Colobomatous microphthalmia-rhizomelic dysplasia syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Colobomatous microphthalmia-rhizomelic dysplasia syndrome. ORPHA:424099

Colobomatous microphthalmia-rhizomelic dysplasia syndrome is a rare, genetic developmental defect during embryogenesis characterized by a range of developmental eye anomalies (including anophthalmia, microphthalmia, colobomas, microcornea, corectopia, cataract) and symmetric limb rhizomelia with short stature and contractures of large joints. Intellectual disability with autistic features, macrocephaly, dysmorphic features, urogenital anomalies (hypospadia, cryptorchidism), cutaneous syndactyly and precocious puberty may also be present.