Identification of a novel homozygous frameshift mutation in SLC29A3 gene in a case with H syndrome from Iran

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A B S T R A C T

H syndrome is a rare monogenic autosomal recessive disease with characteristic cutaneous findings and multisystem involvement. The aim of this study is to present an Iranian patient with H syndrome and to describe a novel frameshift mutation in SLC29A3 gene. The patient was diagnosed with a few small areas of hyperpigmentation and accompanying hypertrichosis in the lumbar area of her back. Her clinical phenotypes included short stature, hepatosplenomegaly, facial widespread bilateral telangiectatic lesions, bilateral hypertrophy of the parotid gland, upper extremity flexion contracture, elevated inflammatory markers (ESR, CRP) and diabetes mellitus. The identification of a novel homozygous frameshift mutation (c.307_308delTT, p.F103Ter) in SLC29A3 gene, together with the characteristic clinical manifestations of H syndrome, provided accurate diagnosis for this patient.