International Journal of Clinical Obstetrics and Gynaecology

ISSN (P): 2522-6614
ISSN (E): 2522-6622

www.gynaecologyjournal.com
2025; 9(2): 110-112
Received: 03-02-2025
Accepted: 08-03-2025

Montacer Hafsi
Departement of Gynecology and Obstetrics Menzel, Temim, Nabeul, Tunisia

Houssem Ragmoun
Departement of Gynecology and Obstetrics Menzel, Temim, Nabeul, Tunisia

Sarra Rihani
Departement of Gynecology and Obstetrics Menzel, Temim, Nabeul, Tunisia

Eya Kristou
Departement of Gynecology and Obstetrics Menzel, Temim, Nabeul, Tunisia

Amina Abaab
Departement of Gynecology and Obstetrics Menzel, Temim, Nabeul, Tunisia

Meriem Bezzine
Departement of Gynecology and Obstetrics Menzel, Temim, Nabeul, Tunisia

Arina Jbari
Departement of Gynecology and Obstetrics Menzel, Temim, Nabeul, Tunisia

Achref Ouadday
Departement of Gynecology and Obstetrics Menzel, Temim, Nabeul, Tunisia

Ikram Ben Abdallah
Departement of Gynecology and Obstetrics Menzel, Temim, Nabeul, Tunisia

Elaa Sassi
Departement of Gynecology and Obstetrics Menzel, Temim, Nabeul, Tunisia

Meriem Rahmani
Departement of Gynecology and Obstetrics Menzel, Temim, Nabeul, Tunisia

Sawssen Fenni
Departement of Gynecology and Obstetrics Menzel, Temim, Nabeul, Tunisia

Corresponding Author:
Montacer Hafsi
Departement of Gynecology and Obstetrics Menzel, Temim, Nabeul, Tunisia

Cerebellar hypoplasia: Diagnosis and etiological considerations: A case report

Montacer Hafsi, Houssem Ragmoun, Sarra Rihani, Eya Kristou, Amina Abaab, Meriem Bezzine, Arina Jbari, Achref Ouadday, Ikram Ben Abdallah, Elaa Sassi, Meriem Rahmani and Sawssen Fenni

DOI: https://www.doi.org/10.33545/gynae.2025.v9.i2b.1604

Abstract
Introduction: Cerebellar hypoplasia (CH) is a rare developmental abnormality of the posterior fossa, characterized by underdevelopment of the cerebellum. Its prenatal detection is critical for prognostic and genetic counseling, as it may be isolated or associated with a wide spectrum of syndromes and chromosomal anomalies.
Methods: We report the case of a fetus evaluated during the second trimester for suspected posterior fossa abnormalities. Standard obstetric ultrasound and targeted neurosonography were performed, followed by fetal MRI for further clarification.
Results: Ultrasound revealed a small posterior fossa with reduced cerebellar volume, particularly involving the vermis. The transverse cerebellar diameter was below the 5th percentile for gestational age. The cisterna magna was preserved. Fetal MRI confirmed cerebellar hypoplasia with intact brainstem structures and no supratentorial anomalies. No extracranial malformations were noted. Amniocentesis was performed, and chromosomal microarray was normal. TORCH screening was negative. The diagnosis of isolated cerebellar hypoplasia was retained.
Discussion: Cerebellar hypoplasia can result from genetic mutations, congenital infections (such as CMV), vascular insults, or metabolic disorders. When isolated, prognosis is variable and depends on the degree of hypoplasia and postnatal development. Detailed imaging and exclusion of infectious and genetic causes are crucial for prenatal counseling.
Conclusion: This case illustrates the importance of a systematic approach in evaluating posterior fossa abnormalities. Accurate diagnosis and etiological exploration of cerebellar hypoplasia enable appropriate counseling and guide perinatal management decisions.

Keywords: Cerebellar hypoplasia, posterior fossa abnormalities, fetal neurosonography

Introduction
Cerebellar hypoplasia (CH) is a rare developmental anomaly characterized by incomplete growth of the cerebellum, either affecting the entire structure or localized to specific lobes or the vermis. Prenatal diagnosis has become more frequent due to improvements in neurosonography and fetal MRI. However, interpretation of cerebellar biometry and distinguishing between hypoplasia and other posterior fossa malformations (such as Dandy-Walker malformation or vermian agenesis) remains challenging.
CH may present as an isolated finding or be part of complex syndromes. Identifying its etiology-genetic, infectious, metabolic, or vascular-is essential for assessing prognosis and recurrence risk.
We present a case of prenatally diagnosed isolated cerebellar hypoplasia, discussing the diagnostic approach and differential diagnoses.

Case Presentation
A 30-year-old primigravida with no significant personal or family medical history was referred to our prenatal diagnostic unit at 22 weeks of gestation following suspicion of a posterior fossa abnormality identified during routine second-trimester ultrasound. The initial scan revealed a reduced transverse cerebellar diameter measuring 19 mm, which was below the 5th percentile for gestational age. The cerebellar vermis appeared present but notably small, raising concerns for cerebellar hypoplasia. The cisterna magna measured within normal limits, and there were no

signs of hydrocephalus or enlargement of the lateral ventricles. The supratentorial structures appeared normal, and no extracranial malformations were observed at that time.

To further evaluate the suspected anomaly, a targeted fetal neurosonographic assessment was conducted using both axial and midsagittal views of the fetal brain. This examination confirmed hypoplasia of the cerebellar vermis, with reduced height and surface area. The fourth ventricle was visualized and appeared anatomically normal, without evidence of upward rotation or distortion. The brainstem was intact and exhibited a normal appearance and continuity with adjacent structures. While the cerebellar hemispheres were present bilaterally, they were slightly reduced in volume, consistent with a global cerebellar hypoplasia. No abnormalities were detected in the cerebral hemispheres or other intracranial compartments.

Given the isolated nature of the cerebellar findings, an etiological work-up was initiated. Maternal serologic screening for TORCH infections, including cytomegalovirus (CMV) and toxoplasmosis, returned negative, effectively ruling out the most common congenital infectious causes of posterior fossa anomalies. An amniocentesis was performed at 23 weeks of gestation, and chromosomal microarray analysis revealed a normal female karyotype with no pathogenic copy number variations. The couple denied consanguinity, and there was no known history of genetic or neurologic disorders in either family.

Following a multidisciplinary consultation involving specialists in maternal-fetal medicine, clinical genetics, and pediatric neurology, the diagnosis of isolated cerebellar hypoplasia of uncertain etiology was made. The parents were counseled about the findings, the current limitations of prenatal imaging in predicting neurodevelopmental outcomes, and the potential range of postnatal prognoses. They were also informed of the importance of long-term neurological follow-up after birth. Serial ultrasounds were scheduled to monitor cerebellar growth and overall fetal development, which remained stable for the duration of the pregnancy.

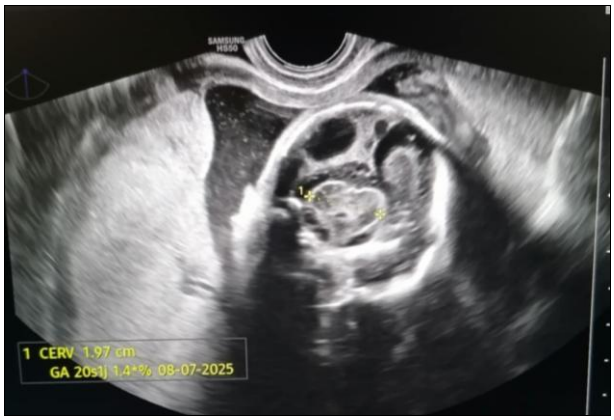

Fig 1: Reduced transverse cerebellar diameter - Cerebellar Hypoplasia

Discussion

Cerebellar hypoplasia (CH) represents a heterogeneous group of malformations that can be classified based on:

- Anatomic distribution (focal vs. diffuse) [1]
- Temporal progression (static vs. progressive) [2]
- Clinical context (isolated vs. syndromic) [3]

Prenatal Diagnostic Challenges

The prenatal differentiation of CH from other posterior fossa anomalies is critical for prognostic counseling. Key differential diagnoses include:

1. **Dandy-Walker malformation (DWM):** Characterized by an enlarged cisterna magna, vermis rotation, and cystic dilatation of the 4th ventricle [4]. In this case, the preserved cisterna magna and normal fourth ventricle excluded DWM.
2. **Vermian agenesis/dysplasia:** May present with partial or complete absence of the vermis but without the cystic posterior fossa enlargement seen in DWM [5].
3. **Pontocerebellar hypoplasia (PCH):** A progressive neurodegenerative disorder with small pons and cerebellum, often associated with severe neurodevelopmental impairment [6].

Advanced neurosonography and fetal MRI improve diagnostic accuracy by assessing vermian foliation, brainstem anatomy, and associated supratentorial anomalies [7].

Etiologic Spectrum

CH arises from diverse pathogenic mechanisms:

- **Genetic causes (40-50% of cases)**
  - Recessive syndromes (e.g., *Joubert syndrome* [*TMEM67*, *CEP290* mutations], *PCH* [*TSEN54*, *EXOSC3* mutations]) [8, 9].
  - Chromosomal anomalies (trisomy 18, 13, or 9q deletions) [10].
- **Prenatal infections (15-20%): CMV** (most common), toxoplasmosis, or Zika virus, which disrupt cerebellar granule cell migration [11].
- **Vascular insults:** Ischemic strokes (e.g., from twin demise in monochorionic pregnancies) or hemorrhages (e.g., fetal thrombocytopenia) [12].
- **Metabolic/mitochondrial disorders:** Rare prenatally but should be considered in progressive CH with elevated lactate or abnormal MR spectroscopy [13].

Neurodevelopmental Outcomes

- **Isolated CH:** Outcomes range from normal cognition to mild motor/speech delays, particularly with unilateral or focal involvement [14].
- **Syndromic CH:** Poor prognosis, especially in PCH or Joubert syndrome, with high rates of intellectual disability, epilepsy, and respiratory failure [15].

Postnatal evaluation must include

- 3T MRI (superior for assessing foliation and brainstem anatomy) [16].
- Genetic testing (karyotype, CMA, and exome sequencing) [17].
- Long-term neurodevelopmental follow-up to monitor for ataxia, hypotonia, or autism spectrum traits [18].

Conclusion

This case highlights the value of combining ultrasound, fetal MRI, and etiological investigations in the prenatal assessment of cerebellar hypoplasia. Accurate diagnosis, even in isolated cases, allows for personalized counseling and informed decision-making. A multidisciplinary approach is essential to guide management and anticipate potential outcomes.

Consent for publication

Written informed consent was obtained from the patient for publication and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of

this journal on request.

Availability of supporting data
Not applicable

Acknowledgements
None

Ethical approval
Not applicable. Our institution requires no ethical approval for case reports

Competing interests
All authors declare that they have no conflicts of interest.

Funding
Not applicable.
All authors read and approved the final manuscript.

References

1. Poretti A, Boltshauser E, Doherty D. Cerebellar hypoplasia: differential diagnosis and diagnostic approach. Am J Med Genet C Semin Med Genet. 2014;166C(2):211-226.
2. Namavar Y, Barth PG, Poll-The BT, Baas F. Classification, diagnosis and potential mechanisms in pontocerebellar hypoplasia. Orphanet J Rare Dis. 2011;6:50.
3. Parisi MA, Dobyns WB. Human malformations of the cerebellum. Handb Clin Neurol. 2018;155:85-101.
4. Bosemani T, Orman G, Boltshauser E, Tekes A, Huisman TA, Poretti A. Congenital abnormalities of the posterior fossa. Radiographics. 2015;35(1):200-220.
5. Adamsbaum C, Moutard ML, André C, *et al*. MRI of the fetal posterior fossa. Pediatr Radiol. 2005;35(2):124-140.
6. Rudnik-Schöneborn S, Barth PG, Zerres K. Pontocerebellar hypoplasia. Am J Med Genet C Semin Med Genet. 2014;166C(2):173-183.
7. Malinger G, Lev D, Lerman-Sagie T. The fetal cerebellum. Prenat Diagn. 2009;29(4):312-320.
8. Bachmann-Gagescu R, Dempsey JC, Phelps IG, *et al*. Joubert syndrome: A model for untangling recessive disorders with extreme genetic heterogeneity. J Med Genet. 2015;52(8):514-522.
9. Graham JM, Spencer AH, Grinberg I, *et al*. Molecular and neuroimaging findings in pontocerebellar hypoplasia type 2 (PCH2): is prenatal diagnosis possible? Am J Med Genet A. 2010;152A(9):2268-2276.
10. Sztriha L, Al-Gazali LI, Wanders RJ, *et al*. Abnormalities of the fetal cerebellum in a case of maternal diabetes mellitus: a case report. Neuropediatrics. 2003;34(1):38-41.
11. Fink KR, Thapa MM, Ishak GE, Pruthi S. Neuroimaging of pediatric central nervous system cytomegalovirus infection. Radiographics. 2010;30(7):1779-1796.
12. Ghi T, Simonazzi G, Perolo A, *et al*. Outcome of antenatally diagnosed intracranial hemorrhage: case series and review of the literature. Ultrasound Obstet Gynecol. 2003;22(2):121-130.
13. van der Knaap MS, Valk J, de Neeling N, Nauta JJ. Pattern recognition in magnetic resonance imaging of white matter disorders in children and young adults. Neuroradiology. 1991;33(6):478-493.
14. Limperopoulos C, Robertson RL, Estroff JA, *et al*. Diagnosis of inferior vermian hypoplasia by fetal MRI: potential pitfalls and neurodevelopmental outcome. Am J Obstet Gynecol. 2006;194(4):1070-1076.
15. Romani M, Micalizzi A, Valente EM. Joubert syndrome: congenital cerebellar ataxia with the molar tooth. Lancet Neurol. 2013;12(9):894-905.
16. Aldinger KA, Doherty D. The genetics of cerebellar malformations. Semin Fetal Neonatal Med. 2016;21(5):321-332.
17. Parrini E, Conti V, Dobyns WB, Guerrini R. Genetic basis of brain malformations. Mol Syndromol. 2016;7(4):220-233.
18. Bolduc ME, Limperopoulos C. Neurodevelopmental outcomes in children with cerebellar malformations: A systematic review. Dev Med Child Neurol. 2009;51(4):256-267.

How to Cite This Article
Hafsi M, Ragmoun H, Rihani S, Kristou E, Abaab A, Bezzine M, *et al*. Cerebellar hypoplasia: Diagnosis and etiological considerations: A case report. International Journal of Clinical Obstetrics and Gynaecology. 2025;9(2):110-112.