

Opis choroby *

Definicja

A rare syndromic intellectual deficiency characterized by psychomotor delay, severe progressive spastic quadriplegia, microcephaly, and a Hallerman-Streiff-like phenotype including absence of eyebrows and eyelashes, glaucoma, and small, beaked nose. Structural central nervous system abnormalities (cervical spinal cyst, occipital cranium bifidum occulatum) were additional findings. There have been no further descriptions in the literature since 1974.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Absent eyebrows and eyelashes-intellectual disability syndrome

Brak brwi i rzęs - niepełnosprawność intelektualna

Zespół Hala, Berga i Rudolpha

Hal-Berg-Rudolph syndrome

Kod ORPHA

2985

Kod OMIM

200130

Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet