Opis choroby *

Definicja

A group of genetic variants of mendelian susceptibility to mycobacterial diseases (MSMD) comprised of MSMD due to complete interferon gamma receptor 1 (IFN-gammaR1) deficiency, complete IFN-gammaR2 deficiency, complete interleukin-12 subunit beta (IL12B) deficiency, complete interleukin-12 receptor subunit beta-1 (IL-12RB1) deficiency and complete ISG15 deficiency.

Dane

Klasyfikacja
Kategoria

Synonimy
Autosomal recessive MSMD due to a complete deficiency
Autosomalna recesywna MSMD z powodu całkowitego niedoboru

Kod ORPHA
319535

Kod OMIM
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Kod ICD10
D84.8

Kod ICD11
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*Źródło

orphanet