Case Study: Gaucher Disease

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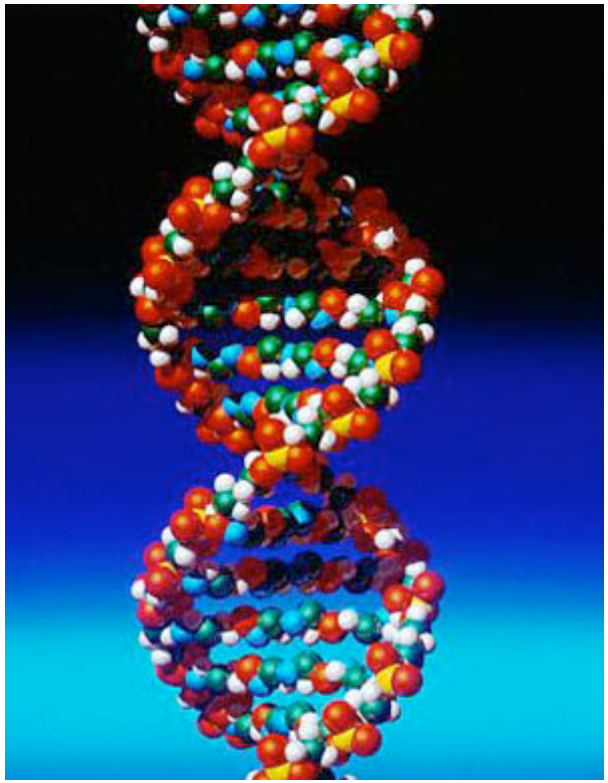
Genetics of Gaucher Disease

- Recessive autosomal gene mutation in the GBA gene
  - Gene cannot produce beta-glucocerebrosidase, which breaks down the fatty substance, glucocerebroside
  - Buildup of glucocerebroside causes symptoms of Gaucher disease



How was Gaucher disease recognized/diagnosed classically?

- Associated Symptoms:
  - Skeletal weakness
  - Spleen and liver enlargement, sometimes resulting in a protruding abdomen
  - Fatty deposit on the eye
  - Anemia
  - Low platelet count
  - Low white blood cell count
  - Neurological symptoms (Type II and Type III)

How was Gaucher disease recognized/diagnosed classically?

- Chemical analysis of the sediment from a 24-hour urine collection
- Detection of Gaucher cells or cells where there is an abnormal storage of glucocerebroside
- Measuring the amount of a certain enzyme in white blood cells
- MRI, CT, or X-ray of the skeleton

How was Gaucher disease treated classically?

- Removal of spleen
- Liver transplant
- Transfusion of blood products for severe anemia and bleeding
- Pain medication for bone problems
- Joint replacement surgery for relief from chronic pain and restoration of function
- Oral bisphosphonates and calcium for osteopenia (low bone mineral density)

Has genetic knowledge of Gaucher disease led to new diagnoses?

- Genetic testing
  - Target groups: Ashkenazi Jews, Swedes in certain regions, and some African tribes
- Prenatal genetic testing when there is a high risk factor

Has genetic knowledge of Gaucher disease led to new treatments?

- Enzyme replacement therapy, using recombinant glucocerebrosidase
- Future: Gene therapy has shown promising results for treating Gaucher disease