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Autosomal recessive 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 recessive mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency. ORPHA:319569*

Autosomal recessive (AR) mendelian susceptibility to mycobacterial diseases (MSMD) due to partial IFN-gammaR1 deficiency is a genetic variant of MSMD (see this term) characterized by a partial deficiency in IFN-gammaR1, 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).