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

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal dominant mendelian susceptibility to mycobacterial diseases due to a partial deficiency. ORPHA:319543

Autosomal dominant (AD) mendelian susceptibility to mycobacterial diseases (MSMD) due to a partial deficiency describes a group of variants of MSMD (see this term) due to dominantly inherited partial deficiencies in interferon gamma receptor 1 (IFN-gammaR1), IFN-gammaR2, signal transducer and activator of transcription 1 (STAT1) or interferon regulator factor 8 (IRF8) (see these terms).