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Tricho-Rhino-Phalangeal Syndrome is a Rare Presentation of Brachydactyly

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Abstract

Mutations of *TRPS1* cause tricho-rhino-phalangeal syndrome (TRPS), characterized by brachydactyly, sparse hair, and specific facial dysmorphism including a bulbous nose. Lab findings regarding parathormone, calcium and phosphate are normal. Thus, TRPS is usually mistaken for acrodysostosis types. The severity of brachidactyly is various, one or more metacarpals can be affected. Herein we report a Turkish female with phenotype consistent with TRPS. In this patient with severe brachydactyly and short stature, we identified a novel heterozygous missense mutation in exon 6 (c.2782T<G. p.Tyr928Asp). located within the GATA DNA-binding domain. These patients usually refer to dermatologist due to sparse hair. Brachydactyly is less noticed feature for patients. Our findings emphasize that physician should think all syndromes that can cause brachydactyly.

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