Novel ANKRD11 gene mutation in an individual with a phenotype of KBG syndrome presenting like West syndrome

Aslı Kübra Atasever1, Sibğatullah Ali Orak1, Çisil Çerçi Kubur1, Celil Yılmaz1, Muzaffer Polat1,

1 Celal Bayar University Faculty of Medicine Department of Pediatric Neurology,

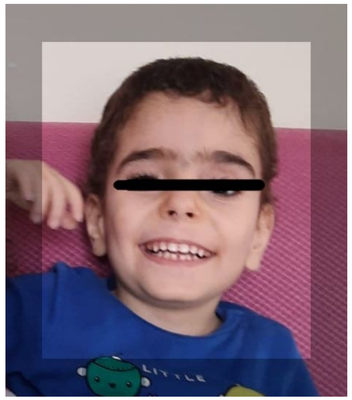
ICNC 2022
17th INTERNATIONAL CHILD NEUROLOGY CONGRESS
ANTALYA, TURKEY | OCTOBER 3-7, 2022

INTRODUCTION AND ABSTRACT

KBG syndrome is a rare autosomal dominant disorder, typically characterized by macrodontia, craniofacial findings, short stature, neurological involvement including intellectual disability, and epilepsy.

MATERİALS AND METHODS

The 2-year-old male patient is followed up in pediatric neurology with epilepsy. . The patient was born by cestio at 39 weeks, 2515 gr, 45 cm, head circumference 32 cm. The patient's first seizure started at the age of 5 months, in the form of epileptic spasms. that repeated 100 times a day In the examination of the patient at that time, pathologically; central hypotonicity, flat philtrum, and 2/6 murmur were present. He was admitted to the neonatal intensive care unit because of anal atresia. VSD was detected at 20 days of age.



RESULTS

A hypsarrhythmia pattern was detected in the EEGs. Apical muscular VSD was detected in the echo. Brain MRI was normal. We gave ACTH to the patient for epileptic spasms. After 4 months, clobazam and sodium valproate were started, respectively, for non-motor behavior arrest and motor atonic seizure. Because the patient had dysmorphic findings and additional system anomalies, whole-exome sequencing(WES) was performed on the patient. ANKRD11 c.3562C>T p.Arg1188Ter heterozygous pathogenic mutation was detected. Confirmed by Sanger sequencing. No pathogenic variation was observed in his parents. His seizures are under control with triple antiepileptic therapy. He is receiving special education and rehabilitation.

CONCLUSİONS

There are about 100 cases of KGB reported in the literature. Our case has additional phenotypic features since it presented to us with West syndrome and a history of anal atresia. For this reason, we wanted to share our case.

REFERENCES

1. 2. Herrmann J, Pallister PD, Tiddy W, Opitz JM. The KBG syndrome-a syndrome of short stature, characteristic facies, mental retardation, macrodontia and skeletal anomalies. *Birth Defects Orig Artic Ser*. 1975;11(5):7-18
2. Alves RM, Uva P, Veiga MF, et al. Novel ANKRD11 gene mutation in an individual with a mild phenotype of KBG syndrome associated to a GEFS+ phenotypic spectrum: a case report. *BMC Med Genet*. 2019;20(1):16. Published 2019 Jan 14. doi:10.1186/s12881-019-0745-7