

Cerebellar and brainstem malformations

The development of the cerebellum and brainstem is a highly complex process that involves a large number of genes. Mutations in these genes may cause an abnormal development of the cerebellum and brainstem and result in several congenital morphological/anatomical abnormalities also known as “malformations” (Fig. 1). In the last few decades, progress in neuroimaging, genetic analysis, and mouse model research led to a significant improvement in the classification of cerebellar and brainstem malformations as well as in the recognition of novel disorders. An accurate classification and diagnosis of cerebellar and brainstem malformations is important for therapy, prognosis, and genetic counseling. Some cerebellar and brainstem malformations are associated with simultaneous affection of body regions other than the brain, e.g. liver and kidneys. It is important to identify the involvement of other organs as early as possible to initiate the best possible therapy before irreversible injury may occur. A precise diagnosis of cerebellar and brainstem malformations allows physicians to decide whether a checkup of other body organs is needed and how often it should be performed. In addition, a precise diagnosis may help physicians to provide patients and families with more detailed information about the prognosis and long-term outcome, e.g. how the child will develop with respect to motor, cognitive, language, and behavioral functions. Finally, an accurate diagnosis of cerebellar and brainstem malformations will allow physicians to determine the recurrence risk, e.g. the chance that a future child in the same family may have the same abnormality.

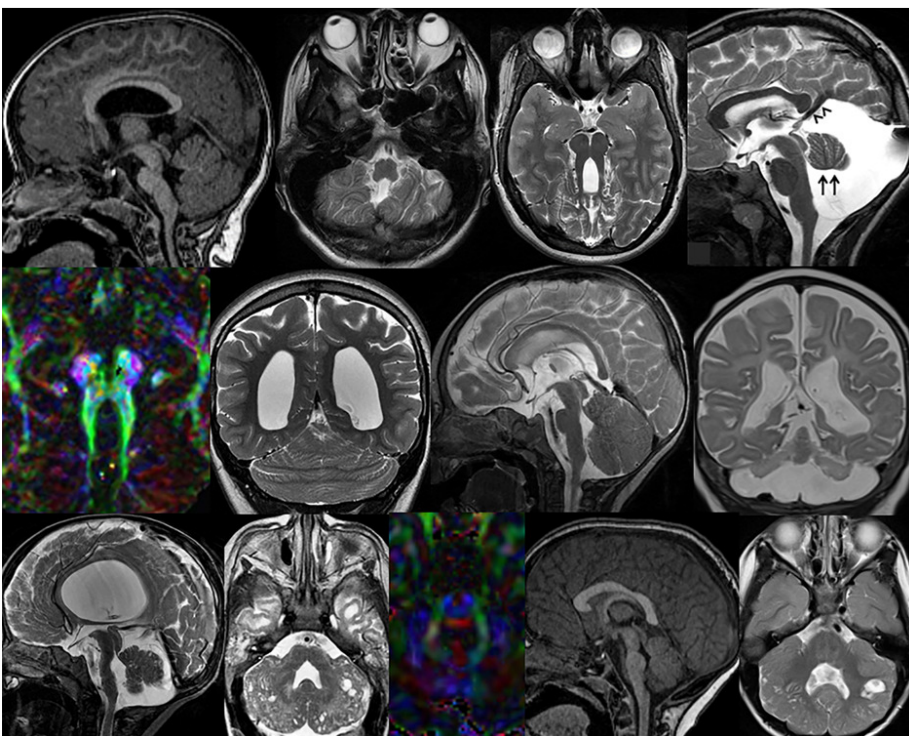


Fig. 1. Spectrum of cerebellar and brainstem malformations including from the left top to the right bottom: tubulinopathy, cerebellar dysplasia of unknown cause, Dandy-Walker malformation, Joubert syndrome (conventional image and diffusion tensor imaging), rhombencephalosynapsis, macrocerebellum, pontocerebellar hypoplasia type 2, muscle eye brain disease, pontine tegmental cap dysplasia (diffusion tensor imaging and conventional image), and Poretti-Boltshauser syndrome.

Neuroimaging, particularly magnetic resonance imaging (MRI) plays a key role in the diagnostic work-up of malformations of the cerebellum and brainstem. Diagnostic criteria for cerebellar and brainstem malformations are based on neuroimaging findings on both pre- and postnatal MRI studies. Conventional MRI sequences allow a detailed evaluation of the anatomy and morphology of the cerebellum and brainstem and play a key role in the diagnosis of cerebellar and brainstem malformations. Advanced MRI sequences such as diffusion tensor imaging (DTI) may provide additional important information that remains undetected by conventional MRI sequences. In particular, DTI allows to study the course of various white matter tracts within the brain. As example, DTI studies of children with selected cerebellar and brainstem malformations such as Joubert syndrome or pontine tegmental cap dysplasia revealed that specific white matter tracts within the cerebellum and brainstem do not cross the midline as expected. This information helps physicians to better understand the cause of these malformations.

To date, there are several different malformative diseases that affects the cerebellum and brainstem. Depending on the most predominant involvement, malformations may be divided into 3 groups: malformations with predominant cerebellar involvement, malformations with cerebellar and brainstem involvement, and malformations with predominant brainstem involvement. This is possible with a detailed evaluation of the anatomical changes within the cerebellum and brainstem that characterize the various malformations. In addition, a detailed evaluation of the MRI findings may allow a correlation between the imaging features and the underlying genetic cause of the malformation. Finally, in some cerebellar and brainstem malformations, a correlation between neuroimaging findings and neurodevelopmental outcome has been shown. In children with Joubert syndrome, for example, a smaller size of the cerebellar vermis correlates with a less favorable neurodevelopmental outcome and lower cognitive functions.

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