Bone Response to Enzyme Replacement in Gaucher's Disease

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The purpose of this study is to examine how the skeleton responds to repeated doses of enzyme replacement therapy in patients with type I Gaucher's disease who have had their spleens removed.

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.

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.

In this study patients will be divided into three groups. Each group will receive different doses of enzyme replacement (Ceredase). In addition, two of the three groups will also receive doses of a form of vitamin D (calcitriol). Researchers believe the groups receiving vitamin D will have an improved response as compared to those patients only receiving enzyme replacement.

Patients in each group who respond to enzyme replacement with increases in bone density will be compared to the other treatment groups.