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2-hydroxyglutaric aciduria

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base*. 2-hydroxyglutaric aciduria. ORPHA:19

2-Hydroxyglutaric aciduria is a group of neurometabolic disorders with a wide clinical spectrum ranging from severe neonatal presentations to progressive forms, and asymptomatic cases, characterized biochemically by increased levels of 2-hydroxyglutaric acid in the plasma, cerebrospinal fluid and urine.