Combined immunodeficiency due to CRAC channel dysfunction

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Combined immunodeficiency due to CRAC channel dysfunction. ORPHA:169090

Combined immunodeficiency (CID) due to Ca2+ release activated Ca2+(CRAC) channel dysfunction is a form of CID characterized by recurrent infections, autoimmunity, congenital myopathy and ectodermal dysplasia. It comprises two sub-types that are due to mutations in the ORAI1 and STIM1 genes: CID due to ORAI1 deficiency and CID due to STIM1 deficiency.