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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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 Mobius 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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