Opis choroby *

Definicja

A rare, primary bone dysplasia characterized by rhizomelic limb shortening, punctate calcifications in cartilage with epiphyseal and metaphyseal abnormalities (chondrodysplasia punctata) and coronal cleft vertebrae associated with profound postnatal growth deficiency, early-onset cataracts, severe intellectual disability and seizures.

Dane

Klasyfikacja
Choroba

Synonimy
RCDP
RCDP

Kod ORPHA
177

Kod OMIM
600121

Kod ICD10
Q77.3

Kod ICD11
LD24.04

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*Źródło

orphanet