



INFECTIOUS DISEASES (BACTERIAL, FUNGAL, VIRAL, PARASITIC, INFESTATIONS)

CHRONIC MUCOCUTANEOUS CANDIDIASIS: MORE THAN A SKIN DISEASE

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Background: Chronic mucocutaneous candidiasis (CMC) is characterized by susceptibility to candida Infections: skin, nails, and mucous membranes. Host factors that result in increased susceptibility to infections to Candida species are AD hyper-IgE syndrome, gain of function (GOF) STAT1 defects, CARD9 defects, IL17 deficiency, and AR autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy syndrome type 1 (APS-1 or APECED), caused by AIRE deficiency. In addition, other fungal infections, staphylococcal and mycobacterial disease were described in these patients

Observations: We present 7 patients with CMC, belonging to 5 unrelated and non-consanguineous families

All patients exhibit oral mucosa and recurrent fungal cutaneous infections. 4/7 patients had nail dystrophy and eczema.

Other infections were identified in these patients, been staphylococcus aureus abscess the most frequent. 2 patients had meningitis (Cryptococcus and Enterococcus were identified), all patients presented recurrent respiratory tract infections, pulmonary Tuberculosis, as well as, extrapulmonary involvement in one patient. Endocrinopathies were observed in 2 of them.

Other findings were: hemolytic autoimmune anemia, renal insufficiency and hepatitis.

Molecular diagnosis showed mutations: STAT 1 GENE: 4p, IL 17RA: 2p, AIRE 1p.

Key message: Clinical recognition of CMC is important as well as the genetic study in order to recognized the host immunological risk factors for CMC hand give the correct family genetic counseling and also in the development of new targeted treatments

