CLINICAL CASE OF STING-ASSOCIATED VASCULOPATHY WITH EARLY ONSET (SAVI) - DIFFICULTIES IN DIAGNOSIS

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STING-associated early-onset vasopathy (SAVI) is an auto-inflammatory syndrome classified as a type 1 interferonopathies, part of the spectrum of primary immune deficiencies (PID).

We present a 2-year-old boy who has had tachypnea since birth. At 6 months of age during episode with rotavirus gastroenteritis, an anemic syndrome (Hbg 80 g/l) was found and was attributed to iron deficiency anemia and iron therapy was started. Autoimmune diseases and cystic fibrosis have been discussed and rejected in the differential diagnostic plan. After performing rigid bronchoscopy pavement of erythrocytes and hemosiderophages were seen and the diagnosis of idiopathic hemosiderosis has been accepted. Due to frequent skin infections, the immune status was examined and elevated IgG and IgA levels were found with a reduced absolute CD4+ T-lymphocyte count. HIV infection in the patient and the parents was excluded. Due to the delay in physical development and unstable stools, antibodies for celiac disease were additionally tested and despite the unconvincing data, the child had to follow strict gluten-free and diary-free diet. At subsequent follow-up hospitalization in the clinic a progressive decline in helper-induced T-lymphocytes and B-lymphocytes and a relative increase in NK cells were observed. NGS analysis for selected PID genes revealed pathogenic mutation in the STING1 gene (TMEM173, c.461 A> G (p.Asn154Ser), heterozygote), which contributed to a definitive diagnosis of SAVI. Currently we have planned to initiate therapy with JAK kinases inhibitor with close follow-up. In some cases, diagnosis of PID is a long and multi-step process, requiring collaboration of teams of different specialists.