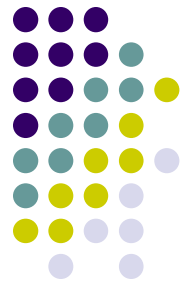


Galactosemia

- Inability to convert galactose to 



Galactosemia

- Inability to convert galactose to glucose



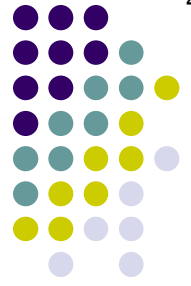
Galactosemia

- 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



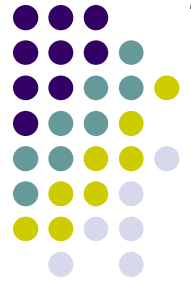
Galactosemia

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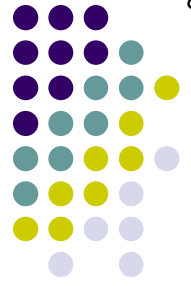
Galactosemia

- 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 vs least common and most vs least severe form

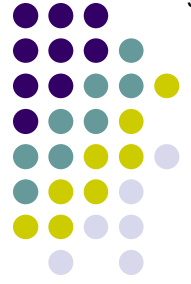


Galactosemia

- Inability to convert galactose to glucose
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- *Classic galactosemia*:
 - Most common and most severe form



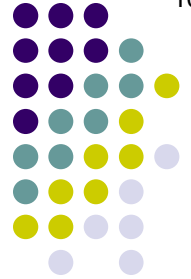
Galactosemia



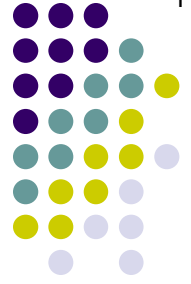
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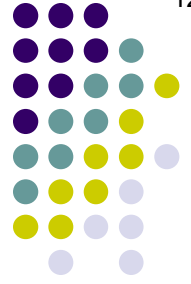


Galactosemia



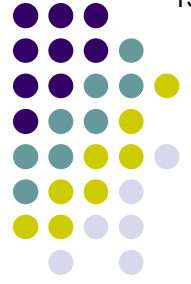
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Galactosemia



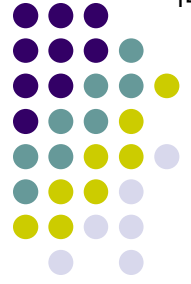
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 - Caused by defect in the **uridyltransferase** enzyme
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 - Starts as classic two-word description

Galactosemia



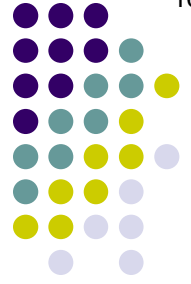
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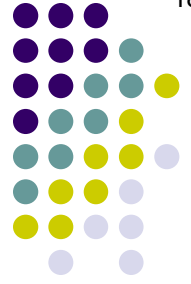
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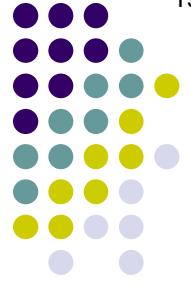
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 - To diagnosis classic galactosemia: Check UA for presence of two words *after* important, often overlooked

Galactosemia



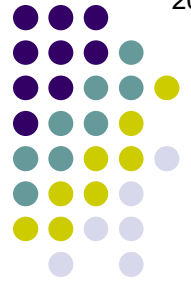
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Galactosemia

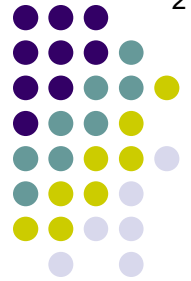


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Failure to thrive; hepatomegaly with jaundice; impaired cognitive development

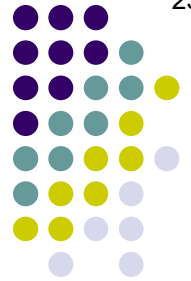
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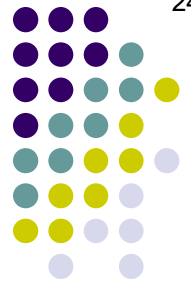
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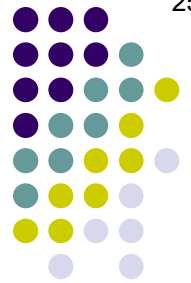
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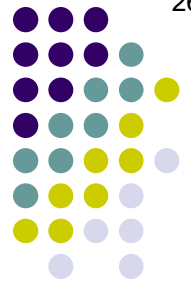
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It is uniformly fatal





Galactosemia

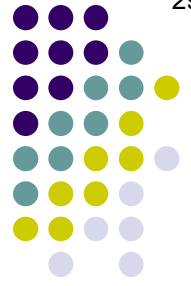
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