

## 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

<b>Klasyfikacja</b>	<b>Synonimy</b>
Zespół wad wrodzonych CDO syndrome	
	Zespół rogówkowo-skórno-kostny
	Stern-Lubinsky-Durrie syndrome

<b>Kod ORPHA</b>	<b>Kod OMIM</b>	<b>Kod ICD10</b>
3194	122440	H18.5

### Kod ICD11

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### \*Źródło

orphanet