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# N-Acetylglutamate Synthase Deficiency

National Cancer Institute

## Source

National Cancer Institute. *N-Acetylglutamate Synthase Deficiency*. NCI Thesaurus. Code C129307.

An autosomal recessive disorder caused by mutation(s) in the NAGS gene, encoding N-acetylglutamate synthase, mitochondrial. It may be characterized by failure to thrive, hyperammonemia, lethargy, seizures, and coma.