Opis choroby *

Definicja

A rare, congenital muscular dystrophy due to dystroglycanopathy characterized by early onset muscular dystrophy, severe muscular hypotonia, severe mental retardation and typical brain and eye malformations, including pachygyria, polymicrogyria, agyria, brainstem and cerebellar structural anomalies, severe myopia, glaucoma, optic nerve and retinal hypoplasia. Patients may present with seizures, macrocephaly or microcephaly, microphthalmia, and congenital contractures. Depending on the severity, limited motor function is acquired. Less severe cases have been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
MEB syndrome
Wrodzona dystrofia mięśniowa Santavuori
Zespół MEB
Zespół mięsień-oko-mózg
Muscle-eye-brain syndrome
Santavuori congenital muscular dystrophy

Kod ORPHA
588

Kod OMIM
615350

Kod ICD10
G71.0

Kod ICD11
8C70.6

---

*Źródło

orphanet