Recurrent Torticollis- A rare ADHPRL2 mutation

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Introduction

Torticollis is involuntary bending of the head and neck to one side as a result of the contraction of the neck muscles. Although torticollis can occur at any age, it is seen especially in childhood. Many different causes may play roles in the etiology of torticollis in children, ranging from diseases with a good prognosis to conditions with high mortality rates(1). Adp-ribosylhydrolase-like 2 (ADHPRL2) mutation is an extremely rare disorder and characterized by paroxysmal torticollis, ataxia episodes, and serious progressive neurodegeneration.

This report describes an unusual case of ADHPRL2. The purpose of this

The case is to highlight the key characteristics of ADHPRL2 despite its initial appearance resembling benign torticollis attacks.

Case

A three-year-old boy presented with recurrent torticollis attacks, which occurred typically once a month and resolved without medical.The intervention had progressive neurological regression after about 6 months. Cyproheptadine for benign paroxysmal vertigo attacks and carbamazepine for movement disorder did not benefit. However, as the frequently recurring condition continued, ataxic gait started. It was then decided to conduct a genetic analysis. The patient, whose psychometric development was considered to be normal by his age, had a pause in psychomotor development within 6 months. During the follow-up, the patient's ataxia increased, he lost the ability to walk and became immobile. His nystagmus started. EEG showed epileptic encephalopathic changes. He had refractory epilepsy after 1 year of follow-up. Atrophic changes developed on cranial MRI. Meanwhile, the patient's genetics resulted as a mutation in ADPHRL2. After a period of follow-up in an intubated mechanical ventilator.

Initial complaint/age	Torticollis	Time from first torticollis attack
Regression initiating event/age	Prolonged duration of recurrent torticollis attacks	+6 months
EEG abnormality / age		+8 months
Age at nystagmus onset		+8 months
Neuropathy/ age at onset	Severe diffuse sensorimotor neuropathy	unknown
Hearing loss	Normal on initial examination	
Eye examination	First examination was normal.	unknown
Facial myoclonia	None	
Serious Infection	Yes(followed at an intensive care unit)	
Mechanical Ventilator Requirement	Yes after 18 months	
Refractory seizures	Yes after 12 months	

Key words

Ataxia, Torticollis, Neurodegenerative Disorders, Adphrl2 Mutation

Conclusions

Torticollis has various causes, including intracranial pathologies, medications affecting vision and balance, ocular diseases, and reflux-related syndromes. In rare cases, it may be caused by ADPHRL2 mutation. Although there is no definitive cure for the ADPHRL mutation, genetic studies may be necessary for progressive degeneration and recurrent episodes of torticollis. It is still crucial to diagnose the underlying pathology to ensure proper treatment and recovery.

References

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