Giant anterior enchephalocele. Successful multidisciplinary surgical resolution in a neonate. A case report

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Background: Encephaloceles are rare congenital neural tube defects characterized by the herniation of intracranial structures through a bony defect in the cranium. Frontonasal/frontoethmoidal subtype accounts for 13–15% of cases and is particularly rare when presenting as a giant mass exceeding cranial dimensions. We present the case of a 20-day-old male neonate with a giant frontal mass measuring 18 × 10 cm, diagnosed antenatally via structural ultrasound and characterized postnatally using computed tomography imaging. Successful multidisciplinary surgical repair and dural closure were achieved with optimal functional and cosmetic outcomes.

Key words: Encephalocele, Frontoethmoidal Encephalocele, Neural Tube Defects, Neuroimaging, Pediatric Neurosurgery.

Case Report

Radiology

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Encephalocele is a congenital malformation of the neural tube characterized by the herniation of brain parenchyma, meninges, and cerebrospinal fluid (CSF) through a focal osseous defect in the calvaria or skull base. Embryologically, it results from a failure in the separation of the neuroectoderm from the surface ectoderm during primary neurulation (around the fourth gestational week), preventing proper mesenchymatous interposition and normal craniocervical osification.

Globally, encephalocele occurs in approximately 1 in 4,000 to 10,000 live births. While occipital lesions predominate in European and American populations (75–80%), sincipital and frontoethmoidal variants represent 13–15% of cases. Giant frontoethmoidal encephaloceles present a critical diagnostic and neurosurgical challenge due to high risks of CSF fistula, meningitis, and cosmetic/functional morbidity. Advanced neuroimaging modalities, including high-resolution computed tomography (CT) and magnetic resonance imaging (MRI) are essential for precise structural mapping and surgical planning.

Case report

A 20-day-old male neonate was evaluated at our institution due to a progressive frontal mass present since birth. Antenatal history revealed an initial ultrasound detection of a frontal lesion at 12 weeks of gestation, followed by a detailed structural ultrasound at 21 weeks that confirmed a predominantly cystic frontal mass. The infant was born via cesarean delivery at 31 weeks of gestation due to premature labor. Initial dismal prognosis led to a discharge against medical advice; however, secondary to progressive enlargement of the mass over 20 days, the parents reconsulted at our tertiary care center.

On physical examination, the neonate was hemodynamically stable (HR: 135 bpm, RR: 35 bpm, SpO_2 : 93% on room air). Physical inspection demonstrated a giant, soft-tissue frontal mass measuring 18 × 10 cm with prominent superficial blood vessels, covered by a thin, semitransparent membrane. Pupils were equal and reactive to light. Neurological, cardiopulmonary, and abdominal examinations were unremarkable. Laboratory parameters (complete blood count, coagulation profile, metabolic panel, and C-reactive protein) were within normal limits for gestational age.

Advanced neuroimaging (multidetector CT with 3D bone reconstructions) confirmed a giant anterior frontoethmoidal meningoencefalocele protruding through a midline frontal calvarial defect.

The patient underwent single-stage multidisciplinary surgical intervention consisting of herniated sac resection, meticulous dural repair (duraplasty), and cranial reconstruction without intraoperative complications. The patient achieved complete clinical recovery with satisfactory skin closure, normal oral feeding, and preserved neurological function.

Discussion

Anterior frontoethmoidal encephaloceles emerge through bony defects between the frontal and ethmo id bones (frontonasal, nasoethmoidal, or

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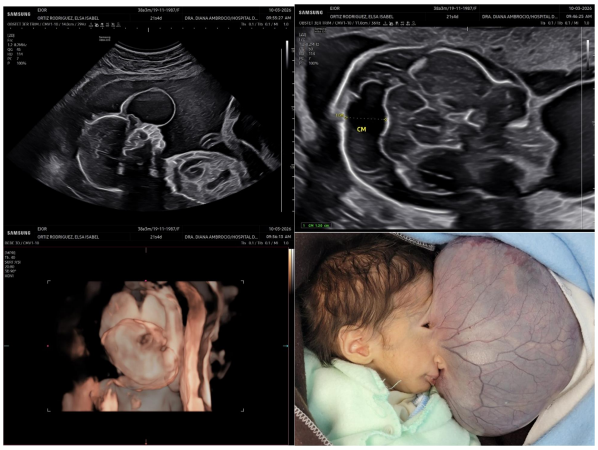


Figure 1. Antenatal structural ultrasound and clinical presentation. (A–C) Second-trimester structural ultrasound (21 weeks of gestation) showing a well-defined cystic lesion (measurements demonstrating posterior fossa cisterna magna at 1.20 cm) and a prominent anterior frontal herniation defect labeled as frontal encephalocele. (D) Clinical photograph at 20 days post-birth depicting a giant frontal mass measuring 18 × 10 cm covered by a translucent membrane with prominent vascularity causing severe facial distortion.

nasoorbital subtypes). Neuroimaging is pivotal in defining the contents, vascular relationships, and skull base bony anatomy.

Ultrasonography serves as the primary screening tool during the second trimester, demonstrating an extra-cranial cystic or complex mass associated with a defect in calvaria. Computed tomography with 3D reconstructions provides detailed delineation of the osseous margins, crucial for operative planning of craniofacial osteotomies. MRI remains the gold standard to evaluate brain parenchyma integrity, ventricular involvement, and dural venous sinus relationship (via MR venography) to prevent accidental vascular injury during sac ligation.

Differential diagnosis for pediatric midline cranial masses includes cephalhematoma, subgaleal hematoma, epidermoid/dermoid cysts, sinus pericranii, nasal glioma, and cystic hygroma. Demonstration of direct bony defect and intracranial continuity on CT/MRI excludes non-congenital scalp and facial masses.

Surgical correction aims for early excision of non-functional dysplastic brain tissue, water-tight dural closure to prevent CSF leakage and secondary bacterial meningitis, and structural repair of the bony defect. In this case, early surgical management prevented life-threatening complications such as CSF fistula, hydrocephalus, and refractory seizures, achieving optimal cosmetic and neurodevelopmental outcomes.

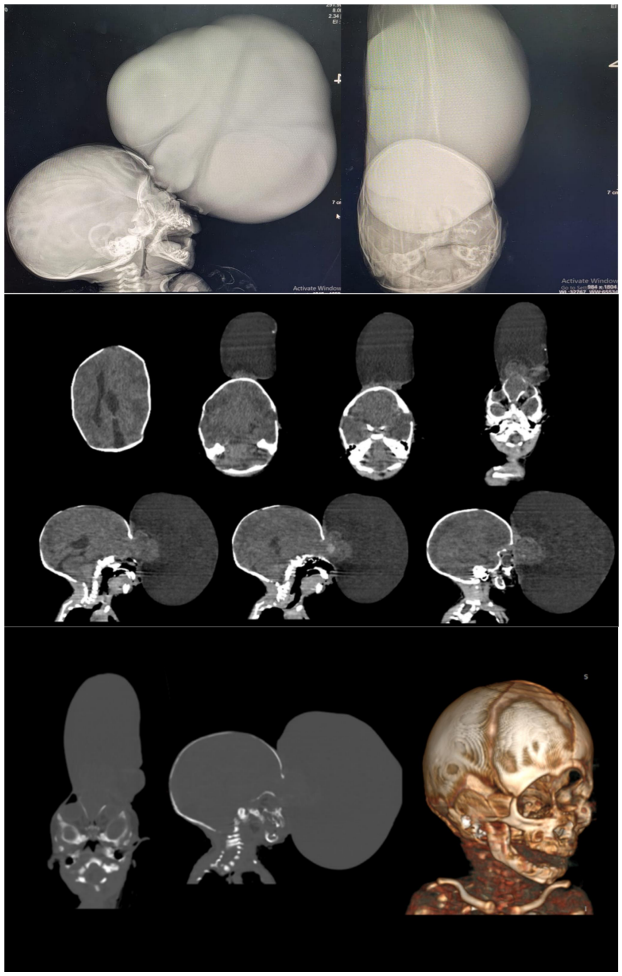


Figure 2. Postnatal skull Radiography and Multidetector Computed Tomography (CT). *(A–B)* Lateral and anteroposterior skull radiographs showing a massive soft-tissue mass superior to the facial skeleton. *(C–E)* Axial, coronal, and sagittal non-contrast brain CT views demonstrating a large anterior skull defect with herniation of brain tissue and fluid-filled sac. *(F)* 3D bone CT reconstruction showing the precise midline frontal bony defect (bony rim defect) used for surgical repair planning.

Conclusion

High-resolution CT with 3D multiplanar reconstructions are indispensable diagnostic modalities for characterizing giant frontoethmoidal encephaloceles, allowing precise definition of the bony defect, parenchymal involvement, and vascular anatomy.

Differential diagnosis must systematically rule out non-communicating cranial lesions such as dermoid cysts, nasal gliomas, and subgaleal collections through demonstration of intracranial continuity.

Early multidisciplinary surgical intervention featuring herniated tissue excision and meticulous

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Figure 3. Intraoperative and postoperative resolution. *(A–D)* Intraoperative photographs showing the surgical dissection of the giant sac, exposure of the frontal bony defect, resection of non-functional dysplastic herniated tissue, and watertight duraplasty. *(E)* Immediate postoperative photograph demonstrating complete reconstruction of the frontal contour, facial symmetry, and primary skin closure. *(F)* Follow-up photograph showing the patient active, feeding comfortably, and exhibiting excellent surgical recovery.

water-tight duraplasty resolves life-threatening risks of CSF infection and provides favorable clinical, neurological, and aesthetic outcomes.

Conflicts of interests

None declared by the authors.

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