Opis choroby *

Definicja

A rare benign eye tumor characterized by the presence of glial cells, vascular tissue, and sheets of pigment epithelial cells lacking the distribution and organization of the normal retina and retinal pigment epithelium. The lesion is most commonly found unilaterally as a slightly elevated mass in a peripapillary location but can also occur in the macula or the retinal periphery. It is sometimes associated with neurofibromatosis type 1 or 2, nevoid basal cell carcinoma syndrome, or branchio-oculo-facial syndrome. Patients may be asymptomatic or present with progressive loss of vision.

Dane

Klasyfikacja
Choroba

Synonimy
CHR-RPE
Hamartoma siatkówki i RPE
Combined hamartoma of the retina and RPE

Kod ORPHA
440727

Kod OMIM
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Kod ICD10
D31.2

Kod ICD11
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*Źródło

orphanet