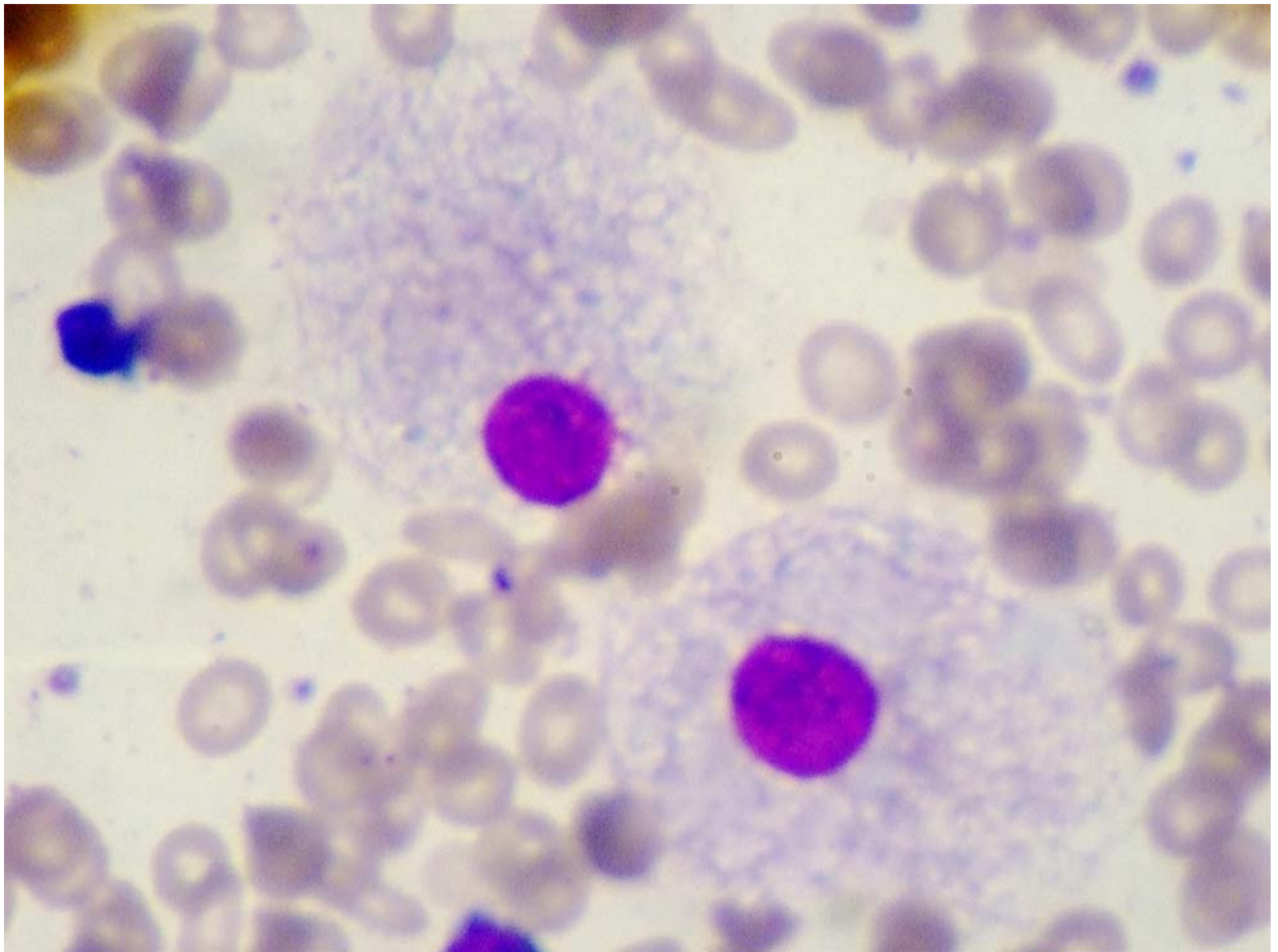






Gaucher disease

- 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 vertebrae or ribs
 - infiltration of the lung capillaries and parenchyma
 - Fibrosis and pulmonary hypertension are rare.