Gaucher's Disease

Gaucher disease is an inherited metabolic disorder. With this disorder, harmful quantities of a fatty substance called glucocerebroside build up in the:

- Spleen.
- Liver.
- Lungs.
- Bone marrow.
- Brain (rare).

Three clinical forms (*phenotypes*) of Gaucher disease are commonly recognized.

- The first category is called *type 1*.

It is is by far the most common. Patients in this group usually bruise easily and experience fatigue due to:

- Anemia.
- Low blood platelets.
- Enlargement of the liver and spleen.
- Weakening of the skeleton.
- Lung and kidney impairment (in some cases).

There are no signs of brain involvement. The onset of clinical problems may be early in life. Or they may be delayed until adulthood.

- The second group is classified as *type 2*.

In this form, liver and spleen enlargement appear by 3 months of age. Also, there is extensive and progressive brain damage. These patients usually die by 2 years of age.

- The third category is called *type 3*.

In this form, liver and spleen enlargement is variable. Signs of brain involvement such as seizures (*convulsions*) gradually appear.

All of these patients lack an enzyme called *glucocerebrosidase*. It catalyzes the first step in the biodegradation of glucocerebroside. Except for the brain, glucocerebroside arises mainly from the biodegradation of old red and white blood cells. In the brain, glucocerebroside arises from the turnover of complex lipids. This happens during brain development and the formation of the myelin sheath of nerves.

TREATMENT

- Highly effective enzyme replacement therapy is available for patients with type 1 form. This therapy:
  - Decreases liver and spleen size.
  - Reduces skeletal abnormalities.
  - Reverses other effects of the disorder including abnormal blood counts.
- There is currently no effective treatment for severe brain damage that may occur in patients with types 2 and 3.

There is no permanent cure for this disorder. Enzyme replacement therapy is helpful for type 1 patients and most of the type 3 patients with this condition.

RESEARCH BEING DONE

The NINDS is currently conducting a study to examine the skeletal response to enzyme replacement therapy in patients with type 1 whose spleens have been removed and its effect in patients with type 3 Gaucher disease with severe seizures.

Research is continually being done to learn more about this problem.

Provided by: The National Institute of Neurological Disorders and Stroke

Document Released: 12/08/2003 Document Re-Released: 03/16/2010

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