Beta-ureidopropionase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Beta-ureidopropionase deficiency. ORPHA:65287

Beta-ureidopropionase deficiency is a very rare pyrimidine metabolism disorder described in fewer than 10 patients to date with an extremely wide clinical picture ranging from asymptomatic cases to neurological (epilepsy, autism) and developmental disorders (urogenital, colorectal).