Mendelian susceptibility to mycobacterial diseases due to complete IFNgammaR1 deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Mendelian susceptibility to mycobacterial diseases due to complete IFNgammaR1 deficiency. ORPHA:99898

Mendelian susceptibility to mycobacterial diseases (MSMD) due to complete interferon gamma receptor 1 (IFN-gammaR1) deficiency is a genetic variant of MSMD (see this term) characterized by a complete deficiency in IFN-gammaR1, leading to impaired IFN-gamma immunity and, consequently, to severe and often fatal infections with bacillus Calmette-Guérin (BCG) and other environmental mycobacteria (EM).