Galactosemia

- Inability to convert galactose to glucose
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- Inability to convert galactose to **glucose**
- Inheritance: [Abb.]
Galactosemia

- Inability to convert galactose to *glucose*
- Inheritance: *AR*
Galactosemia

- Inability to convert galactose to glucose
- Inheritance: AR
- Results from a defect in one of the enzymes involved in galactose metabolism
Galactosemia

- Inability to convert galactose to glucose
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Galactosemia

- Inability to convert galactose to **glucose**
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**Classic galactosemia:**
- Most vs least common and severe form
Galactosemia

- Inability to convert galactose to glucose
- Inheritance: AR
- Results from a defect in one of the three enzymes involved in galactose metabolism
- Classic galactosemia:
  - Most common and most severe form
Inability to convert galactose to glucose

Inheritance: AR

Results from a defect in one of the three enzymes involved in galactose metabolism

Classic galactosemia:
- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
Inability to convert galactose to glucose
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Classic galactosemia:
- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
Galactosemia

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- **Classic galactosemia:**
  - Most common and most severe form
  - Caused by defect in the uridyltransferase enzyme
  - 75% develop cataracts within time unit to time unit of birth
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Classic galactosemia:
- Most common and most severe form
- Caused by defect in the uridylyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as 'oil droplet' classic two-word description
Inability to convert galactose to glucose

Inheritance: AR

Results from a defect in one of the three enzymes involved in galactose metabolism

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Classic galactosemia:

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- 75% develop cataracts within **days** to **weeks** of birth
  - Starts as ‘oil droplet’; progresses to...
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- Most common and most severe form
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- 75% develop cataracts within days to weeks of birth
  - Starts as ‘oil droplet’; progresses to total opacification
- To diagnosis classic galactosemia: Check UA for presence of two words, important, often overlooked
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- 75% develop cataracts within **days** to **weeks** of birth
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- To diagnosis classic galactosemia: Check UA for presence of reducing substances **after milk ingestion**
Galactosemia

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- To diagnosis classic galactosemia: Check UA for presence of **reducing substances after milk ingestion**
- Treatment: Elimination of dietary **two words**
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**What are the systemic manifestations of classic galactosemia?**

- Failure to thrive; hepatomegaly with jaundice; impaired cognitive development
- 75% of affected infants develop cataracts within days to weeks of birth:
  - Starts as ‘oil droplet’; progresses to total opacification
- To diagnosis classic galactosemia: Check UA for presence of reducing substances after milk ingestion
- Treatment: Elimination of dietary milk products

**How soon do these findings begin to manifest?**

- Within a few weeks after birth

**What is the prognosis if classic galactosemia goes untreated?**

- It is uniformly fatal
Inability to convert galactose to glucose
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Treatment: Elimination of dietary milk products

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- Caused by defect in the uridylyltransferase enzyme

75% of children with classic galactosemia develop cataracts within days to weeks of birth.

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Failure to thrive; hepatomegaly with jaundice; impaired cognitive development

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- In the two non-classic forms...
  - Disease more/less severe than in classic disease
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In the two non-classic forms…
- Disease less severe than in classic disease
- Cataracts present earlier vs later in life
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