L-Arginine:glycine amidinotransferase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. L-Arginine:glycine amidinotransferase deficiency. ORPHA:35704

L-Arginine:glycine amidinotransferase (AGAT) deficiency is a very rare type of creatine deficiency sydrome characterized by global developmental delay, intellectual disability, and myopathy.