Galactosemia

- Inability to convert galactose to [redacted]

Galactosemia

- Inability to convert galactose to glucose

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- Inheritance: abb.

Galactosemia

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  - most vs least common and most vs least severe form

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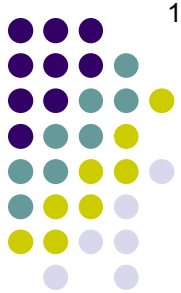
Galactosemia



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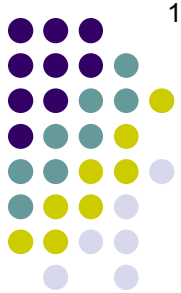
What simple exam maneuver is the best way to pick up an oil droplet cataract?

Galactosemia



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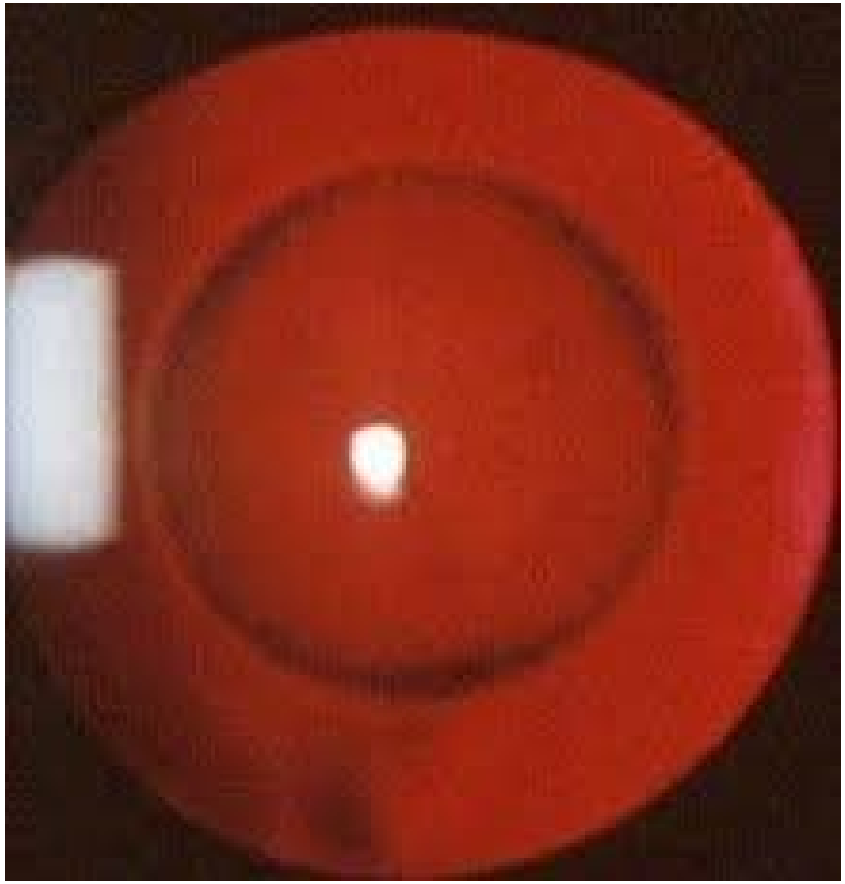
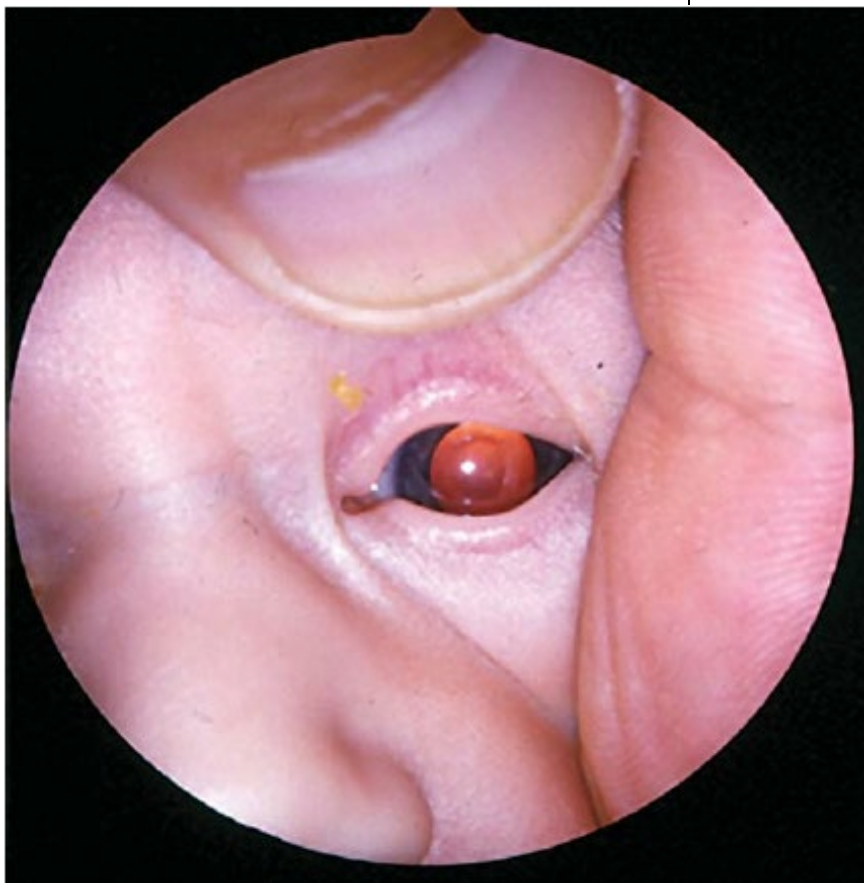
What simple exam maneuver is the best way to pick up an oil droplet cataract?
Retroillumination

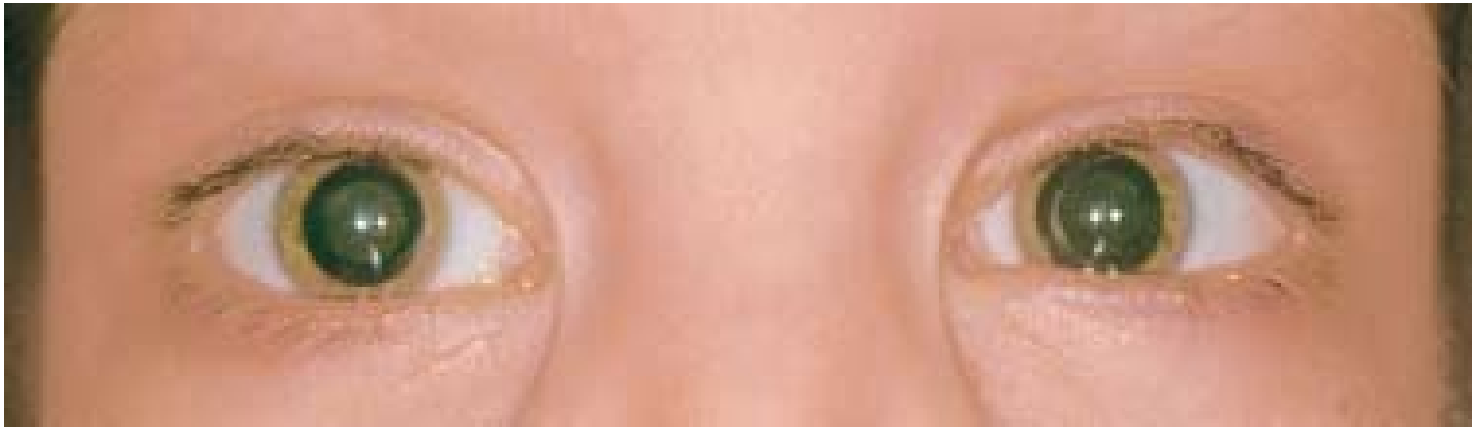
Galactosemia





Oil-droplet cataract: Retroillumination

Galactosemia



Galactosemia: Oil-droplet cataracts

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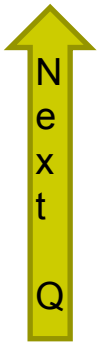
    - Starts as 'oil droplet'; progresses to classic two-word description



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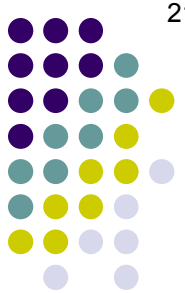
Next Q

Galactosemia



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Anterior and posterior

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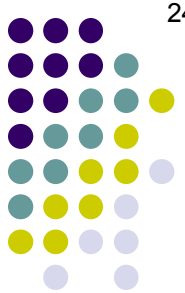
Anterior and posterior lenticonus

Galactosemia



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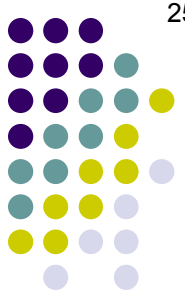
Anterior and posterior **lenticonus**

In one (unsurprising) word, what is the shape of the affected lens surface in lenticonus?

Galactosemia



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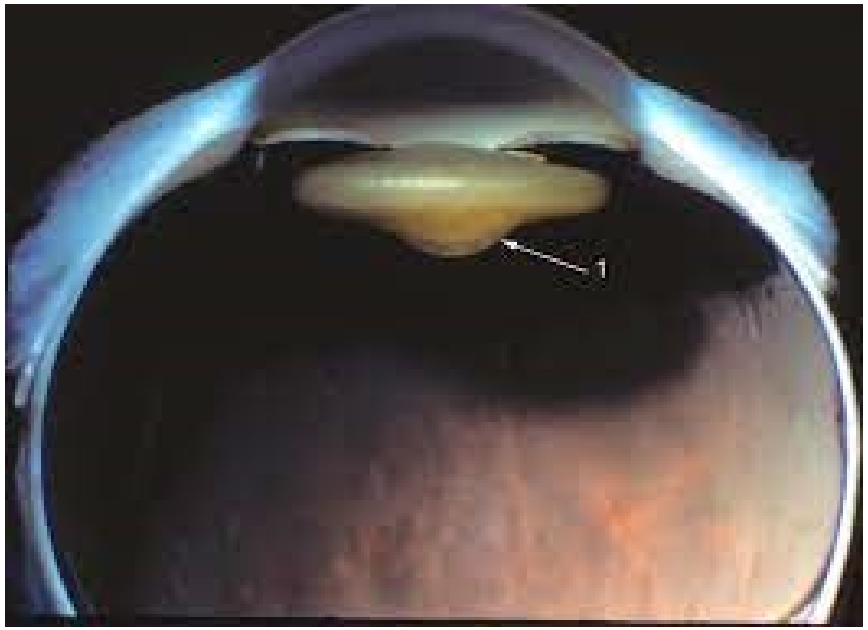
In one (unsurprising) word, what is the shape of the affected lens surface in lenticonus?
'Conical.'

Galactosemia



The conical shape of a lenticonus lens

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Anterior and posterior **lenticonus**

In one (unsurprising) word, what is the shape of the affected lens surface in lenticonus? 'Conical.' So, there's a fundamental difference between the cause of the oil-droplet appearance in lenticonus (= increased central lenticular power) vs that of galactosemia (= the accumulation of galactitol and fluid in the central lens).

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Most cases of anterior lenticonus are unilateral vs bilateral *and* inherited vs sporadic

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for
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Anterior and posterior **lenticonus**

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Anterior and posterior **lenticonus**

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Anterior and posterior **lenticonus**

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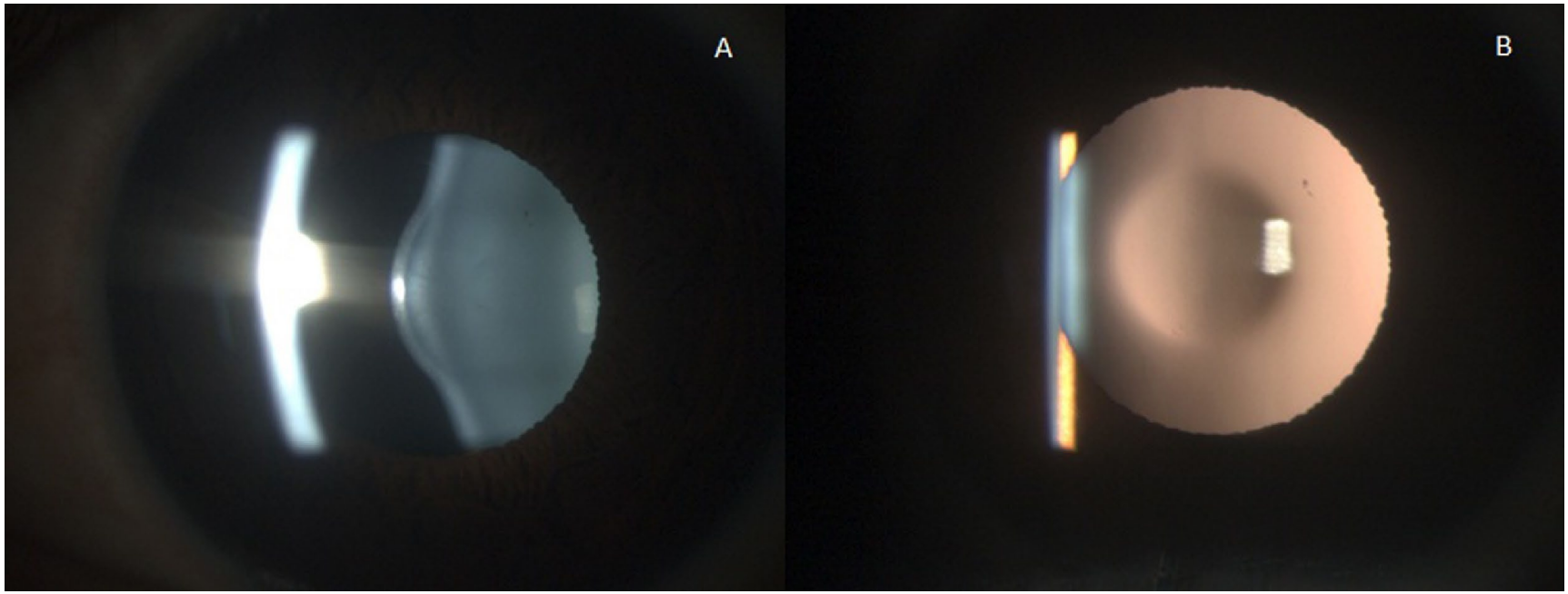
Anterior and posterior **lenticonus**

Galactosemia



Anterior lenticonus

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Alport syndrome

Anterior and posterior **lenticonus**

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Anterior and **posterior lenticonus**

Most cases of posterior lenticonus are unilateral vs bilateral *and* inherited vs sporadic

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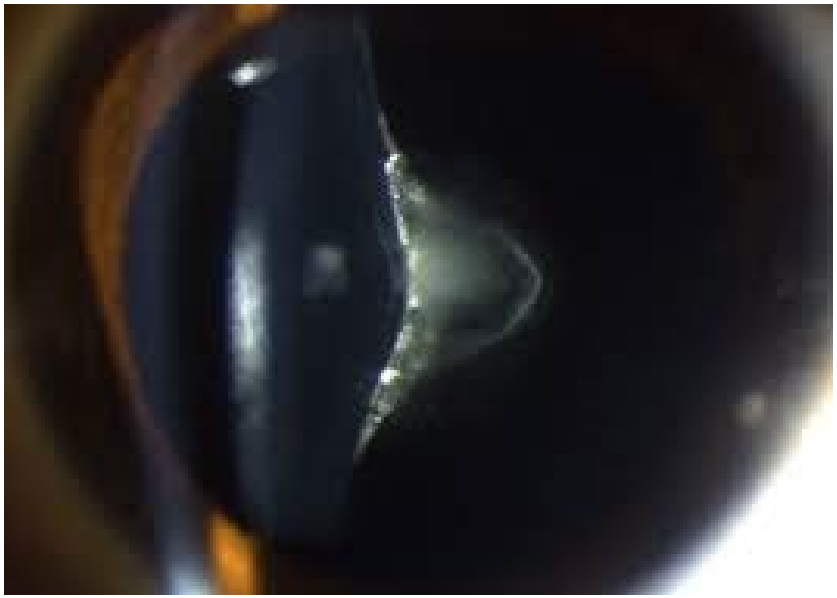
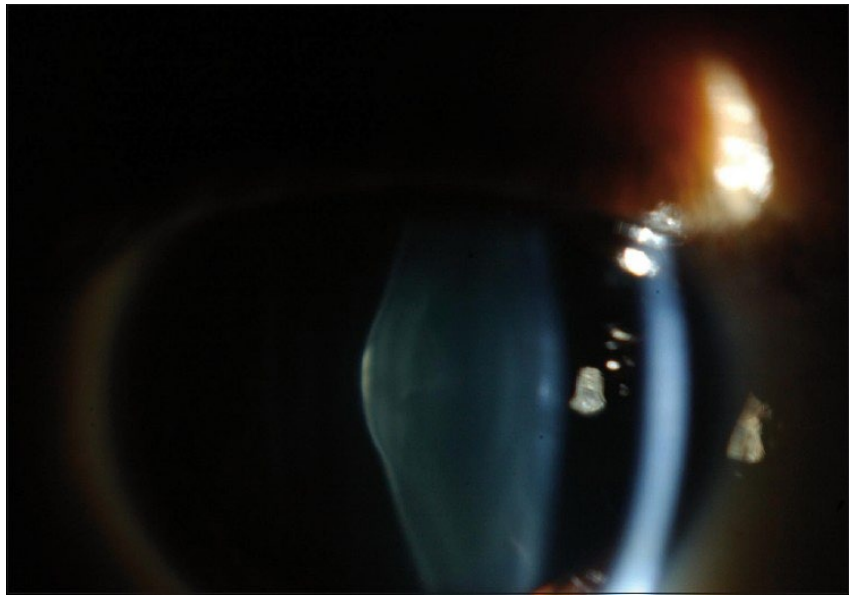
Anterior and **posterior lenticonus**

Most cases of posterior lenticonus are unilateral and sporadic .

Galactosemia





Posterior lenticonus

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Anterior and **posterior lenticonus**

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Anterior and **posterior lenticonus**

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Lowe syndrome

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Alport and Lowe syndromes are examples of what class of condition?

Most cases of anteric… it is usually sporadic

Alport syndrome

Anterior a…

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Familial oculorenal syndromes

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Hematuria

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Alport syndrome

Anterior a…

Of the two relationships (Alport<->anterior lenticonus; Lowe<->posterior lenticonus), which is stronger?

*Most cases o… usually syndromic . With what syndrome is **posterior** lenticonus strongly associated?*

Lowe syndrome

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Alport syndrome

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Lowe<->posterior lenticonus. Anterior lenticonus occurs in Alport syndrome in only about % of cases.

Anterior a…

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  - 75% develop cataracts within days to weeks of birth
    - S… …ion

Alport and Lowe syndromes are examples of what class of condition?
Familial oculorenal syndromes

Most cases of anteric… it is usually sporadic…

Alport syndrome

Of the two relationships (Alport<->anterior lenticonus; Lowe<->posterior lenticonus), which is stronger?
Lowe<->posterior lenticonus. Anterior lenticonus occurs in Alport syndrome in only about 25% of cases.

Anterior a…

*Most cases o… usually syndromic . With what syndrome is **posterior** lenticonus strongly associated?*

Lowe syndrome

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For more on lenticonus and Alport/Lowe syndromes, see slide-set L4

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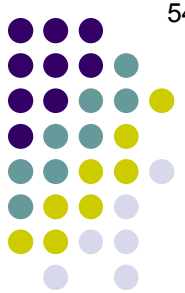
--?
--?
--?

enzyme
ks of birth
cification

Galactosemia



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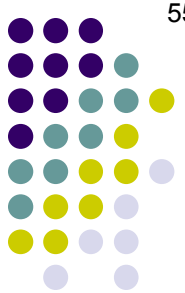

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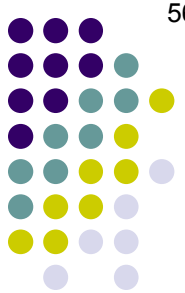

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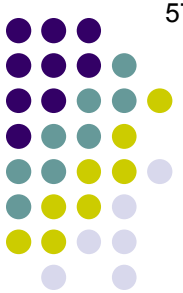

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enzyme
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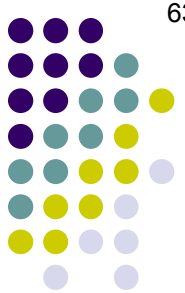

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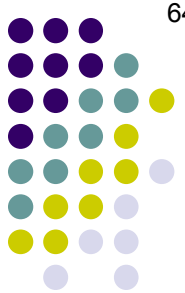
Will the cataractous changes reverse with treatment?

Galactosemia



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Will the cataractous changes reverse with treatment?
Yes, if it is started in a timely fashion

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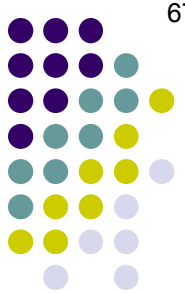

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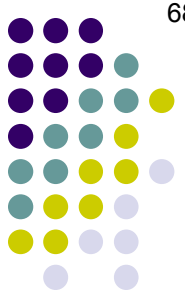

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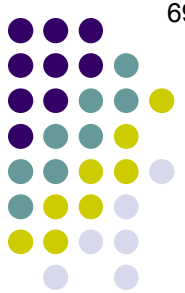

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