

Opis choroby *

Definicja

A rare genetic syndromic intellectual disability characterized by global developmental delay, intellectual disability, infantile or childhood onset of progressive ataxia, and bilateral sensorineural hearing impairment. Variable features include signs of upper and lower motor neuron disease, peripheral neuropathy, myopathic facies, lower limb muscle wasting, and heel contractures. There have been no further descriptions in the literature since 1993.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Ataxia-hearing loss-intellectual disability syndrome Zespół Reardona i Baraitsera Reardon-Baraitser syndrome

Kod ORPHA	Kod OMIM	Kod ICD10
1188	208850	G11.1

Kod ICD11
LD2H.Y

*Źródło

orphanet