

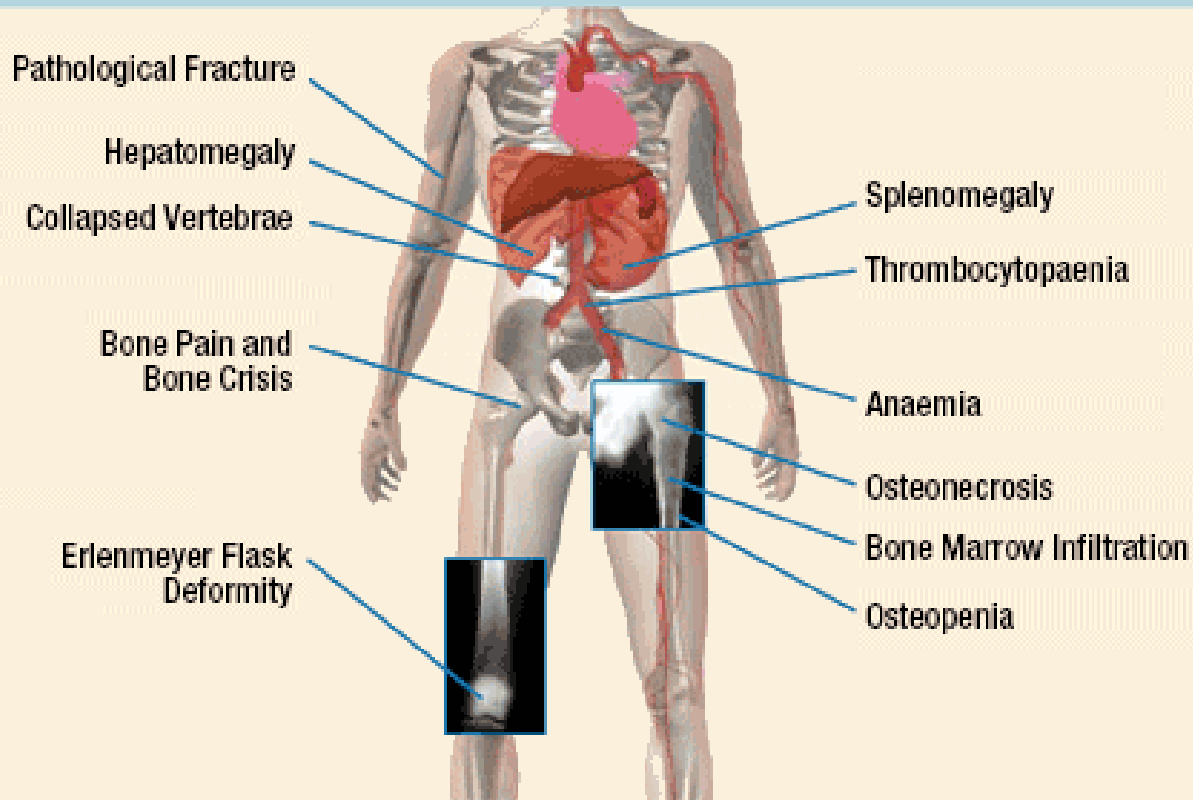
Gaucher Disease

Maya Kornberg

Symptoms and Types

- 3 types
- Type 1 Gaucher's most common
- Symptom include:
 - Bone abnormality
 - Easy bruising
 - Enlarged liver/spleen
 - Lung disease
 - Brain damage/seizure (type 2-3)

Multisystem Involvement in Type 1 Gaucher Disease Can Manifest at Any Age



GBA GENE

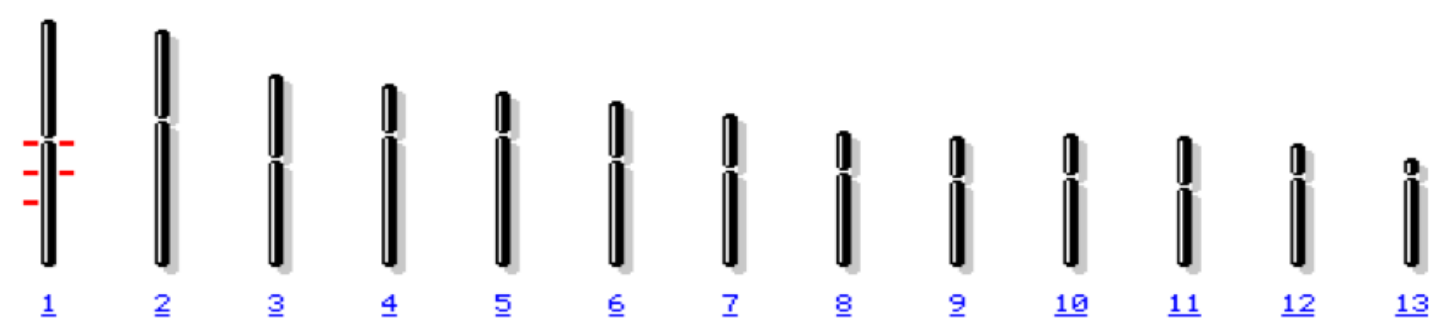
Entrez Genome view

http://www.ncbi.nlm.nih.gov/mapview/map_search.cgi?taxid=9606&build=current&advsrch=off&q Google

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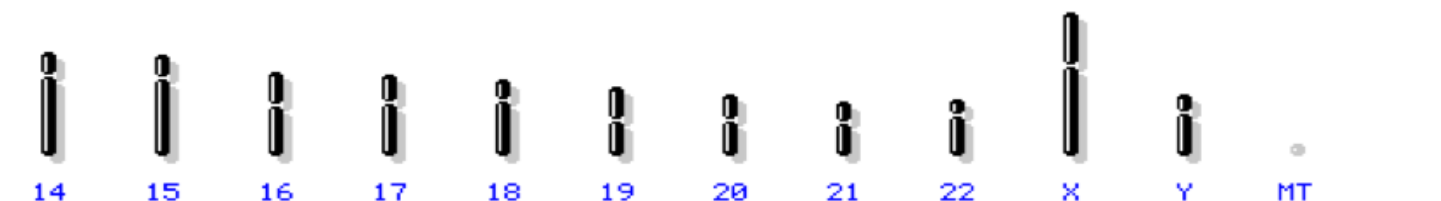
Search for on chromosome(s) assembly

***Homo sapiens (human)* genome view** **BLAST search the human genome**
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Classic Diagnosis

- Deficient glucosylomadise enzyme activity
- Bone marrow examination

Classic Treatment

- Enzyme Replacement Therapy
- Substrate Reduction Therapy
- Chemical Chaperone Therapy
- Bone Marrow transplantation

Novel Diagnosis

- Targeted mutation analysis
- Sequence analysis

Novel Treatment

- Gene Therapy
- Genetic Counseling

