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
Zespół wad wrodzonych

Synonimy
Ayazi syndrome
Zespół Ayazi
Del(X)(q21)
Monosomy Xq21

Kod ORPHA
1435

Kod OMIM
303110

Kod ICD10
Q93.5

Kod ICD11
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*Źródło

orphanet