KAT6A Syndrome

https://pubmed.ncbi.nlm.nih.gov/34748993/

Pathogenic variants in KAT6A, encoding a histone acetyltransferase, have been identified as a cause of a developmental disorder with a definite clinical spectrum including intellectual disability, speech delay, dysmorphic facial features, microcephaly, cardiac and gastrointestinal defects. Seizures have been described in a minority of patients without a detailed characterization. In this work we focus on epilepsy in KAT6A syndrome, reporting two affected girls with history of seizures, bearing a KAT6A de novo heterozygous variant, of which one is novel. We describe the different epilepsy phenotypes of these two patients and compare them to the other individuals in literature presenting with epilepsy.