

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

Choroba

Synonimy

Oculo-skeleto-dental syndrome

Oculo-skeleto-dental syndrome

Kod ORPHA

557003

Kod OMIM

618440

Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet