PAEDIATRIC DERMATOLOGY

FROM SEVERE COMBINED IMMUNODEFICIENCY TO OMENN'S SYNDROME

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Background: Severe combined immunodeficiency (SCID) is the most severe form of primary immunodeficiency disease, and is very heterogeneous. Omenn syndrome (OS) is a particular variant of SCID.

Observation: A 3-month-old boy, born to consanguineous parents, was admitted to our dermatology department with an exfoliative erythroderma associated with eczematous patches and alopecia of the scalp, eyelashes and eyebrows, without lymphadenopathy or hepatosplenomegaly. He displayed mild respiratory symptoms and chronic diarrhea since birth and was treated for an acute otitis media caused by Escherichia coli at the age of 2 months. A complete blood count showed marked leukocytosis with eosinophilia and lymphocytosis. These clinical and biological findings however changed within a few days. The patient no longer had erythroderma and showed regrowth of hairs, eyelashes and eyebrows. Biology revealed less marked eosinophilia with mild lymphopenia and no leukocytosis. Immunoglobulin analysis showed undetectable IgG, IgA, IgM and IgE levels and immunological investigations concluded to the diagnosis of SCID T-B-NK+. Mutation analysis revealed a homozygous c.1338C>G (p.Cys446Trp) deleterious mutation in the RAG2 gene. A hematopoietic stem cell transplantation is planned in the near furture.

Key message: SCID T-B-NK+ typically presents as recurrent opportunistic infections, chronic diarrhea, eczematous dermatitis, very low immunoglobulin blood levels, lymphopenia and inconstantly eosinophilia. However, clinical and biological symptoms firsty objectified in our patient (severe exfoliative erythroderma, loss of eyelashes and eyebrows, lymphocytosis and severe eosinophilia) were rather suggestive of OS. The absence of lymphadenopathy, splenomegaly and hepatomegaly as well as the low Ig E blood level are all arguments against the diagnosis of OS. Our patient had seemingly an intermediate form of SCID also called Omenn-like, which associate criteria of typical SCID T-B-NK+ with some features of OS. The RAG2 deficiency is common to both SCID T-B-NK+ and OS, which explains the phenotypic overlap displayed by our patient.