Opis choroby *

Definicja

A rare neurologic disease characterized by bilateral cataract, Dandy-Walker malformation, and childhood onset of distal spinal muscular atrophy. Patients present with progressively deteriorating symmetrical distal muscle weakness and atrophy of the lower limbs (and, to a much lesser degree, also the upper limbs) and decreased tendon reflexes in the lower and upper limbs.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
73245

Kod OMIM
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Kod ICD10
G12.8

Kod ICD11
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*Źródło

orphanet