

Opis choroby *

Definicja

Combined immunodeficiency (CID) due to *Orai1* deficiency is a form of CID due to Calcium release activated Ca²⁺ (CRAC) channel dysfunction (see this term) characterized by recurrent infections, congenital myopathy, ectodermal dysplasia and anhydrosis.

Dane

Klasyfikacja

Podtyp kliniczny

Synonimy

CID due to Orai1 deficiency

CID z powodu niedoboru Orai1

Kod ORPHA

317428

Kod OMIM

612782

Kod ICD10

D81.8

Kod ICD11

4A01.1Y

[*Źródło](#)

orphanet