Opis choroby *

Definicja

A rare syndromic retinitis pigmentosa characterized by pigmentary retinopathy, diabetes mellitus with hyperinsulinism, acanthosis nigricans, secondary cataracts, neurogenic deafness, short stature mild hypogonadism in males and polycystic ovaries with oligomenorrhea in females. Inheritance is thought to be autosomal recessive. It can be distinguished from Alstrom syndrome (see this term) by the presence of intellectual disability and the absence of renal insufficiency. There have been no further descriptions in the literature since 1993.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Retinitis pigmentosa-intellectual disability-labyrinthine deafness-hypogenitalism syndrome
Retinitis pigmentosa-intellectual disability-sensorineural hearing loss-hypogenitalism syndrome

Kod ORPHA
3085

Kod OMIM
268020

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet