

## Opis choroby \*

### Definicja

Chondroectodermal dysplasia with night blindness is a rare genetic bone development disorder characterized by proportionate short stature, nail dysplasia (enlarged, convex, hypertrophic nails), hypodontia and night blindness. Osteopenia, a tendency to present fractures, talipes varus with abnormal gait, ear infections, and watering eyes due to narrow tear ducts are frequently associated. Radiologically patients present delayed bone age on wrist X-rays, platyspondyly, and broad metaphyses of humeri with dense and thickened growth plates.

### Dane

### Klasyfikacja

Choroba

#### Kod ORPHA

319195

#### Kod OMIM

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#### Kod ICD10

Q77.6

#### Kod ICD11

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### \*Źródło

orphanet