Open Peer Review on Qeios

Hyperammonemia due to Nacetylglutamate synthase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hyperammonemia due to N-acetylglutamate synthase deficiency. ORPHA:927

N-acetylglutamate synthase (NAGS) deficiency is a urea cycle disorder leading to hyperammonaemia.