

Inheritance of Gaucher Disease



STRAIGHT TALK FOR PATIENTS AND FAMILIES

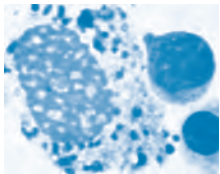
Gaucher patients can't produce enough of an important enzyme

Glucocerebrosidase (GC) is an enzyme that helps break down glucocerebroside, a type of fat or lipid found in certain parts of your body that is the result of the breakdown of worn-out red and white blood cells. Without this important enzyme, special cells called *macrophages*, responsible for picking up and recycling glucocerebroside, fill up with the undigested fat. These cells are referred to as *Gaucher cells*.

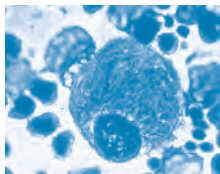
Your bones and organs are at risk even when you're not in pain

Gaucher cells, bloated with undigested fat, accumulate primarily in the liver, spleen and bone marrow, and sometimes in the lungs. When this happens, the affected organs, especially the spleen and liver, can become enlarged and distended so that they protrude from the abdomen. This condition is called *organomegaly* (literally, big organs), or *hepatosplenomegaly* (big liver and spleen).

Normal Cell



Gaucher Cell



This type of family history is particularly important in the case of an autosomal recessive disorder such as Gaucher disease where generations of people within a family may carry a single copy of the defective gene and never know it. As a result, the diagnosis of Gaucher disease in one person (or the identification of an ancestor with significant Gaucher symptoms) may be the first indication that siblings, aunts, uncles and cousins may be Gaucher disease carriers.

If you suspect that you or someone close to you either has Gaucher disease or is a carrier, talk to your doctor, or refer to the resources listed in this booklet.



A family story

This chart is designed to help you gather and organize relevant information about your family members and their status as potential carriers of Gaucher disease. Together with a knowledge of inheritance patterns outlined in the *Straight Talk* booklet, it will help you to better understand how Gaucher disease occurs and how it is passed along.

genzyme