Say-Barber-Biesecker Syndrome

https://pubmed.ncbi.nlm.nih.gov/28857140/

The Say-Barber-Biesecker-Young-Simpson variant of Ohdo syndrome (SBBYSS) and Genitopatellar syndrome (GTPTS) are 2 rare but clinically well-described diseases caused by de novo heterozygous sequence variants in the KAT6B gene. Both phenotypes are characterized by significant global developmental delay/intellectual disability, hypotonia, genital abnormalities, and patellar hypoplasia/agenesis. In addition, congenital heart defects, dental abnormalities, hearing loss, and thyroid anomalies are common to both phenotypes. This broad clinical overlap led some authors to propose the concept of KAT6B spectrum disorders. On the other hand, some clinical features could help to differentiate the 2 disorders. Furthermore, it is possible to establish a genotype-phenotype correlation when considering the position of the sequence variant along the gene, supporting the notion of the 2 disorders as really distinct entities.