Opis choroby *

Definicja

A rare, genetic, congenital muscular alpha-dystroglycanopathy with brain and eye anomalies disease characterized by a severe muscle-eye-brain disease-like phenotype associated with intellectual disability, muscular dystrophy, macrocephaly and extended bilateral multicystic white matter disease.

Dane

Klasyfikacja
Choroba

Synonimy
MEB disease with bilateral multicystic leucodystrophy
Choroba MEB z obustronną wielotorbielowatą leukodystrofią

Kod ORPHA
370997

Kod OMIM
616538

Kod ICD10
G71.0

Kod ICD11
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*Źródło

orphanet