Opis choroby *

Definicja

An early-onset distal osteolysis characterised by severe resorption of the hands and feet and absence of the distal and middle phalanges. It has been described in a son and daughter born to consanguineous parents. Other manifestations include distal muscular hypertrophy, flexion contractures, short stature, mild intellectual deficit and characteristic facies (maxillary hypoplasia, exophthalmos, and a broad nasal tip). It is transmitted as an autosomal recessive trait.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Distal osteolysis-short stature-intellectual disability syndrome
Osteoliza dystalna - niski wzrost - niepełnosprawność intelekualna
Zespół Petit i Frynsa
Petit-Fryns syndrome

Kod ORPHA
2776

Kod OMIM
259610

Kod ICD10
M89.5

Kod ICD11
FB86.2

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*Źródło

orphanet