THE CHALLENGE PID: RECOGNITION IN CLINICAL PRACTICE

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Primary immune deficiencies are rare diseases characterized by increased incidence of infections, unusual causative agents and unusual sites of infections, and an unusual combination of autoimmune manifestations and/or malignancies. The characteristic of the infectious manifestations could be not only an alarming sign for the workup for primary immune deficiency, but also a guiding criterion for its' subtype.

The experience of our center on the diagnostic challenges and the disease course of 26 patients with immunologically proven and genetically specified in most of them PID will be presented: X-linked agammaglobulinemia – 3, hyper-IgM syndrome – 1, neutrophils dysfunction - 2, Severe congenital neutropenia of Kostmann - 2, defect in innate immunity - 5, Common variable immune deficiency - 7, Severe combined immune deficiency - 3 and unspecified - 3.

The clinical scenarios leading to the diagnosis are considered, the proven causative agents of infections are analyzed and a comparison with the literature data to date is presented.