of NAA (NAA/Cr ratio) within the cerebellum and the pons have been well correlated with disability scores in patients [4]. A recent study in ataxia-telangiectasia (A-T) patients has revealed significantly lower NAA/Cho and higher Cho/Cr ratios in the cerebellum as compared to controls, suggesting altered membranes turnover within the cerebellum [5]. In our case, the significantly higher Cho/Cr in the BG shows that the disease is not restricted to the posterior fossa and may affect supratentorial structures by impairing membranes turnover in areas corresponding to the cerebellar-thalamic pathways. Within the cerebellum, the significant increase of total glutamine-glutamate (GlxT/Cr, GlxT/NAA) suggest an imbalance between excitatory (Glx) and inhibitory (gamma-aminobutyric acid [GABA]) neurotransmitters. Our patient showed a clinical positive response to gabapentin. This drug acts upon the GABAergic pathways [6]. The positive response to this treatment and the abnormally high GlxT/Cr and GlxT/NAA ratios within the cerebellum support the hypothesis of a disorder involving the GABA neurotransmission and likely induced by anti-GAD Ab. MRS may be useful to detect and monitor specific changes in brain biochemistry in such cases while morphology remains unremarkable on MRI.

Conflicts of interest

We declare no conflict of interest.

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References


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Möbius syndrome and epilepsy

Syndrome de Möbius et épilepsie

A 3420 g infant girl was delivered through caesarean section at 39 weeks of gestation, with an Apgar score of 7 at 1 min and 10 at 5 min. On examination, bilateral facial nerve paralysis, right abducens nerve paralysis and craniofacial asymmetry with microtretrognathia were noted. A diagnosis of Mobius syndrome was therefore suggested. Patient presented tonic seizures in the first 24 h of life, which were initially controlled with phenobarbital. Soon after, she started to have frequent focal and multifocal clonic seizures. Despite medication with clonazepam and phenobarbital, the child continued to have clinical seizures. A history of epilepsy in the family (the mother and maternal grandfather) was present. Also of note, the mother took misoprostol in the first trimester of pregnancy in an attempt to induce abortion. Brain MRI revealed alterations in brainstem morphology, including hypoplasia of the dorsal aspect of the lower pons with straightening of the fourth ventricular floor. The middle cerebellar peduncles were also hypoplastic and only their rostral segment was present. The rostral pontine tegmentum was protruding in the fourth ventricle. The superior cerebellar peduncles and the mesencephalon appeared normal (Figs. 1 and 2).

The hallmark of Mobius syndrome is facial diplegia and abducens nerve palsy, as described by Mobius in 1888. The aetiology is not well established. Most cases are sporadic, but there are reported familial cases, with the data indicating a gene for Mobius syndrome on chromosome 3, 10 and 13 [1,2]. It was proposed that sporadic cases are causally related with vascular impairment in a watershed zone of the brainstem, between the territories of the segmental para-median penetrating arteries and the long circumferential arteries, branches of the basilar artery [3]. The abnormality of the brainstem that occurs in Mobius syndrome could be the result of a transient interruption in fetal blood.

Several teratogens have been implicated in the aetiology of Mobius syndrome. One of the most reported teratogenic agent is misoprostol [4]. When taken early in the pregnancy, misoprostol is a potent uterine stimulant that induces abortion. Induced uterine contractions can cause vascular disruption, resulting in fetal hypoxia and ischemia. MR imaging findings vary, according to the degree of rhombencephalic maldevelopment. Imaging study frequently shows a variable degree of pontine hypoplasia with straightening of the fourth ventricular floor [5].

We describe a neonate with Mobius syndrome presenting pontine and middle cerebellar peduncles hypoplasia. The differential diagnosis includes other brainstem development disorders. Ventral tegmental changes are similar to those found in pontine tegmental cap dysplasia, which also presents middle cerebellar peduncles hypoplasia [6]. However, in this case, we did not find an abnormal shape and orienta-
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Figure 1  Sagittal T1-weighted image (A) demonstrates hypoplasia of the dorsal aspect of the pons with straightening of the fourth ventricular floor. Coronal T1-weighted image (B) shows hypoplasia of the lower pons and middle cerebellar peduncles.

Figure 2  Axial T2-weighted images show hypoplasia of the caudal (A) and middle pons (B). Only the rostral middle cerebellar peduncles are present (C). Superior cerebellar peduncles appear normal, without elongation (D).

...tion of superior cerebellar peduncles, which give the molar tooth appearance. Recurrent seizures are not usually described in patients with Möbius syndrome. We may assume that the same mechanism responsible for brainstem abnormality can lead to brain dysfunctions by damaging the cerebral cortex.

Conflict of interest

No potential conflict of interest relevant to this article was reported.

References


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