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L-2-hydroxyglutaric aciduria

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base*. [L-2-hydroxyglutaric aciduria](#). ORPHA:79314

L-2-hydroxyglutaric aciduria is a primarily neurological form of 2-hydroxyglutaric aciduria (see this term) characterized by psychomotor retardation, cerebellar ataxia and variable macrocephaly or epilepsy.