Dysmorphism-cleft palate-loose skin syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Dysmorphism-cleft palate-loose skin syndrome. ORPHA:1779

Dysmorphism-cleft palate-loose skin syndrome is a rare, genetic developmental defect during embryogenesis characterized by severe psychomotor delay, intellectual disability, congenital, symmetrical circumferential skin creases of arms and legs, cleft palate, and facial dysmorphism (incl. elongated face, high forehead, blepharophimosis, short palpebral fissures, microphthalmia, microcornea, epicanthic folds, telecanthus, microtia, posteriorly angulated ears, broad nasal bridge, microstomia and micrognathia). Additional features reported include short stature, microcephaly, hypotonia, pectus excavatum, severe scoliosis, hypoplastic scrotum, and mixed hearing loss.