

## Opis choroby \*

### Definicja

Congenital muscular alpha-dystroglycanopathy with brain and eye anomalies (MDDGA) is a cobblestone lissencephaly characterized by and considered to be pathognomonic of a continuum of recessive autosomal disorders with brain, ocular and muscular involvement. MDDGA includes Walker-Warburg syndrome, muscle-eye-brain disease, Fukuyama muscular and cerebral dystrophy and muscle eye brain disease with bilateral multicystic leukodystrophy.

### Dane

#### Klasyfikacja

Grupa fenomenów

#### Synonimy

Lissencephaly type 2 with muscular and ocular involvement

Lizencefalia typu 2 z zaangażowaniem mięśni i oczu

MDDGA

MDDGA

Kod ORPHA

352687

Kod OMIM

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Kod ICD10

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Kod ICD11

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#### \*Źródło

orphanet