

MRI depiction of fetal brain abnormalities

Cory M Pfeifer¹ , Scott D Willard² and Patricia Cornejo³ 

Acta Radiologica Open
8(12) 1–6
© The Foundation Acta
Radiologica 2019
Article reuse guidelines:
sagepub.com/journals-permissions
DOI: 10.1177/2058460119894987
journals.sagepub.com/home/arr



Abstract

Intracranial abnormalities are commonly suspected findings on antenatal ultrasound that require evaluation by magnetic resonance imaging. This review depicts multiple abnormalities imaged as a means to guide clinicians in proper diagnosis.

Keywords

Congenital brain malformations, fetal magnetic resonance imaging, midline abnormalities

Received 13 September 2019; accepted 22 November 2019

Introduction

Magnetic resonance imaging (MRI) is an important adjunctive tool used to evaluate the fetus in cases of suspected anomalies detected by ultrasound (US). Recent improvements in magnetic field strength and scan time in addition to parallel imaging techniques result in decreased acquisition times, optimized specific absorption rates (SAR), improved signal-to-noise ratio, and improved dielectric/non-uniformity artifacts. While 1.5-T imaging was long-considered the standard in fetal imaging, MRI of the fetus performed at 3-T is no longer uncommon (1–3).

The most common indications for fetal brain MRI are ventriculomegaly (VM), agenesis/dysgenesis of the corpus callosum, and posterior fossa abnormalities. Antenatal management of many of these disorders is typically expectant, but fetal MRI may provide information that can guide appropriate follow-up in the post-partum setting. Presented here are a series of intracranial abnormalities commonly encountered in mothers presenting with abnormal antenatal ultrasound exams.

Case series

Agenesis of the corpus callosum

The corpus callosum (CC) is the largest white matter commissure in the brain comprising the rostrum and genu anteriorly, the body centrally, and the splenium posteriorly. Since multiple chromosomal loci contribute to the formation and myelination of the corpus callosum, callosal dysgenesis and agenesis may be

caused by an extensive list of genetic syndromes and mutations. There is a known association with Dandy-Walker malformation (DWM), fetal body anomalies, and aneuploidy (4). Often, callosal dysgenesis occurs in association with interhemispheric cyst/lipoma. In addition, metabolic derangements, infection, and toxicity are known to contribute.

The reported incidence of callosal agenesis is approximately 1:4000 live births (5) with a male predominance. A child with agenesis of the corpus callosum may develop normally; however, the morbidity of callosal disorders is worsened in the setting of other anatomic abnormalities and/or syndromes.

Imaging findings are characteristic, with parallel orientation of the ventricles, “longhorn” sign on coronal imaging, and radially oriented midline sulci (Fig. 1). The third ventricle is high in location and communicates with the interhemispheric fissure. White matter loss and colpocephaly are common.

¹Department of Radiology, University of Texas Southwestern Medical Center, Dallas, TX, USA

²Department of Radiology, Nemours Children’s Health System, Jacksonville, FL, USA

³Department of Medical Imaging, Phoenix Children’s Hospital, Phoenix, AZ, USA

Corresponding author:

Cory M Pfeifer, Department of Radiology, University of Texas Southwestern Medical Center, 5323 Harry Hines Boulevard, Dallas, TX 75390, USA.

Email: Cory.Pfeifer@UTSouthwestern.edu



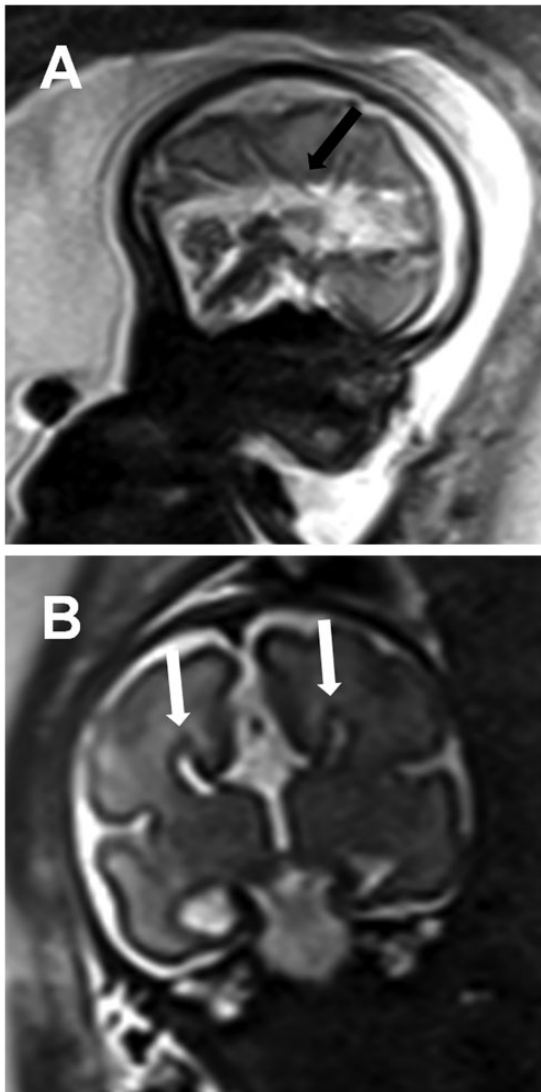


Fig. 1. MRI findings of agenesis of the corpus callosum in a 30-week, 0-day fetus. (a) Sagittal HASTE image demonstrates absence of the corpus callosum (black arrow) and radial pattern of the parasagittal gyri. (b) Coronal HASTE image shows the “longhorn” configuration of the lateral ventricles (white arrows).

Aqueductal stenosis

Aqueductal stenosis (AS) causes VM of the lateral and third ventricles due to obstruction of outflow from the third ventricle into the fourth ventricle, which is normal in size. AS is the second most common cause of VM after Chiari malformation. It has been associated with neonatal mortality in up to 25% of affected fetuses (6).

On fetal MRI, the cerebral aqueduct is effaced (Fig. 2). The remaining findings relate to hydrocephalus, notably prominent lateral VM and, to a lesser extent, third VM. The septum pellucidum is absent or incomplete and the corpus callosum is often thinned and/or superiorly displaced.

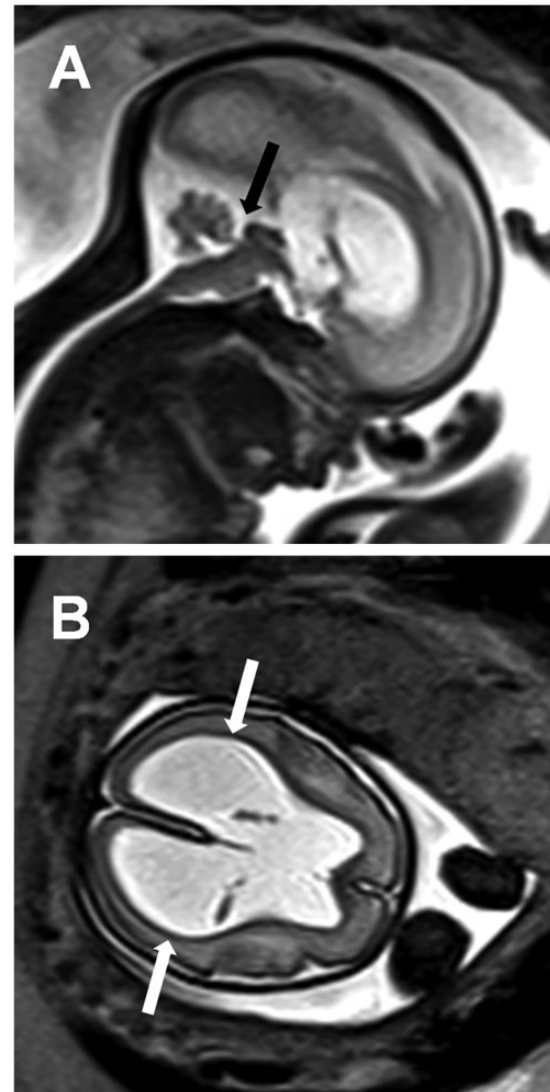


Fig. 2. MRI findings of aqueductal stenosis in a 38-week, 1-day fetus. (a) Sagittal HASTE image displays lack of a normal cerebral aqueduct and normal size of the fourth ventricle in the posterior fossa (black arrow). (b) Axial HASTE image demonstrates marked ventriculomegaly (white arrows).

Chiari 2 malformation

Chiari 2 malformation is characterized by downward translation of the cerebellar tonsils in the setting of a small posterior fossa and open neural tube defect with a lumbar myelomeningocele (Fig. 3). This condition is a common cause of antenatal and neonatal hydrocephalus and occurs in 0.5–1 per 1000 live births (7). MRI can demonstrate inferiorly displaced posterior fossa contents, small cerebellum with mass effect, hydrocephalus, frontal bone concavity, and myelomeningocele. Also better evaluated at MRI are the many associated anomalies which include agenesis of the corpus callosum and gray matter heterotopia. Fetal MRI has been shown to provide

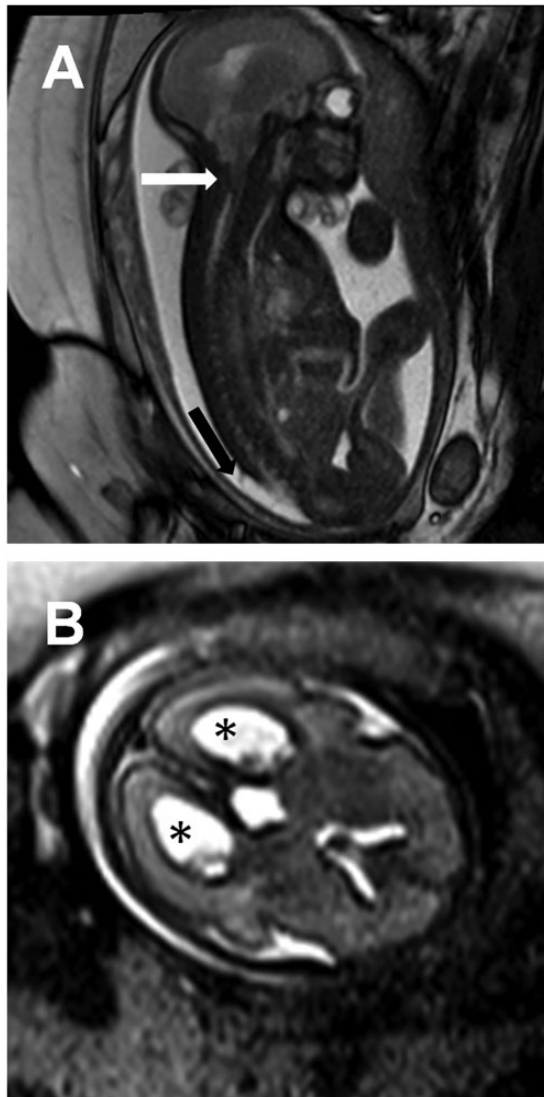


Fig. 3. MRI findings of Chiari 2 malformation in a 26-week, 5-day fetus. (a) Sagittal HASTE image reveals an open neural tube defect (black arrow) and downward translation of the cerebellar tonsils (white arrow). (b) Axial HASTE image shows enlarged ventricular atria (asterisks).

increased sensitivity and specificity compared to US with significant influence on management decisions (8).

Dandy-Walker continuum

Dandy-Walker continuum is a disorder of the posterior fossa with classic DWM representing the most severe end of the spectrum (Fig. 4). In classic DWM, there is complete or partial vermian agenesis, cystic dilatation of the fourth ventricle, large posterior fossa, and elevated torcula. Mild vermian hypoplasia may be considered the least severe end of the continuum. The term “Dandy Walker variant” is vague and thus less utilized. Dandy-Walker continuum disorders are often components of

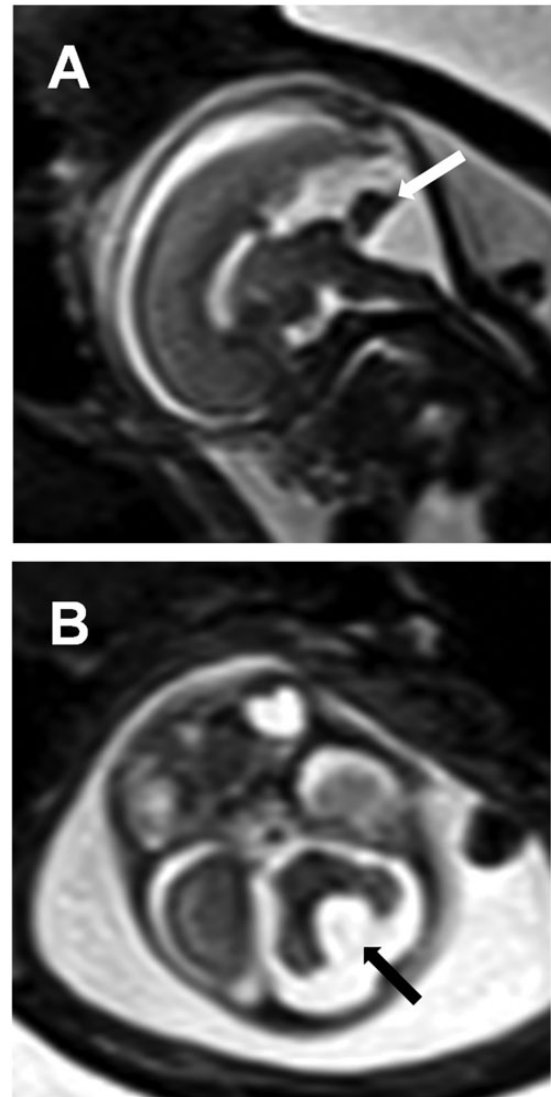


Fig. 4. MRI findings of Dandy-Walker continuum in a 21-week, 5-day fetus. (a) Sagittal HASTE image exhibits a small vermian (white arrow) with expansion of the posterior fossa. (b) Axial HASTE image reveals communication between the fourth ventricle and prominent posterior fossa cerebrospinal fluid (black arrow).

other syndromes (e.g. Walker-Warburg, Meckel-Gruber, etc.). The reported incidence is 1:30,000 (9).

Elements in describing Dandy-Walker continuum disorders in utero include determining the extent of vermian maturity and assessing the tegmento-vermian angle. Differential considerations include persistent Blake pouch cyst and mega cisterna magna, which some consider even milder forms of the continuum, as well as arachnoid cyst (10).

Cephalocele

Cephaloceles occur when there is a calvarial defect with extracranial extension of intracranial structures

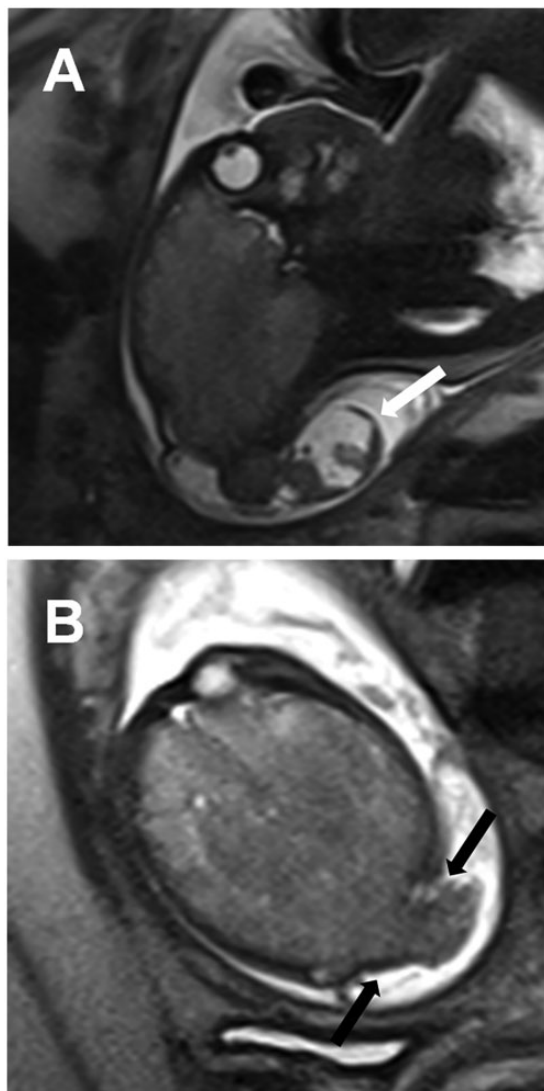


Fig. 5. MRI findings of encephalocele in a 35-week, 1-day fetus. (a) Sagittal HASTE image demonstrates meninges and brain tissue (white arrow) protruding through a posterior calvarial defect. (b) Axial HASTE image exhibits extension of the supratentorial brain through the occipital calvarial defect (black arrows).

covered by skin (Fig. 5). Meningoencephaloceles (extracranial extension of brain, meninges, and cerebrospinal fluid [CSF]) are a common subtype. Cephaloceles account for 10%–20% of dysraphisms, and the estimated prevalence is 0.8–4.0 per 10,000 live births (11). Most cases are sporadic; however, there are genetic and non-genetic syndromes associated with cephaloceles in addition to environmental exposures. The majority (80%) are occipital in location, though parietal and midline frontal cephaloceles have been seen (12).

Prognosis relates to the amount and type of herniated intracranial contents. MRI has proven a useful adjunct to US to evaluate contents of the sac, including

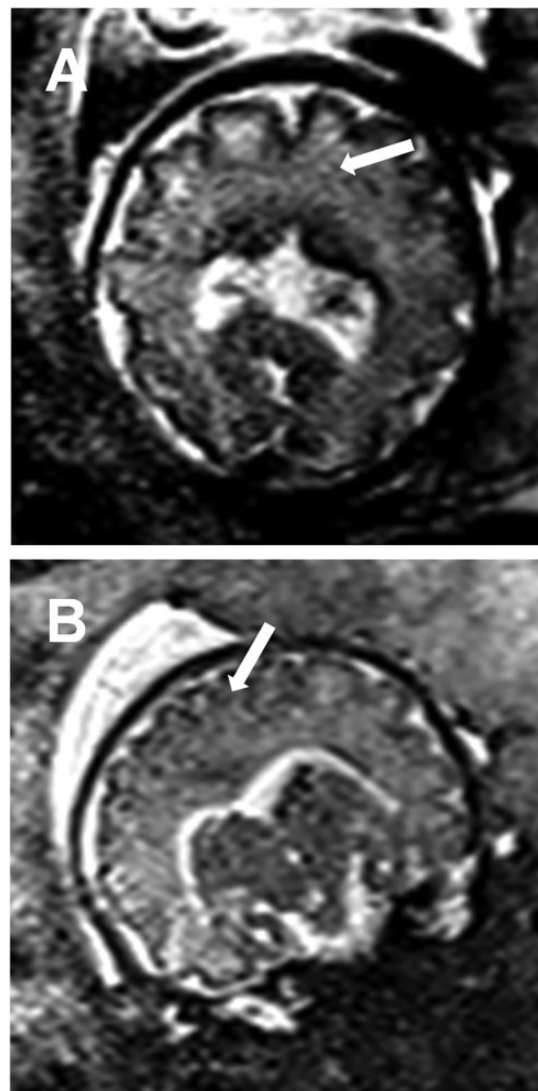


Fig. 6. MRI findings of holoprosencephaly in a 34-week, 2-day fetus. (a) Axial HASTE image reveals fusion of the frontal lobes across the midline (white arrow). (b) Coronal HASTE image also depicts fusion of the frontal lobes across the midline (white arrow).

herniated dural venous sinuses, and to search for associated anomalies, which are present in 50% of cases. Diffusion-weighted imaging should be included to evaluate for ischemia.

Holoprosencephaly

Holoprosencephaly occurs when there is failed or incomplete cleavage of the prosencephalon (Fig. 6). MRI is indicated to define the severity of fusion and associated anomalies. The most severe form is alobar holoprosencephaly, which manifests as a primitive monoventricle, absent interhemispheric fissure, and thalamic fusion. In semilobar holoprosencephaly, the

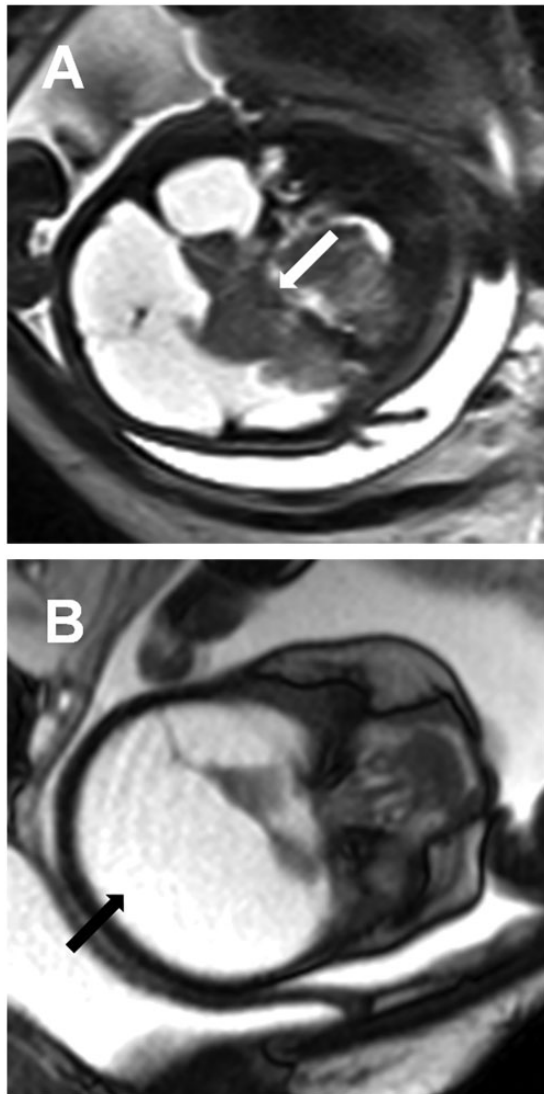


Fig. 7. MRI findings of hydranencephaly in a 31-week, 5-day fetus. (a) Coronal HASTE image reveals intact thalami (white arrow). (b) Axial HASTE image shows near complete absence of the cerebral hemispheres (black arrow).

anterior frontal lobes are fused, but the posterior interhemispheric fissure is present, and there is at least partial fusion of the thalami. Rudimentary frontal horns are present. In lobar holoprosencephaly, only the basal frontal lobes are fused and the thalami are separated. There is also a form known as middle interhemispheric fusion (syntelencephaly) in which the poster frontal and parietal lobes are fused (13).

Holoprosencephaly has a prevalence of approximately 1 in 250 conceptions and an incidence of 1 in 10,000–20,000 live births (14). Multiple genetic mutations are known in this disorder, and many affected patients have an additional syndromic condition (15). Midline facial anomalies are common. In utero diagnosis should prompt karyotyping and genetic counseling.

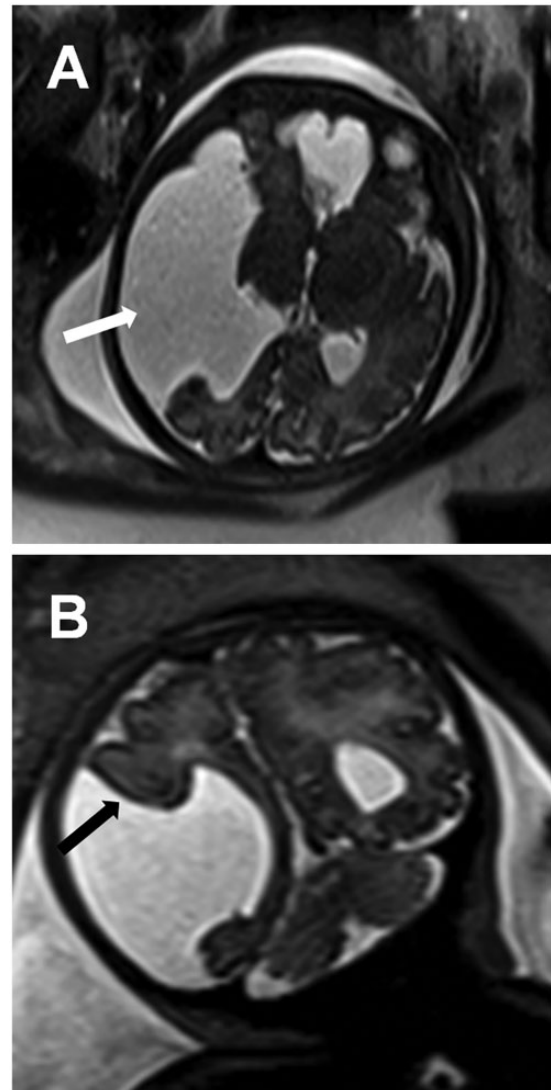


Fig. 8. MRI findings of schizencephaly in a 37-week, 0-day fetus. (a) Axial HASTE image demonstrates prominent cerebrospinal fluid within a cleft in the right cerebral hemisphere which communicates with the right lateral ventricle (white arrow). (b) Coronal HASTE image reveals that the parenchymal cleft is lined by gray matter (black arrow).

Hydranencephaly

Hydranencephaly is reflected by a complete or near absence of cerebral cortex, generally due to circulatory compromise (e.g. anoxia, thrombus, toxin, twin–twin transfusion) (Fig. 7). The posterior fossa structures, thalami, and interhemispheric falx remain intact, and macrocephaly is common due to the persistent production of CSF. The majority of cases are sporadic, and the incidence is 1 in 10,000 births (16). MRI can aid in distinguishing this entity from hydrocephalus, schizencephaly, and holoprosencephaly, which are differential considerations.

Schizencephaly

Schizencephaly refers to a cleft in the brain parenchyma extending from the ependymal surface to the pia mater and is commonly segregated into two types: open-lipped and closed-lipped. Open-lipped schizencephaly refers to a schizencephalic defect in which the cleft walls are separated by a CSF-containing cavity (Fig. 8). The cleft walls are apposed in closed-lipped schizencephaly. The walls of the cleft are lined by gray matter. Schizencephaly is distinguished from encephaloclastic porencephaly in which the cavity is lined by white matter. This can often be discerned by MRI. It is caused by an abnormality of neuronal migration and may be the result of an insult early in development; however, some associations with genetic mutations have been identified.

The incidence of schizencephaly is approximately 1 in 65,000 live births (17). There are associations with other abnormalities including septo-optic dysplasia (17). It can be unilateral or bilateral with the frontal and parietal lobes as the most common sites (18). As is the case with other brain malformations, the extent of symptoms depends on the location and extent of the disease. Seizures, motor disorders, and developmental delay are common.

In conclusion, MRI is an important complementary tool in the evaluation of fetal brain malformations. Precise and accurate diagnoses in these patients is important for treatment planning and parental counseling.

Declaration of conflicting interests


The author(s) declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

Funding

The author(s) received no financial support for the research, authorship, and/or publication of this article.

ORCID iDs

Cory M Pfeifer  <https://orcid.org/0000-0002-2220-9230>

Patricia Cornejo  <https://orcid.org/0000-0001-5096-6047>

References

- Victoria T, Jaramillo D, Roberts TPL, et al. Fetal magnetic resonance imaging: jumping from 1.5 to 3 tesla (preliminary experience). *Pediatr Radiol* 2013;44:376–386.
- Tocchio S, Kline-Fath B, Kanal E, et al. MRI evaluation and safety in the developing brain. *Semin Perinatol* 2015;39:73–104.
- Sans Cortes M, Bargallo N, Arranz, A et al. Feasibility and success rate of a fetal MRI and MR spectroscopy research protocol performed at term using a 3.0-tesla scanner. *Fetal Diagn Ther* 2017;41:127–135.
- Santo S, D'Antonio F, Homfray T, et al. Counseling in fetal medicine: agenesis of the corpus callosum. *Ultrasound Obstet Gynecol* 2012;40:513–521.
- Paul LK, Brown WS, Adolphs R, et al. Agenesis of the corpus callosum: genetic, developmental and functional aspects of connectivity. *Nat Rev Neurosci* 2007;8:287–299.
- Levitsky DB, Mack LA, Nyberg DA, et al. Fetal aqueductal stenosis diagnosed sonographically: how grave is the prognosis? *AJR Am J Roentgenol* 1995;164:725–730.
- Bulas D. Fetal evaluation of spine dysraphism. *Pediatr Radiol* 2010;40:1029–1027.
- Saleem SN, Said AH, Abdel-Raouf M. Fetal MRI in the evaluation of fetuses referred for sonographically suspected neural tube defects (NTDs): impact on diagnosis and management decision. *Neuroradiology* 2009;5:761–772.
- Hirsch JF, Pierre-Kahn A, Renier D, et al. The Dandy-Walker malformation. A review of 40 cases. *J Neurosurg* 1984;61:515–522.
- Tortori-Donati PI, Fondelli MP, Rossi A, et al. Cystic malformations of the posterior cranial fossa originating from a defect of the posterior membranous area. Mega cisterna magna and persisting Blake's pouch: two separate entities. *Childs Nerv Syst* 1996;12:303–308.
- Simpson DA, David DJ, White J. Cephaloceles: treatment, outcome, and antenatal diagnosis. *Neurosurgery* 1984;15:14–21.
- Goldstein RB, LaPisus AS, Filly RA. Fetal cephaloceles: diagnosis with US. *Radiology* 1991;180:803–808.
- Simon EM, Hevner RF, Pinter JD. The middle interhemispheric variant of holoprosencephaly. *AJNR Am J Neuroradiol* 2002;23:151–156.
- Orioli IM, Castilla EE. Epidemiology of holoprosencephaly: prevalence and risk factors. *Am J Med Genet C Semin Med Genet* 2010;154C:13–21.
- Raam MS, Solomon BD, Muenke M. Holoprosencephaly: a guide to diagnosis and clinical management. *Indian Pediatr* 2011;48:457–466.
- Taori KB, Sargar KM, Disawal A, et al. Hydranencephaly associated with cerebellar involvement and bilateral microphthalmia and colobomas. *Pediatr Radiol* 2011;41:270–273.
- Howe DT, Rankin J, Draper ES. Schizencephaly prevalence, prenatal diagnosis and clues to etiology: a register-based study. *Ultrasound Obstet Gynecol* 2012;39:75–82.
- Braga VL, da Costa MDS, Riera R, et al. Schizencephaly: a review of 734 patients. *Pediatr Neurol* 2018;23:29.