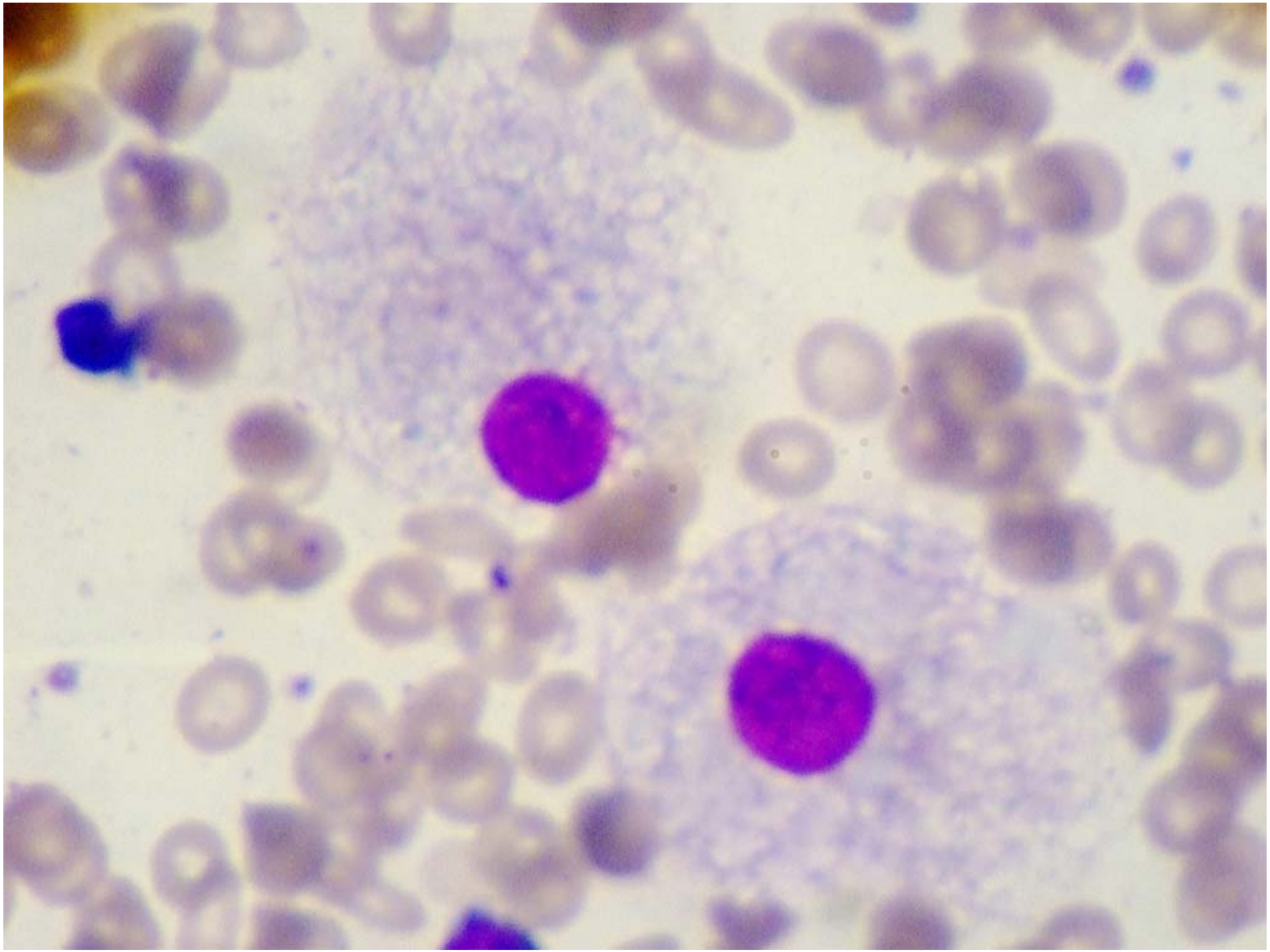

Gaucher disease

- Gaucher disease is an inherited autosomal recessive metabolic defect due to a deficiency in the lysosomal enzyme β-glucocerebrosidase
  - which leads to accumulation of glucocerebroside in the body, predominantly in the liver, spleen and bone marrow

- Gaucher disease is the most common lysosomal disease.
- The prevalence is highest in Ashkenazi Jews, about 1/855 compared to 1/100,000 in other populations

Main clinical manifestations

- Skeletal manifestations of Gaucher disease
  - osteopenia with an increased risk of fractures
  - decreased osteoclast activity with a reduction in shaping and remodeling of young bones
  - blood vessel alterations with avascular necrosis and acute attacks of pain (bone crisis)
  - increased risk of osteoarticular infection

- Extra-osseous involvement in Gaucher disease
  - 95% Splenomegaly cause cytopenia
  - 80% Hepatomegaly cause cirrhosis
  - Lung involvement
    - compression by other organs
    - fractures of the vertebras or ribs
    - infiltration of the lung capillaries and parenchyma
    - Fibrosis and pulmonary hypertension are rare.