Autosomal recessive chorioretinopathy-microcephaly syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive chorioretinopathy-microcephaly syndrome. ORPHA:2518

Autosomal recessive chorioretinopathy-microcephaly syndrome is a rare neuro-opthalmological disease characterized by severe microcephaly of prenatal onset (with diminutive anterior fontanelle and sutural ridging), growth retardation, global developmental delay and intellectual disability (ranging from mild to profound), dysmorphic features (sloping forehead, micro/retrognathia, prominent ears) and visual impairments (including microphthalmia to anophtalmia, generalized retinopathy or multiple punched-out retinal lesions, retinal folds with retinal detachment, optic nerve hypoplasia, strabismus, nystagmus). Brain MRI may show reduced cortical size, cerebral hemispheres, corpus callosum, pachygyria, symplified gyral folding or normal pattern. Other associated features include epilepsy and neurological deficits.