

Open Peer Review on Qeios

Argininemia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. <u>Argininemia</u>. ORPHA:90

Arginase deficiency is a rare autosomal recessive amino acid metabolism disorder characterized clinically by variable degrees of hyperammonemia, developing from about 3 years of age, and leading to progressive loss of developmental milestones and spasticity in the absence of treatment.

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