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Revised recommendations for the management of Gaucher disease in children.

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Abstract

Gaucher disease is an inherited pan-ethnic disorder that commonly begins in childhood and is caused by deficient activity of the lysosomal enzyme glucocerebrosidase. Two major phenotypes are recognized: non-neuropathic (type 1) and neuropathic (types 2 and 3). Symptomatic **children** are severely affected and manifest growth retardation, delayed puberty, early-onset osteopenia, significant splenomegaly, hepatomegaly, thrombocytopenia, anemia, severe bone pain, acute bone crises, and fractures. Symptomatic **children** with types 1 or 3 should receive enzyme replacement therapy, which will prevent debilitating and often irreversible **disease** progression and allow those with non-neuropathic **disease** to lead normal healthy lives. **Children** should be monitored every 6 months (physical exam including growth, spleen and liver volume, neurologic exam, hematologic indices) and have one to two yearly skeletal assessments (bone density and imaging, preferably with magnetic resonance, of lumbar vertebrae and lower limbs), with specialized cardiovascular monitoring for some type 3 patients. Response to treatment will determine the frequency of monitoring and optimal dose of enzyme replacement. Treatment of **children** with type 2 (most severe) neuropathic **Gaucher disease** is supportive. Pre-symptomatic **children**, usually with type 1 **Gaucher**, increasingly are being detected because of affected siblings and screening in high-prevalence communities. In this group, annual examinations (including bone density) are recommended. However, monitoring of asymptomatic **children** with affected siblings should be guided by the age and severity of manifestations in the first affected sibling. Treatment is necessary only if signs and symptoms develop.

CONCLUSION: Early detection and treatment of symptomatic types 1 and 3 **Gaucher disease** with regular monitoring will optimize outcome. Pre-symptomatic **children** require regular monitoring. Genetic counseling is important.

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