HYPER-IGM SYNDROME - CLINICAL CASE OF A CHILD WITH LATE DIAGNOSIS

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We present a clinical case of a patient with frequent infections mainly affecting the respiratory system, including pneumocystis pneumonia at 6 months of age, when primary humoral immune deficiency is diagnosed. Initially, intravenous immunoglobulin replacement therapy was started, but irregularly - every 2-4 months, and there is a normal frequency of infectious episodes. Subsequently, at the age of 12, elevated IgM levels were found, including in periods free of acute infection, and genetic studies proved a variant of CD40LG. At the age of 15, regular subcutaneous replacement therapy was initiated, serum IgG levels >5 g/l are maintained, and a reduction in the number of infections is reported. The dilemma for the need of hematopoietic stem cell transplantation as a definitive treatment is logical.