

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	Kod OMIM	Kod ICD10
79345	602497	Q77.3
Kod ICD11		
LD24.04		

*Źródło

orphanet