

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

Podtyp kliniczny

Synonimy

CID due to STIM1 deficiency

CID z powodu deficytu STIM1

Kod ORPHA

317430

Kod OMIM

612783

Kod ICD10

D81.8

Kod ICD11

4A01.1Y

*Źródło

orphanet