# Fabry Disease

by

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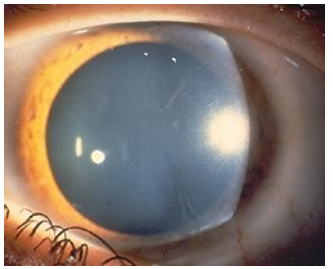
# Fabry Disease

- 1:117k in general population
- 1:40k in males
- genetic disease
- inherited (x-chromosomal recessive)
- progressive

# Symptoms / Phenotype

- Pain and burning in the hands and feet
- Impaired sweating
- Psychological and social issues
- Low tolerance for exercise
- Eye abnormalities

children



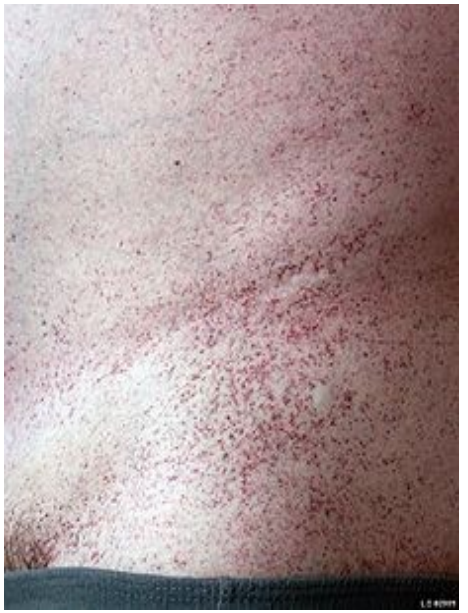
http://medschool.ucsf.edu/

- Dark red skin rashes (angiokeratomas)
- Fatigue
- Gastrointestinal problems

adolescents

- Heart problems
- Kidney problems
- Nervous system problems
- Hearing problems

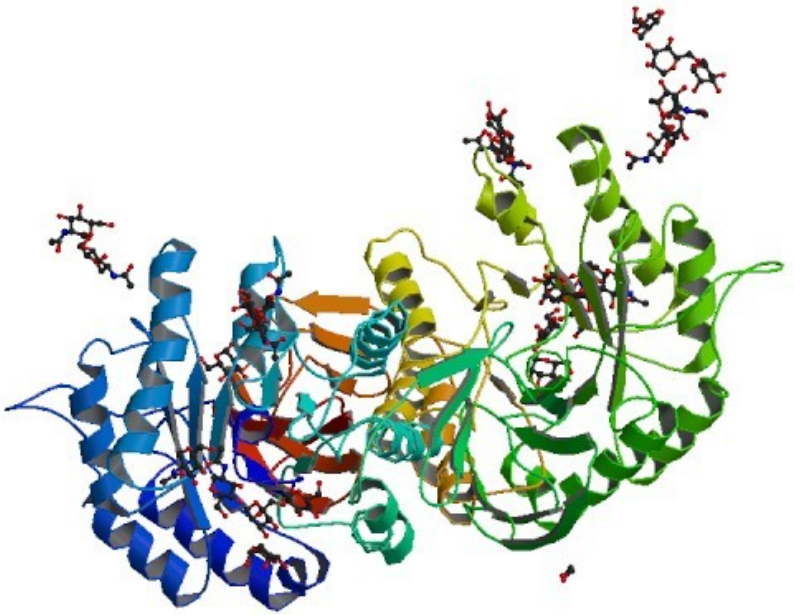
adults



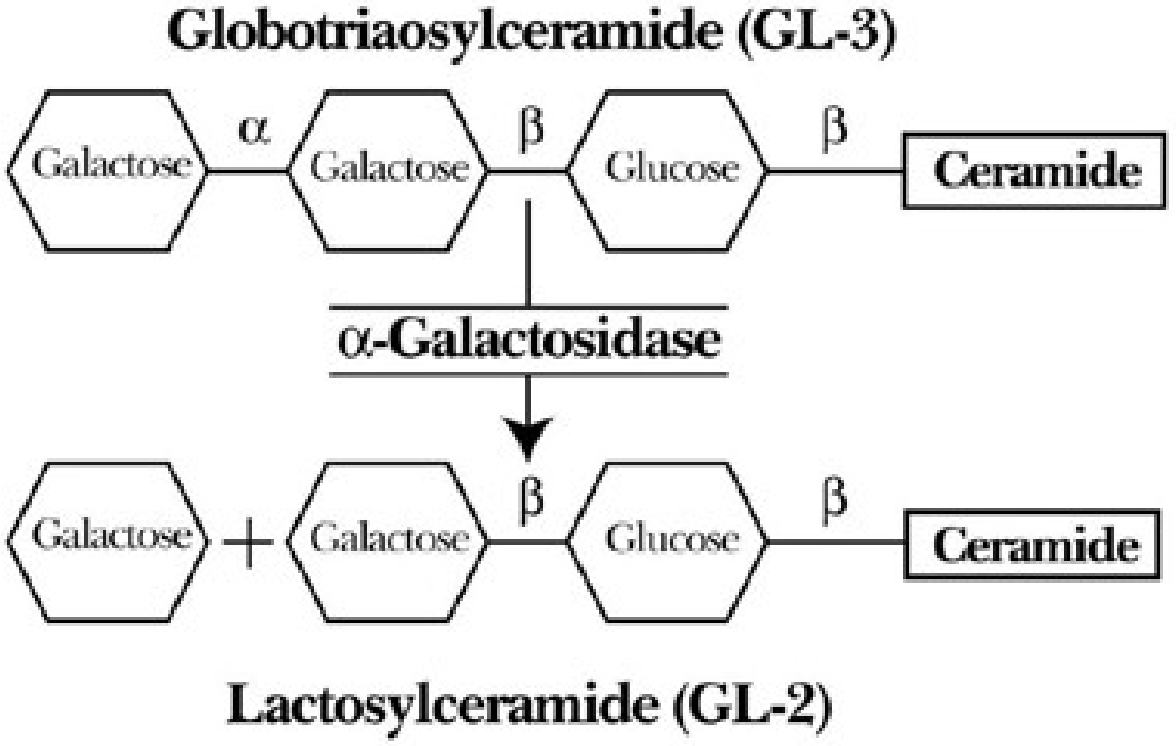
http://en.wikipedia.org/wiki/

# Gene / Protein

- Associated gene: GLA
  - Located on the X chromosome
  - Consists of
    - 7 exons (1290 nts)
    - 6 introns
  - Encodes the enzyme alpha-Galactosidase



# Biochemical Mechanism




http://www.genzyme.co.il

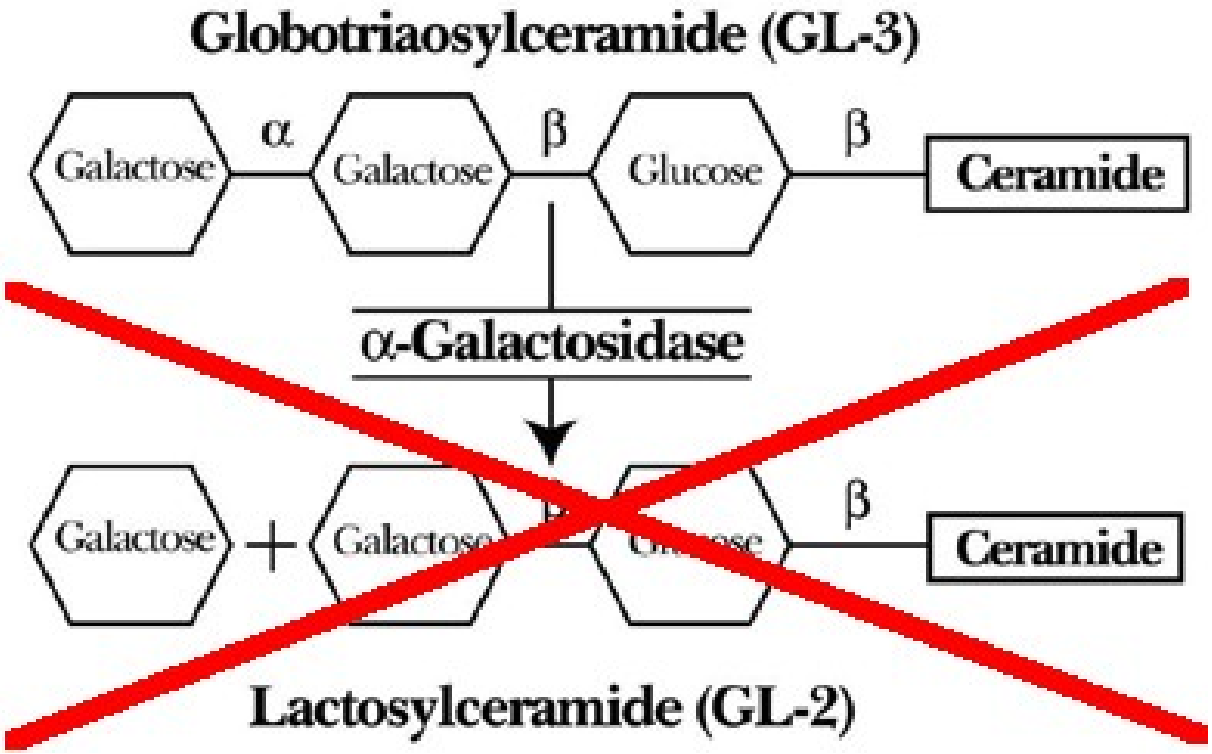
# Mutations

- 492 known mutations
  - Great majority nonsense/missense

- Leads to malfunction of the protein
  - Due to changes of kinetic properties and stability

    → decreased enzyme activity

# Biochemical Disease Mechanism

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Thanks for your attention!

Check our wiki for resources and further information!