
Pediatric Cephaloceles: A Multimodality Review

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Cephaloceles are complex brain malformations that can be further characterized by the content of the herniated tissue. They can be classified by location, which is important for family counseling and surgical planning. Although imaging is vital for characterizing cephaloceles, and fetal MRI is becoming more commonly utilized for prenatal characterization, little appears in the radiology literature on this complex topic. This review illustrates the four major types of cephaloceles using a multimodality approach with prenatal and postnatal correlation. A brief overview of epidemiology and embryology is provided, and associated anomalies and distinguishing features of associated syndromes are discussed. By utilizing the location-based system and understanding the commonly associated features, the radiologist may provide a more comprehensive description of cephaloceles to better facilitate clinical management.

Cephaloceles are one of the most common forms of neural tube defects, ranking only behind myelomeningocele and anencephaly. *Cephalocele* is a generic term defined as a protrusion of the meninges with or without brain tissue through a defect in the skull.¹⁻² A *meningocele* is a protrusion of only meninges and cerebrospinal fluid (CSF). An *encephalocele* is a protrusion of meninges, CSF, and brain tissue. An *encephalocystocele* contains meninges, CSF, brain tissue and ventricle. The term *atretic cephalocele* (also called meningocele manqué) describes a small, midline subscalp nodule that contains meninges, fibrous tissue, and dysplastic brain tissue.³ The term *fronto-ethmoidal* denotes the involvement of the upper anterior cranium and is synonymous with the less commonly used *sincipital* (Table 1).

The incidence of cephaloceles is 0.8-4 of 10,000 live births.⁴ Cephaloceles account for 10-20% of all craniospinal

dysraphisms.⁴ Geographical variation exists, with occipital subtypes representing 66-89% of all cephaloceles in the Caucasian populations of North America and Western Europe.⁵⁻⁷ Anterior subtypes are more common in Southeast Asia.⁶ Most cases of isolated cephaloceles (not associated with other congenital anomalies) are sporadic, with genetic and non-genetic factors involved in the pathogenesis.⁸ Cephaloceles may be associated with myriad genetic syndromes, most commonly Meckel-Gruber, as well as the Chiari III malformation, holoprosencephaly, and Dandy-Walker malformation.^{2,9-10} A cephalocele detected prenatally warrants a detailed diagnostic assessment and characterization for an underlying syndrome.¹¹

Embryology

The central nervous system begins forming in the third week of embryonic life as thickened ectoderm called the neural plate. Elevation of the lateral edges of the neural plate forms the neural folds, which fuse to form the neural tube. Fusion begins in the cervical region, proceeding in both the rostral and caudal directions until closure between days 25-27 postconception. The mechanism resulting in cephaloceles is uncertain but presumably involves defective closure of the anterior neural tube. Anterior

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Table 1. Definitions of Terms Characterizing Herniated Tissue in Cephaloceles with Imaging Correlates

Term	Meningocele	Encephalocele	Encephalocystocele	Atretic cephalocele	Fronto-ethmoidal cephalocele
Content	Only meninges and CSF	Meninges, CSF, and brain tissue	Meninges, CSF, brain tissue and ventricle	Small midline sub-scalp nodule containing meninges, fibrous tissue, and dysplastic	Involving the upper anterior cranium; also referred to as sincipital cephalocele
Example					

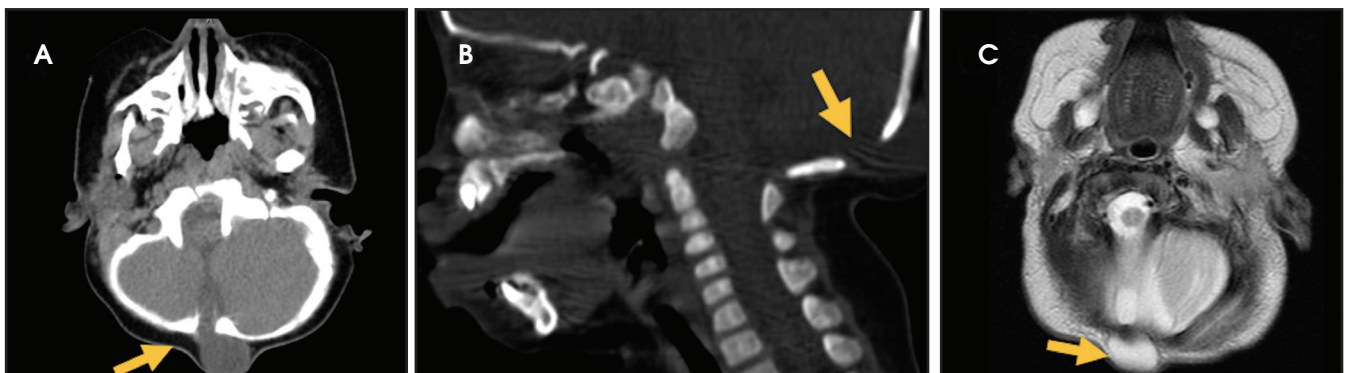


FIGURE 1. Occipital meningocele. Axial (A) and sagittal (B) CT images demonstrate an occipital bone defect (arrow) with protrusion of tissue. Axial (C) T2 images depict a small, CSF-intensity sac (arrows) representing the meningocele protruding through the defect. No brain tissue is identified in the sac.

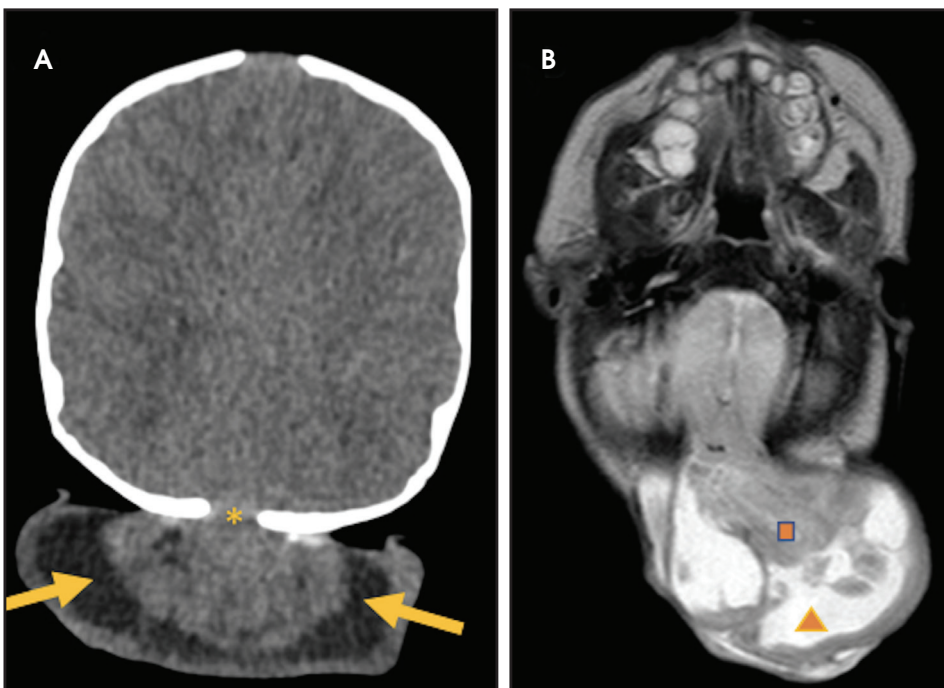


FIGURE 2. Occipital encephalocele. Axial CT image through the supratentorial brain parenchyma (A) shows a fluid- and soft-tissue attenuation sac (arrows) herniating through a midline occipital skull defect (*), consistent with an occipital encephalocele. Axial T2 MRI through the cerebellum (B) demonstrates that the hernia sac contains CSF (triangle) and dysplastic neural tissue (square). Irregular margins of the neural tissue and lack of definite connection to the ventricles argues against encephalocystocele in this case.

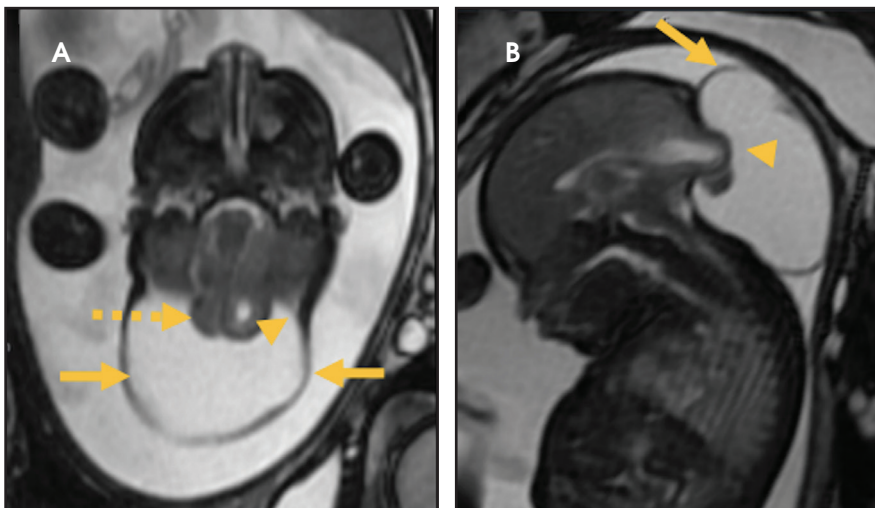


FIGURE 3. Occipital encephalocystocele. Axial (A) and sagittal (B) fetal MRI images demonstrate an occipital cranial defect with a large hernia sac (solid arrows) consisting of meninges and supratentorial neural tissue (dashed arrow) containing a portion of the occipital horns of the lateral ventricles (arrowhead) consistent with an encephalocystocele.

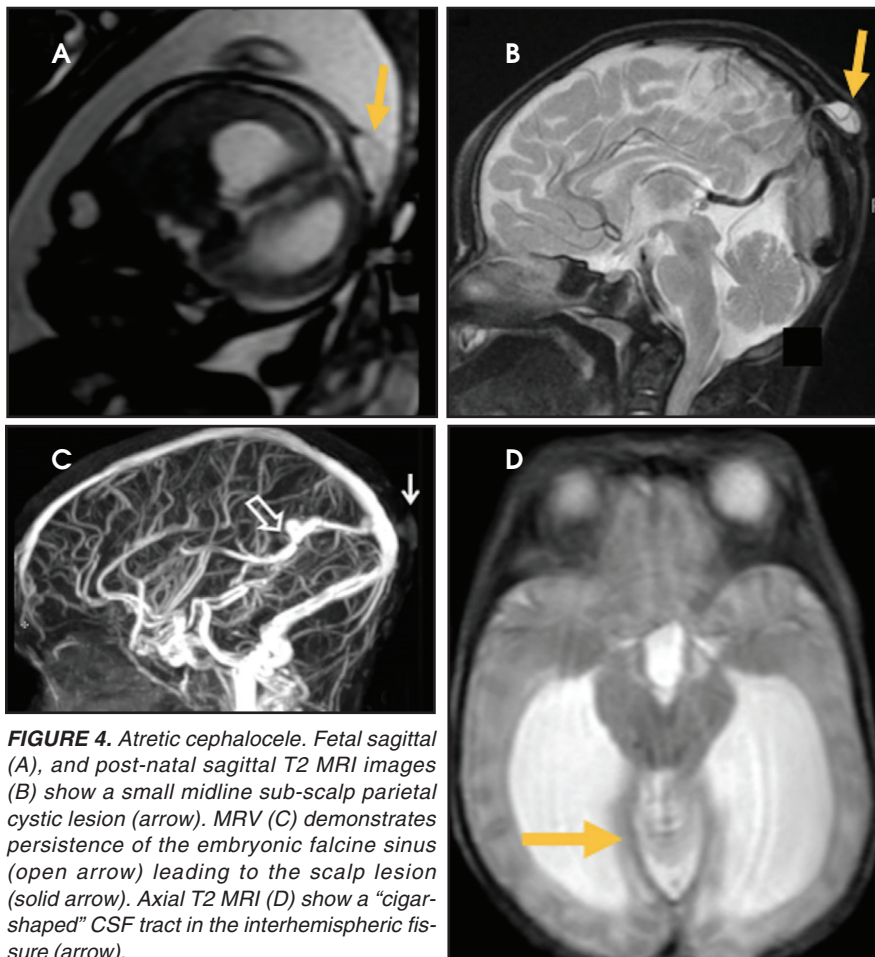


FIGURE 4. Atretic cephalocele. Fetal sagittal (A), and post-natal sagittal T2 MRI images (B) show a small midline sub-scalp parietal cystic lesion (arrow). MRV (C) demonstrates persistence of the embryonic falcine sinus (open arrow) leading to the scalp lesion (solid arrow). Axial T2 MRI (D) show a "cigar-shaped" CSF tract in the interhemispheric fissure (arrow).

cephaloceles (fronto-ethmoidal and basal) are thought to arise from defective development of the prosencephalic neural crest tissue.¹² In contrast, occipital

cephaloceles may relate to defective segmentation of the posterior cranial bones.¹³ Some authors believe the etiology of congenital cephalocele

centers on a postneurulation event in which brain tissue herniates through a defect in the mesenchyme that eventually becomes the cranium and dura.¹⁴

Diagnosis and Classification

Most cephalocele can be detected prenatally by ultrasound. Alpha fetoprotein levels may be unreliable given that both maternal serum and amniotic fluid alpha fetoprotein levels may be normal if the cephalocele is covered by skin.¹⁵ On ultrasound these lesions may appear as cystic (meningocele) and/or solid (encephalocele) structures protruding through a calvarial defect. Further evaluation with computed tomography (CT) and magnetic resonance imaging (MRI, including fetal MRI) is useful to determine the extent of herniation as well as the presence of associated anomalies. CT is useful for identifying osseous defects while MRI is superior for defining the portions of herniated tissue, detecting abnormal signal in dysplastic brain tissue, and evaluating the cartilaginous nasofrontal region. An MR or CT angiogram or venogram may provide more detailed evaluation of vascular anatomy and its relation to the cephalocele.

Several classification systems exist. The system proposed by Suwanwela and Suwanwela provides a comprehensive, location-based classification that characterizes cephalocele as 1) occipital, 2) cranial vault, 3) fronto-ethmoidal, and 4) basal. This system has also been found useful for selecting the operative approach.⁶ More extensive cephalocele, however, may encompass more than one type. Clinical features and prognoses of cephalocele depend on location, severity, and presence of dysplastic brain tissue and associated abnormalities.¹⁶⁻¹⁷

Occipital Cephalocele

Occipital cephalocele demonstrate defects involving the occipital bones, with the cephalocele extending posteriorly (Figures 1-3). The herniated tissue may include the supra- and/or infratentorial brain, tentorium, and dural

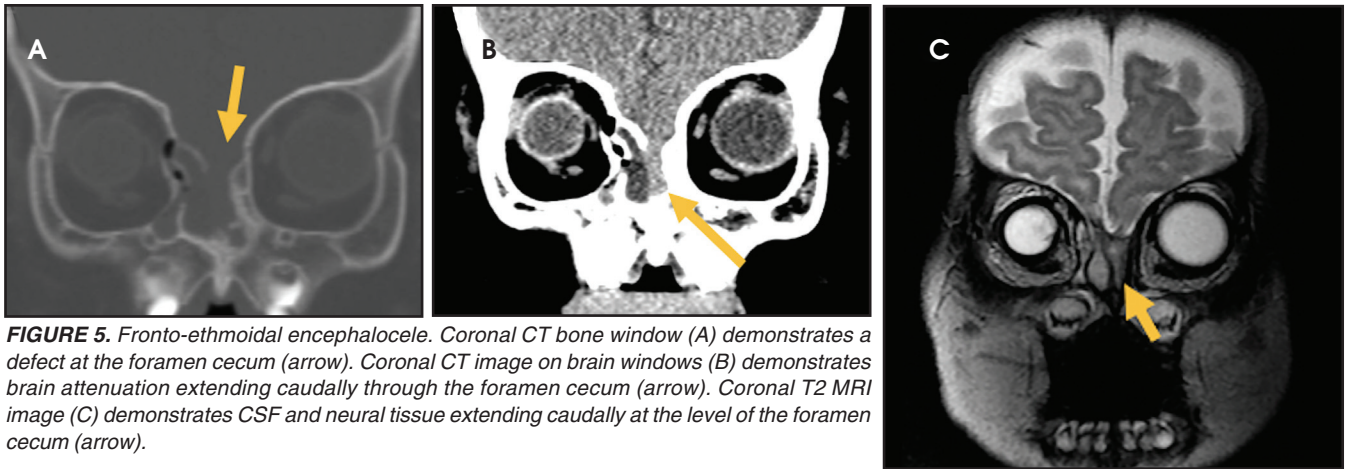


FIGURE 5. Fronto-ethmoidal encephalocele. Coronal CT bone window (A) demonstrates a defect at the foramen cecum (arrow). Coronal CT image on brain windows (B) demonstrates brain attenuation extending caudally through the foramen cecum (arrow). Coronal T2 MRI image (C) demonstrates CSF and neural tissue extending caudally at the level of the foramen cecum (arrow).

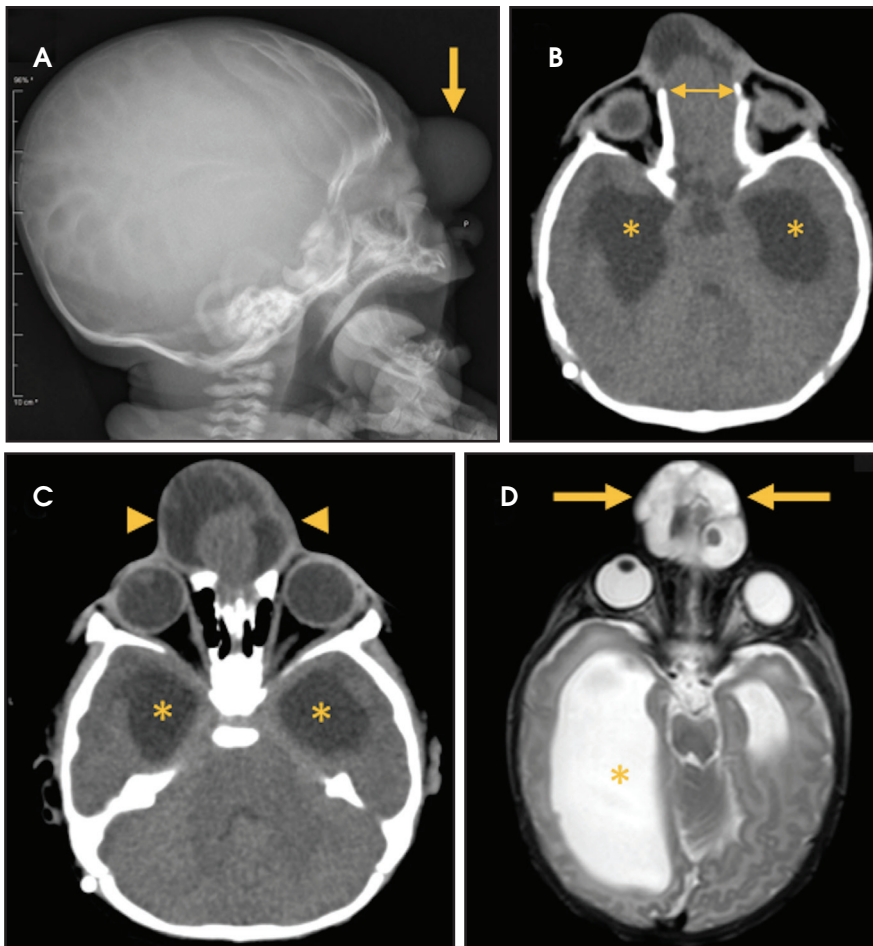


FIGURE 6. Fronto-nasal encephalocele with hydrocephalus. Lateral skull radiograph (A) shows a frontal soft tissue mass (arrows). (B, C) Axial CT images through the glabella demonstrate a frontal bone defect (arrows) and encephalocele (arrowhead). Hydrocephalus is also noted (*). Axial T2 MRI (D) show a midline encephalocele (arrows) at the level of the lower frontal bone. Marked hydrocephalus is also seen (*).

venous sinuses. These cephaloceles are the most common type overall and account for a higher proportion of cephaloceles in Caucasian populations of

Europe and North America.⁵⁻⁷ Occipital cephaloceles are typically apparent on physical exam at birth, and the size of the herniation varies. Ventriculomegaly

is observed in the majority of cases. Prognostic factors include size of herniation, degree of hydrocephalus, and presence of associated anomalies.¹⁶⁻¹⁷ Large occipital encephalocetes may be associated with developmental delay, blindness, poor feeding, cranial nerve deficits, and seizures.¹⁸

Cranial Vault Cephaloceles

Cranial vault cephaloceles occur along the superior cranium within the fontanelles or defects in the parietal, frontal, or temporal bones. They present as a midline posterior scalp mass. The patient often is otherwise clinically normal unless associated anomalies are present.¹⁹ Atretic cephaloceles, the most common form, are small midline subcutaneous scalp masses consisting of dura and dysplastic meninges connected to the intracranial meninges by a fibrous stalk. They are usually located in the parietal lobe; MRI typically demonstrates a fibrous tract and vertical falcine vein, which extend to a subcutaneous scalp mass (Figure 4). Atretic cephalocele may arise through a bone defect or fenestration, or the bone may be closed with completely separated intra- and extracranial contents.²⁰

The embryonic falcine sinus is often positioned vertically, with a cigar-shaped CSF tract in the interhemispheric fissure.^{19,21} Cranial vault cephaloceles are considered abortive or involuted true cephaloceles²⁰; they have a more favorable prognosis than other true cephaloceles.²²

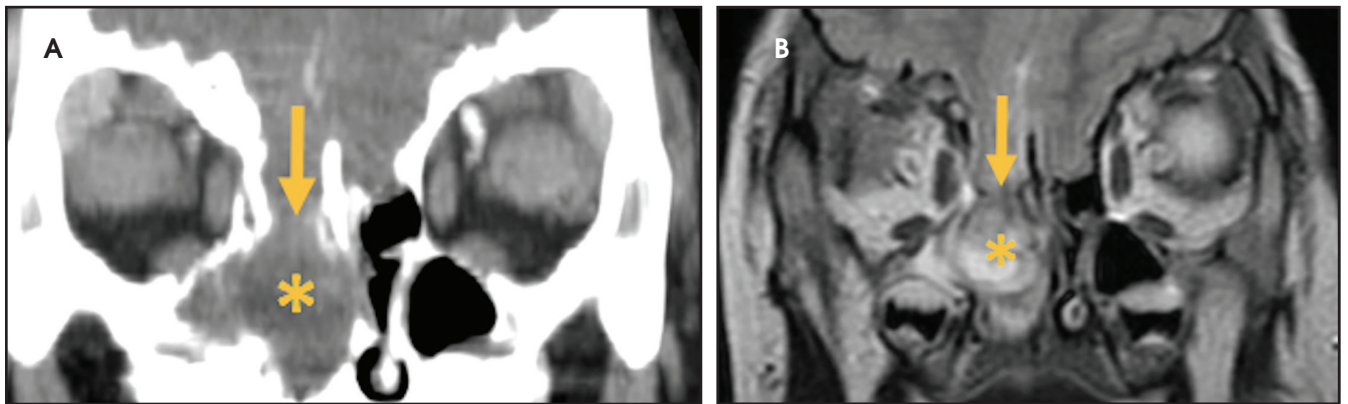


FIGURE 7. Basal encephalocele (A) and T2 MRI images (B). A defect of the cribriform plate (arrow) is seen. There is protrusion of CSF and brain tissue into the nasal cavity and right maxillary sinus (*).

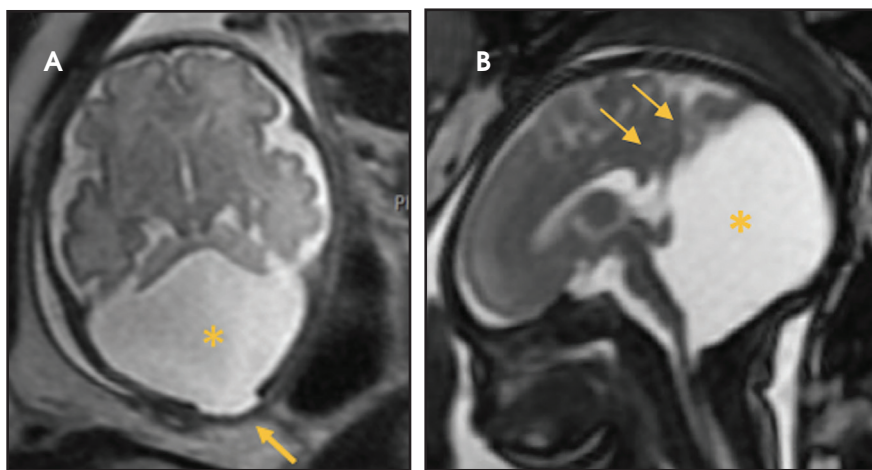


FIGURE 8. Occipital meningocele with Dandy-Walker malformation. Fetal axial T2 image (A) shows a small midline occipital meningocele (arrow) and cystic replacement of an enlarged posterior fossa (*). A fetal sagittal T2 image (B) demonstrates the large posterior fossa cyst (*) and an elevated tentorium (arrows).

Fronto-ethmoidal Cephaloceles

Fronto-ethmoidal cephaloceles (FECs) range from occult lesions to marked craniofacial abnormalities, including microcephaly, telecanthus, hypertelorism, orbital dystopia, or micro/anophthalmos. There is an increased incidence in Southeast Asian populations.⁶ Sagittal and coronal images may be the most helpful in demonstrating contiguity between intracranial contents and the mass.²³ Prior to surgical repair, CT scanning helps to characterize the bone defect. FECs can be classified according to the osseous defect's location.

Naso-ethmoidal cephaloceles are characterized by herniation into the superomedial nasal cavity, with the defect

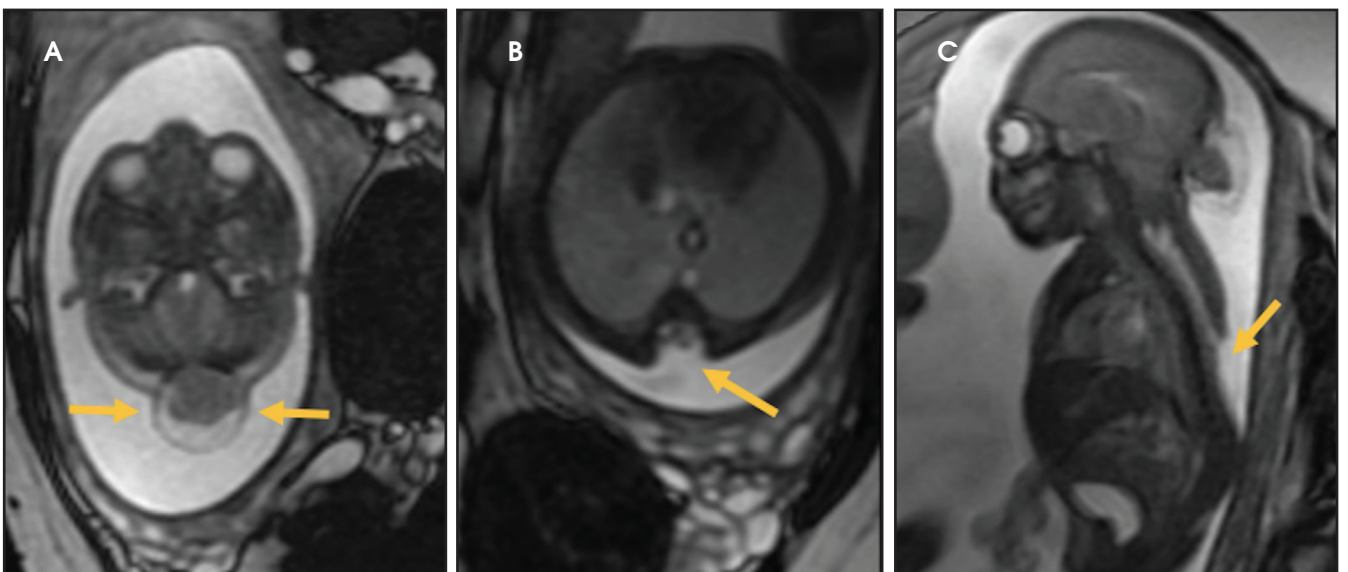


FIGURE 9. Chiari III with thoracic myeloschisis. Axial T2 (A) demonstrates an occipital encephalocele (arrows) with cerebellar crowding and tonsillar herniation. Axial T2 (B) demonstrates open the thoracic dysraphism (arrow). Sagittal T2 (C) demonstrates the open thoracic dysraphism beginning in mid-thoracic spine extending caudally (arrow).

Table 2. Conditions Associated with Cephaloceles

Site	Condition	Distinguishing Features
Frontal	Amniotic Band Syndrome	Ring constrictions and amputations of digits and/or limbs, irregular/asymmetric cephalocele, microcephaly, microphthalmia, distal syndactyly, orofacial clefts
	Frontonasal Dysplasia/ Median Cleft Face Syndrome	Ocular hypertelorism, widow's peak, anterior cranium bifidum occultum, notching of nostrils, widely set nostrils with lack of elevation of the nasal tip
Occipital or high cervical	Chiari III Malformation	Syringomyelia, hydrocephalus, tethered cord, abnormal neuroectodermal tissues
Occipital	Cryptophthalmos/Fraser Syndrome	Ear anomalies, notching of nasal wings, extension of forehead skin to cover one or both eyes, unusual hairline, soft-tissue syndactyly of hands and/or feet, genital anomalies including cryptorchidism, micropenis, and clitoromegaly
	Dandy Walker Formation	Triad of cystic dilation of the fourth ventricle, complete or partial agenesis of the cerebellum and an enlarged posterior fossa with torcular-lamboid inversion
	Dyssegmental Dwarfism	Lethal dwarfism, short broad pelvis with widely flared iliac wings, short broad tubular bones with metaphyseal widening, accelerated carpal bone maturation, bowing of legs, thighs and forearms, vertebral anomalies, small thorax, cleft palate, micrognathia
	Fetal Warfarin Syndrome	Nasal hypoplasia, limb shortening, bone stippling, low birth weight, seizures, hydrocephaly, optic atrophy, intellectual disability
	Knobloch Syndrome	Myopia, vitreoretinal degeneration, retinal detachment, meningocele, normal intelligence
	Meckel-Gruber Syndrome	Classically triad of renal cystic dysplasia, cephalocele or holoprosencephaly, and postaxial polydactyly; microphthalmia, retinal dysplasia, cardiac anomalies, orofacial clefting, ambiguous external genitalia
	Pseudo-Meckel Syndrome	Arhinencephaly, agenesis of corpus callosum, Arnold-Chiari defect, cleft palate, congenital heart defects, accessory spleen, clubfoot, hallucal hammertoes; retinal dysplasia is not a feature
	von Voss-Cherstvoy Syndrome	Corpus callosum dysplasia, hypoplastic olives and pyramids of medulla oblongata, phocomelia, urogenital anomalies, thrombocytopenia
	Walker-Warburg Syndrome	Muscle weakness and atrophy early in life, developmental delay, hydrocephalus, microphthalmia, buphthalmos, cataracts

centered at the foramen cecum (Figure 5).⁶ They protrude through the foramen cecum into the prenasal space. They are positioned inferior to the nasal bones.

Naso-frontal cephaloceles have a midline frontal defect, often with mass at the glabella (root of the nose) (Figure 6).⁶ They protrude through an unobliterated fonticulus frontalis.

Naso-orbital cephaloceles are characterized by an inferomedial orbital defect. They protrude into the inferomedial orbit through a defect in the

maxillary bones at the lacrimal/frontal process. They can induce proptosis and globe displacement.

FECs are more commonly associated with craniofacial clefts.⁶ FECs also generally have a better prognosis than occipital cephaloceles because the protruding mass in the FEC tends to contain scarred, nonfunctional neural tissue.¹⁸

Basal Cephaloceles

Basal cephaloceles occur when there is a defect in skull base (Figure 7). They

are rare and may present even later in the first decade of life with recurrent meningitis.²⁴ Basal cephaloceles may be occult or present with midface anomalies such as cleft lip/palate, hypertelorism, or a nasal epipharyngeal mass. Immediate surgical repair is indicated because of the elevated risk of meningitis.¹⁸

Associated Anomalies and Neurologic Manifestations

Many cases of cephalocele are associated with additional congenital

anomalies, which are important to identify for prognostic purposes (Table 2). In one of the largest studies on cephaloceles, the following associated anomalies and neurologic manifestations were identified in order of decreasing frequency: hydrocephalus, seizure disorder, corpus callosum abnormalities, cerebral dysgenesis, and migrational disorders, including gray matter heterotopia, microcephaly, and myelomeningocele.⁴

In this study, 52% of patients experienced at least mild developmental delay, with hydrocephalus and other associated intracranial abnormalities identified as predictors of developmental delay.⁴ Lesion location notably was not found to be a significant predictor of outcome. Myriad genetic syndromes and conditions are associated with cephaloceles, including Meckel-Gruber syndrome, the middle interhemispheric variant of holoprosencephaly, Dandy-Walker malformation (Figure 8) and Chiari III malformation (Figure 9).^{2,9-10} Meckel Gruber is the most commonly associated syndrome.²⁵ With the presence of associated malformations affecting cognitive outcome, imaging is a critical component of the workup of patients with cephaloceles and often serves as the primary basis for prenatal counseling.⁴

Conclusion

Cephaloceles are complex cranial malformations that can be classified by

location, each with differing clinical presentation and associated anomalies. Prenatal and postnatal imaging is important to delineate the relevant anatomy. By describing cephaloceles via a location-based classification system, the radiologist may facilitate more accurate presurgical planning and prenatal counseling.

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