Opis choroby *

Definicja

A rare ciliopathy characterized by congenital moderate-to-severe deafness, retinitis pigmentosa developing in the first or second decade, and normal vestibular function. Congenital bilateral sensorineural hearing loss is mild to moderate in the low frequencies and severe to profound in the higher frequencies. Additional manifestations include night blindness, constricted visual field (tunnel vision), and later on decreased visual acuity sometimes ending with bare light perception.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
USH2
USH2

Kod ORPHA
231178

Kod OMIM
611383

Kod ICD10
H35.5

Kod ICD11
LD2H.4

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*Źródło

orphanet