Niemann-Pick Disease, Type B

National Cancer Institute

Source

National Cancer Institute. Niemann-Pick Disease, Type B. NCI Thesaurus. Code C126866.

An autosomal recessive lysosomal storage disease caused by mutations in the SMPD1 gene, encoding sphingomyelin phosphodiesterase. The condition is characterized by hepatosplenomegaly and interstitial lung disease, but with little neurological involvement. It is part of a continuum of disease resulting from decrease activity of sphingomyelin phosphodiesterase, with Type B being the milder form.