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JSM Pediatric Neurology

Case Report

Cerebellar Agenesis with developmental delay case report: -Negelle Arsi General Hospital and Medical College, Ethiopia

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Submitted: 26 March 2025

Accepted: 11 October 2025

Published: 13 October 2025

Keywords
- Ethiopia
- Negelle Arsi
- Developmental Delay
- Cerebellar Agenesis

Abstract

Background: Cerebellar agenesis (CA) is a rare congenital disorder characterized by the complete absence of the cerebellum, leading to severe motor and cognitive impairments. Its varied symptoms often mimic other neurological conditions, making diagnosis challenging, especially in resource-limited settings like Ethiopia. This highlights the need for greater awareness and improved diagnostics for better management.

Case: An 8-year-old girl presented with a history of inability to walk since birth. She was delivered at home and hospitalized for two days shortly after birth. Her developmental milestones were significantly delayed; she achieved head control at 3 years of age and began speaking at 4 years. Her vaccinations were up to date.

On physical examination, she appeared healthy, with stable vital signs and good nutritional status. There were no abnormalities in the chest, and her heart sounds (S1 and S2) were normal, with no murmurs or gallops. The abdomen was soft and non-tender. Neurological examination revealed a normal mental status, with a Glasgow Coma Scale (GCS) score of 15/15. However, spastic tone and muscle atrophy were noted in her lower extremities. Despite this, motor strength was preserved at 5/5 in all extremities. The combination of her clinical presentation and developmental history pointed to an underlying neurological condition, requiring further investigation.

Conclusion: Early recognition of neurological disorders in children with developmental delays and timely evaluation and imaging are essential to diagnosis rare abnormalities such as Cerebellar agenesis. Early diagnosis can guide appropriate management, im prove outcomes, and prevent further complications.

INTRODUCTION

Cerebellar agenesis is a very rare condition characterized by the complete absence of cerebellar tissue [1-3]. Cerebellar agenesis results from developmental malformation or prenatal/perinatal disruption, though its exact cause remains unclear [2-4]. Age at the time of diagnosis of cerebellar agenesis has been from a few weeks of life and up to 71 years of age [1].

Cerebellar agenesis presents with highly variable symptoms, ranging from no noticeable effects to significant motor and cognitive impairments [1-5]. Individuals with cerebellar agenesis may experience mild to moderate motor issues, such as ataxia, dysarthria, and delayed motor development [1-4]. In severe cases, cerebellar agenesis can lead to significant motor and cognitive impairments, including intellectual disability, epilepsy, and hydrocephalus. Some cases are discovered incidentally during imaging for unrelated conditions or at autopsy [3-6].

Imaging, particularly magnetic resonance imaging (MRI), plays a key role in diagnosing cerebellar agenesis [1-6]. In cerebellar agenesis, imaging studies, reveal the absence of cerebellar tissue within the posterior fossa, which may be filled with cerebrospinal fluid (CSF) [1-4].

Differential diagnosis of this condition from other pathologies of the cerebellum includes Dandy-Walker malformation and Chiari malformation [2-6]. All these conditions may present with similar clinical features as Cerebellar agenesis but result from structural abnormalities of the cerebellum without its complete absence [1-3]. We report a case of a child with cerebellar agenesis accompanied by profound developmental delays.

CASE PRESENTATION

This case involves an 8-year-old girl with a history of inability to walk since birth. She was born at home and hospitalized for two days shortly after birth, with no major neonatal complications reported. Her developmental milestones were significantly delayed, as she achieved head control at 3 years and began speaking at 4 years. Her vaccinations were up to date.

On examination, the patient appeared healthy and was in stable condition with normal vital signs. She showed no signs of systemic distress. Chest examination was clear, and heart sounds (S1 and S2) were normal, with no murmurs or gallops. The abdomen was soft and non-tender, with no palpable masses. Neurological assessment revealed a Glasgow Coma Scale (GCS) score of 15/15. Notable findings included spasticity and muscle atrophy in the lower extremities, although motor strength remained intact with a muscle power rating of 5/5 across all extremities. Tests for balance and coordination, including the Romberg test, rapid alternating movements, finger-to-nose, and tandem walking, were impaired. Meningeal signs were absent.

Laboratory investigations, including a complete blood count, organ function tests, and random blood sugar levels, were within normal ranges. A pelvic X-ray was unremarkable. However, a non-contrast head CT scan revealed a complete absence of both cerebellar hemispheres, replaced by a cystic fluid component extending along the medulla oblongata. Minimal remnants of cerebellar peduncles were noted (Figure 1).

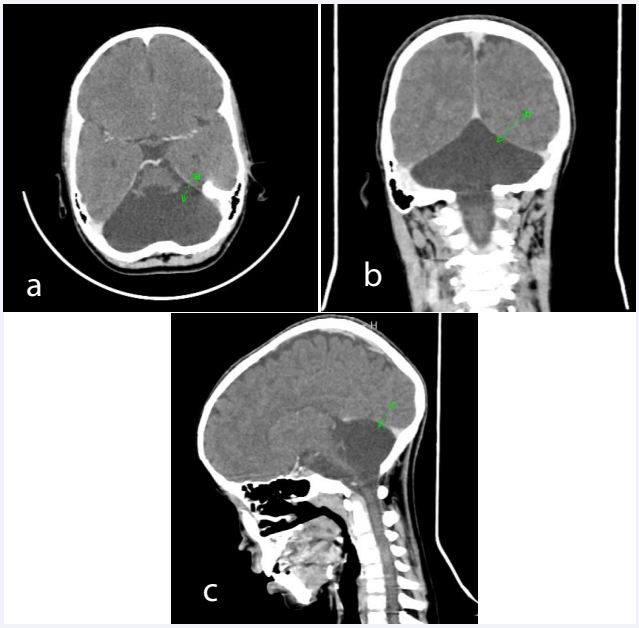

Figure 1 (a) Axial plane of contrast-enhanced CT scan of the brain complete absence of both cerebellar hemispheres replaced with cystic fluid component extending along the medullar oblongata (b) Coronal plane of contrast-enhanced CT scan of the brain (c) Sagittal plane of contrast enhance CT scan of the brain.

DISCUSSION

Cerebellar agenesis (CA) is an extremely rare condition characterized by the complete absence of cerebellar tissue [1-3]. The exact cause of Cerebellar agenesis is unknown, but it may be due to a malformation during development or disruption of the cerebellum during the prenatal or perinatal period [2-4].Cerebellar agenesis has been diagnosed in individuals ranging in age from a few weeks old to 71 years old [1].

Clinical presentation of Cerebellar agenesis is highly variable, and some individuals may have no symptoms while others experience a range of motor and cognitive impairments [1-5]. Some individuals with Cerebellar agenesis exhibit mild to moderate motor deficiencies such as ataxia, dysarthria, and delayed motor development [1-6]. Some individuals experience more severe motor and cognitive impairments, such as mental retardation, epilepsy, and hydrocephaly [1-5]. Some cases of Cerebellar agenesis are discovered incidentally, either during imaging for an unrelated condition or at autopsy [3-7].

The fact that some individuals with Cerebellar agenesis can lead relatively normal lives suggests that other areas of the brain, particularly the supra-tentorial brain, may be able to compensate for the missing cerebellum [2-4]. This ability to compensate may be more pronounced when Cerebellar agenesis occurs early in life, allowing for greater neural plasticity [2-3].

Imaging, particularly MRI, is crucial for diagnosing Cerebellar agenesis [1-6]. In Cerebellar agenesis, imaging reveals an absence of cerebellar tissue in the posterior fossa, which may be filled with cerebrospinal fluid [1-4]. The brainstem may also be hypo-plastic in individuals with CA [1-6].

It is important to distinguish cerebellar agenesis from other conditions that affect the cerebellum, such as Dandy-Walker malformation and Chiari malformation [2-7].

These conditions can have similar clinical presentations to cerebellar agenesis, but they involve structural abnormalities of the cerebellum rather than its complete absence.

Because cerebellar agenesis is so rare, there are many unanswered questions about its causes, the range of clinical presentations, and the mechanisms underlying the brain's ability to compensate for the missing cerebellum [1-3]. Further research, particularly studies involving living individuals with Cerebellar agenesis, is needed to better understand this complex condition.

CONCLUSION

We present an uncommon instance of cerebellar agenesis in an 8-year-old female patient, initially presenting with developmental delays and motor impairment, including failure to walk since birth. The condition was suspected based on neurological findings such as spasticity, muscle atrophy, and coordination difficulties. Neuroimaging revealed complete absence of the cerebellar hemispheres, replaced by cystic fluid extending into the medulla oblongata, highlighting the rarity of this congenital abnormality. This case underscores the importance of considering structural brain malformations in the differential diagnosis of developmental and motor delays in children. Early recognition through imaging is crucial for guiding management and prognosis.

DECLARATIONS

Ethics approval and consent to participate

The publication of a single case report does not necessitate ethics approval according to the policies of the authors' institution.

Consent for Publication

The patient provided written informed consent for the publication of this case report and accompanying images.

Author contribution

All authors made a significant contribution to the work reported, whether that is in the conception, study design, execution, acquisition of data, analysis and interpretation, or in all these areas; took part in drafting, revising or critically reviewing the article; gave final approval of the version to be published; have agreed on the journal to which the article has been submitted; and agree to be accountable for all aspects of the work.

ACKNOWLEDGEMENTS

The authors express their gratitude to the patient and their family for granting permission to share this case with fellow colleagues.

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