

Zespół Ushera typu 1

Kod Orpha: 231169 Kod OMIM: 614869

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

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.