Cerebellar and Brainstem Malformations

Andrea Poretti, MD[a,b,*], Eugen Boltshauser, MD[b], Thierry A.G.M. Huisman, MD, EQNR, FICIS[a]

KEYWORDS

• Malformation • Cerebellum • Brain stem • Neuroimaging • Diffusion tensor imaging • Children

KEY POINTS

- Progress in neuroimaging and genetics in the last decades has led to a significant improvement/refinement in the definition of cerebellar and brainstem malformations.
- Neuroimaging plays a key role in the diagnostic work-up of children with cerebellar and brainstem malformations.
- Diagnostic criteria of cerebellar and brainstem malformations are mostly based on neuroimaging findings.
- Neuroimaging findings may elucidate the role of the cerebellum for neurocognitive functions in children with cerebellar malformations and serve as predictive biomarkers for cognitive outcome.
- Advanced neuroimaging techniques such as diffusion tensor imaging may provide additional information that is helpful to better understand the pathogenesis of selected cerebellar and brainstem malformations.

INTRODUCTION

In the last few decades, progress in neuroimaging techniques, genetic analysis, and mouse model research has led to a significant improvement in the definition of cerebellar and brainstem malformations as well as in the recognition of novel disorders. Classifications based on neuroimaging, molecular genetic criteria, and developmental biological criteria have been proposed and include both inherited (developmental) and acquired (disruptive) anomalies.[1–4]

Malformations are defined as nonprogressive, congenital morphologic anomalies of a single organ or body part caused by an alteration of the primary developmental program.[5] Malformations result from intrinsic developmental processes, which refer to the cellular and molecular pathways involved in organogenesis. The molecules in these pathways can be altered by gene mutations, teratogens, or combined effects. The complex development of the cerebellum and brainstem and the high number of involved genes result in a high number of malformations.

Neuroimaging plays a key role in the diagnostic work-up of posterior fossa malformations.[2–4] Diagnostic criteria for posterior fossa malformations are based on neuroimaging findings. For some posterior fossa malformations, the spectrum of neuroimaging findings has been shown to explain the cognitive outcome (eg, in Dandy-Walker malformation [DWM] and rhombencephalosynapsis).[6,7] In addition, the spectrum of neuroimaging findings may suggest the

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[a] Section of Pediatric Neuroradiology, Division of Pediatric Radiology, The Russell H. Morgan Department of Radiology and Radiological Science, Charlotte R. Bloomberg Children's Center, The Johns Hopkins School of Medicine, Sheikh Zayed Tower, Room 4174, 1800 Orleans Street, Baltimore, MD 21287-0842, USA; [b] Department of Pediatric Neurology, University Children's Hospital, Steinwiesstrasse 75, Zurich 8032, Switzerland

* Corresponding author. Section of Pediatric Neuroradiology, Division of Pediatric Radiology, The Russell H. Morgan Department of Radiology and Radiological Science, Charlotte R. Bloomberg Children's Center, The Johns Hopkins School of Medicine, Sheikh Zayed Tower, Room 4174, 1800 Orleans Street, Baltimore, MD 21287-0842.

E-mail address: aporett1@jhmi.edu

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underlying genotype (eg, in pontocerebellar hypoplasias [PCHs]).[8] Conventional magnetic resonance (MR) imaging, including two-dimensional and three-dimensional T1-weighted, T2-weighted, and fluid attenuated inversion recovery (FLAIR) sequences, plays a key role in the evaluation/characterization of posterior fossa malformations and many diagnostic criteria have been based on conventional MR imaging findings.[2,3] In the last decade, advanced MR imaging techniques became increasingly available in the clinical setting and have been applied to explore posterior fossa malformations in more detail.[4,9] Diffusion tensor imaging (DTI) has been shown to provide noninvasive detailed qualitative and quantitative information on white matter tracts in children with brain malformations.[4,9] In addition, the detailed internal neuroarchitectural exploration of the brain by DTI has allowed clinicians to elucidate certain aspects of the pathogenesis of selected posterior fossa malformations, such as Joubert syndrome (JS) and pontine tegmental cap dysplasia (PTCD).[10,11]

This article discusses the normal anatomy of the posterior fossa followed by a discussion of the characteristic neuroimaging features of a variety of cerebellar and brainstem malformations. In this context, we classify posterior fossa malformations based on the neuroimaging pattern into (1) predominantly cerebellar, (2) cerebellar and brainstem, and (3) predominantly brainstem malformations as previously suggested.[2,3]

NORMAL ANATOMY OF THE POSTERIOR FOSSA

Conventional MR imaging sequences allow detailed evaluation of the anatomy of the posterior fossa and its contents.[2,3] A midline sagittal T1-weighted or T2-weighted sequence is ideal for showing the size of the posterior fossa, the shape and size of the vermis, and the size and morphology of the fourth ventricle and brainstem (**Fig. 1**). The vermis is divided into 3 parts by the primary and prepyramidal fissures. The rostrocaudal length of the ventral pons should be approximately twice that of the midbrain from the isthmus (ventral midbrain-pons junction) to the third ventricle, whereas the rostrocaudal length of the midbrain should be roughly the same as that of the medulla (from the level of the obex to the level of the ventral pontomedullary junction).[2,3] The posterior margin of the brainstem extending from the caudal sylvian aqueduct to the obex should be a straight line. The fastigium, or summit of the fourth ventricle, should lie just below the midpoint of the ventral pons on sagittal images. The cerebellar hemispheres and peduncles can be well assessed on parasagittal images, whereas the size and morphology of the vermis, cerebellar hemispheres, dentate nuclei, and superior and middle cerebellar peduncles can be best evaluated on axial images. In addition, the cerebellar folia run parallel to the calvaria (onionlike configuration) (see **Fig. 1**). Coronal images show fissures

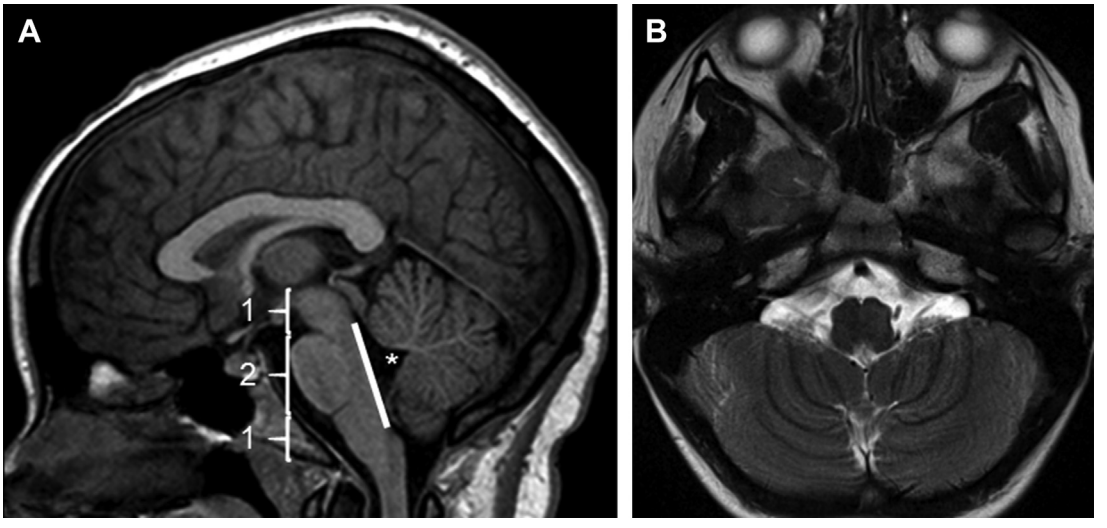

Fig. 1. Normal anatomy of the posterior fossa in a 12-year-old boy. (*A*) Midsagittal T1-weighted MR image shows a normal-sized posterior fossa, a normal vermis, an appropriate-sized pons (the rostrocaudal length of the pons is approximately twice [2] that of the midbrain [1] and medulla [1]), a flat dorsal surface of the brainstem (*line*), and a normal position of the fastigium just below the midpoint of the ventral pons (*asterisk*). (*B*) Axial T2-weighted MR image of the posterior fossa shows normal orientation of the cerebellar folia, which run parallel to the calvaria (onionlike orientation).