Chronic mucocutaneous candidiasis

GENERAL INFORMATION

Description:

CMC often is associated with defects in cell-mediated immunity and can be accompanied by endocrinopathies, thymoma, or multiple autoimmune conditions. CMC does not represent a specific disease but rather a phenotypic presentation spectrum of immunologic, endocrinologic, and autoimmune disorders. CMC can be divided broadly into the following 3 categories: CMC without associated endocrinopathy, CMC with associated endocrinopathy, CMC associated with thymoma.

Alternative names:

- CMCT
- CMC
- Candidiasis, chronic mucocutaneous, with thyroid disease

Classification:

- Other well-defined immunodeficiency syndromes

Inheritance:

Autosomal recessive/Autosomal dominant

OMIM:

- %606415 Candidiasis, familial chronic mucocutaneous, autosomal dominant, with thyroid disease
- %114580 Candidiasis, familial chronic mucocutaneous, autosomal dominant
- %212050 Candidiasis, familial chronic mucocutaneous, autosomal reccesive

Incidence:

Incidence is not known yet.

CLINICAL INFORMATION

Description:

Patients present with an early onset of superficial candidiasis affecting nails and mouth and occasionally the oesophagus. Generally, a history of recurrent or persistent superficial candidal infections of the oral cavity (thrush) or intertriginous or periorificial areas. Infants often present with recalcitrant thrush and/or candidal diaper dermatitis. These infections can be followed by more extensive scaling of skin lesions, as well as thickened nails and red swollen periungual tissues. Infections are typically unresponsive to routine topical antifungal preparations. Some patients also have an endocrinopathy causing hypocalcaemia due to parathyroid insufficiency, hypothyroidism, and adrenal insufficiency. Increased susceptibility to bacterial infections, tuberculosis, herpesviruses, and toxoplasmosis. Severe forms may progress to a more generalized combined immunodeficiency.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Candidiasis familial chronic, ORPHANET
- Candidiasis, Chronic Mucocutaneous, eMedicine

Therapeutic options:

- Treatment with topic and systemic antimycotic drugs (fluconazole or itraconazole) results in temporary recovery, but is unable to reach complete remission. Promising results with Amphotericine B. Resistence to these antifungals may occur. Intravenous immunoglobulins should be used for patients with recurrent bacterial infections. Continuous antibiotics tend to exacerbate the candidiasis. A regular surveillance for significant endocrine disease is essential to mantain.
- Candidiasis, Chronic Mucocutaneous, eMedicine

Research programs, clinical trials:

- European Initiative for Primary Immunodeficiencies

GENE INFORMATION

Names:

HUGO name:

Localization:

Maps:

(Map View)

Other gene-based resources:

Ensembl: , GENATLAS: , GeneCard: , UniGene: , LocusLink: , euGenes: , GDB:

PROTEIN INFORMATION

Description:

Other features:

Expression pattern for human:

Tissue	Exp. (%)	Clones

OTHER RESOURCES

Societies:

General:

- International Patient Organization for Primary Immunodeficiencies
- Immune Deficiency Foundation
- European Society for Immunodeficiencies
- NIH/National Institute of Allergy and Infectious Diseases