

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

*Źródło

orphanet