



Downbeat nystagmus and progressive ataxia in adults: consider Chiari malformation type 1

Nistagmo downbeat e ataxia progressiva em adultos: considere malformação de Chiari tipo 1

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Downbeat nystagmus (DBN) is present in between 4 and 6% of patients with Chiari malformation type 1 (CM1). It is present in primary gaze and is characterized by a pathological phase which drifts the eyes in the upward direction followed by a downward quick-phase.^{1–3} Chiari malformation type 1 is one of the most prevalent craniocervical junction abnormalities⁴ and is more frequent in Northeastern Brazilians, due to pre-historic ancestors (previously thought to be caused by the Dutch colonization).⁵ The herniation of cerebellar tonsils through the foramen magnum is radiologically characterized by the distance between the apex of the odontoid and Chamberlain line of at least 5 mm.⁶

We present a four-case series of CM1 patients who developed progressive cerebellar ataxia and DBN. Brain magnetic resonance imaging (MRI) revealed the abnormal projected cerebellar tonsils. Evaluation of the posterior fossa through brain imaging looking for structural abnormalities such as CM1 is mandatory in cases of progressive ataxia combined with DBN, headache, and pyramidal signs (–**Video 1** and –**Fig. 1**).

Note

The patients have consented with the publication of the manuscript and its accompanying figures and videos.

Authors' Contributions

BKM: data curation, formal analysis, investigation, methodology, writing – original draft; TCV: conceptualization, formal analysis, methodology, project administration, validation, visualization, writing – original draft, writing – review & editing; FMRF: data curation, formal analysis, investigation, methodology; OGPB: conceptualization, data curation, formal analysis, investigation, methodology, project administration, supervision, validation, visualization, writing – original draft, writing – review & editing; JLP: conceptualization, data curation, formal analysis, methodology, supervision, writing – original draft, writing – review & editing.

Conflict of Interest

The authors have no conflict of interest to declare.

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Video 1

Video of the patients with progressive ataxia and downbeat nystagmus related to Chiari malformation type 1. Online content including video sequences viewable at: <https://www.thieme-connect.com/products/ejournals/html/10.1055/s-0043-1771171>.



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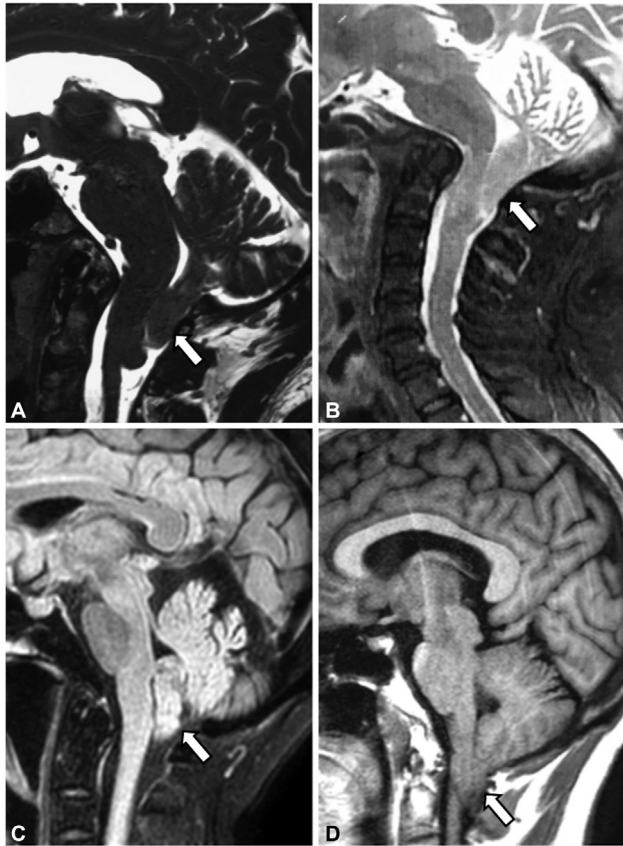


Figure 1 (Case A-D) Cranio-cervical MRI showing Chiari malformation type 1 in the four patients with progressive ataxia and downbeat nystagmus, which is defined by herniation of cerebellar tonsils through the foramen magnum (located at least 5 mm below this structure) (arrows).

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