

Opis choroby *

Definicja

An X-linked retinal dystrophy characterized by choroideremia, causing in affected males progressive nyctalopia and eventual central blindness. Obesity, moderate intellectual disability and congenital mixed (sensorineural and conductive) deafness are also observed. Female carriers show typical retinal changes indicative of the choroideremia carrier state.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Ayazi syndrome
	Zespół Ayazi
	Del(X)(q21)
	Monosomy Xq21

Kod ORPHA	Kod OMIM	Kod ICD10
1435	303110	Q93.5

Kod ICD11
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*Źródło

orphanet