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Case Report

Total cerebellar agenesis: A case report of a very rare condition ☆

*Zineb Bouanani**, *Insaf Rhalem*, *Ghita Lahnine*, *Amal Akammar*, *Nizar El Bouardi*, *Badreddine Alami*, *Youssef Alaoui Lamrani*, *Mustapha Maaroufi*, *Meryem Boubbou*

University of Sidi Mohammed Benabdallah, Radiology Department, CHU Hassan II, Fès, Morocco

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ABSTRACT

Complete cerebellar agenesis is an extremely rare condition characterized by the complete absence of cerebellar tissue. Only a small number of cases have been reported, with varying motor and cognitive deficits. We describe a case of an 11-month-old baby with developmental delay, whose CT scan evaluation showed the complete absence of the cerebellum with no other associated cerebral malformation.

Introduction

Complete cerebellar agenesis is an uncommon condition [1]. It can manifest as an anatomically isolated anomaly or be a part of a more complex brain malformation [1]. It has been documented in patients ranging in age from a few weeks old to 71 years old [2]. Patients might present with a wide range of symptoms; they can be asymptomatic or present several symptoms such as ataxia, dysarthria, or motor abnormalities [3]. MRI and CT scans can be used to establish the diagnosis and rule out alternative causes. This article describes another case of complete cerebellar agenesis in an 11-month-old baby.

Case presentation

An 11-month-old baby girl, born from a nonconsanguineous marriage, presented to our hospital for developmental delay. The pregnancy was full term and it was described as uneventful. There was no history of perinatal hypoxia. The parents and siblings were clinically normal.

She had a delayed development of head control and sitting positions. The physical examination showed hypotonia and the routine hematological investigations were normal.

A CT scan showed total agenesis of the cerebellum, the tentorium was shifted downwards, the posterior fossa was small in size and fluid-filled. The brainstem was hypoplastic

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* Corresponding author.
E-mail address: zineb.bouanani1@usmba.ac.ma (Z. Bouanani).

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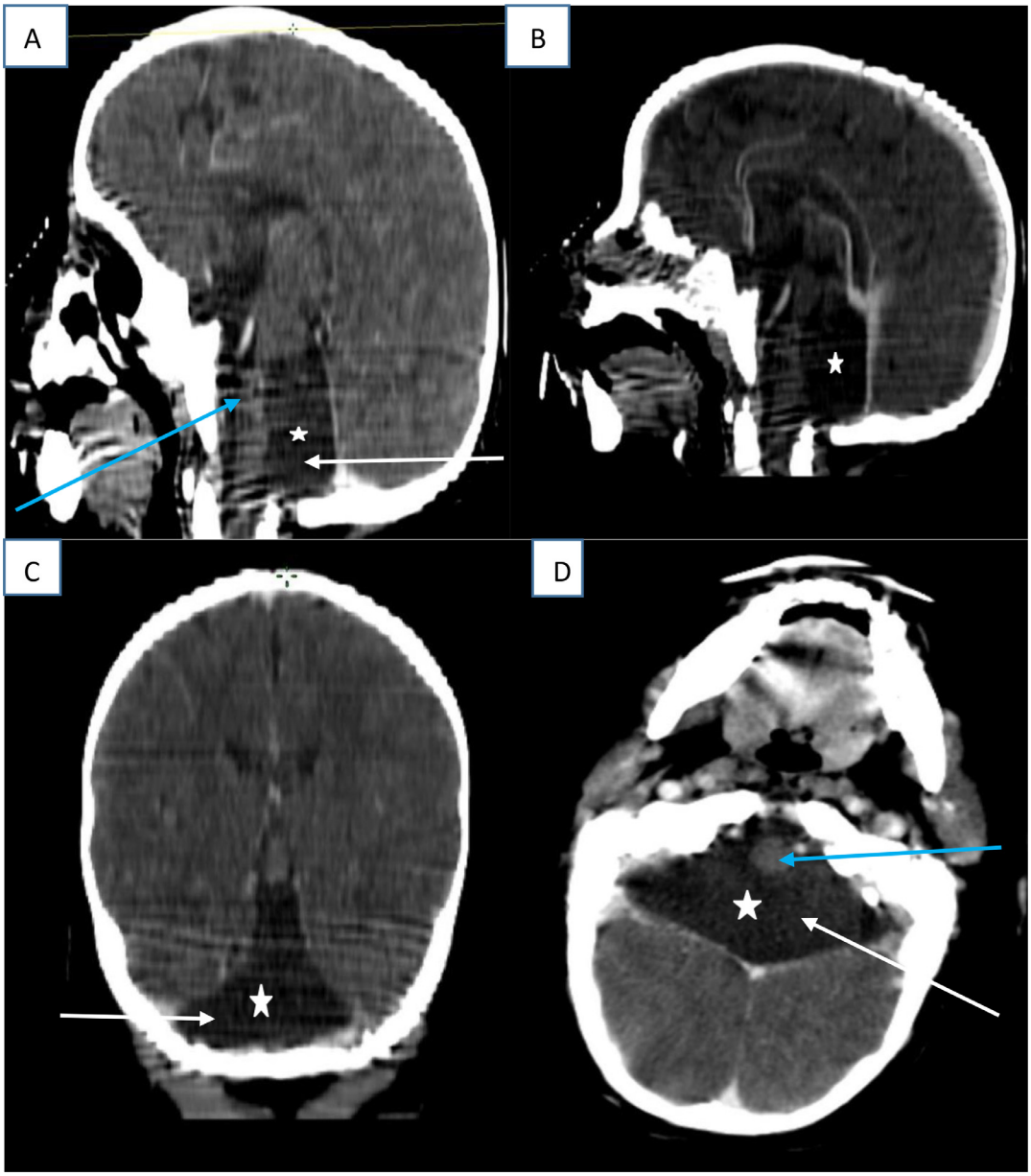

Fig. 1 – Computed tomography with contrast injection on sagittal (A, B), coronal (B), and axial (C) plans, showing complete absence of cerebellar tissue (white arrow) with a small size cerebellar fossa filled by CSF (asterix). The pons was hypoplastic (blue arrow).

(Fig. 1). There were no clearly identified associated cerebral malformations on CT-scan, and unfortunately, no MRI was performed for the patient.

With these findings, the patient was diagnosed with complete cerebellar agenesis and no further imaging follow-up was done.

Discussion

Primary cerebellar agenesis is the term for total or complete agenesis of the cerebellum. It is a very uncommon disorder [4].

The pathophysiology and molecular mechanisms of cerebellar agenesis remain unclear [1,5].

During embryogenesis, the dorsal part of the metencephalon's alar plates gives rise to the cerebellum. At each side a lip differentiates into a primordium of the cerebellum that projects into the fourth ventricle [6]. Near the end of the second month, the two cerebellar rudiments join at the midline, enlarging the extra-ventricular portion of the cerebellum. The cerebellum and its peduncles are absent in the propositus, but the mesencephalon, pons, and medulla oblongata are present.

It appears that the cerebellum stopped developing at an early age. The onset of this disability should have started no later than the second month following conception [1]. Brainstem-associated hypoplasia has been previously described in the literature, as described in our case [6]. This hypoplasia is believed to be associated with the loss of

cerebellar radiation, including the afferent and efferent fibers of the cerebellum [7].

Cerebellar agenesis can be diagnosed at any age, ranging from a few days to several decades [1,8–10]. Our patient's total cerebellar agenesis was diagnosed at the age of 11 months.

Clinical manifestations vary significantly, ranging from developmental delay and varying degrees of cerebellar dysfunction to early mortality. Patients might grow up to adulthood, but they usually exhibit a significant level of mental, physical, and neurobehavioral abnormalities [4]. Although the exact findings vary from patient to patient, most of them generally exhibit a nonprogressive syndrome characterized by an inability to coordinate movements [2].

After examining every case of total or almost complete cerebellar agenesis that has been reported, Glickstein [11] found evidence of a substantial motor deficit. In all these cases, individuals eventually learned to stand, walk, and speak but were profoundly retarded in acquiring these normal motor functions [8].

In some cases, cerebellar agenesis has been accidentally discovered during an autopsy [2].

Cerebral imaging (CT or MRI) is used to make the diagnosis, showing an absence of cerebral parenchyma in the posterior cerebral fossa. It also allows the detection of other associated cerebral malformations. CT, in our case, allowed the diagnosis.

Cerebellar agenesis must be distinguished from incomplete congenital cerebellar abnormalities such as Dandy-Walker deformity, Joubert syndrome, and congenital pontoneocerebellar atrophy [1].

In the Dandy-Walker malformation, there is hypoplasia of the cerebellar vermis, cystic dilation of the fourth ventricle, and an enlarged posterior fossa with upward displacement of the tentorium and torcular [14].

Congenital pontoneocerebellar atrophy was initially reported by Brun [12] and evaluated by Gadisseux et al. It is characterized by hypoplastic lateral cerebellar lobes and hypoplasia of the ventral section of the pons [13].

Conclusion

In conclusion, total cerebellar agenesis is an extremely rare condition. It can manifest either as an anatomically isolated anomaly or as part of a more complex cerebral malformation.

Patients present with a variety of developmental, clinical, and mental abnormalities, and imaging helps make the diagnosis, rule out other etiologies, and look for associated cerebral malformations.

Patient consent

Informed written consent was obtained from the patient for publication of the case report and all imaging studies.

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