Opis choroby *

Definicja

A rare primary immunodeficiency characterized by increased susceptibility to infections with candida albicans and weakly pathogenic mycobacteria, such as mycobacterium bovis. Patients present in infancy with chronic mucocutaneous candidiasis of varying severity, disseminated mycobacterial disease, absence of palpable axillary and cervical lymph nodes, reduced thymus size, and variable hepatosplenomegaly. The immunological phenotype comprises mild T-cell lymphopenia, absence of type 1 natural killer T-cells and mucosal-associated invariant T-cells, and low levels of type 3 innate lymphoid cells.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive MSMD due to complete RORgamma receptor defiency
Autosomalna recesywna MSMD z powodu całkowitego niedoboru receptora RORgamma
Autosomalny recesywny niedobór odporności spowodowany mutacją RORC
Autosomal recessive primary immunodeficiency due to RORC mutation

Kod ORPHA
477857

Kod OMIM
616622

Kod ICD10
D84.8

Kod ICD11
-

---

*Źródło

orphanet