

DE NOVO MUTATION IN SETD1B IS ASSOCIATED WITH INTELLECTUAL DISABILITY, EPILEPSY, AND AUTISTIC BEHAVIOR

^aSedef Terzioglu Ozturk, ^bHande Dilber, ^cMehmet Ali Ekenel, ^dBahtiyar Sahinoglu, ^aCengiz Dilber

Kahramanmaras Sutcu Imam University Faculty of Medicine, ^aPediatric Neurology, ^bPediatrics, ^cNeurology, ^dGenetics, Turkey

INTRODUCTION

SETD1B (SET domain containing 1B) is a component of the SET1 histone methyltransferase complex, which mediates the methylation of histone H3 on lysine 4 (H3K4) (1,2). We described the clinical features of a patient with a rare SETD1B gene mutation in the literature.

CASE REPORT

A Turkish boy was born to consanguineous healthy parents after an uneventful 39-week pregnancy. At the age of five months, he had seizures in the form of a myoclonic spasm and eyelid myoclonia. Her seizures' were refractory to multiple antiepileptic drugs and developed into frequent myoclonic absence seizures at three years of age.

He had developmental delay, language delay, autistic behavior, and mildly tapering fingers in his physical examination. No abnormality was detected in metabolic screening tests, urine and blood amino acids, lactic acid, ammonia, and thyroid function tests. Growth parameters and brain imaging were essentially normal. EEG taken at different times showed frequent repetitive generalized sharp slow-wave activity. The microarray analysis result was normal. In the WES examination performed for resistant epilepsy, the patient was SETD1B (NM_015048), c.544+2T>G, p. variant detected. Oxcarbazepine, clonazepam, and ethosuximide treatments were gradually added to the patient whose valproic acid treatment was continuing. Although the patient's seizures decreased, myoclonic seizures rarely continued.

CONCLUSION

We identified de novo SETD1B variants in one sporadic case of seizures, developmental delay, intellectual disability, and autistic behavior. The long-term studies will be more supportive in detecting pathogens in the SETD1B gene.

REFERENCES

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