Acalvaria

Case report

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Acalvaria is a rare congenital malformation characterized by an absence of skin and skull. The authors describe a newborn at an estimated 38 weeks gestational age who was delivered via cesarean section from a 32-year-old mother. Upon delivery, the child was noted to have a frontal encephalocele and an absence of calvaria including skull and skin overlying the brain. A thin membrane representing dura mater was overlying the cortical tissue. After multiple craniofacial operations, including repair of the encephalocele and application of cultured keratinocytes over the rostral defect, the patient demonstrated significant closure of the calvarial defect and was alive at an age of more than 17 months with near-average development.

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**Key Words** • acalvaria • congenital malformation • spina bifida • acrania

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Examination. Apgar scores at 0, 5, 10, and 15 minutes from delivery were 0, 2, 4, and 5, respectively. The neonate’s weight was 2.5 kg (< 10th percentile), length was 47.2 cm (15th percentile), head circumference was 31.9 cm (10th percentile), temperature was 36.8°C, pulse was 162 bpm, respiratory rate was 78 breaths per minute, and mean arterial blood pressure was 40 mm Hg. The prenatally diagnosed 4 × 4 × 4–cm fonticulus frontalis encephalocele was noted at the glabella. The rostral portion of the head was notable for a large scalp and skull defect with exposed thin dura (Fig. 1A). Bilateral epicanthal folds, hypertelorism, and bilateral red reflexes were present. The nares were patent, the palate was intact, neck supple, and chest symmetric, and there were bilateral lung breath sounds. Cardiovascular exam revealed regular rhythm, normal heart rate, and prominent S2 but no murmur. The abdomen was soft and nondistended, without organomegaly or masses. No overt stigmata for cervical or thoracolumbar spinal dysraphisms were noted, and the anus appeared to be patent and normally placed. Neurological exam revealed spontaneous eye opening, movement of all extremities, multiple beats of clonus upon heel

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History. This female infant of an estimated 37 and 6/7 weeks gestational age was delivered from a 32-year-old gravida 2, para 0-0-1-0 mother who had received excellent prenatal care. The prenatal course was complicated by elevated alpha-fetoprotein and weakly positive acetylcholine esterase, and a fetal nasal encephalocele was diagnosed at 20 weeks’ gestation. The mother had undergone two fetal MRI studies, which revealed a soft tissue mass in the region of the fetal glabella consistent with an encephalocele. Amniocentesis showed normal karyotype. The child was delivered via scheduled cesarean section. Delivery was noteworthy for bradycardia and hypoxia requiring chest compressions, epinephrine therapy, and intubation.
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dorsiflexion, normal muscle tone, and intact grasp and suck reflexes. Postnatal chromosomal microarray analyses revealed no genomic imbalance.

Treatment. Broad-spectrum antibiotics were initiated, and the defect was immediately covered with a sterile, wet nonstick dressing. Magnetic resonance imaging of the brain confirmed the absence of calvaria and skin as well as the presence of a fonticulus frontalis encephalocele (Fig. 1B). On Day 2 of life, the exposed dura was gently debrided, and an Integra dermal regeneration template was applied at the bedside in the neonatal intensive care unit. Small wedge-type closing incisions were fashioned in the graft to improve the contour, and the graft was sutured to the scalp. The dermal regeneration template was intended to create a soft tissue layer between the dura and developing skin. If skin were directly grafted onto the dura, future procedures on the scalp could threaten dural competency. In addition to soft tissue augmentation, the Integra graft protected the wound from infection and moisture loss and served as scaffolding for the generation of skin cells. On Days 25 and 40 the defect was explored, and new skin generation was observed over many regions of the exposed dura. During each of these subsequent procedures, the Integra graft was reapplied to further augment the soft tissues. By 2 months of age, the defect had closed significantly. The Integra graft was removed, and the remaining defect was reconstructed with the transposition of a small flap of redundant skin on the occiput and the application of cultured keratinocytes. After spending the initial 4 months of her life in the hospital, the patient was discharged to her parents’ care. At 6 months of age, the patient underwent resection of the frontonasal encephalocele defect and reconstruction with a parietal bone graft. Pathology of the lesion showed complex cortical malformation consistent with encephalocele.

Posttreatment Course. At the time of her last follow-up clinic visit, the child was 17 months of age and had experienced no major complications. The encephalocele was repaired, and the acalvaria skin defect was completely healed with partial skin pigmentation (Fig. 1C and D). Palpation and CT studies of the head (Fig. 2) provided evidence of osteogenesis and reduction of the skull defect. Developmental evaluation using Bayley Scales of Infant and Toddler Development revealed average age-adjusted performance in cognitive, language, and motor skills; however, the child demonstrated developmental delay for expression, performing at the 9-month-old level. She was therefore referred for speech therapy.

Discussion
Acalvaria is a rare congenital disorder reported to be fatal in most neonates. Although three surviving cases

Fig. 1. Acalvaria. A: Photograph of acalvaria in a newborn. B: MR image of the brain demonstrating acalvaria and encephalocele. C and D: Postoperative photographs after repair of acalvaria and encephalocele.
have been reported, there is no consensus on the management of this disease. Some authors have reported success with conservative nonsurgical management. However, the children in these cases had scalp overlying their skull defect, providing protection from meningeal infection. The child featured in the present report lacked scalp and skull, leaving a thin dural layer exposed to the outside elements and prone to physical damage and infection. For this case, a multidisciplinary approach among pediatric neurosurgery, plastic surgery, and neonatal intensivists allowed correction of the patient’s encephalocele and acalvaria. In addition to closure of the soft tissue defect, spontaneous bone growth over some of the defect and near-average neurodevelopment were revealed at the follow-up evaluations. Modern reconstructive technologies were used to protect underlying tissue and facilitate skin growth. Although skin substitutes and adjuvants are used in both neurosurgery and plastic surgery, the present case represents the first application of these methods for acalvaria. In summary, the present case is among the few instances of surviving acrania and demonstrates the presentation and successful operative management of an infant with the congenital absence of a calvarial vault and overlying scalp.

**Disclosure**

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

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