Oculoauricular syndrome, Schorderet type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Oculoauricular syndrome, Schorderet type. ORPHA:157962

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).