

Opis choroby *

Definicja

Retinal degeneration-nanophthalmos-glaucoma syndrome is characterized by progressive pigmentary retinal degeneration (with nyctalopia and visual field restriction), cystic macular degeneration and angle closure glaucoma. It has been described in seven members of one family. Patients also have hyperopia and nanophthalmos. The mode of transmission is autosomal recessive.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Mackay-Shek-Carr syndrome
	Zespół Mackay'a, Sheka i Carra

Kod ORPHA	Kod OMIM	Kod ICD10
1574	267760	H35.5

Kod ICD11
9E1Y

*Źródło

orphanet