Abstract

Poster Title: Neuropsychological profile of patients with pathogenic variant in CHD3 and KAT6B

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Abstract

The discovery of rare de novo heterozygous pathogenic variants in the CHD3 and KAT6B genes has unveiled a novel neurodevelopmental disorder coined Snijders Blok-Campeau syndrome (SNIBCPS, caused by CHD3 variants) and identified the genetic basis of Genitopatellar syndrome (GPS) and Say-Barber-Biesecker-Young-Simpson variant syndrome (SBBYSS, a variant of Ohdo syndrome), both now known to be caused by KAT6B variants. These genes encode crucial enzymes that regulate chromatin structure, gene transcription and DNA repair during the development of embryos' nervous system. CHD3 and KAT6B pathogenic variants cause global neurodevelopmental delays and characteristic physical malformations. This study serves a duality of functions as it aims to define the range of neuropsychological features of these newly discovered disorders and to introduce them to clinicians and researchers in the field. This work in progress presents a glimpse of the neuropsychological profile of individuals affected by the CHD3 and KAT6B variants. Patient recruitment has mainly been conducted through referrals from international geneticists and social media posts on the Facebook page CHD3 Families. Data was gathered from 13 participants using neuropsychological assessments and parent-report questionnaires evaluating functioning on intellectual, behavioral, emotional and social domains. Severely impaired global intellectual development, adaptive functioning, health-related quality of life and coordination as well as varied social impairment and nonapparent emotional malfunction seem to characterize the neuropsychological profile of both SNIBCPS and GPS/SBBYSS. Clinicians' limited knowledge of the symptomology of these syndromes as well as worldwide and rare occurrence of these cases (≤ 60 cases with CHD3 disorders and 90 with KAT6B disorders) challenged data collection. Additional individuals with data from other neurodevelopmental questionnaires are yet to be assessed. This study will further expand knowledge on these rare disorders and aid in their diagnosis, ultimately benefitting both clinical and research fields.