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