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Novel Pathogenic Variants of TMPRSS6 Gene in a Girl with Iron Refractory Iron Deficiency Anemia (IRIDA)

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Abstract

Iron refractory iron deficiency anemia (IRIDA) is an autosomal recessive disorder caused by mutations in the TMPRSS6 gene, which impairs iron homeostasis. We report a 4-year-old girl presented with a one-year history of IDA. Hemoglobin, transferrin saturation, and hepcidin levels were 6.5 g/dL, 1.6%, and 112.17 ng/mL, respectively. High-dose oral iron therapy partially corrected hemoglobin levels, and they declined after decreasing or stopping iron therapy. Genetic analysis of the TMPRSS6 gene identified compound heterozygotes of two novel pathogenic variants of c.811C>T in exon 7 (p.R271X) and c.1254C>G in exon 11 (p.Y418X). Her parents carried a heterozygous mutation of each variant.

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