Opis choroby *

Definicja

Autosomal recessive mendelian susceptibility to mycobacterial diseases (MSMD) due to partial IFNgammaR2 deficiency is a genetic variant of MSMD (see this term) characterized by a partial deficiency in IFN-gammaR2, leading to a residual response to IFN-gamma and consequently to recurrent, moderately severe infections with bacillus Calmette-Guérin (BCG) and other environmental mycobacteria (EM).

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive MSMD due to partial IFNgammaR2 deficiency
Autosomalna recesywna mendlowska podatność na choroby mykobakteryjne z powodu częściowego niedoboru receptora 2 interferonu gamma
Autosomalna recesywna MSMD z powodu częściowego niedoboru IFNgammaR2
Autosomalna recesywna MSMD z powodu częściowego niedoboru receptora 2 interferonu gamma
Autosomal recessive MSMD due to partial interferon gamma receptor 2 deficiency
Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial interferon gamma receptor 2 deficiency

Kod ORPHA
319574

Kod OMIM
614889

Kod ICD10
D84.8

Kod ICD11
4A00.2

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*Źródło

orphanet