Gaucher Disease

National Cancer Institute

Source

National Cancer Institute. Gaucher Disease. NCI Thesaurus. Code C61268.

An inherited lysosomal storage disease caused by deficiency of the enzyme glucocerebrosidase. It results in the accumulation of a fatty substance called glucocerebroside in mononuclear cells in the bone marrow, liver, spleen, brain, and kidneys. Signs and symptoms include hepatomegaly, splenomegaly, neurologic disorders, lymphadenopathy, skeletal disorders, anemia and thrombocytopenia.