

Opis choroby *

Definicja

A rare genetic disease characterized by frequent infections associated with neutropenia and IgA deficiency, in combination with osteoporosis and skeletal anomalies, such as posterior spinal arch fusion defect, metacarpal subluxation, syndactyly, and camptodactyly. Reported dysmorphic features include synophrys, anteverted nostrils, and single palmar crease. There have been no further descriptions in the literature since 1972.

Dane

Klasyfikacja

Choroba

Kod ORPHA

2390

Kod OMIM

246550

Kod ICD10

D70

Kod ICD11

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*Źródło

orphanet