Inherited Classical and Alternative Pathway Complement Deficiencies in Children: A Single Center Experience

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ABSTRACT

Background: Primary complement deficiencies are rare diseases. **Objective:** To retrospectively evaluate the clinical and laboratory findings and complications of patients to increase awareness of pediatricians about complement deficiencies, which are rarely encountered. **Methods:** In this study, the clinical and immunological characteristics of 21 patients who consulted the Immunology Department of our hospital between 2003 and 2017 and were diagnosed with classical or alternative pathway complement deficiency were obtained from the file records. **Results:** Ten patients with C1 inhibitor deficiency, four patients with factor I deficiency, three patients with properdin deficiency, two patients with C8 deficiency, one patient with C1q deficiency, and one patient with C4B deficiency were assessed. The mean age of the patients at diagnosis was 11.4±4.7 years, the age of onset of symptoms was 7.9±3.9 years, and the follow-up period was 6.7±3.9 years. Fourteen cases had a similar medical history in the family. All patients with C1q, factor I, properdin, C8, and C4B deficiencies presented with an infection, and vasculitic rash was present in two patients with factor I deficiency. In addition, immune complex glomerulonephritis was present in one patient with factor I deficiency. Meningococcal, Haemophilus influenzae type B, and pneumococcal vaccines were administered and prophylactic antibiotic treatment was initiated in all patients except patients with C1 inhibitor deficiency. **Conclusions:** Early diagnosis of complement deficiencies can facilitate prevention of life-threatening complications such as severe bacterial infections by considering prophylactic antibiotics and vaccines. In patients with C1 inhibitor deficiency, achieving an acurate early diagnosis will assist in the management and timely treatment of life-threatening attacks such as upper airway obstruction and improve outcomes.

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