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Amnion Rupture Sequence with 'Exencephaly': MR Findings in a Surviving Infant

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Summary: In an infant who survived the amnion rupture sequence, MR findings included a large defect in the cranial vault, a Chiari II malformation, rotation of the hemispheres relative to the skull base, and a cleft lip and palate.

Index terms: Exencephaly; Brain, herniation; Fetus, abnormalities and anomalies

The amnion rupture sequence is a heterogeneous group of congenital deformities in which rupture of the amnion occurs early in gestation. Fragments of the amnion may adhere to, or encircle and constrict, embryonic parts. This may interrupt normal morphogenesis and cause deformity or even mutilation of formed structures; large cranial defects are a common feature. In contrast to other causes of cranial defects, the amnion rupture sequence carries no increased risk of recurrence in subsequent pregnancies.

Case Report

A 15-week-old male infant with craniofacial dysmorphism was referred for magnetic resonance (MR) evaluation of the cranial contents as part of an assessment for reconstructive surgery.

An antenatal ultrasound performed elsewhere at 28 weeks' gestation, because of excessive maternal weight gain, had been interpreted as showing fetal exencephaly. Subsequently, chorioamnionitis developed, and the fetus was delivered by cesarean section at 34 weeks. At birth, absence of the cranial vault above the supraorbital ridges was noted, but the scalp was intact. The orbits were asymmetrical, with hypertelorism. On the left, the eyelids were absent, and a cataract was noted; there was no left pupillary reaction. The right ear was abnormally low-set. There was bilateral clefting of the lip and palate (Tessier types 3 and 11), more severe on the left, where there was an associated band of scar tissue passing from the labial cleft to the region of the medial canthus. There was no apparent visceral malformation and no clinical evidence of

spinal dysraphism. Cardiac and renal ultrasound performed elsewhere were said to be normal. Examination of the limbs revealed syndactyly of the right middle and ring fingers, a constriction band around the distal middle finger on the left, and syndactyly of the left first to fourth toes.

At 6 weeks after delivery, the parents had noted an episode of limb jerking, possibly representing seizure activity; an electroencephalogram subsequently confirmed seizure activity.

Plain films (not shown) confirmed the extensive cranial defect; part of the occipital bone was present, closing the foramen magnum posteriorly.

MR of the head showed abnormal orientation of the intracranial structures relative to the craniofacial bones. The cerebral hemispheres were deformed and asymmetric, with the interhemispheric fissure lying almost perpendicular to the midfacial plane (Figs 1A and 1B). Extracranially, bilateral palatal clefts were present (Fig 1C).

Intracranially, the right cerebral hemisphere lay predominantly anteriorly and inferiorly within the cranial cavity, whereas the (left) hemisphere overlapped it posteriorly and superiorly. Only a few hemispheric sulci were seen, and several abnormal fissures were noted. The corpus callosum was largely absent, only a few fibers being identified anteriorly. The lateral ventricles were deformed but present (Fig 1B); the third ventricle was small and difficult to identify. The hemispheres, especially the left, protruded through the cranial defect, surrounded by cerebrospinal fluid; the meninges seemed fused to the skin superiorly, with no intervening subcutaneous fat. The cerebellum lay exclusively to the left of the craniofacial midplane and showed no definite hemisphere formation; no definite vermis was identified. The cerebellar midplane was perpendicular to that of the face. There was gross angulation of the tentorium, secondary to the asymmetry of the posterior fossa structures (Fig 1D). Sagittal images demonstrated an extensive Chiari malformation, with the tip of the cerebellum lying at the C5–6 level, and a thin tubular fourth ventricle (Fig 1E).

The patient returned 3 weeks later with a sudden increase in head size. Repeat MR demonstrated gross dilatation of the lateral and third ventricles, consistent with aqueduct obstruction (Fig 1F). Several abnormal diverticula

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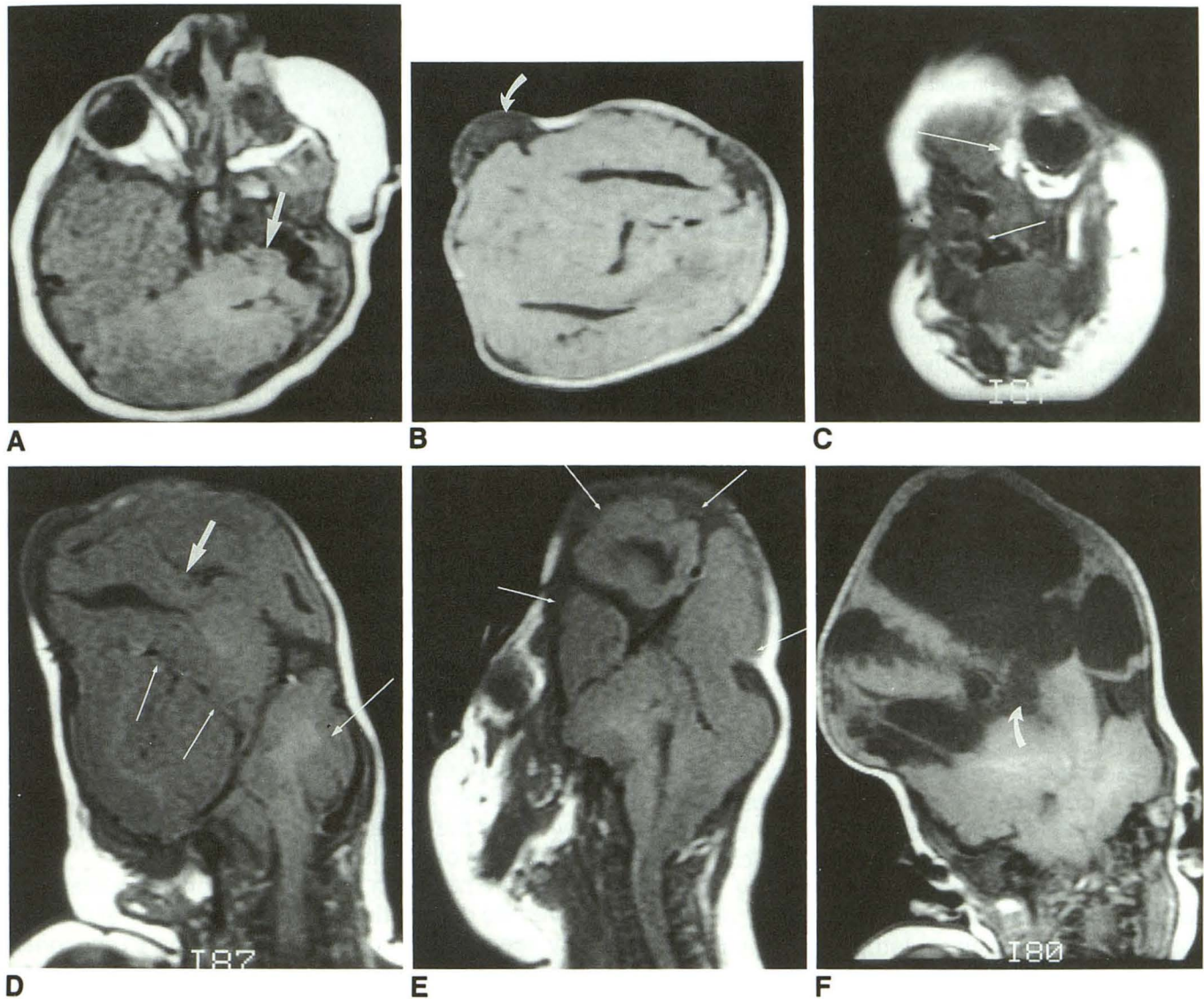


Fig. 1. A and B, Axial T1-weighted (450/20/1 [repetition time/echo time/excitations]) images of the head at the levels of the nose and right eye (A) and the lateral ventricles (B), showing the rotation of the cerebral hemispheres relative to the facial structures. Deformity of the nares on the left is seen in A. Note partial absence of cranial vault in B (curved arrow), with the skin fused to the protruding meninges, and transverse orientation of the brain stem (A, arrow).

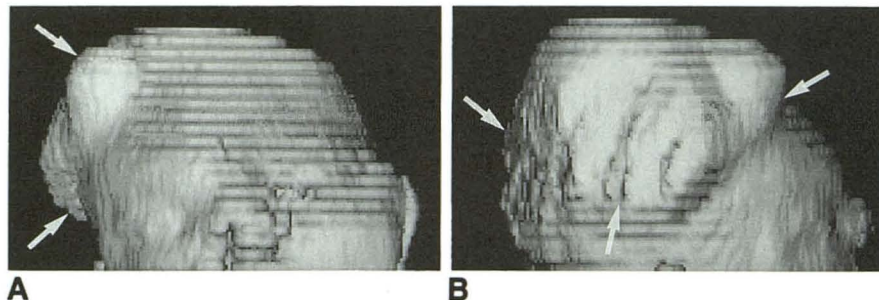
C, Coronal T1-weighted (400/20/1) image of the face anteriorly demonstrates bilateral palatal clefting (small arrow). The bony walls of the left orbit are malformed, with orbital fat lying immediately adjacent to the left cerebral hemisphere (large arrow).

D, Coronal T1-weighted (400/20/1) image of the head at the level of the foramen magnum shows a rudimentary cerebellum lying to the left of the brain stem (arrow); the cerebellar midplane is parallel to the coronal imaging plane. The cerebral hemispheres are seen in cross-section, the left hemisphere lying more superiorly and overlapping the right. There is angulation of the interhemispheric fissure (small arrows) relative to the sagittal plane, and of the tentorium relative to the transverse plane. A deformed left lateral ventricle is visible (thick arrow).

E, Sagittal T1-weighted (600/20/1) image of the head at the level of the foramen magnum shows an extensive Chiari malformation, with the cerebellar vermis extending down to the C5–6 disk level. There is a thin, elongated fourth ventricle. The extent of the cranial defect is apparent (arrows). Rotation of the cerebral hemispheres and angulation of the interhemispheric fissure are again seen.

F, Coronal T1-weighted (500/20/1) image of the head, immediately anterior to the foramen magnum, at the second presentation of the patient, demonstrates massive lateral ventricular dilatation, more marked on the left. An irregular area, centrally within the head and isointense with cerebrospinal fluid, may represent a deformed and dilated third ventricle (curved arrow). Protrusion of the hemispheres through the cranial defect is now more prominent; the fused meningeal and cutaneous covering of the protruding brain remains intact.

Fig. 2. Three-dimensional reformation images of the patient's face and head from the spin-echo images. The intersection gaps create *stripes* on the images. The bulging brain (*arrows*) can be appreciated from straight anteroposterior projection (A) and right posteriorlateral projection (B). Deformed facial structures can be appreciated.



were seen to arise from the dilated and deformed lateral ventricles. Protrusion of the hemispheres through the cranial defect was now more marked.

Computed tomographic ventriculography, performed the day before ventricular shunting, demonstrated intercommunication between the lateral ventricles, their diverticula, and the third ventricle (Fig 2).

Subsequently, the labial clefts were repaired and the band of scar tissue on the left side of the face excised. An assessment of speech and language development at the age of 7 months was most consistent with a developmental age of 4 to 5 months.

Discussion

The amnion rupture sequence (or "amniotic band syndrome") is a heterogeneous group of congenital defects believed to be the result of amnion rupture during early embryonic life. Damage to the embryo may be caused by adhesion of part of the amnion to the embryo, disruption of the embryonic vasculature, or constriction of part of the embryo by a band of amniotic tissue (1). The prevalence of amnion rupture sequence at birth is 1.16 per 10 000 live births (2). The anatomic pattern of the defect or defects is highly variable but is thought by some to be partly dependent on the time of amnion rupture (3). The principle features are limb-reduction defects, craniofacial clefts and defects, and thoracoabdominal defects. Seven subgroups of the sequence were identified by Ossipoff and Hall (4), based on the seven possible combinations of one or more of the three principle features. A majority of the reported cases with cranial or visceral defects have been described from fetal or neonatal autopsy material.

The salient findings in this patient—a large cranial vault defect, palatal and facial clefting, syndactylies, and constriction rings—are typical of the amnion rupture sequence group II (4). In this case, the palatal and labial clefting was of a type commonly seen in other conditions; more

bizarre clefts may be seen in the amnion rupture sequence.

In many of the reported cases of these defects, part of the amnion (sometimes much of the placenta as well) has been found to be adherent to the skin of the head, in the region of the defect, or to an associated encephalocele. It is uncertain whether the amniotic adhesion causes the defect or whether the amnion attaches to tissue already damaged by a vascular insult (1). Similar defects, without persistence of adherent amnion, previously have been described (1, 5) in the amnion rupture sequence.

In the present case, the malalignment of the cranial contents relative to the craniofacial skeleton suggests that the cerebral structures may have been anchored by an amniotic adhesion, whereas the remainder of the embryo rotated 90° about its long axis.

There has been some confusion in the literature over the description of the craniocerebral deformity seen in this condition, the terms *exencephaly* and *encephalocele* having both been used. An encephalocele can be defined as an extracranial herniation of brain tissue through a defect in the cranium. In the present case, there is a large cranial defect, with brain lying within, and extending somewhat beyond, this defect; however, the brain and meninges retain approximately normal shapes and anatomic relationships (allowing for their misorientation), with no true extension of tissue through the cranial defect into a discrete hernial sac. Perhaps this should be described as an "encephalocele in situ."

In their recent review of cephaloceles, Naidich et al (6) included the cranial defects of the amnion rupture sequence in their description of *exencephaly*, which was defined as acrania with persistence of a substantial part of the central nervous system, the central nervous system remnant protruding into the amniotic cavity. The extent of the cranial defect in the illustrative case seems similar to that in the present case.

However, there are two important differences between the present case and exencephaly as usually described. First, relatively normal (though distorted and malformed) hemispheres were present, in contrast to the grossly dysplastic and often degenerate neural tissue found in true exencephaly. More severe cerebral disorganization has been reported in the amnion rupture syndrome (7). Second, the amnion rupture sequence is believed to be a sporadic disorder, with no increased risk of recurrence in a subsequent pregnancy (8). In contrast, exencephaly is thought to be part of the spectrum of neural tube defects (9, 10), in which there is a significantly increased risk of recurrence (2–5% [11]). Hence, the distinction between these conditions may be of considerable importance for counseling of the affected parents. Antenatal sonographic features distinguishing the amnion rupture sequence from the exencephaly/anencephaly sequence have been described and the importance of the distinction emphasized (12).

Because of the normal volume of brain tissue (at least in some cases), and the low risk of recurrence, the cranial defects seen in amnion rupture sequence should be distinguished from those seen in other causes of exencephaly. Multiplanar MR of surviving affected neonates may be very helpful in defining the neural structures present and facilitating this distinction. It is as yet uncertain how helpful MR assessment will be in

determining the neurologic prognosis of these severely affected infants.

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