BCGitis in IPEX syndrome

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Abstract

We present the case of a male infant diagnosed with IPEX (immune dysregulation, polyendocrinopathy, enteropathy, and X-linked) syndrome who presented a generalized rash with a BCG site reaction several months after vaccination. In our patient this dermatological manifestation might be secondary to immune dysregulation, given the important role of Tregs and Th17 cells in mycobacterial immunity, similar to that observed in Kawasaki disease patients.

A 7-month-old boy presented with chronic diarrhea, failure to thrive and a generalized dermatitis with diffuse erythema. His family history was relevant for a brother deceased at 1 year of age with a similar phenotype. Physical examination revealed erythema, edema, induration and ulceration, on the BCG vaccination site applied at birth. A 207-gene primary immunodeficiency NGS panel identified a pathogenic variant c.1099T>C (p.Phe367Leu) in the forkhead domain of the *FOXP3* gene, confirming the diagnosis of IPEX syndrome.

To our knowledge, BCG-site reactions have not previously been described as a clinical manifestation of IPEX syndrome. In summary, BCGitis adds to the plethora of clinical manifestations of IPEX syndrome, but its pathogenesis may be different from other IEI.

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