Opis choroby *

Definicja

Helicoid peripapillary chorioretinal degeneration is a rare autosomal dominantly inherited chorioretinal degeneration disease, presenting at birth or infancy, characterized by progressive bilateral retinal and choroidal atrophy, appearing as lesions on the optic nerve and peripheral ocular fundus and leading to central vision loss. Congenital anterior polar cataracts are sometimes associated with this disease.

Dane

Klasyfikacja
Choroba

Synonimy
Atrophia areata
Atrofia miejscowa
SCRA
Zanik siatkówki i naczyniówki Sveinssona
SCRA
Sveinsson chorioretinal atrophy

Kod ORPHA
86813

Kod OMIM
108985

Kod ICD10
H31.2

Kod ICD11
9B70

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*Źródło

orphanet