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Mandibulofacial dysostosis-macroblepharon-macrostomia syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Mandibulofacial dysostosis-macroblepharon-macrostomia syndrome. ORPHA:357158

Mandibulofacial dysostosis-macroblepharon-macrostomia syndrome is a rare developmental defect during embryogenesis disorder characterized by macroblepharon, ectropion, and facial dysmorphism which includes severe hypertelorism, downslanting palpebral fissures, posteriorly rotated ears, broad nasal bridge, long and smooth philtrum, and macrostomia with thin upper lip vermilion border. Other features may include large fontanelles, prominent metopic ridge, thick eyebrows, mild synophrys, increased density of upper eyelases, anterverted nares, abnormal dentition and capillary hemangioma.