**Condition:** Gaucher disease

**Inheritance:**
Autosomal recessive

**Genetic etiology:**
Mutation in \textit{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.