Pyrimidine Metabolism Disorders as Rare Cause of Psycho-motor Retardation, Dysmorphism and Epilepsy

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OBJECTİVES

We describe 2 patients with psycho-motor developmental delay, dysmorphic feautures and epilepsy

diagnosed with pyrimidine metabolism disorder by detecting mutations in *UPB1* and *DPYS* with whole exome sequencing (WES) analysis.

METHODS

The data of the cases were obtained retrospectively from the pediatric neurology records of Kocaeli University Faculty of Medicine.

RESULTS

Five-moth-old boy presented with severe psycho-motor retardation, dysmorphism and epileptic spasms. EEG examination revealed hypsarrhytmia. The patient was diagnosed as West syndrome. Now, he is 2-year-old, and still has severe psycho-motor retardation, partially controlled seizures with polytherapy. Routine diagnostic tests were uninformative. WES analysis showed pathogenic, compund heterozygous variants in *UPB1,* for each variant parents were heterozygous.

16-month-old boy presented with mild psycho-motor retardation, dysmorphism and seizures. The etiology could not be identified with routine diagnostic tests. Now, he is 14-year-old, needs special education support, and seizures are partially controlled with monotherapy. WES analysis showed a likely-pathogenic, homozygous variant of *DPYS.*

Both patients are diagnosed with pyrimidine metabolism disorder. The diagnosis was confirmed by urine pyrimidine metabolite analysis.

CONCLUSION

Psycho-motor retardation, dysmorphism and seizures are frequently observed in patients with inborn errors of metabolism disorders. Urine pyrimidine metabolites should be screened in patients with negative metabolic disorders screening before performing more complicated diagnostic tests.