Sandhoff disease is a lipid storage disorder characterized by a progressive deterioration of the central nervous system. The clinical symptoms of Sandhoff disease are identical to Tay-Sachs disease. Sandhoff disease is an autosomal recessive genetic disorder caused by an abnormal gene for the beta subunit of the hexosaminidase B enzyme. This gene abnormality results in a deficiency of hexosaminidase A and B that results in accumulation of fats (lipids) called GM2 gangliosides in the neurons and other tissues. Sandhoff disease is a very rare disorder that affects males and females in equal numbers. This disorder occurs in people of many different ethnic backgrounds. Sandhoff disease may be more common in the Creole population of northern Argentina, the Metis Indians in Saskatchewan, Canada and individuals of Lebanese ancestry. Sandhoff disease can be diagnosed by performing an enzyme assay to determine activity of the hexosaminidase A and B enzymes. Affected individuals have absent or reduced activity of both enzymes. Molecular genetic (DNA) testing is available to determine the specific gene mutation that is present in the beta subunit of the Hexosaminidase B gene and confirm the diagnosis.