

## Opis choroby \*

### Definicja

Combined immunodeficiency (CID) due to *Orai1* deficiency is a form of CID due to Calcium release activated Ca<sup>2+</sup> (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

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#### \*Źródło

orphanet