Facial dysmorphism-macrocephaly-myopia-Dandy-Walker malformation syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Facial dysmorphism-macrocephaly-myopia-Dandy-Walker malformation syndrome. ORPHA:1970

Facial dysmorphism-macrocephaly-myopia-Dandy-Walker malformation syndrome is characterised by Dandy-Walker malformation, severe intellectual deficit, macrocephaly, brachytelephalangy, facial dysmorphism and severe myopia. Three cases have been described. Transmission appears to be autosomal recessive.