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Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency. ORPHA:319581

Autosomal dominant (AD) mendelian susceptibility to mycobacterial diseases (MSMD) due to partial interferon gamma receptor 1 (IFN-gammaR1) deficiency is a genetic variant of MSMD (see this term) characterized by a partial deficiency leading to impaired IFN-gamma immunity and, consequently, recurrent, moderately severe infections with bacillus Calmette-Guérin (BCG) and other environmental mycobacteria (EM).