

Opis choroby *

Definicja

A rare ophthalmic disorder characterized by mostly unilateral failure of the regression of a fetal ocular vessel component, the tunica vasculosa lentis and/or the hyaloid system, resulting in an anterior (presenting with microphthalmia, leukocoria, cataract, glaucoma, elongated ciliary processes, shallow anterior chamber, and retrolental fibrovascular membranes, among others) or posterior disease subtype (with microphthalmia, leukocoria, presence of a retinal fold or detachment, hypo- or dysplastic optic nerve, and vitreous membranes and stalk), respectively. Most patients present with a combination of the two subtypes.

Dane

Klasyfikacja

Choroba

Synonimy

Congenital retinal detachment

PFVS

PHPV

Choroba NCRNA

Wrodzone odwarstwienie siatkówki

Zespół przetrwałego unaczynienia płodowego

NCRNA disease

Non-syndromic congenital retinal non-attachment

PFVS

PHPV

Persistent fetal vasculature syndrome

Kod ORPHA

91495

Kod OMIM

611308

Kod ICD10

Q14.0

Kod ICD11

LA13.Y

*Źródło

orphanet