

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

-

Kod ICD10

-

Kod ICD11

-

*Źródło

orphanet