

Opis choroby *

Definicja

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.

Dane

Klasyfikacja

Choroba

Synonimy

Immune dysfunction due to T-cell inactivation due to calcium entry defect

Dysfunkcja odporności z powodu inaktywacji komórek T spowodowanych defektem wejścia wapnia

Kod ORPHA

169090

Kod OMIM

612783

Kod ICD10

D81.8

Kod ICD11

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*Źródło

orphanet