PUF60-related developmental disorder (Verheij syndrome)

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Background:

Verheij syndrome is a rare microdeletion syndrome of chromosome 8q24.3 that harbors PUF60, SCRIB, and NRBP2 genes. Subsequently, loss of function mutations in PUF60 have been found in children with clinical features significantly overlapping with Verheij.

Case presentation:

Here we present the first Chinese Han patient with a de novo nonsense variant (c.1357C > T, p.Gln453*) in PUF60 by clinical whole exome sequencing. The 5-year-old boy presents with dysmorphic facial features, intellectual disability, and growth retardation but without apparent cardiac, renal, ocular, and spinal anomalies.

Conclusions:

Our finding contributes to the understanding of the genotype and phenotype in PUF60 related disorder.