## 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