Oculoauriculofrontonasal syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Oculoauriculofrontonasal syndrome. ORPHA:398156

Oculoauriculofrontonasal syndrome is a rare dysostosis syndrome characterized by vertical, median craniofacial clefting of fronto-naso-maxillary structures associated with auriculo-mandibular malformations, manifesting with highly variable craniofacial features which include hypertelorism, eyelid colobomas, orbital dystopia, epibulbar dermoids, nasal anomalies (e.g. wide nasal bridge, bifid nose, widely separated, slit-like nares, nasal bone dysplasia), auricular and middle ear dysplasia (microtia, aural stenosis, pre-auricular skin tags/pits), cleft lip/palate, mandibular/maxillary hypoplasia and facial asymmetry. Intracranial abnormalities and extra-craniofacial features are frequently associated.