Anophthalmia-megalocornea-cardiopathy-skeletal anomalies syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Anophthalmia-megalocornea-cardiopathy-skeletal anomalies syndrome. ORPHA:1101

Anophthalmia-megalocornea-cardiopathy-skeletal anomalies syndrome is a multiple congenital anomalies syndrome, reported in the offsprings of a consanguineous couple and characterized by multiple congenital skeletal (dolichocephaly, skull asymmetry, camptodactyly, clubfoot), muscular (muscle hypoplasia), ocular (anophthalmia, buphthalmos, retinal detachment, aniridia (see this term)) and cardiac (prolapse of tricuspid valves, mitral and tricuspid insufficiency) abnormalities. An autosomal recessive inheritance with variable expressivity was suspected. There have been no further descriptions in the literature since 1992.