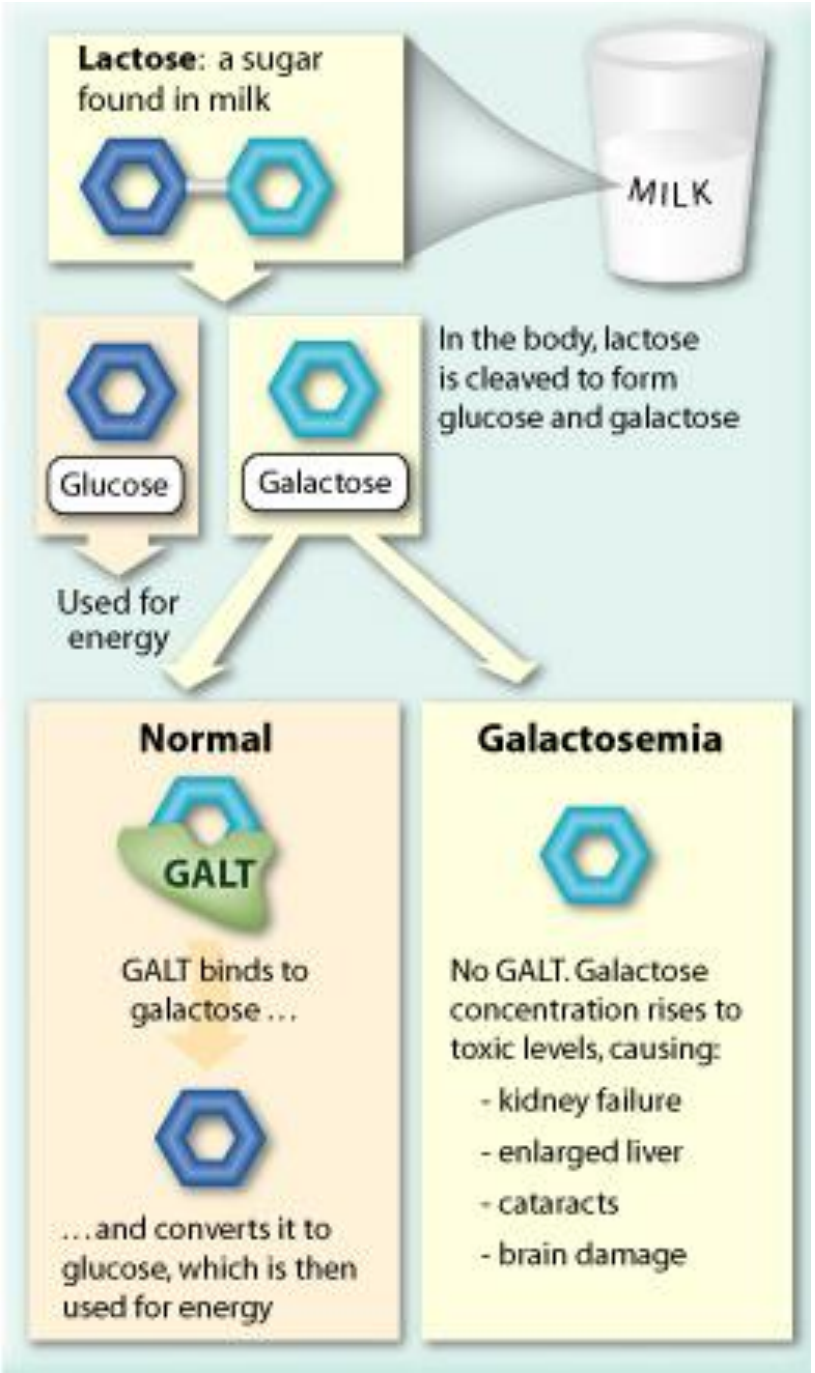
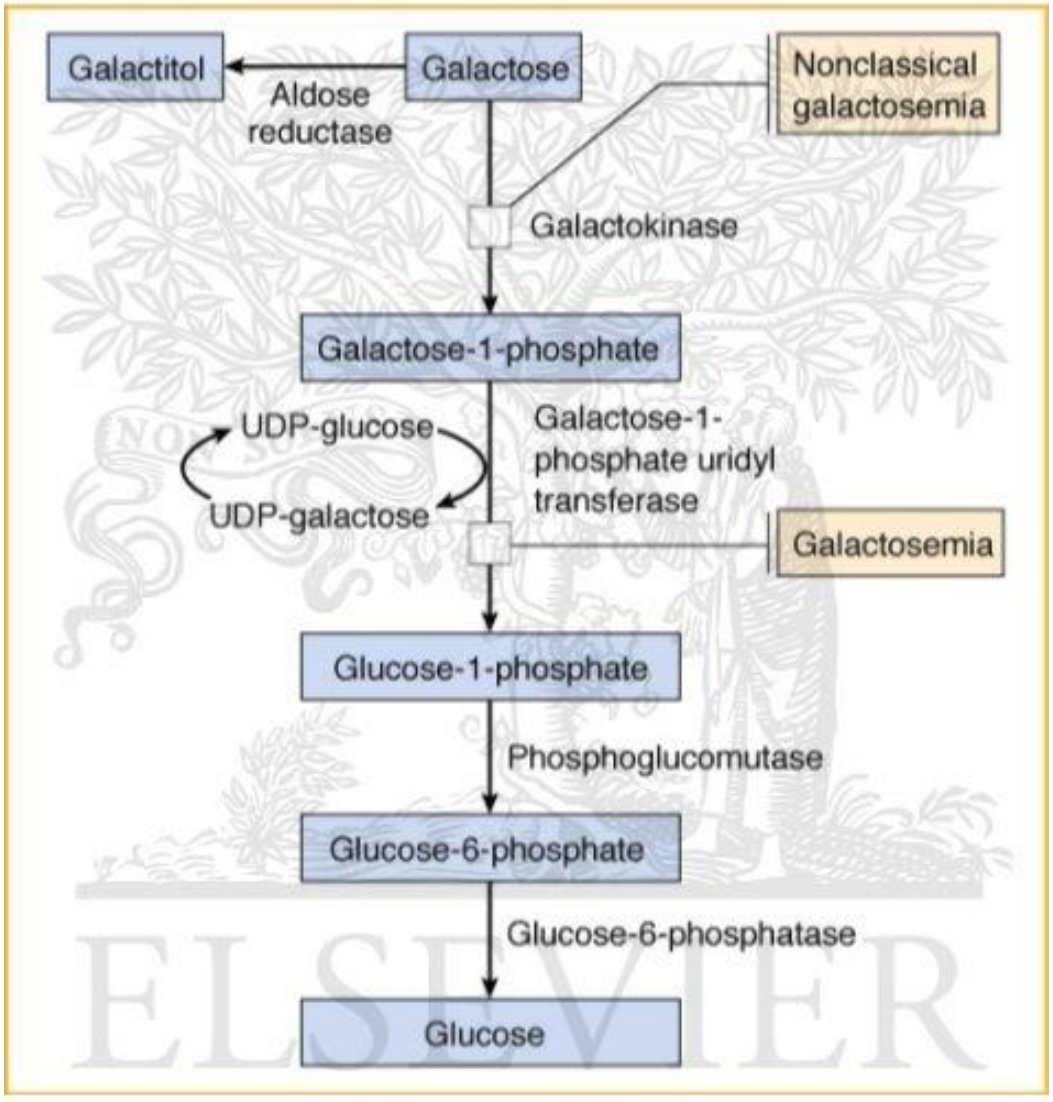


Galactosemia

Amal Alamri

➢**Galactosemia** is an inherited disorder marked by the inability to metabolize galactose , it is one of the carbohydrate metabolic disorder .

➢Individuals with galactosemia are unable to metabolize galactose derived from lactose(milk sugar) to glucose metabolites

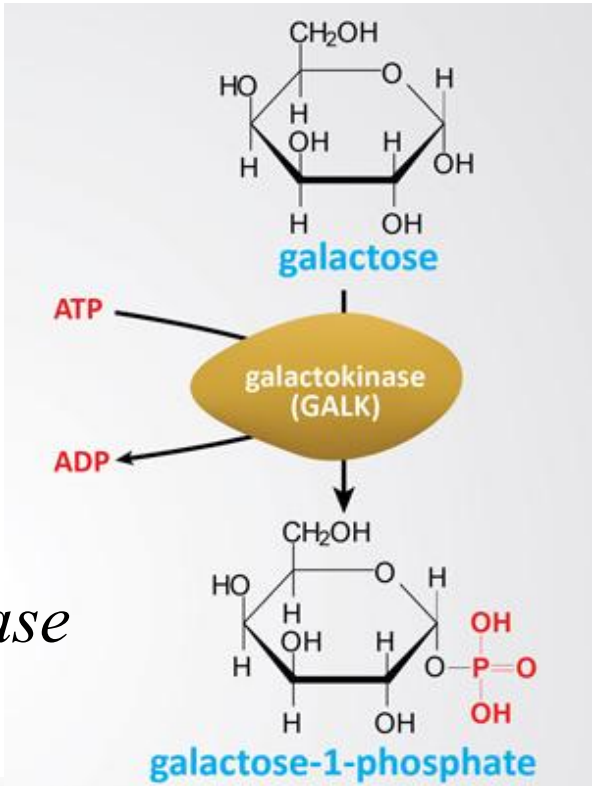


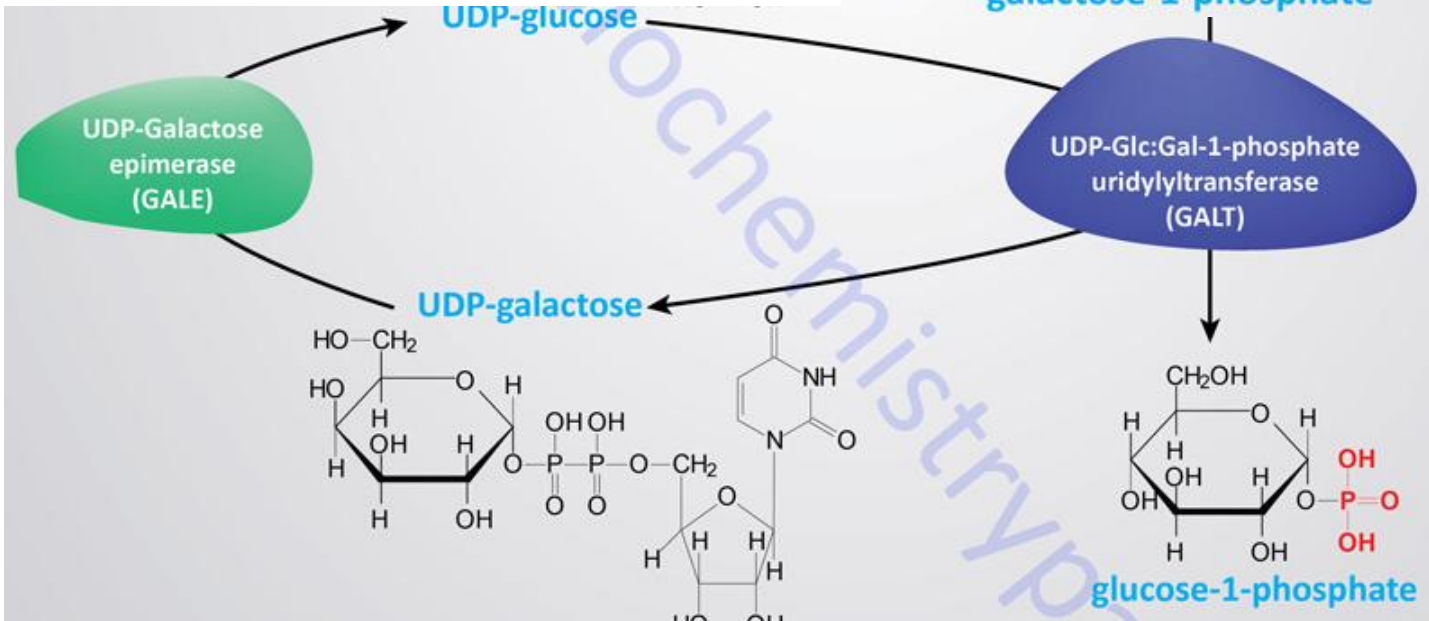
➢There are three forms of the disease:

1-*Galactose-1 phosphate uridylyltransferase* deficiency (classic galactosemia, the most common and most severe form)

2-Deficiency of *galactose kinase*\

3-Deficiency of *galactose-6-phosphate epimerase*





Clinical presentation :

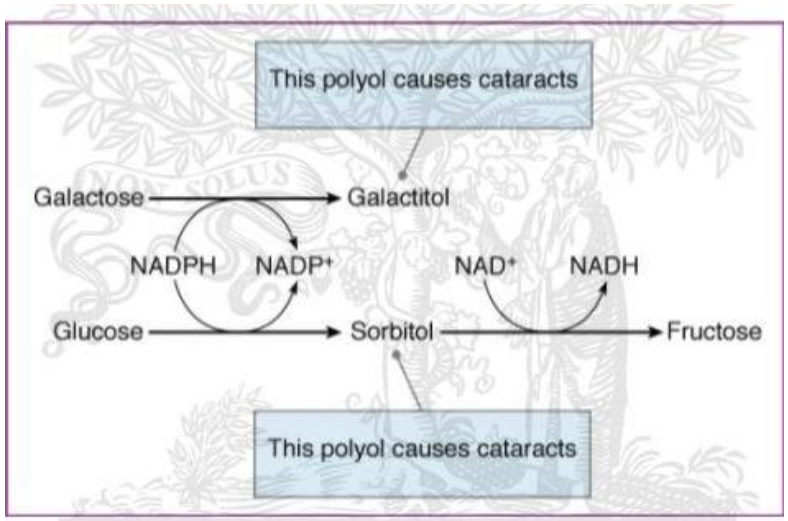
1-If an infant with galactose-1 phosphate uridylyltransferase deficiency .. galactose-1 phosphate accumulate inside liver cells and causes hepatocellular damage and rapid liver failure.

2-Galactose-1 phosphate accumulation causes acute renal tubular failure and tubular loss of glucose ,phosphate,and amino acids.

3-The loss of glucose in cooperation with the liver damage result in sever hypoglycemia .

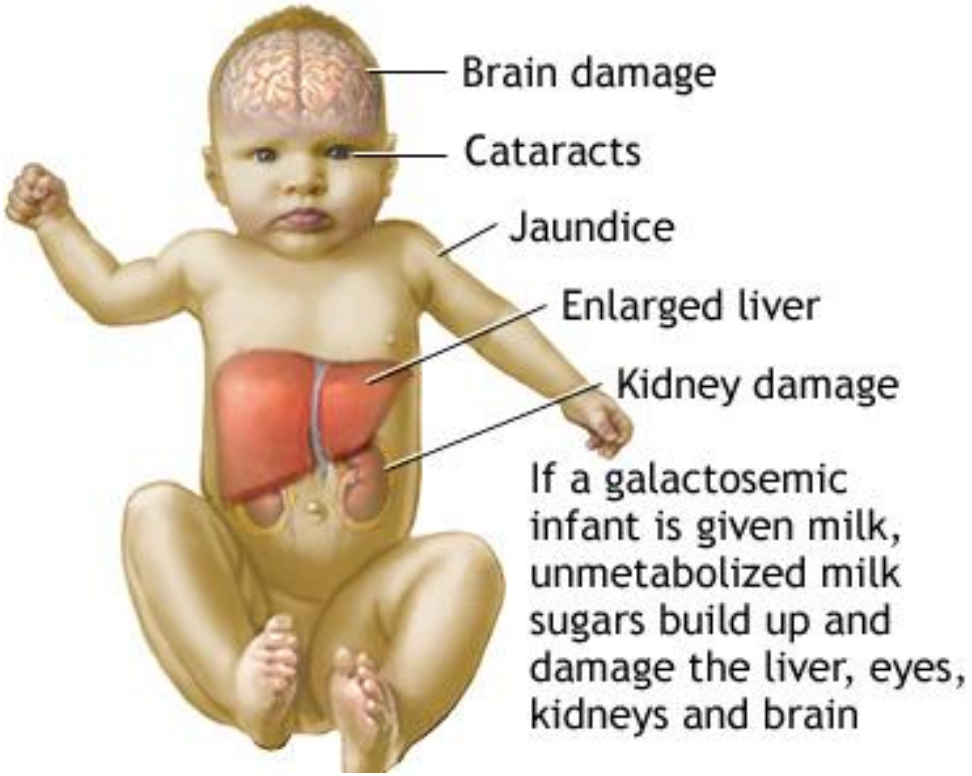
4- Accumulation of galactose in the eye results in cataract formation.

Cataract formation : the accumulation of Gala in eyes will converted to reduced metabolite (Galactitol) by aldo reductase .



Complications

- Cataracts
- hepatocellular damage and liver failure.
- Death (if there is galactose in the diet)
- Delayed speech development
- Intellectual disability
- Severe infection with bacteria (*E. coli sepsis*)
- Tremors and uncontrollable motor functions



Treatment

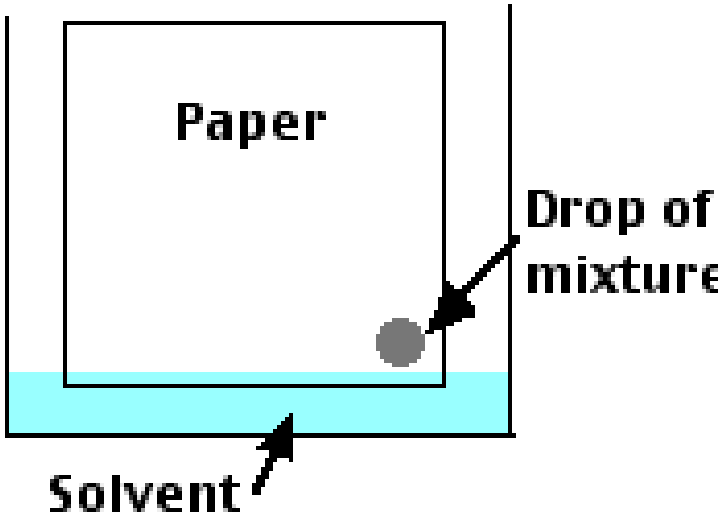
- People with this condition must avoid all milk, milk-containing products (including dry milk), and other foods that contain galactose for life. It is essential to read product labels and be an informed consumer.
- Infants can be fed with:
- Soy formula
- Another lactose-free formula
- Calcium supplements are recommended.

Objective:

To screen for galactosemia

Principle:

Separation of mixture depends on the relative affinity of Compounds towards stationary phase and mobile phase by using paper chromatography .



paper chromatography

Material

- -Paper chromatography
- -standard galactose solution 10%.
- - patient urine sample .
- - normal urine sample.
- -Dinitrosalicylic acid reagent:
- 5 % 3,5 dinitrosalicylic acid in 4 % of NaOH solution (warm).
- - chromatography jar
- -Solvent contains isopropanol : water(80:20)

Method:

•You will be provided with a paper Standard of galactosemia , serum samples.

•With a pencil draw a line about 2 cm . Draw the line gently so as not to break the surface of the paper.

•At equally spaced intervals mark the line at three different places using the tip of the pencil on the silica gel plate.

• Carefully spot the standard , urine patient and normal urine with a 5 µl pipette or spot it by capillary tube .

•Place the gel coated plate in the chamber. Cover the glass chamber tightly with aluminum foil. The solvent will rise up the silica gel by capillarity.

• When the solvent front is between 1 and 2 cm from the top of the paper , remove the chromatogram from the chamber and mark the position of the solvent front. Dry the plate in a stream of cold air.

•Locate the spot by spraying the plate with DNS reagent and heating the plate briefly at 100^0C in an oven.

Results:

Relative flow value is constant for a particular compound, solvent system and insoluble matrix
Calculate the Relative flow value for each spot .

$$\text{Relative flow} = \frac{\text{Distance of migration of solute}}{\text{Distance moved by solvent}}$$

Sample	Relative flow
Standard of galactosemia	
Serum Sample1	
Serum Sample2	

References

- **1- Bishop .M; Fody. E and Schoeff .L.(2005) .Clinical chemistry .Principle , procedures and correlations .Fifth edition. Lippincott Williams &Wilkins.**
- 2- http://www.nlm.nih.gov/medlineplus/ency/article/000366.htm
- 3- Tietz, N. W. (1987) .Fundamentals of Clinical Chemistry . W.B.
- Saunders. Philadelphia. third Edition .
- **4-Clark,Jr. J.M and Switzer .R.(1977).Experimental biochemistry .**
- W.H.Freeman and company ,second edition .
- 5- Arneson,W and Brickell ,J.(2007).Clinical chemistry .F.A.Davis company
- 6- http://www.nlm.nih.gov/medlineplus/ency/article/000366.htm