Abstract

Barakat syndrome is an autosomal dominant rare genetic disease caused by haploinsufficiency of the GATA binding protein 3 (*GATA3*) gene. It is also known as HDR syndrome, and is characterized by varying degrees of hypoparathyroidism, sensorineural deafness and renal disease. This is the first report of a heterozygous *GATA3* whole gene deletion causing HDR syndrome in a Sri Lankan family.

A 13-year-old boy with an acute febrile illness, hypocalcaemia and bilateral carpopedal spasm was referred for evaluation. A past medical history of treatment for persistent hypocalcaemic symptoms since the age of 7 months was obtained. Biochemical investigations showed persistent low serum corrected calcium levels with hyperphosphataemia, hypomagnesaemia, low parathyroid hormone levels, hypercalciuria, and low total 25-hydroxy vitamin D levels. His renal functions and renal sonography were normal. Audiometry showed bilateral moderate to severe sensorineural hearing loss. On screening, his mother was also found to have asymptomatic hypocalcaemia, hypomagnesaemia, hyperphosphataemia, hypercalciuria and low total 25-hydroxy vitamin D levels. She had impaired renal functions and chronic parenchymal changes in the renal scan. Audiometry showed bilateral profound sensorineural hearing loss. Genetic analysis using multiplex-ligation dependent probe amplification showed a reduced gene dosage for *GATA3* that is consistent with a heterozygous whole gene deletion in both the child and mother. This report demonstrates the wide intra-familial phenotypic variability observed in HDR syndrome and adds further to the existing scientific literature on the genotype-phenotype correlation of this syndrome. It highlights the need for HDR syndrome to be considered in the differential diagnosis of persistent hypocalcaemia with sensorineural deafness and/or renal involvement, and for appropriate genetic evaluation to be done to confirm the diagnosis.