

Opis choroby *

Definicja

Combined immunodeficiency (CID) due to *STIM1* deficiency is a form of CID due to Calcium release activated Ca²⁺(CRAC) channel dysfunction (see this term) characterized by recurrent infections, autoimmunity, congenital myopathy and ectodermal dysplasia.

Dane

Klasyfikacja	Synonimy
Podtyp kliniczny	CID due to STIM1 deficiency CID z powodu deficytu STIM1

Kod ORPHA	Kod OMIM	Kod ICD10
317430	612783	D81.8

Kod ICD11
4A01.1Y

[*Źródło](#)

orphanet