2-aminoadipic 2-oxoadipic aciduria

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 2-aminoadipic 2-oxoadipic aciduria. ORPHA:79154

2-aminoadipic 2-oxoadipic aciduria is a rare disorder of lysine and hydroxylysine metabolism characterized by variable clinical presentation including hypotonia, developmental delay, mild to severe intellectual disability, ataxia, epilepsy and behavioral disorders, most commonly attention deficit hyperactivity disorder. Frequently, individuals are completely without clinical phenotype.