Alazami Syndrome

https://pubmed.ncbi.nlm.nih.gov/32888391/

Biallelic mutations in the LARP7 gene have been recently shown to cause Alazami syndrome, a rare condition characterized by short stature, intellectual disability, and peculiar facial dysmorphisms. To date, only 24 cases have been reported. Here, we describe two brothers initially suspected to have Smith-Lemli-Opitz syndrome, in whom clinical exome sequencing detected a novel homozygous truncating variant in LARP7. These cases expand the phenotypic spectrum of Alazami syndrome to include toes syndactyly and adaptive behavior, and confirm the power of "genotype first" approach in patients with syndromic presentations overlapping distinct rare conditions.