SIN3A Witteveen-Kolk syndrome WITKOS

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Witteveen-Kolk syndrome (WITKOS; OMIM #613406) is a recently described, rare neurodevelopmental syndrome characterized by mild intellectual disability and a recognizable facial gestalt. WITKOS is caused by heterozygous loss-of-function variants in SIN3A. It shares some features with 15q24 deletion syndrome but to date has only been described in a limited number of patients mostly of Northern European ancestry. Here, we report the first patient with Hispanic ancestry to our knowledge diagnosed with WITKOS, who has a novel, truncating variant in the SIN3A gene. Clinical exome sequencing performed in-house using a custom bioinformatics pipeline identified a de novo heterozygous, nonsense variant in SIN3A, c.1015C>T (p.Gln339Ter) that has not been previously described in the literature. This 3-year-old boy with WITKOS demonstrated classic features including mild developmental delay and triangular facies with hypotelorism and deep-set, hooded eyes. This patient supports the currently described phenotype for WITKOS in more diverse populations.