Opis choroby *

Definicja

Oculoauricular syndrome, Schorderet type is a rare, genetic developmental defect during embryogenesis syndrome characterized by various ophthalmic anomalies (including congenital microphthalmia, microcornea, cataract, anterior segment dysgenesis, ocular coloboma and early onset rod-cone dystrophy) and abnormal external ears (low-set pinna with crumpled helix, narrow intertragic incisures, abnormal bridge connecting the crus of the helix and the antihelix, narrow external acoustic meatus, and lobule aplasia).

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
157962

Kod OMIM
612109

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet