2 de novo heterozygous variants in SON gene are associate with Zhu-Tokita-Takenouchi-Kim syndrome

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Abstract

Zhu-Tokita-Takenouchi-Kim (ZTTK) is a rare disorder caused by heterozygous variants of SON gene, which is an autosomal dominant genetic disease, with only 32 cases and 25 causative variants in SON have been reported to date since the first report in 2015. Herein, we reported 2 additional sporadic cases with clinical features strikingly similar to cases having been reported. Notably, through penetration of left palm print and growth hormone deficiency in our Patient #1 has not been mentioned in reported literature. Whole-exome sequencing revealed 2 novel variants, c.5297 delC (p.S1766Leufs*7) and c.5230 delC (p.Arg1744Valfs*29) in SON resulting in ZTTK syndrome. Our report expands the mutant spectrum of SON gene and refine the genotype-phenotype map of ZTTK syndrome.

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