Opis choroby *

Definicja

A rare, genetic, X-linked syndromic intellectual disability disorder characterized by severe intellectual disability, microcephaly, post-natal growth retardation, severe visual impairment or blindness (due to optic atrophy), severe hearing defect, spasticity, epileptic seizures, restricted large-joint movements and early death (in infancy or early childhood). Facial dysmorphic features (large dysplastic ears and short broad nose) are additionally observed. There have been no further descriptions in the literature since 1993.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
3078

Kod OMIM
309555

Kod ICD10
F72.9

Kod ICD11
LD90

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*Źródło

orphanet