

Type 2 Gaucher Disease (Acute Infantile Gaucher Disease or Neuropathic Type)

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Gaucher disease is a relatively frequent recessive disease affecting 1 in 400,000 to 1 in 200,000 persons and 1 in 400 to 1 in 2000 persons among Ashkenazi Jews. The gene coding for beta-glucocerebrosidase is located on chromosome 1q21-q31.

Type 1 Gaucher disease is the most common type but is only occasionally observed in children. There is no involvement of the CNS except in rare cases.

Type 2 Gaucher disease or neuropathic type is also due to glucosyl ceramide beta-glucosidase deficiency.

Clinical features:

Symptom onset in infants with Gaucher disease type II is usually before 6 months of age and frequently before 3 months of age. The initial features are motor regression and cranial nerve dysfunction.

Children are first hypotonic and then spastic.

Head retraction is an early and characteristic sign that probably is due to meningeal irritation.

Difficulties are sucking and swallowing, trismus, and oculomotor palsies are typical. Mental deterioration is rapid but seizures are uncommon.

Type 3 Gaucher disease becomes apparent during the first decade of life the major features at this period being slowly progressive hepatosplenomegaly, rapidly associated with intellectual deficiency, cerebellar ataxia and extrapyramidal signs frequently develop.

The most suggestive features include supranuclear ophthalmoplegia and in some myoclonic epilepsy.

Diagnosis

Assay of acid B-glucosylceramidase enzyme activity in peripheral blood leukocytes is reliable for diagnosis which is 0 to 15% of normal. Carrier detection and prenatal diagnosis are available.

Management

Enzyme therapy reverses the hematologic and visceral manifestation of Gaucher's disease but does not influence the ultimate progression of neurologic complications in patients with type 2 Gaucher's disease.

Keywords: Acute infantile Gaucher disease; Motor regression; Hypotonia

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