IMMUNE-FUNCTION DISTURBANCES IN POLISH CHILDREN WITH GENETICALLY DETERMINED FORM OF HLH.

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Hemophagocytic lymphohistiocytosis (HLH) is a very severe systemic syndrome associated with hyperactivation of macrophages and impaired immune regulation. The most typical clinical manifestation include: pancytopenia, hepatosplenomegaly, hyperferritynemia, hypertriglicerydemia, hipofibrynogenemia and impaired immune cells function. Two forms of HLH are currently recognized: genetically determined HLH, and HLH developed in course of primary diseases like autoimmune disorders, cancers or infections. In polish population genetically determined cases constitute the minority of patients with HLH.

The aim of our study was to assess the presence of immune function defects in group of children with clinical suspicion of HLH.

In the whole group of 45 children only 3 cases with UNC13D mutation were identified. The other four children with recurrent disease and unidentified mutation were classified as genetically determined form.

In all cases with congenital HLH, cytotoxic abilities of cells were impaired compared to control group (p=0,005). The perforin expression was normal or higher then observed in controls (p=0,002). In patients with mutation in MUNC protein gene, degranulation was lower than in healthy children (<5%).

Immune function of children with known mutations is depressed. Currently unknown mutations resulting in similar clinical presentation and parallel immune disturbances have to be determined.