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PIK3R1mutation associated with Hyper IgM syndrome and APDS2: First case from the national Iranian registry and review of the literature

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Abstract

Background: APDS [Activated phosphoinositide 3-kinase (PI3K) δ Syndrome] is a newly found special form of primary immunodeficiency caused by mutations in genes encoding PI3Kδ subunits and over-activation of the PI3K signaling pathway. Gain-of-function and loss-of-function mutations in PIK3CD (encoding P110δ) and PIK3R1 (encoding p85α, p55α and p50α) lead to APDS1 and APDS2, respectively. The subsequent irregular PI3K downstream signaling cascade is associated with abnormalities in B cells and T cells and the consequent heterogeneous clinical manifestations including respiratory tract infections, autoimmunity, lymphoproliferation and not to mention primary antibody deficiency. In this study, we report a 12-year-old girl with a mutation in the PIK3R1 gene who manifested immunological phenotypes resembling hyper IgM syndrome along with a review of the literature of the previously reported patients.

Methods: Whole exome sequencing was performed to detect the underlying genetic mutation in this patient.

Results: A de novo heterozygous splice site mutation in the hot spot of the PIK3R1 gene within the intron 10 was found (c.1425+1G>A).

Conclusion: Further investigations are required for evaluation of the underlying genetic defects and the possible associations between genetic underpinning and heterogeneous severity and features of the disease.

Biography

Fateme Babaha is a newly M.Sc. graduate in medical immunology from Tarbiat Modares university, Tehran, Iran. Her thesis focused on the investigation of epigenetic phenomena in CVID patients who carried no genetic defects. During her masters she joined the Iranian research center for immunodeficiency, participated in national and international congresses. She also engaged in writing articles to boost her versatility in scientific writing. She has 3 published articles and a few in the line to be published.

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