Opis choroby *

Definicja

A rare progressive autosomal recessive tapetoretinal degeneration disease, occurring in the third decade of life, characterized by small sparkling crystalline deposits in the posterior retina and corneal limbus in addition to sclerosis of the choroidal vessels and manifesting as nightblindness, decreased vision, paracentral scotoma, and, in the end stages of the disease, legal blindness.

Dane

Klasyfikacja
Choroba

Synonimy
BCD
BCD
Retinopatia krystaliczna Biettiego
Bietti crystalline corneoretinal dystrophy
Bietti crystalline retinopathy

Kod ORPHA
41751

Kod OMIM
210370

Kod ICD10
H35.5

Kod ICD11
9B61

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*Źródło

orphanet