11q terminal deletion and combined immunodeficiency (Jacobsen syndrome): Case report and literature review on immunodeficiency in Jacobsen syndrome

Abstract

Antibody deficiency is common finding in patients with Jacobsen syndrome (JS). In addition, there have been few reports of T-cell defects in this condition, possibly because most of the reported patients have not been specifically evaluated for T-cell function. In this article, we present a child with an 11q deletion and combined immunodeficiency and we perform a literature overview on immunodeficiency in JS. Our patient presented with recurrent bacterial and prolonged viral infections involving the respiratory system, as well as other classic features of the syndrome. In addition to low IgM, IgG4, and B-cells, also low recent thymic emigrants, helper and naïve T-cells were found. We propose that patients with Jacobsen syndrome need thorough immunological evaluations as T-cell dysfunction might be more prevalent than previously reported. Patients with infections consistent with T-cell defects should be classified as having combined immunodeficiency. © 2016 Wiley Periodicals, Inc.

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