Opis choroby *

Definicja

A rare, genetic, ectodermal dysplasia syndrome characterized by corneal epithelial changes (ranging from roughening to nodular irregularities), diffuse palmoplantar hyperkeratosis with thickened, erythematous, scaly lesions affecting the elbows, knees and knuckles, distal onycholysis, brachydactyly accompanied by a single transverse palmar crease, short stature, premature birth, and increased susceptibility to tooth decay. Ocular symptoms include photophobia, reduced night vision, burning and watery eyes, and varying visual acuity. There have been no further descriptions in the literature since 1984.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
CDO syndrome
Zespół rogówkowo-skórno-kostny
Stern-Lubinsky-Durrie syndrome

Kod ORPHA
3194

Kod OMIM
122440

Kod ICD10
H18.5

Kod ICD11
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*Źródło

orphanet