Cerebrofacioarticular syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Cerebrofacioarticular syndrome. ORPHA:314679

Cerebrofacioarticular syndrome is a rare multiple congenital anomalies syndrome characterized by mild to severe intellectual disability, a distinctive facial gestalt (blepharophimosis, maxillary hypoplasia, telecanthus, microtia and atresia of the external auditory meatus) as well as skeletal and articular abnormalities (e.g. camptodactyly of the fingers, cutaneous syndactyly, talipes equinovarus, flexion contractures of the proximal interphalangeal joints, hip or elbow subluxation, joint laxity). Affected individuals also present neonatal hypotonia, variable respiratory manifestations, chronic feeding difficulties and gray matter heterotopia.