Lysosomal Storage Disease

National Cancer Institute

Source

National Cancer Institute. Lysosomal Storage Disease. NCI Thesaurus. Code C61250.

A group of autosomal recessive or X-linked inherited metabolic disorders caused by defects in the function of the lysosomes. Signs and symptoms include hepatomegaly, splenomegaly, nervous system manifestations, skeletal abnormalities, and mental deterioration. Representative examples include Gaucher disease, Niemann-Pick disease, Wolman disease, and Fabry disease.