Opis choroby *

Definicja

A rare genetic syndromic intellectual disability of broad phenotypic range characterized by developmental delay and variable clinical features which most commonly, but not consistently, include aplasia or hypoplasia of the distal phalanx or nail of the fifth digit, and coarse facial features.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
CSS
CSS

Kod ORPHA
1465

Kod OMIM
618362

Kod ICD10
Q87.1

Kod ICD11
LD27.0Y

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*Źródło

orphanet