Open Peer Review on Qeios

Argininemia

National Cancer Institute

Source

National Cancer Institute. <u>Argininemia</u>. NCI Thesaurus. Code C84568.

A rare autosomal recessive metabolic disorder characterized by a deficiency of the enzyme arginase resulting in accumulation of arginine and urea in the blood and cerebrospinal fluid. Signs and symptoms include developmental delays, spasticity, ataxia, seizures and mental retardation.