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.

Kod ICD10

H35.5

Dane

Klasyfikacja Podtyp kliniczny	Synonimy USH2 USH2
Kod ORPHA	Kod OMIM

611383

Kod ORPHA 231178

Kod ICD11 LD2H.4

<u>*Źródło</u>

orphanet