Opis choroby *

Definicja

A rare syndromic intellectual disability characterized by severe intellectual disability and calcification of the choroid plexus, associated with elevated cerebrospinal fluid protein concentration. Additional signs and symptoms include strabismus, increased deep tendon reflexes, and foot deformities, among others. There have been no further descriptions in the literature since 1993.

Dane

Klasyfikacja
Choroba

Kod ORPHA
1313

Kod OMIM
215480

Kod ICD10
G93.8

Kod ICD11
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*Źródło

orphanet