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Chronic mucocutaneous candidiasis with STAT1 mutation in early childhood

B.A. Lögering[1]; P. Weisenseel[1]; B. Kunz[1]; J. Hiller[2]; C. Traidl-Hoffmann[3]; J.-L. Casanova[4]; K. Reich[1]

[1]Dermatologikum Hamburg, Hamburg, Germany, [2]Institute of Environmental Medicine, Medical Faculty of the Technische Universität München, Munich, Germany, [3]Institute of environmental medicine, Medical Faculty of the Technische Universität München, Munich, Germany, [4]St. Giles Laboratory of Human Genetics of Infectious Diseases, The Rockefeller University, New York, United States

Chronic mucocutaneous candidiasis (CMC) is a rare disease which is characterized by an increased tendency to persistent or recurrent infections of the skin and mucous membranes caused by Candida albicans.

We report a case of a four-year-old boy who had been suffering from recurrent Candida infections of the oral mucosa and from erythematous papules on his face since early infancy whereas his overall development was mostly normal. The family anamnesis was negative.

Perlèche, a white coating of the mucous membranes, swelling of the lips and erosions of the mucosal lower lip were visible. Additionally,

there was an onychomycosis caused by Candida. Regularly Candida albicans was isolated from the oral mucosa, once also Candida dubliensis.

A weak reaction to miconazole and itraconazole was seen in the determination of resistance. Local antimycotics were insufficiently effective. Repeatedly bacterial infections occurred, especially infections of the skin by Staphylococcus aureus, further more our patient suffered a few times from bronchitis and once from pneumonia. A stable condition could be achieved on prophylaxis with fluconazole 3 mg/kg body weight daily.

A gain-of-function mutation in STAT1 allel could be identified in our patient. Newer investigations revealed that these gain-of-function STAT1 alleles may cause a CMC by impairing IL-17 immunity. The cause for this defect seems to be an enhanced cellular response to STAT1 dependent repressors of IL-17-producing T-cells.

The term chronic mucocutaneous candidiasis disease (CMCD) best describes patients who show characteristics of CMC without any underlying illness or immune deficiency but affected IL-17 immunity. The occurrence of recurrent Candida infections beyond early infancy is seldom perceived among healthy children. The hereby presented young patient shows that innate immune deficiency has to be taken into consideration when being confronted with chronic recurrent infections of the skin and mucous membranes in childhood.

By means of further genetic clearance the pathogenesis of so far predominantly clinically defined dermatoses will be identified, better understood and seen in a completely new context.