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Niemann-Pick Disease, Type A

National Cancer Institute

Source

National Cancer Institute. *Niemann-Pick Disease, Type A*. NCI Thesaurus. Code C126561.

An autosomal recessive inherited lysosomal storage disease caused by mutations in the SMPD1 gene. It manifests with hepatosplenomegaly, failure to thrive, psychomotor regression, and interstitial lung disease.