Gaucher disease is the most common lipid-storage disorder, and is the most common genetic disease affecting Jewish people of Eastern European ancestry. The disease is an autosomal recessive trait. Gaucher disease results from a glucocerebrosidase enzyme deficiency, caused by a single-base mutation (adenosine to guanosine transition) in exon 9 of the glucocerebrosidase gene, resulting in an accumulation of glucocerebroside in macrophages.

Gaucher specialists divide the disease into 3 classifications based on the particular symptoms and course of the disease. Type 1, adult Gaucher disease is the most common form, and occurs in 1 in 100,000 people in the general population. Type 1 differs from types 2 and 3 by having non-neurological symptoms.

Besides managing the symptoms of Gaucher disease, the most direct and logical treatment is enzyme replacement therapy. However, natural glucocerebrosidase infusions are ineffective.

A drug called, Cerezyme, is very effective and has had FDA approval since 1996.

Current research involves looking into investigation of biophosphonate group of drugs for osteoporosis and bone disease, attempts to do gene therapy, and trial of the drug OGT 918.

- [http://www.gaucherdis...5e00]. The Human Gene Map, Gaucher Disease.