

Opis choroby *

Definicja

Brachytelephalangi chondrodysplasia punctata (BCDP) is a form of non-rhizomelic chondrodysplasia punctata, a primary bone dysplasia, characterized by hypoplasia of the distal phalanges of the fingers, nasal hypoplasia, epiphyseal stippling appearing in the first year of life, as well as mild and non-rhizomelic shortness of the long bones.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

79345

Kod OMIM

602497

Kod ICD10

Q77.3

Kod ICD11

LD24.04

*Źródło

orphanet