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Combined immunodeficiency due to ORAI1 deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Combined immunodeficiency due to ORAI1 deficiency. ORPHA:317428

Combined immunodeficiency (CID) due to ORAI1 deficiency is a form of CID due to Calcium release activated Ca^{2+} (CRAC) channel dysfunction (see this term) characterized by recurrent infections, congenital myopathy, ectodermal dysplasia and anhydrosis.