Opis choroby *

Definicja

A rare ciliopathy characterized by progressive hearing and visual loss in the first decades of life and, in some cases, vestibular dysfunction. Patients have normal hearing at birth. Onset of hearing loss is usually in late childhood or adolescence after development of speech. Profound deafness is mostly reported by middle age. Retinitis pigmentosa related visual loss also develops in late childhood or adolescence. Developmental motor milestones are generally normal but vestibular dysfunction may occur in adulthood.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
USH3
USH3

Kod ORPHA
231183

Kod OMIM
614504

Kod ICD10
H35.5

Kod ICD11
LD2H.4

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*Źródło

orphanet