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Title: A family with hypoparathyroidism, sensorineural deafness and renal disease (Barakat syndrome) caused by a reduced gene dosage in *GATA3*

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Abstract (Max 250 words):

Background: Barakat syndrome is an autosomal dominant rare genetic disease caused by haploinsufficiency in 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 *GATA3* gene deletion causing Barakat syndrome in a Sri Lankan family.

Case presentation: 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 in *GATA3* that is consistent with a heterozygous deletion of exons 3-6 in both the child and mother.

Conclusions: This case demonstrates the wide intra-familial phenotypic variability observed in Barakat syndrome. It highlights the need for Barakat syndrome to be considered in the differential diagnosis of persistent hypocalcaemia and for appropriate genetic evaluation to be done to confirm the diagnosis.