Open Peer Review on Qeios

Congenital muscular alpha-dystroglycanopathy with brain and eye anomalies

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Congenital muscular alpha-dystroglycanopathy with brain and eye anomalies. ORPHA:352687

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.