Opis choroby *

Definicja

A rare, genetic, neurological disorder characterized by the association of slowly progressive spinocerebellar degeneration and corneal dystrophy, manifesting with bilateral corneal opacities (which lead to severe visual impairment), mild intellectual disability, ataxia, gait disturbances, and tremor. Additional manifestations include facial dysmorphism (i.e. triangular face, ptosis, low-set, posteriorly angulated ears, and micrognathia), as well as mild upper motor neuron involvement with hypertonia, lower limb hyperreflexia and extensor plantar responses. There have been no further descriptions in the literature since 1985.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Der Kaloustian-Jarudi-Khoury syndrome
Degeneracja móżdżkowo-rdzeniowa - dystrofia rogówki
Zespół Der Kaloustiana, Jarudi i Khoury'ego

Kod ORPHA
3177

Kod OMIM
271310

Kod ICD10
G11.1

Kod ICD11
9A70.Y

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*Źródło

orphanet