

## Opis choroby \*

### Definicja

The Immunodeficiency, Centromeric region instability, Facial anomalies syndrome (ICF) is a rare autosomal recessive disease characterized by immunodeficiency, although B cells are present, and by characteristic rearrangements in the vicinity of the centromeres (the juxtacentromeric heterochromatin) of chromosomes 1 and 16 and sometimes 9.

### Dane

#### Klasyfikacja

Zespół wad wrodzonych Immunodeficiency-centromeric instability-facial anomalies syndrome  
Niedobór odporności - niestabilność centromeryczna - anomalie twarzy

#### Synonimy

#### Kod ORPHA

2268

#### Kod OMIM

614069

#### Kod ICD10

D84.8

#### Kod ICD11

4A01.00

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#### \*Źródło

orphanet