Formiminoglutamic aciduria

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Formiminoglutamic aciduria. ORPHA:51208

A rare disorder of folate metabolism and transport characterized, biochemically, by elevated formiminoglutamate in urine and plasma due to glutamate formiminotransferase deficiency, associated with a highly variable clinical phenotype, ranging from developmental delay, intellectual disability and anemia to normal development without anemia. Increased hydantoin-5-propionic acid and/or folate in plasma may also be associated.