Autosomal recessive mendelian susceptibility to mycobacterial diseases due to a complete deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive mendelian susceptibility to mycobacterial diseases due to a complete deficiency. ORPHA:319535

Autosomal recessive (AR) mendelian susceptibility to mycobacterial diseases (MSMD) due to a complete deficiency describes a group of genetic variants of MSMD (see this term) 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 (see these terms).