Prenatal MR Imaging in Fetal Cerebellar Abnormalities

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Posterior fossa malformations are a common indication for fetal MRI. Despite recent advances in both US and MRI, false-positive and false-negative prenatal diagnoses still occur.

**Main indications for fetal MRI:**
1. Abnormal US findings (biometry, cisterna magna increase >10 mm)
2. Feto-maternal infection (toxoplasmosis, CMV, HIV,)
3. Family history of genetic disorders (eg, pontocerebellar hypoplasia)
4. High-risk pregnancy (deceased twin, coagulopathy, hypoxia, trauma)
5. Presence of fetal malformations possibly associated with cerebellar abnormalities (eg, renal, digital, ocular)

**In general, these abnormalities may be approached as:**
- Abnormalities with normal or increased posterior fossa size
- Abnormalities with small posterior fossa (Chiari malformation)
The cerebellum is one of the first structures originating in utero.

The long pre- and postnatal development makes it vulnerable.

The cerebellar primordium differentiates between 28-32 days of gestation at the isthmus, in the dorsal neural tube, between midbrain and hindbrain.

The middle and anterior portions of the primordium result in the vermis, whereas the lateral and posterior portions lead to the hemispheres.

The posterior membranous area invaginates caudal to the vermis in the primitive meninx, forming a diverticulum (Blake pouch), whose inner and outer layers are comprised of ependyma and pia-arachnoid, respectively.

- Blake pouch fenestrates in the midline at 7-8 weeks, forming the **foramen of Magendie**
- Blake pouch fenestration in the lateral portion around 14-17 weeks leads to the **foramen of Lushka**
- Growth of the caudal portion of the vermis leads to progressive closure of the fourth ventricle, usually concluding towards 18 weeks’ pregnancy.

- As the limit, the fourth ventricle should be completely closed around 22-24 weeks’ gestation.
ANATOMIC COMMENTS ON THE NORMAL FETAL CEREBELLMUM

- Primary fissure visible on MR at 28-30 wks
- Other fissures are not so constantly visualized at 30-32 wks' gestation
- Cranio-caudal diameter (perpendicular to the fastigium/decline)
- Superior vermis/Inferior vermis 1:2

- In common practice, the evaluation of the vermis is based on both biometric and morphologic data
- The primary fissure delineates the anterior and the posterior lobes, the latter being twice as large as the former

### BIOMETRY: VERMIS CRANIO-CAUDAL DIAMETER

<table>
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<th>Gestational age (weeks)</th>
<th>CC Diameter (mms)</th>
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**TEGMENTAL-VERMIAN ANGLE**

- Calculated as the angle formed by drawing a line along the dorsal aspect of the brainstem and another line along the ventral vermis.
- An angle near 0° is considered normal.
- Definitely abnormal if >40°.

- Prenatal diagnosis of posterior fossa dysgenesis is still associated with false positives and false negatives.
- Recent reports show limitations of MR in sensitivity and specificity, with only 60% of prenatal Dx being confirmed by postnatal MRI.
- Early gestational age is the most decisive factor in the discrepancy between pre- and postnatal RMs.

**Megacisterna magna (34 wks)**

- **Megacisterna magna** might be a normal variant (enlarged cisterna magna with normal cerebellar structures)
- **BUT** the presence of a cisterna magna of more than 10 mm raises the possibility of an anomaly such as Dandy Walker malformation, cerebellar hypoplasia or posterior fossa arachnoid cyst

- Late closure (Blake pouch cyst)
- Vermian hypoplasia
- Vermian agenesis
- Dandy-Walker malformation
- Posterior fossa arachnoid cyst
- “Molar tooth”-related syndromes
- Pontocerebellar hypoplasia
- Rhombencephalosynapsis
Twin gestation

At 22 wks abnormal vermis is seen in fetus 2 that normalizes in follow-up MRI at 30 wks
- **Blake pouch cyst** may be misdiagnosed with a partial vermian agenesis.
- Midsagittal plane is crucial depicting the upward displaced otherwise normal vermis.
Suspected Gorlin syndrome

22 wks  Mother had Gorlin syndrome
MRI at 31 weeks.

Blake pouch remnant.

Follow-up MRI at 5 days postnatal age.

Right perisylvian polymicrogyria.
Vermian agenesis
Severe cerebellar hypoplasia or agenesis

Prenatal MRI. 29 wks

Postnatal MRI. 21 months
Vermian hypoplasia may be partial or complete. The tentorium cerebelli has a normal insertion with normal size of the posterior fossa.
Vermian hypoplasia associated with corpus callosum dysgenesis (20 wks)
Final diagnosis was Wolf-Hirschorn syndrome (4p-)

- Cerebellar hypoplasia may involve hemispheres, vermis or both
- Diagnosis is based on biometric evaluation of the different parts of the cerebellum
- Cerebellar hypoplasia may be observed in the setting of aneuploidy
Cerebellar hypoplasia, CC dysgenesis & heterotopia
Cerebellar hypoplasia, CC dysgenesis & heterotopias

- Other brain malformations
- Infection (cytomegalovirus)
- Metabolic disorders and syndromes (eg, PHACE syndrome)

- corpus callosum dysgenesis
- gray matter heterotopias
- brainstem hypoplasia

Prenatal MRI (22 wks)
PHACE syndrome

- In this case, the right cerebellar hemisphere hypoplasia was diagnosed prenatally
- During the postnatal follow-up, multiple infantile head and neck hemangiomas developed

PHACE syndrome
(Posterior fossa abnormalities, Hemangioma, Arterial lesions, Cardiac abnormalities/aortic coarctation, and Eye anomalies)

Prenatal MRI (34 wks)

Postnatal MRI
DD: Cerebellar hemorrhage

RH isoimmunization
Prenatal (24 s)

Prenatal (28 s)
Postnatal (21 days)
Dandy-Walker malformation

Prenatal MRI (32 wks)

- Elevation of the tentorium cerebelli
- Increased posterior fossa volume
- Vermian dysgenesis

Postnatal MRI
DD: Posterior fossa arachnoid cyst

Prenatal MRI (37 wks)

- Increased posterior fossa volume
- NORMAL vermis and hemispheres (may be compressed)

Postnatal MRI (6 d)
DD: Posterior fossa arachnoid cyst

Prenatal MRI (31 wks)

Postnatal MRI (3 months)
Ponto-cerebellar hypoplasia

Suspected lissencephaly 2
(34 wks)
Rombencephalosynapsis

- Characterized by fusion of the cerebellar hemispheres with absent vermis
- On axial view there is an absent posterior clefting between the cerebellar hemispheres, being round, and the cerebellar folia may traverse the midline
- Ventriculomegaly is usually associated
Rombencephalosynapsis

Prenatal US and MRI (21 wks)
Partial rhombencephalosynapsis

- Prenatal (22 wks)
- Prenatal (31 wks)
- Postnatal (5 days)
- Prenatal US correctly identifies the vertebral and CNS anomalies
- Although complementary, fetal MRI provides data influencing prognosis
- Biochemical test to detect alpha-fetoprotein in amniotic fluid is useful only in open dysraphism, not in the closed condition

**SMALL POSTERIOR FOSSA: CHIARI II MALFORMATION**

Prenatal MRI (35 wks)
Fetal intervention may improve hydrocephalus and hindbrain herniation associated with Chiari II malformation and reduce the need for VP shunting.
Prenatal post-fetal surgery 28 s
Close dysraphism may be confused with open dysraphism. These abnormalities are not associated with Chiari II malformation, have better prognosis than open forms and are not candidates for prenatal surgery. Therefore, their recognition is critical for appropriate advice and management.


