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CHARACTERIZATION OF NOVEL BTK MUTATION IN AN IRANIAN PATIENT PRESENTING VACCINE-ASSOCIATED PARALYTIC POLIOMYELITIS

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Objectives: X-linked agammaglobulinemia (XLA) is a prototypic humoral deficiency caused by mutations in the Bruton's tyrosine kinase (BTK) gene. In addition to susceptibility to bacterial infections, patients with XLA are especially prone to enteroviruses. Here, we describe the occurrence of VAPP in a 15-month old Iranian boy. The child had received four doses of OPV, administered at birth, 2, 4, and 6 months of age. The patient's infectious history was unremarkable.

Methods: Laboratory evaluation revealed low levels of immunoglobulin G and CD19(+) B cells of less than 1% of the lymphocyte population. Genomic DNA was isolated from peripheral blood and DNA sequencing using Big dye termination method was performed in ABi 310 capilary sequencer.

Results: A novel insertion (c.685_686insTTAC) in the SH3 domain of the BTK gene was detected as the underlying cause.

Conclusion: Immunodeficient recipients of OPV can excrete poliovirus vaccine strains for a long period and are at risk of developing flaccid paralysis. They could also serve as a source of reverted virulent poliovirus to be reintroduced into the general population. This patient presented for the first time with VAPP, without any history of other major infections in 15 months. This suggests that a negative history for recurrent infections does not exclude the presence of a primary defect in the immune system.

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