Opis choroby *

Definicja

A rare ciliopathy characterized by profound congenital deafness, retinitis pigmentosa and vestibular dysfunction. Retinitis pigmentosa results in visual loss and generally manifests as night blindness, progressively constricted visual fields, and impaired visual acuity. Vestibular dysfunction a defining feature of this form, manifests as delayed motor development with affected infants taking longer to sit independently and to walk. Later on, vestibular dysfunction results in difficulty with activities requiring balance.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
USH1
USH1

Kod ORPHA
231169

Kod OMIM
614869

Kod ICD10
H35.5

Kod ICD11
LD2H.4

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*Źródło

orphanet