Opis choroby *

Definicja

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.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Autosomal recessive chorioretinopathy-microcephaly-intellectual disability syndrome
Zespół toksoplazmozy rzekomej

Kod ORPHA
2518

Kod OMIM
616335

Kod ICD10
Q87.8

Kod ICD11
9B61

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*Źródło

orphanet