

Clinical Manifestations of Type 1 Gaucher Disease

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Patients with type 1 Gaucher disease commonly present with painless splenomegaly, anemia, or thrombocytopenia at onset. They may also have chronic fatigue, hepatomegaly (with or without abnormal liver function test findings), bone pain, or pathologic fractures and may bruise easily because of thrombocytopenia. Bleeding secondary to thrombocytopenia may manifest as nosebleeds, bruising, or both.

In symptomatic patients, splenomegaly is progressive and can become massive. Children with massive splenomegaly may be short stature because of the energy expenditure required by the enlarged organ.

Most patients with type 1 Gaucher disease have radiologic evidence of skeletal involvement, including an Erlenmeyer flask deformity of the distal femur, which is an early skeletal change. Clinically apparent bony involvement, which occurs in more than 20% of patients with Gaucher disease, can present as bone pain or pathologic fractures. In patients with symptomatic bone disease, lytic lesions can develop in the long bones, ribs, and pelvis, and osteosclerosis or osteopenia may be evident at an early age. Bone crises with severe pain and swelling can occur in individuals with type 1 Gaucher disease and are frequently mistaken for synovitis or osteomyelitis until other symptoms become apparent.

Occasional patients with type 1 Gaucher disease develop pulmonary involvement, parkinsonism, or portal hypertension.

Patients with milder presentations of Gaucher disease are diagnosed later in life during evaluations for hematologic or skeletal problems or are found to have splenomegaly during routine examinations. Some patients are overtly asymptomatic, and a diagnosis is made incidentally after evaluation for other medical problems.

Keywords: Gaucher Type 1; Clinical manifestations; Hepatosplenomegaly

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Fig: Huge Hepatosplenomegaly

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