

## 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

<b>Klasyfikacja</b>	Synonimy
Podtyp kliniczny	USH1
	USH1

<b>Kod ORPHA</b>	<b>Kod OMIM</b>	<b>Kod ICD10</b>
231169	614869	H35.5

**Kod ICD11**  
LD2H.4

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### \*Źródło

orphanet