A CASE OF VANISHING WHITE MATTER DISEASES WITH ATYPICAL NEUROIMAGING FINDINGS

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INTRODUCTION

Vanishing white matter disease (VWMD) is a recessive neuro-degenerative disease caused by eIF2B gene mutations(1). The disease is characterized by progressive loss of white matter in both hemispheres of the brain. The resulting axonal degeneration causes progressive deterioration of neurological functions. An important feature of the disease is the worsening of clinical symptoms when exposed to physiological and environmental stress factors (2).

OBJECTIVES

We aimed to present our patient because of atypical localized brain Magnetic resonance imaging(MRI) results and different electromyography (EMG) results.

CASE PRESENTATION

A 5-year-old boy from a consanguineous marriage applied with complaints of developmental delay and cognitive retardation. At 3 years he had his first generalızated tonıc clonıc seizure. After that he continued to had focal and generalizated seizures. Symptoms such as cerebellar and motor dysfunction, loss of ambulation, and dysphagia progress insidiously.According to the results of MRI; there were hyperintense lesions in T2A- FLAİR in both cerebellar hemispheres, especially at the dentate nucleus and mesencephalon-pons-bulbus level, along the corticospinal tract , in both putaminal regions and globus pallidus (Figure1-2). We perform a whole exome sequence analysis to this patient: EIF2B2 (NM_014239.3)Exon 6 Chr14:75473412 c.826C>T(p.Pro276Ser) homozygot mutation was identified. EMG was found to be compatible with axonal type polyneuropathy-myopathy.

CONCLUSION

The typical brain MRI of VWMD patients shows that the cerebral white matter is widely sparse, whereas the cortex is relatively well pre-served (3). While peripheral nervous system involvement is not expected in VWM disease, the EMG of our case was compatible with axonal type polyneuropathy-myopathy. It should be kept in mind that patients may present with different neuroimaging and EMG findings in contrast to the classical presentation to VWMD.

REFERENCES

1.Leegwater, P., Vermeulen, G., Könst, A. *et al.* Subunits of the translation initiation factor eIF2B are mutant in leukoencephalopathy with vanishing white matter. *Nat Genet* **29,** 383–388 (2001). https://doi.org/10.1038/ng764

2. van der Knaap, M.S., Leegwater, P.A.J., Könst, A.A., Visser, A., Naidu, S., Oudejans, C.B., Schutgens, R.B. and Pronk, J.C. (2002), Mutations in each of the five subunits of translation initiation factor eIF2B can cause leukoencephalopathy with vanishing white matter. Ann Neurol., 51: 264-270. https://doi.org/10.1002/ana.10112

3. Meoded A, Poretti A, Yoshida S, Huisman TA. Leukoencephalopathy with vanishing white matter: serial MRI of the brain and spinal cord including diffusion tensor imaging. Neuropediatrics. 2011;42(2):82-85.

4. Jabbehdari, Sayena et al. "The clinical features and diagnosis of Metachromatic leukodystrophy: A case series of Iranian Pediatric Patients." *Iranian journal of child neurology* vol. 9,3 (2015): 57-61.

Figure1-2: There were hyperintense lesions in T2A- FLAİR in both cerebellar hemispheres, especially at the dentate nucleus and mesencephalon-pons-bulbus level, along the corticospinal tract , in both putaminal regions and globus pallidus