

Opis choroby *

Definicja

X-linked (XR) Mendelian susceptibility to mycobacterial diseases (MSMD; see this term) describes a rare group of immunodeficiencies due to specific mutations in the inhibitor of kappa light polypeptide gene enhancer in B-cells, kinase gamma (*IKBKG*) or the cytochrome b-245, beta polypeptide (*CYBB*) genes. They are characterized by mycobacterial infections, occurring in males.

Dane

Klasyfikacja Choroba	Synonimy X-linked MSMD MSMD sprzężona z chromosomem X	
Kod ORPHA 319605	Kod OMIM 300645	Kod ICD10 D84.8
Kod ICD11 4A00.2		

*Źródło

orphanet