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

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*Źródło

orphanet