Open Peer Review on Qeios

Muscle-eye-brain disease with bilateral multicystic leucodystrophy

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Muscle-eye-brain disease with bilateral multicystic leucodystrophy. ORPHA:370997

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.