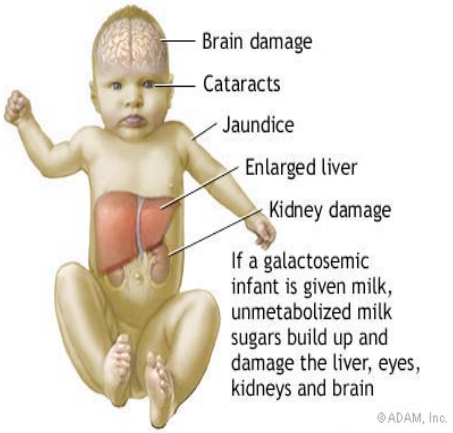
Galactosemia



Case Presentation

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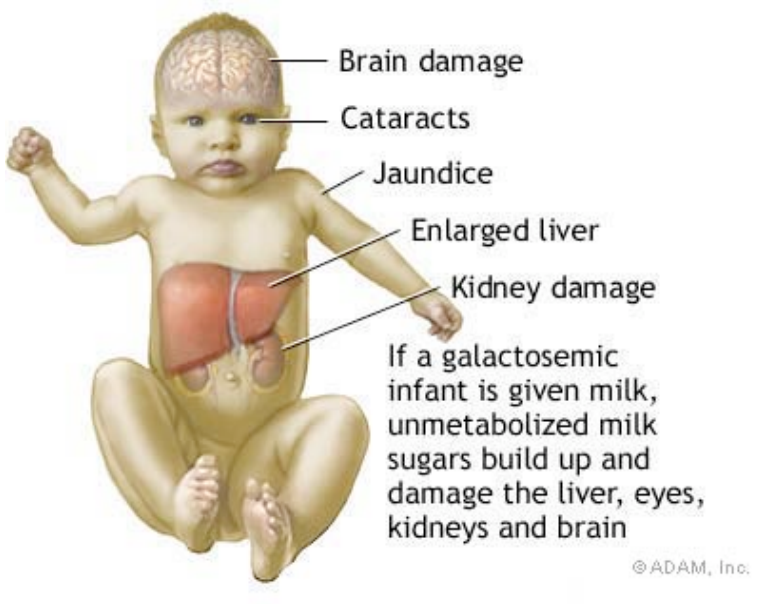
General Overview

- Inability to break down and absorb galactose
- 1 out of 60,000 Caucasian births
- Galactose makes up half of lactose
- Inherited disease
- There is no definite cure
- Three forms of the disease
  - Galactokinase Deficiency
  - Galactose Epimerase Deficiency

Symptoms and Complications



- Convulsions
- Poor weight gain
- Vomiting
- E. coli sepsis
- Cataracts
- Mental Retardation
- Liver Damage (Cirrhosis)
- Death

Classical Treatment of Galactosemia

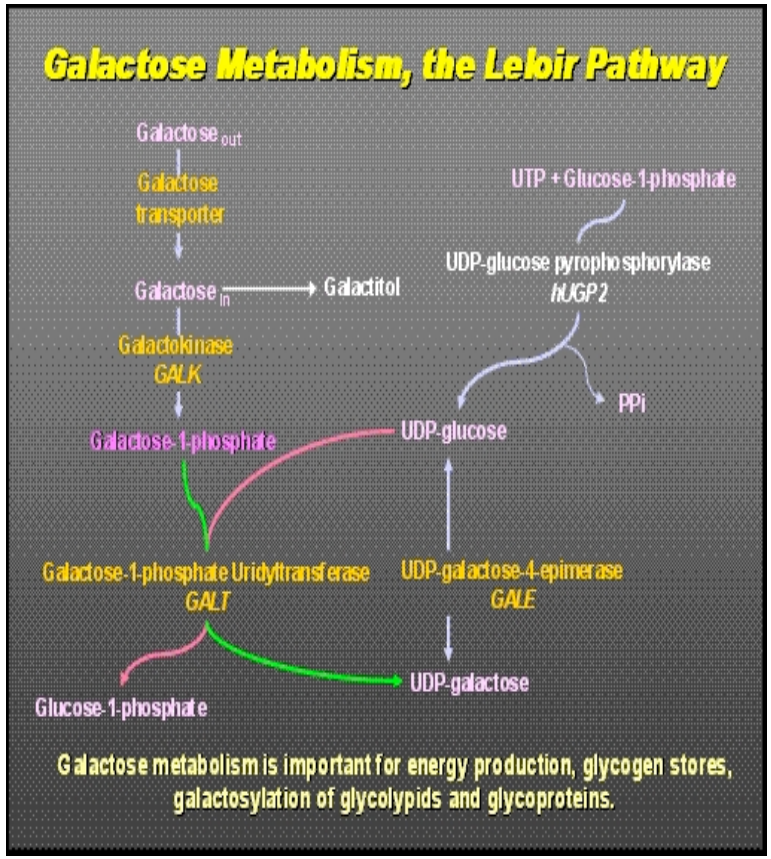
Classical Diagnostic Techniques

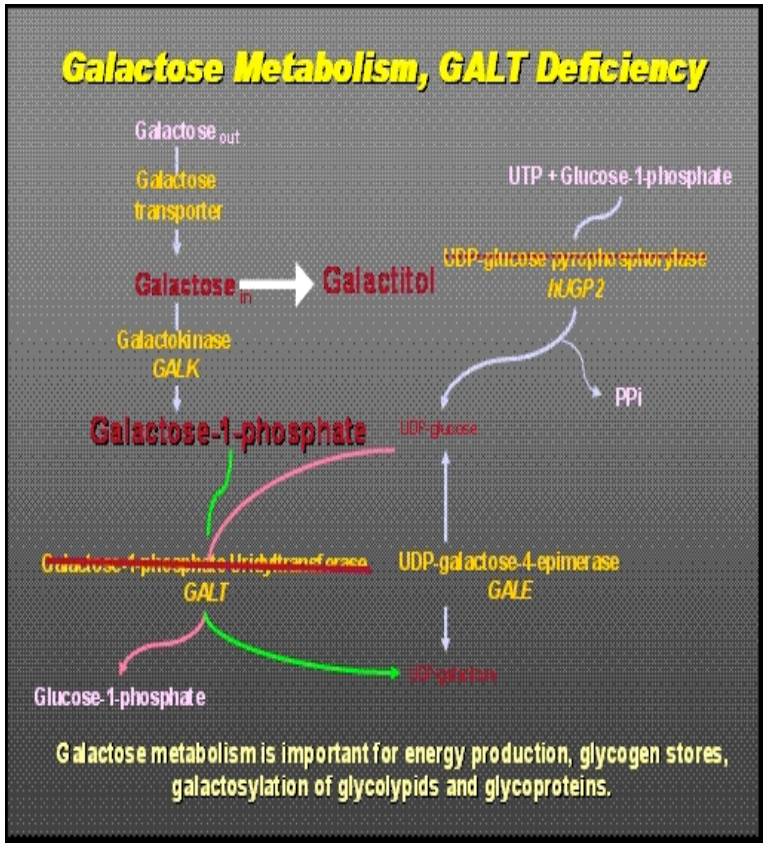
- Enlarged liver
- Jaundice
- Cataracts (10% of cases)
- Failure to thrive
- Urine tests that yield excessive amounts of ammonia
- Sepsis following aforementioned symptoms indicates high likelihood of Galactosemia

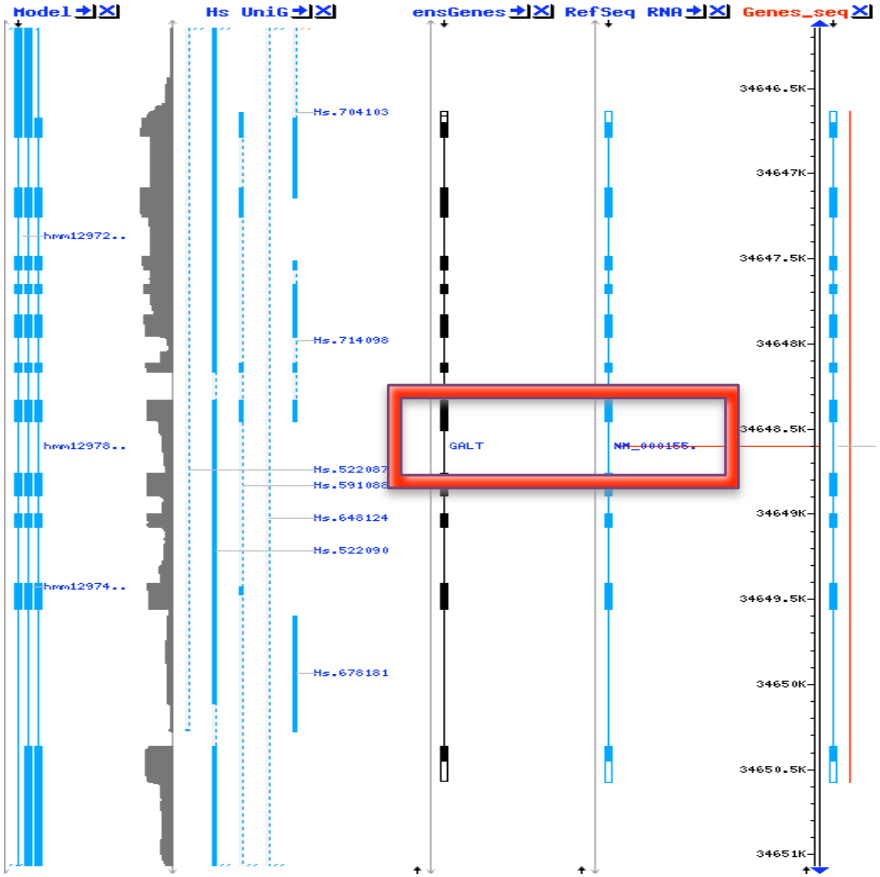
Classical Treatment Techniques

- Dietary Intervention
  - Strict Avoidance of All Dairy Products
  - Calcium Supplementation
- There is no cure for Classic Galactosemia

Galactose-1-phosphate uridyltransferase enzyme (GALT)







Chromosome 9
Location: 9p13

Gene Mapping

Genetic Information: Autosomal Recessive

- **Homozygotes** for the Classic Galactosemia allele (G/G) exhibit 5% of typical GALT enzyme activity

- **Heterozygotes** for the Classic Galactosemia allele (G/g) exhibit 50% of standard GALT enzyme activity

Genetic Diagnostic Tools

- Elimination in breath of less than 5% of ^{13}C-galactose as $^{13}CO_2$ two hours after administration of ^{13}C-D galactose can diagnose disease
- Mutation analysis for the eight common *GALT* Galactosemia (G) mutations (p.Gln188Arg, p.Ser135Leu, p.Lys285Asn, p.Leu195Pro, p.Tyr209Cys, p.Phe171Ser, Δ5kb, IVS2-2A>G.
- Prenatal and neonatal blood work to measure concentration of GALT enzyme. Less than 5 units of GALT per gram of Hemoglobin indicates Galactosemia.

Novel Therapies

- While screening techniques continue to be improved upon due to the genetic understanding of the disease, no viable therapies have been developed to cure Galactosemia.

Citations

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