Effects of Enzyme Replacement in Gaucher's Disease

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Gaucher disease is a lysosomal storage disease resulting from glycocerebroside accumulation in macrophages due to a genetic deficiency of the enzyme glucocerebrosidase. It may occur in adults but occurs most severely in infants, in whom cerebroside also accumulates in neurons. Patients with Gaucher's disease experience enlargement of the liver and spleen and bone destruction. The condition is passed from generation to generation through autosomal recessive inheritance. There are actually three types of Gaucher's disease.

Type I is the most common form. It is a chronic non-neuronopathic form, meaning the disease does not affect nerve cells. The symptoms of type I can appear at any age.

Type II appears in infancy and usually results in death for the patient. Type II is an acute neuronopathic form and can affect the brain stem. It is the most severe form of the disease.

Type III is also neuronopathic, however it is subacute in nature. This means the course of the illness lies somewhere between long-term (chronic) and short-term (acute).

The purpose of this study is to examine the effects of enzyme replacement therapy on patients with Gaucher's disease, specifically those types directly affecting the nervous system (neuronopathic).

Patients with Gaucher's disease types II and III will be selected to participate in the study and receive enzyme replacement therapy. Patients participating will undergo a variety of tests to measure levels of hemoglobin concentration, liver volume, and spleen volume. Improvements in these measures will be compared other laboratory tests measuring the involvement of the nervous system.