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
Zespół wad wrodzonych

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

Kod ORPHA
1188

Kod OMIM
208850

Kod ICD10
G11.1

Kod ICD11
LD2H.Y

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*Źródło

orphanet