## **Condition:** Gaucher disease

---

### Inheritance:

Autosomal recessive

### Genetic etiology:

Mutation in *GBA* gene, encoding glucosylceramidase.

### Frequency:

Varies in different populations; 1/855 in Ashkenazi Jewish population (for type 1 GD).

### Clinical features:

Gaucher disease is a lysosomal disorder in which there is accumulation of glucosylceramide. There are three major clinical subtypes. Type 1 is characterized by bone disease (lytic lesions, osteopenia), hepatosplenomegaly, anemia, thrombocytopenia, and lung disease. The nervous system is not involved in Type 1. Types 2 and 3 have progressive neurological impairment, with type 3 characterized by later age of onset and slower progression.

### Management:

Supportive care and monitoring; enzyme replacement therapy by intravenous infusion benefits those with type 1, and, to some extent type 3.

### Genetic counseling:

Based on autosomal recessive inheritance; genetic testing is available and can be used for carrier detection.

*Human Genetics and Genomics*, Fourth Edition. Bruce R. Korf and Mira B. Irons.