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

Synonimy
Immunodeficiency-centromeric instability-facial anomalies syndrome
Niedobór odporności - niestabilność centromeryczna - anomalie twarzy

Kod ORPHA
2268

Kod OMIM
614069

Kod ICD10
D84.8

Kod ICD11
4A01.00

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*Źródło

orphanet