

[Open Peer Review on Qeios](#)

Sialic Acid Storage Disease

National Cancer Institute

Source

National Cancer Institute. *Sialic Acid Storage Disease*. NCI Thesaurus. Code C85067.

A rare, autosomal recessive lysosomal storage disease caused by mutations in the SLC17A5 gene. It primarily affects the nervous system. Signs and symptoms include developmental delay, intellectual disability, hypotonia, failure to thrive, seizures, and ataxia.