Anophthalmia plus syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Anophthalmia plus syndrome. ORPHA:1104

Anophthalmia plus syndrome is a very rare multiple congenital anomaly syndrome characterized by the presence of anophthalmia or severe microphthalmia, cleft lip/palate, facial cleft and sacral neural tube defects, along with various additional anomalies including congenital glaucoma, iris coloboma, primary hyperplastic vitreous, hypertelorism, low-set ears, clinodactyly, choanal atresia/stenosis, dysgenesis of sacrum, tethering of spinal cord, syringomyelia, hypoplasia of corpus callosum, cerebral ventriculomegaly and endocrine abnormalities. An autosomal recessive inheritance has been suggested.