

Opis choroby *

Definicja

Parana hard skin syndrome is a rare genetic skin disorder characterized by very early-onset of progressive skin thickening over the entire body (except for eyelids, neck and ears), progressively limited joint mobility with gradual freezing of joints, and eventual severe chest and abdomen movement restriction, manifesting with restrictive pulmonary disease, which may lead to death. Additional features include severe growth restriction and osteoporosis. There have been no further descriptions in the literature since 1974.

Dane

Klasyfikacja

Choroba

Synonimy

Hard skin syndrome, Parana type

Zespół twardej skóry, typu Parana

Kod ORPHA

2812

Kod OMIM

260530

Kod ICD10

L91.8

Kod ICD11

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*Źródło

orphanet