Radiologic manifestations in Congenital Brain Anomalies

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Learning objectives

To review the spectrum of radiologic manifestations in congenital brain anomalies. To show the importance and differential diagnosis related to each manifestation.

Background

More than 2000 different congenital brain anomalies have been described in the literature with specific and common radiologic manifestations in each of them. All radiologic modalities can be used for diagnosis of brain anomalies and there are many manifestations in plain radiography, ultrasound, computerized tomography (CT) and magnetic resonance imaging (MRI) for every of brain anomalies.

Imaging findings OR Procedure details

Introduction

More than 2000 different congenital brain anomalies have been described in the literature with specific and common radiologic manifestations in each of them. All radiologic modalities can be used for diagnosis of brain anomalies and there are many manifestations in plain radiography, ultrasound, computerized tomography (CT) and magnetic resonance imaging (MRI) for every of brain anomalies. Neural tube closure defects, disorders of diverticulation and cleavage, neuronal migration and sulcus abnormalities, posterior fossa malformations and neurocutaneous syndromes (phakomatoses) are the famous brain anomalies in children. Some of brain anomalies can be diagnosed by ultrasound in embryonic life like Alobar holoprosencephaly but there are a spectrum of radiologic features in plain radiography, ultrasound, CT and MRI in some of them like chiari II malformation. Specific manifestations can help radiologist to diagnosis certain brain anomalies like Luckenschadel skull, petrous scalloping, cervicomedullary kink, tectal beaking, towering cerebellum, simian appearance, U-shaped curvilinear calcification, molar tooth appearance and etc.

- **Anencephaly**

Anencephalia; congenital defective development of the brain, with absence of the bones of the cranial vault, the cerebral and cerebellar hemispheres, a rudimentary brainstem, and traces of basal ganglia. (Fig1)

- **Cephalocele**
A. Occipital Meningoencephalocele

**Fig2.** Sagittal T1-weighted (a) and axial T2-weighted (b) MR images and MR venogram (c) show an occipital meningoencephalocele with no evidence of involvement of venous structures. The lesion consists predominantly of herniated meninges, but a small amount of brain tissue was found at surgery.

B. Basal (sphenoidal) cephalocele

**Fig3.** Sagittal T1-weighted MR image showing basal (sphenoidal) cephalocele. Note defect in the sphenoid bone, including the sella turcica (large white arrows), with downward herniation of the third ventricle (small white arrows) into the nasal cavity, and absence of the corpus callosum (black arrow).

C. Naso-orbital frontoethmoidal encephalocele

**Fig4.** Naso-orbital frontoethmoidal encephalocele in a 2-year-old patient. (a) Three-dimensional shaded-surface-display image from CT data shows a large, left-sided fronto-orbital mass. (b) Axial CT scan shows a left frontal lobe encephalocele that extends through the ethmoid bone into the orbit. (c) Axial T2-weighted MR image obtained at the same level as b helps confirm the presence of a frontal lobe encephalocele. The contents of the orbital vault were formed normally.

D. Transsphenoidal encephalocele

**Fig5.** Transsphenoidal encephalocele. Coronal T2-weighted MR image shows a transsphenoidal encephalocele anterior to the dorsum sella that projects into the nasopharynx and causes downward displacement of the optic apparatus, hypothalamus, and anterior recess of the third ventricle.

E. Vertically positioned straight sinus with persistent fetal anatomy

**Fig6.** Vertically positioned straight sinus with persistent fetal anatomy. Sagittal T2-weighted MR image (a) and sagittal (b) and coronal (c) MR venograms demonstrate a vertically positioned straight sinus (black arrow in a, arrow in b) and a fenestrated superior sagittal sinus (arrowheads in c) resulting from deflection around the tract of a histologically proved atretic parietal encephalocele (white arrows in a).

- Chiari malformations

A. Chiari I malformation

**Fig7.** Sagittal MR images show cerebellar tonsils greater than 6 mm below the skull base.
B1) LUCKENSCHADEL-SKULL:

Fig8.

Fig9. Deep scalloping between the bony septations that characterize the lacunar skull (luckenschadel) (arrows) are best appreciated on an axial computed tomography section, as in this patient with a Chiari II malformation.

B2) Petrous scalloping

Fig10. Posterior fossa bone findings in a 10-year-old boy. Axial CT scan shows scalloped petrous ridges (arrows) and clivus (arrowhead).

B3) Cervicomedullary kink

Fig11. Sagittal T1-weighted magnetic resonance image of posterior fossa abnormalities in Chiari II malformation: (1) colpocephaly; (2) beaked tectum; (3) cascade of an inferiorly displaced vermis behind the medulla; (4) elongated, tubelike fourth ventricle; (5) low-lying torcular herophili; (6) cerebellar hemispheres wrapping around the brainstem anteriorly; (7) concave clivus; (8) medullary spur; and (9) medullary kink.

B4) Heart-shaped tentorial incisura

Fig12. a) Axial computed tomography scan in a patient with a Chiari II malformation. This image shows a gaping, somewhat heart-shaped tentorial incisura (large arrowheads) that appears to be completely plugged with the upwardly herniating cerebellum. The cerebellar hemispheres extend anteromedially (small arrowheads) and almost completely engulf the brainstem. The petrous ridges are concave (arrows). b) This T1-weighted axial magnetic resonance image demonstrates heart-shaped incisura and a petrous ridge in a patient with a Chiari II malformation.

B5) Large massa intermedia & a beaked tectum

Fig13. Sagittal midline T1-weighted magnetic resonance image in a patient with a Chiari II malformation. This image shows a large massa intermedia (long arrow) and a beaked tectum (short arrow). Other posterior fossa abnormalities are also seen in this patient.

Fig14. Sagittal midline ultrasonogram shows a large massa intermedia (long arrow) and a beaked tectum (short arrow). This image also shows obliteration of the cisterna magna and the fourth ventricle, as well as compression of the pons and brainstem.

B6) Low-lying transverse sinuses & Towering cerebellum

Fig15. Coronal T1-weighted magnetic resonance image in a patient with a Chiari II malformation. This image shows low-lying transverse sinuses (arrows), hydrocephalus,
and a small posterior fossa. A hypoplastic tentorium cerebelli with gaping incisura (arrowhead) is present with a towering cerebellum (small arrows).

**Fig16.** Cerebellum extending above tentorium (“towering cerebellum”).

Elevation of the cerebellum superiorly through the wide incisura results in the formation of a heart-shaped mass.

**B7) Hypoplastic fenestrated falx cerebri with striking interdigitation of the gyri**

**Fig17.** Axial T2-weighted magnetic resonance image in a patient with a Chiari II malformation. This image shows a hypoplastic fenestrated falx cerebri with striking interdigitation of the gyri (arrows).

**B8) Malrotation of the posterior arches of C1 and C2**

**Fig18.** Inferior operativelike view on 3-dimensional computed tomography scan. This image shows malrotation of the posterior arches of C1 (long arrow) and C2 (short arrow).

**C. Chiari III malformation**

**Fig19.** Large herniated sac (*open arrows*) with enlarged portion of fourth ventricle (*arrowheads*), cerebellum (C), and cervical cord (*black arrow*) are seen.

Osseous defects in the infraoccipital area, posterior rim of foramen magnum, and posterior arch of C1 and C2.

**D. Chiari IV**

**Fig20.** Sagittal T1-weighted MR image showing Chiari IV malformation with thin brainstem. The only part of the cerebellum is a small portion of the superior vermis (arrow). With type IV Chiari malformation, cerebellar hypoplasia is present, without inferior displacement.

  - **Cebocephaly**

Cebocephaly: flattened nose with single or paired nostrils between two orbits resembling monkey.

**Fig21.** Frontal skull radiograph shows simian appearance with hypotelorism.

  - **Holoprosencephaly**

**A. Alobar Holoprosencephaly**
Definition: Failure of prosencephalic cleavage resulting in a spectrum of anomalies ranging from septo-optic dysplasia (mildest) to alobar holoprosencephaly (most severe)

What The Referring Physician Needs To Know

- Ultrasound may provide limited information due to several factors
- MRI can miss subtle cases of alobar holoprosencephaly prenatally
- Associated with genetic syndromes such as Meckel-Gruber, Smith-Lemi-Opitz, trisomy 13, trisomy 18, teratogenic exposure
- Ventriculomegaly in association with other anomalies has worse prognosis
- Diagnosis of alobar holoprosencephaly can be made by antenatal ultrasound or fetal MRI; however, postnatal imaging is rarely done because most affected infants are either stillborn or have short life span
- Holoprosencephaly is the only congenital brain anomaly in which posterior portion of corpus callosum has formed in absence of anterior portion

**Fig 22.** There is absence of the interhemispheric fissure.

There is a single crescentic monoventricle, which leads into a dorsal cyst. The thalami and basal ganglia are fused. Multiple small vessels from the internal carotid and basilar arteries constitute the vascular supply.

**Fig 23.** Alobar holoprosencephaly with **large monoventricle (MV)** and small rim of cerebral tissue (**arrow**) anteriorly (**Anterior Mantle**) without cleavage by an interhemispheric fissure

**B. Semilobar Holoprosencephaly**

In semilobar holoprosencephaly, a single ventricle is present, with absence of the septum pellucidum. The interhemispheric fissure and falx cerebri are partially formed in the posterior parts of the brain.

**Fig 24.** A. T1-weighted MR images of semilobar holoprosencephaly. Coronal image of the face showing midline facial cleft (arrows). **B:** T1-weighted MR image of semilobar holoprosencephaly. Axial image of the brain reveals incomplete formation of occipital horns (black arrows) of the monoventricle and lack of cleavage of the anterior half of the brain (white arrow). **C:** T1-weighted MR image of semilobar holoprosencephaly. Coronal image of the brain shows lack of cleavage of frontal lobes (arrow).

**C. Septo-optic dysplasia**

Absence of septum pellucidum and optic nerve hypoplasia. 70% have hypothalamic/pituitary dysfunction.

Radiographic Findings:
Absence of septum pellocidum

Squared frontal horns of lateral ventricles

Hypoplasia of optic nerve and chiasm


- **Agenesis of the Corpus Callosum**

**Fig26.** a. Sagittal T1-weighted, (b) Coronal Flair, (c) Axial T1 MR images show complete absence of the corpus callosum

- **Agenesis of the Corpus Callosum with Midline Lipoma**

**Synonyms**: ACC

**Definition**: Congenital absence of corpus callosum associated with midline lipoma

**What The Referring Physician Needs To Know**

- Associated with syndromes and other malformations (includes Apert’s syndrome, Chiari II malformation, Dandy-Walker malformation, frontonasal dysplasia, median cleft face, Shapiro syndrome, Smith-Lemli-Opitz syndrome, acrocallosal syndrome, Meckel’s syndrome)
- Intracranial lipomas: asymptomatic thus lesions are noted incidentally
- Interhemispheric lipomas in region of corpus callosum are usually associated with some form of callosal dysgenesis
- Outcome is poorer if associated anomalies are detected such as Dandy-Walker malformation, heterotopia, or encephalocele

**Fig27.** Midline amorphous calcification, with radiolucency representing fat Lipoma. Frontal skull radiograph shows **U-shaped curvilinear calcification** with central radiolucency.

- **Interhemispheric Lipoma**

**Fig28.** Coronal ultrasound scan show an **interhemispheric lipoma** in a neonate.

**Fig29.** Sagittal T1-weighted MR image shows **interhemispheric lipoma** and **partial agenesis of the corpus callosum** in a neonate. The MRI appearance is characteristically hyperintense on T1-weighted images **Chemical shift artifact** may be found with large lipomas resulting in extreme hypointensity and hyperintensity.
• **Dandy-Walker malformation**

  Synonyms: Classic Dandy-Walker, Dandy-Walker continuum, Dandy-Walker malformation, Dandy-Walker complex, Hypoplastic vermis with rotation, Blake pouch cyst, Megacisterna magna

  Definition: Heterogeneous group of posterior fossa cystic malformations with vermian/cerebellar hypoplasia

  **What The Referring Physician Needs To Know**

  - Severity of symptoms and prognosis correlate with associated supratentorial abnormalities, vermian lobulation, and degree of hydrocephalus
  - Overlap between developmental malformations and acquired disorders may occur when external agent interferes with evolving brain development
  - Many central nervous system malformations can be diagnosed in utero; use ultrasonography primarily, then MRI

  1. **Large posterior fossa**

     **Fig30.** Lateral skull radiograph shows a **large posterior fossa** with elevated internal occipital protuberance.

  2. **Dandy-Walker cyst**

     **Fig31.**a) Axial T1-weighted MR image shows a **large posterior fossa cyst** (c) compressing the cerebellar hemispheres *(arrows).* b) Sagittal T1-weighted MR image showing **Dandy-Walker cyst** *(large arrows)* with **absence of the inferior vermis** and a small remnant of the superior vermis *(small arrow)* present. c) Dandy-Walker cyst. Axial noncontrast head CT scan shows **Dandy-Walker cyst** with associated **agensis of the corpus callosum**

  3) **Dandy-Walker variant**

     Dandy-Walker variant is a **mild form** of Dandy-Walker malformation. **Vermian hypoplasia** is less severe and best evaluated by MRI.

     **Fig32.** a) Sagittal T1-weighted MR image showing **Dandy-Walker variant** with a **fourth ventricle** communicating with a **retrocerebellar cyst** *(white arrow), absence** of the inferior vermis, **absence** of the corpus callosum *(black arrow), and lissencephaly.* b) Axial T1-weighted MR image showing **Dandy-Walker variant** with a fourth ventricle *(4), leading into a retrocerebellar cyst *(large arrows)*, and absence of the inferior vermis *(small arrows).*

  4) **Mega cisterna magna**
On imaging, **mega cisterna magna** appears as a fluid collection in the retrocerebellar space. There may be **scalloping** of the inner table of the occipital bone. **The vermis** is **normal** in size and orientation.

**Fig33.** Axial noncontrast CT scan shows a large retrocerebellar cerebrospinal fluid collection.

- **Trapped fourth ventricle**

Occlusion of **the aqueduct of Sylvius** and **the foramina of Luschka and Magendie** results in dilation of the fourth ventricle, which becomes isolated from the ventricular system and circulation of CSF.

**Fig34.** Axial noncontrast CT scan shows significant enlargement of the fourth ventricle. There was a previous history of supratentorial ventriculoperitoneal shunt placement.

- **Joubert's syndrome**

**Fig35.** Axial noncontrast T1-weighted MR image shows a narrow isthmus and large superior cerebellar peduncles (molar tooth appearance of the midbrain).

The "**molar tooth**" appearance of the midbrain on axial MR images is characteristic.

The diagnosis of Joubert syndrome is based on the presence of characteristic clinical features and the "molar tooth sign" on cranial magnetic resonance imaging (MRI), resulting from hypoplasia of the cerebellar vermis and accompanying brainstem abnormalities on axial imaging through the junction of the midbrain and pons (isthmus region).

- **Schizencephaly**

**A.Type 1 Schizencephaly**

MRI is the modality of choice for establishing the diagnosis. The clefts can be identified in multiple planes. **Closed lip types** may not be diagnosed if the imaging plane is parallel to the cleft.

**Fig36.** Axial T1-weighted MR image showing type 1 schizencephaly with gray matter clefts (arrows) extending from the lateral ventricles to the cortex

**B.Type II Schizencephaly**

**Fig37.** Axial T1-weighted MR image showing type II schizencephaly with gray matter-lined clefts (arrows) separated by large cerebrospinal fluid spaces

- **lissencephaly, Total agyria**
MRI is the modality of choice to show characteristic features. The smooth brain with absent or a few broad gyri is well shown, especially in the sagittal plane.

**Fig38. a)** Axial T1-weighted MR images showing lissencephaly, Total agyria.

**b)** Sagittal T1-weighted MR image shows total agyria with **marked increased gray matter**.

- **Pachygyria**

  *Pachygyria* refers to gyri that are focally or diffusely thick, broad and flat, and associated with a thick cortex. **Dysmorphic facies** occur in children with pachygyria unrelated to any syndrome.

**Fig39.** Coronal T1-weighted MR image showing multiple areas of pachygyria (*arrows*).

- **Polymicrogyria**

  It may be unilateral (40%) or bilateral (60%). The cortex surrounding the sylvian fissure is involved in approximately 80% of cases. The frontal lobe is most commonly involved.

**Fig40.** Sagittal T1-weighted MR image showing polymicrogyria that involves the frontal lobe with small irregular gyri (*arrows*) and increased gray matter.

- **Heterotopic gray matter**

  Most patients have heterotopic neurons that are asymmetric and few in number. These are periventricular, mainly located in the trigones and temporal and occipital horns of the lateral ventricles.

**Fig41.** Axial T1-weighted show multiple heterotopic gray matter nodules.

- **Hemimegalencephaly**

  The involved area is enlarged to a varying extent on CT and MRI. The gray-white matter is poorly differentiated. Gray matter heterotopia is common. The ipsilateral lateral ventricle is enlarged.

**Fig42.** Axial T1-weighted MR image showing hemimegalencephaly of the left calvaria and cerebral hemisphere with abnormal pachygyric-appearing cortex (*arrows*).

- **Hydranencephaly**

  CT and MRI show a fluid-filled cranium with the falx cerebri and tentorium. The falx may be midline or deviated, but is usually not thickened. Remnants of the inferomedial and inferofrontal lobes may be present. The brainstem is atrophic.
**Fig43.** Axial noncontrast CT scan showing dilated cerebrospinal fluid sac without intervening cerebral mantle. **Note** preservation of thalamic and posterior fossa structures.

- **Tuberous sclerosis**

**Fig44.**
a) Lateral skull radiograph shows multiple calcific lesions. 
b) Axial T1-weighted MR image shows right frontal cortical tubers. 
c) Axial T1-weighted MR image shows numerous **subependymal tubers** and **hyperintense cortical hamartomas**. 
d) Axial T1-weighted MR image show large mass in region of foramen of Monro representing a **giant cell astrocytoma**. **Note** shunt artifact. Shunt was placed because of obstructive hydrocephalus.

**Giant cell tumors** are enlarging subependymal nodules that usually arise near the foramen of Monro, attached to the adjacent caudate nucleus. They can occur anywhere, however, along the ependymal surface.

- **Neurofibroma**

**A. Neurofibromatosis type 1**

Optic nerve tumors are the **most common** brain tumors encountered in NF1, with an incidence of 1.5% to 15%. They arise exclusively in young children, with a median age of 4 to 9 years.

**Fig45.**
a) Image of the orbits obtained with inversion recovery sequence showing **bilateral optic nerve gliomas**. 
b) Axial T2-weighted MR image of **neurofibromatosis type 1** with buphthalmos of the right orbit, **dysplasia** of the anterior wing of the sphenoid (**black arrow**), and of the temporal area of the scalp (**white arrows**). 
c) Axial contrast-enhanced CT scan shows contrast-enhancing **optic glioma** involving the **optic chiasm**. **Note** calcification of the choroid plexus in the temporal horns bilaterally, indicative of neurofibromatosis. 
d) Axial T2-weighted MR image showing bilateral hyperintense lesions in basal ganglia representing **vacuolar myelination**.

Cerebral abnormalities include myelin vacuolization and neurofibromatosis bright objects. Foci of hyperintensity are found on T2-weighted or FLAIR images in 43% to 93% of children with NF1.

**B. Neurofibromatosis type 2**

Virtually all patients with NF2 develop bilateral vestibular schwannomas. There also may be involvement of other cranial nerves, especially cranial nerve V and less commonly the cranial nerves IX and X, spinal and peripheral nerves.
**Fig 46.** *a)* Axial contrast-enhanced T1-weighted MR image shows bilateral acoustic neuromas. *b)* Non-contrast (above) and contrast-enhanced (below) T1-weighted images show an **intraventricular meningioma.**

- **Sturge-Weber syndrome**

Cortical calcification between layers two and four has a **characteristic serpentine "tram track" gyral pattern.** This is best shown on noncontrast CT. It is most dense in the **occipital region** and may extend into the parietal lobe.

**Fig 47.** *a)* Lateral skull radiograph shows parietal serpentine calcification. *b)* Axial noncontrast CT scan shows serpentine calcification within right cerebral hemisphere, which is decreased in size. *c)* Contrast-enhanced T1-weighted MR images show **enhancement of pial angioma.**

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Conclusion

It is important that radiologists recognize the manifestations of congenital brain anomalies which will allow confident diagnosis on the basis of imaging findings alone or narrowing of the differential diagnosis.

Personal Information

References


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