

## 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