg/ml for our patient.

Repeat ultrasound examination at 20 weeks of gestation demonstrated appropriate growth and amniotic fluid volume for both fetuses. At this time, examination of our patient clearly revealed retroflexion of the head with spinal rachischisis in the cervical region. Mild lateral ventricular dilatation with an apparently normal third ventricle was also demonstrated. The remaining anatomy was unremarkable. An in utero diagnosis of iniencephaly was made and the prognosis discussed with the parents. Partly because of the other normal fetus, the pregnancy was carried to term.

Neonatal Findings. The twins were delivered at 36 weeks of gestation by a Caesarean section following
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spontaneous rupture of membranes. The female twin was normal and followed an uncomplicated course. Our patient had a short broad neck with the head held in a fixed retroflexed position (Fig. 1). There was no open neural tube defect. Radiographic analysis showed pedicle spreading and absence of laminar arches from C-1 to T-4 (Fig. 2). Despite mild hydrocephalus, the patient’s head circumference (35.75 cm) was within normal limits for his gestational age. Cardiovascular and renal examinations were also normal.

The patient was fed by an orogastric tube and kept in an oxygen hood for 3 days. We were uncertain at that time whether the child would survive based upon previously reported cases. He was discharged home at the age of 5 days with plans for further follow-up procedures.

A workup for persistent vomiting at 6 weeks of age demonstrated hypertrophic pyloric stenosis; an uncomplicated pyloromyotomy was subsequently performed. At 12 weeks of age magnetic resonance (MR) images of the head and spine were obtained which demonstrated agenesis of the corpus callosum (Fig. 3). There was no evidence of tethering of the spinal cord.

Operation. The child continued to thrive and we proceeded with repair of the flexion deformity at 14 weeks of age. A transverse incision was made, centered over the junction of the inion with the upper thoracic spine and extending laterally to the base of each mastoid process (Fig. 4A). The subcutaneous fat contained many thick fibrous bands which appeared to contribute to the retroflexion deformity. These fibrous bands, as well as a thickened nuchal ligament, were sectioned. Dissection was carried out laterally along the inferior occiput and lower cervical region to release several tethering fibrous bands that were believed to represent fibrosis and underdevelopment of the trapezius muscles. Further dissection around the enlarged foramen magnum revealed a thickened tectorial membrane.

The tectorial membrane and underlying dura were opened with a 3-cm horizontal incision centered in the midline (Fig. 4B). Subarachnoid exploration demonstrated an inferiorly positioned, sclerotic-appearing vermis which had flattened the cervical spinal cord proximally. The foramen magnum was enlarged 1.5 cm posteriorly to expose a dural edge for suturing a dural graft.

The patient’s head was then flexed forward to a nearly neutral position, rolling the vermis and inferior cerebellar hemispheres up and off the cervical cord and creating an approximate 4-cm vertical dural opening (Fig. 4C). A pericranial graft harvested from the occipi-
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Fig. 4. Schematic representation of the patient's first operative procedure. A: Dorsal view of the initial transverse skin incision extending from mastoid to mastoid just inferior to the inion. B: Dorsal view after sectioning of the thickened nuchal ligament (fused to dorsal dura) and vertically directed fibrous bands. The operative exposure demonstrates the caudally displaced cerebellar tonsils and vermis. C: Lateral view illustrating intraoperative neck flexion enlarging the cervical soft-tissue defect. D: Dorsal view following repositioning of the head with a pericranial graft placed to augment cervical dura. E: Dorsal view illustrating rotation of the trapezius myocutaneous flap to close the cervical defect. F: Dorsal view showing the final appearance of the skin closure.

Fig. 5. Magnetic resonance image after the first surgical procedure demonstrating decompression of the brainstem with generous dorsal and cerebrospinal fluid space now present as well as restoration of the sagittal midline.

ital bones was used to create a watertight dural closure over the defect created by flexion (Fig. 4D). A single large trapezius myocutaneous flap was used to close the underlying skin defect (Fig. 4E and F).

Postoperative Course. Postoperatively, the patient was maintained in a soft cervical collar contoured to maintain the flexed position that had been achieved during surgery. At 7 months of age, MR imaging demonstrated partial correction of the flexion deformity and relief of brainstem compression (Fig. 5).

At 1 year of age, disproportionate head growth coupled with increasing size of the ventricles necessitated ventriculoperitoneal shunt placement. Four months later, a lateral release of the investing fascia of the right levator scapulae muscle and subcutaneous fibrous bands tethering the right mastoid bone to the right scapula was combined with creation of a pectoral myocutaneous flap to straighten the patient's head further and to increase the height of the neck.
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The patient, now 21 months of age, continues to thrive with a stable head and neck position, although neck movements are significantly limited (Fig. 6). Developmentally, he is functioning between a 9- and 11-month level as measured by a recent Bayley Infant Development and Mental Scale test. This test may underestimate his true abilities since limited vision inferiorly due to restricted cervical flexion and limited dexterity of the upper extremities interfere with adequate testing and further diminish his already reduced performance. Only when useful expressive speech develops will a more accurate assessment of his mental development be possible.

**Discussion**

**Historical Review**

Iniencephaly was first described in 1887 by Lewis. Since then the diagnosis has become well recognized, with an incidence ranging from 1:1000 to less than 1:100,000. The features present in all cases include skull base defects, rachischisis, and retroflexion of the head. Although the severity of deformity covers a broad spectrum, the vast majority of cases have resulted in fetal death or stillbirth. Only three cases of long-term survival have been documented. We have described an infant with iniencephaly who has thrived for over 1½ years and has undergone significant surgical correction of the retrofexion and lateral flexion deformity.

**Associated Abnormalities**

Iniencephaly may be associated with additional neurological deformities as well as anomalies of other organ systems. Generally, the malformation is divided into two types: iniencephalus clausus referring to cases without an associated encephalocoele, and iniencephalus apertus describing cases with an encephalocoele in the region of the foramen magnum associated with an occipital bone defect. The occipital mass does not always contain nervous tissue. Aleksic and Budzilovich presented a neuropathological study of iniencephaly and associated central nervous system malformations including anencephaly, hydrocephaly, cyclopia, and encephalocoele. Associated anomalies of the cerebrum include microcephaly, polymicrogyria, holoprosencephaly, and atresia of the ventricular system. These authors also described widespread lack of cortical laminar organization. The spectrum of cerebellar and brain-stem abnormalities includes lesser degrees of dysplasia and/or hypoplasia, as appeared to be the case in our patient. Cerebellar and brain stem structures, when present, may be found within the occipital encephalocoele.

Rachischisis is a common disorder in iniencephaly. The vertebral column may be reduced in number or fused at various locations. Often cleavage is present at the posterior aspect of the occiput, with an enlarged foramen magnum which may extend as a posterior vertebral defect through the cervical region and into the thorax. The pedicles are shortened and laminar arches are absent. The extent of cutaneous covering over this defect may range from complete exposure of the neural elements to closure, as was found in our patient. Fortunately, this aspect of the anatomy is reliably visualized on in utero ultrasound examination and probably contributes significantly to the ultimate prognosis.

In a review of 50 cases of iniencephaly, David and Nixon found that 85% of patients had other significant malformations. The urinary tract was most often coincidentally involved, followed by the cardiovascular and gastrointestinal systems. The etiology of these multi-system abnormalities remains unclear. Gardner and Brever proposed that all of the anomalies in those patients may be related to the initial defect in the occipitocervical region. The extremely lordotic spine may restrict development of the abdominal contents and contribute to the development of the coexisting malformations.

**Neurosurgical Intervention**

The surgical repair of the retrofexion deformity in our patient included several simple but important principles. Multiple thick fibrous bands in the occipitocervical region were found to be largely responsible for the fixed retroflexed position of the head. Identification and sectioning of these structures were essential for achieving flexion. Repositioning of the head into the flexed position was cautiously performed under direct visualization of the cerebellum and brain stem. The forward movement of the head into flexion did not produce symptoms of brain-stem or cord dysfunction despite the low position of inferior brain-stem and

**Fig. 6.** Photograph of the patient at 21 months of age.
cerebellar structures. Neither arachnoidal adhesions nor gliosis and scarring of the cerebellar tonsils were present. Dural augmentation at the occipitocervical junction was necessary to cover the large posterior dural defect created by forward movement of the head into the flexed position. Finally, use of the large myocutaneous flap, which was rotated to cover the defect produced by cervical flexion, requires strict attention to both hemostasis and preservation of the vascular supply to the flap. The bulk of the flap provides some protection for the exposed dura.

The case described here demonstrates that the iniencephalic defect is not uniformly fatal. Furthermore, when these pregnancies are brought to term, neurosurgical intervention may offer significant improvement in the deformity, especially in its more benign form. With advanced prenatal diagnostic capabilities, the ability to detect and delineate the extent of the iniencephalic defect is enhanced. Better prenatal counseling can be provided to parents, and the possibility of surgical correction offers hope for more normal and productive lives for some of these patients.

References


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