Opis choroby *

Definicja

A rare, genetic macular dystrophy disorder characterized by the presence of small yellow-white accumulations of extracellular material under the retinal pigment epithelium in the ocular posterior pole, and affecting multiple members of a family. The disease has a variable clinical presentation ranging from asymptomatic patients to progressive loss of vision and scotomas, possibly associated with subfoveal choroidal neovascularization, extensive pigmentary changes, geographic atrophy and/or subretinal hemorrhage.

Dane

Klasyfikacja
Choroba

Synonimy
DHRD
DHRD
Dominujące druzy tarczy nerwu wzrokowego
Dominujące promieniowe druzy tarczy neru wzrokowego
Dystrofia plamki typu "plastra miodu" Doyne'a
Dominant drusen
Dominant radial drusen
Doyne honeycomb retinal dystrophy
Malattia leventinese

Kod ORPHA
75376

Kod OMIM
126700

Kod ICD10
H35.5

Kod ICD11
9B70

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*Źródło

orphanet