Open Peer Review on Qeios

Argininosuccinic Aciduria

National Cancer Institute

Source

National Cancer Institute. <u>Argininosuccinic Aciduria</u>. NCI Thesaurus. Code C84569.

A genetic inherited disorder caused by mutations in the ASL gene. It is characterized by accumulation of ammonia in the blood. It presents with lethargy in the first few days of life, accompanied by developmental delay and mental retardation.