## Opis choroby \*

## Definicja

A rare ciliopathy characterized by congenital cataract with secondary glaucoma, developmental delay, short stature, multiple skeletal abnormalities (spinal deformities, limb anomalies, delayed bone age), dental anomalies (oligodontia, enamel defects), dysmorphic facial features (including coarse facies, low hairline, epicanthal folds, flat and broad nasal bridges, and retrognathia), and stroke. Other recurrent manifestations are hearing loss and nephrocalcinosis.

Dane

Klasyfikacja Synonimy

Choroba Oculo-skeleto-dental syndrome

Oculo-skeleto-dental syndrome

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 557003
 618440
 Q87.8

**Kod ICD11** 

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## \*Źródło

orphanet