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Autoimmune cytopenia among 382 patients with inborn errors of immunity

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Abstract

Background: Inborn errors of immunity (IEIs) are genetic diseases of impaired host immunity. Although recurrent infections comprise the clinical hallmark of IEIs, other noninfectious manifestations such as autoimmune cytopenia (AIC) are often present.

Objective: We aimed to determine the prevalence of AIC in a cohort of Algerian patients with different categories of IEIs.

Methods: In this study, IEI patients were retrospectively screened for the occurrence of AIC. A questionnaire was filled for all patients to evaluate demographic, laboratory, clinical, and molecular data.

Results: A total of 382 IEI patients (213 (56%) males and 169 (44%) females), with a median age of 12 years were enrolled in this study. The IEIs were distributed as follows: combined immunodeficiencies (CID; n=97); CID with syndromic features (n=50); predominantly antibody deficiencies (AID; n=99); diseases of immune dysregulation (n=24); congenital defects of phagocytes (n=11); defects in intrinsic and innate immunity (n=7), complement deficiencies (n=83), other IEIs (n=11). Autoimmune cytopenia was reported in 44 (11,5%) patients, including 29 (7.6%) with autoimmune hemolytic anemia, 20 (5.2%) with immune thrombocytopenia, 6 (1.6%) with Evans syndrome, and 6 (1.6%) with autoimmune neutropenia. Ten patients (2.6%) showed AIC as the first presentation of the disease. AIC was more common in patients whose IEI were classified as diseases of immune dysregulation (33.3%) and CID (17.5%). Among CID patients, the highest prevalence of AIC was seen in patients with atypical severe combined immunodeficiency (SCID) (42.1%). None of the patients with defects of innate immunity and complement deficiencies showed AIC.

Conclusion: Our study confirms the high prevalence of AIC in IEI patients. AIC should be viewed as a warning sign of PID even in the absence of infectious disease. Patients with IEIs, especially those with diseases of immune dysregulation and atypical SCID should be routinely screened for AIC.