

## **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

|                     |   |
|---------------------|---|
| <b>Klasyfikacja</b> | Synonimy  |
| Grupa fenomenów     | Lissencephaly type 2 with muscular and ocular involvement<br>Lizencefalia typu 2 z zaangażowaniem mięśni i oczu |
|                     | MDDGA   |
|                     | MDDGA   |

**Kod ORPHA**                    **Kod OMIM**                    **Kod ICD10**  
352687

Kod ICD11

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źródło

orphanet