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INTRODUCTION

We presented progressive myoclonic epilepsy with a SEMA6B mutation in this case.

CASE REPORT

A 6-year-old girl had presented with language delay, mental retardation, attention deficit hyperactivity disorder, and drug-resistant epilepsy. She was born at term from a healthy pregnancy. Her parents were not consanguineous, and she did not have any problems at birth or after birth. She was diagnosed with language delay and attention deficit hyperactivity disorder when she was two years old. The patient's first seizure occurred at the age of 4 while asleep. Types of her seizures were generalized tonic-clonic. She was no dysmorphic appearance on physical examination. Amino acids, lactic acid, ammonia, and thyroid function tests were at normal levels in the blood tests. We detected frequent recurrent generalized spike and slow-wave activity in the patient's interictal electroencephalogram. SEMA6B (NM_032108), c.2041del, heterozygous mutation had been detected in the WES examination performed for drug-resistant epilepsy. The patient was receiving valproic acid therapy. We detected SEMA6B gene mutation in the patient and added zonisamide to the treatment. After the zonisamide treatment, the patient's seizures decreased.

CONCLUSION

Progressive myoclonic epilepsy-11 associated with the SEMA6B gene is a neuro-degenerative rare disorder (Hamanaka K., et al). This disorder includes neurodevelopment retardation and various types of seizures around two years of age after relatively normal early development.

REFERENCES

Hamanaka K., Imagawa E., Koshimizu E., Miyatake S., et al. De Novo Truncating Variants in the Last Exon of SEMA6B Cause Progressive Myoclonic Epilepsy, Am J Hum Genet. 2020 Apr 2;106(4):549-558.
doi:10.1016/j.ajhg.2020.02.011