

Q

On LHON



- LHON stands for

[Redacted area]

A

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- LHON is more likely to affect males vs females, despite the fact that transmission is organelle

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Where does LHON rank among inherited mitochondrial diseases in terms of incidence?

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Where does LHON rank among inherited mitochondrial diseases in terms of incidence?
It is #1--the most common

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80-90 are male; 10-20 are female



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As female offspring inherit the same genotype, they would be expected to display the phenotype at rates equal to those of males



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What is the typical time interval between initial and fellow-eye presentation?

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Are there cases in which the interval has been much longer--say, years?



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How old at onset were the youngest and oldest confirmed cases?

Youngest:

Oldest:



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How old at onset were the youngest and oldest confirmed cases?

Youngest: 1 year old

Oldest: 80



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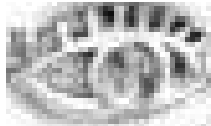
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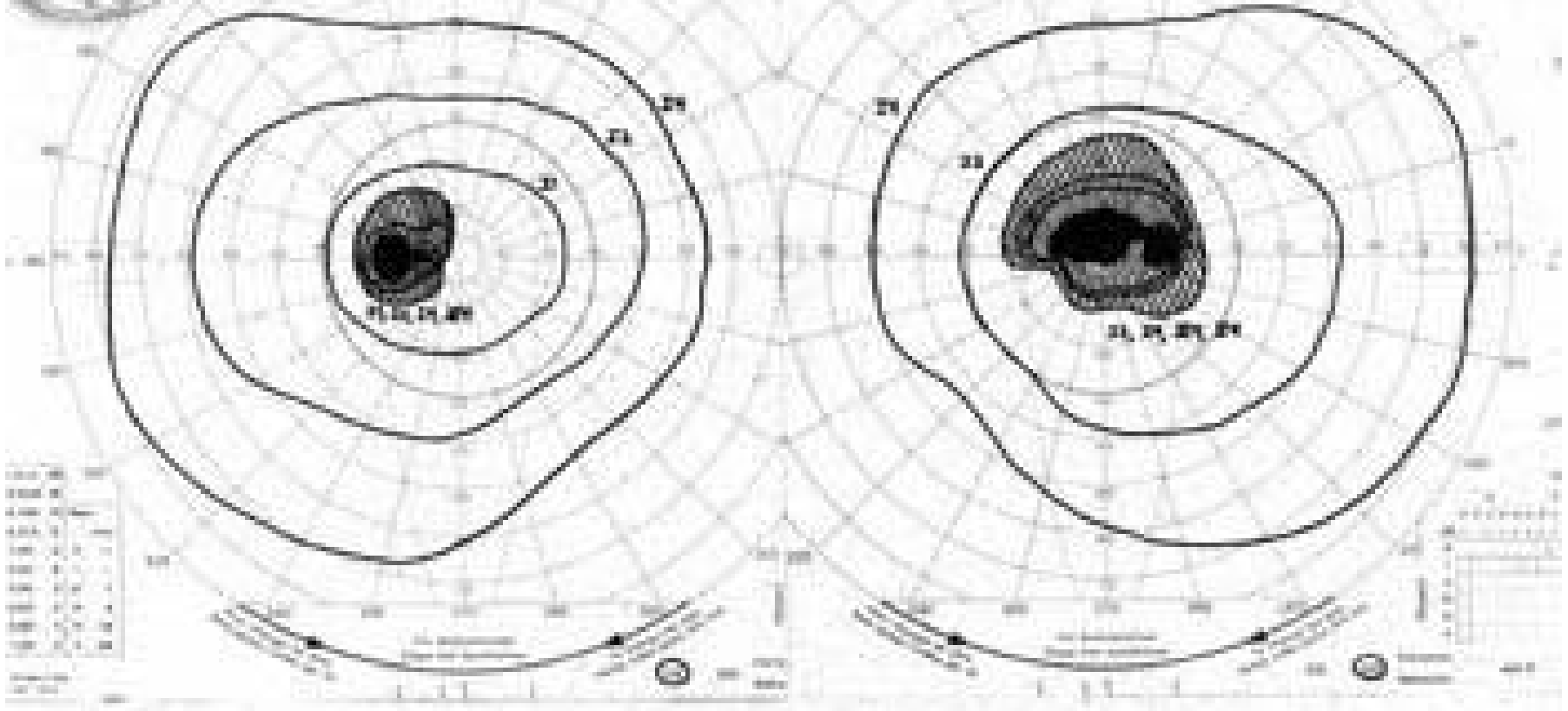
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LHON: Central/cecocentral scotomata



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red - green vs blue - yellow

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- 1)
- 2)
- 3)
- 4)



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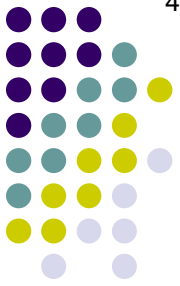
How can you tell if a red-green deficiency is acquired?

- 1) If it is in one eye only
- 2) If the patient is female (females can have inherited red-green defects, but it is highly unusual)
- 3) If it is sectoral (i.e., one portion of the visual field is desaturated compared to others)
- 4) The clinical setting; i.e., if the patient is complaining of decreased acuity, field loss, pain with movement, etc

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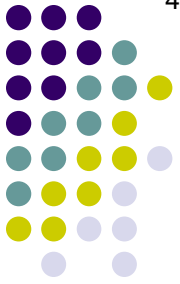
- Classic DFE findings:
 - ONH...
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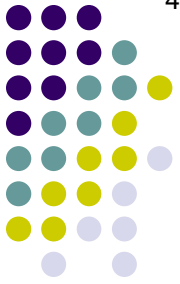
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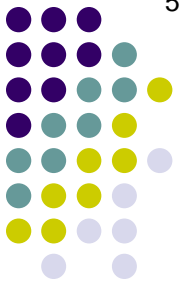
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 - Retinal arteriolar...



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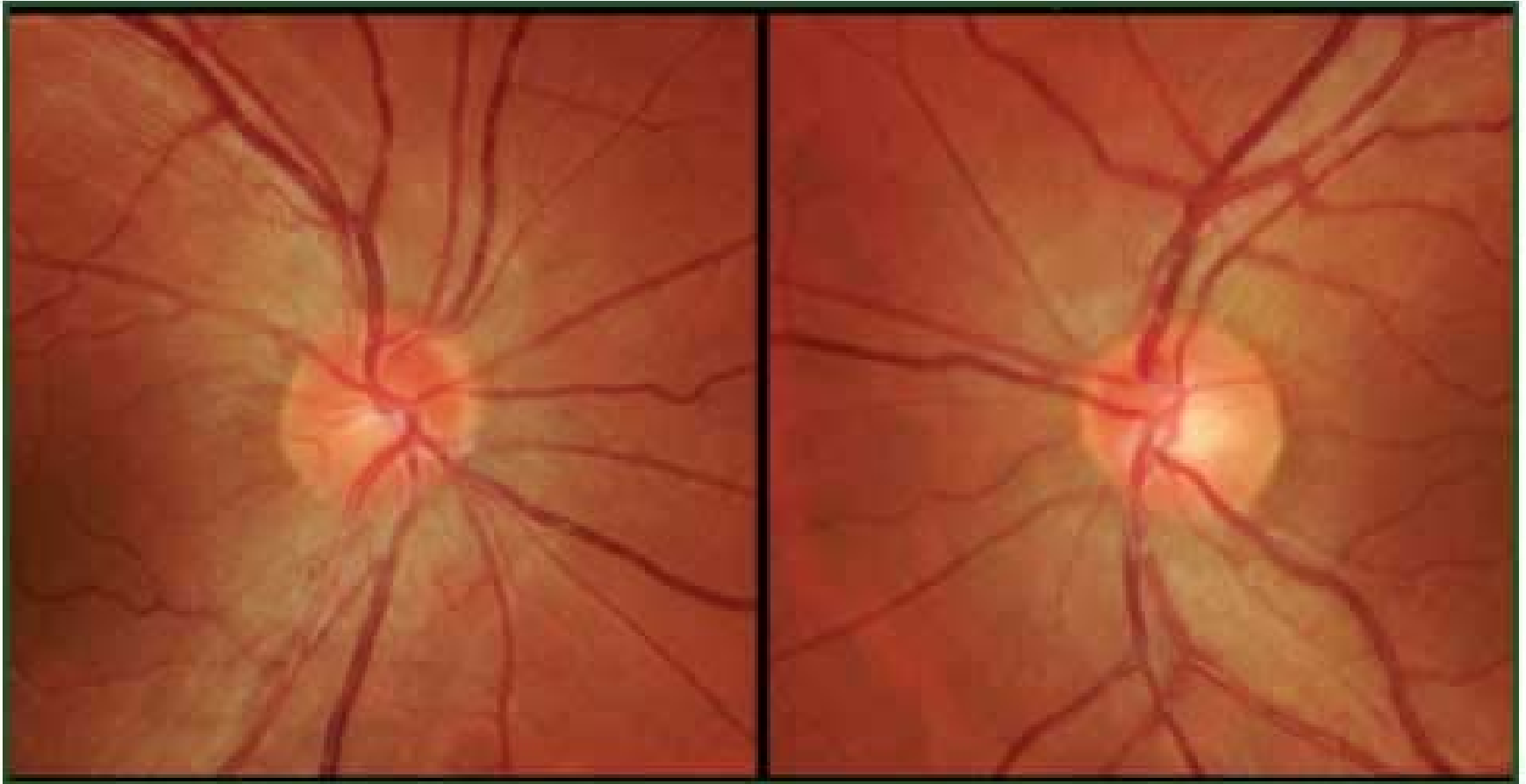
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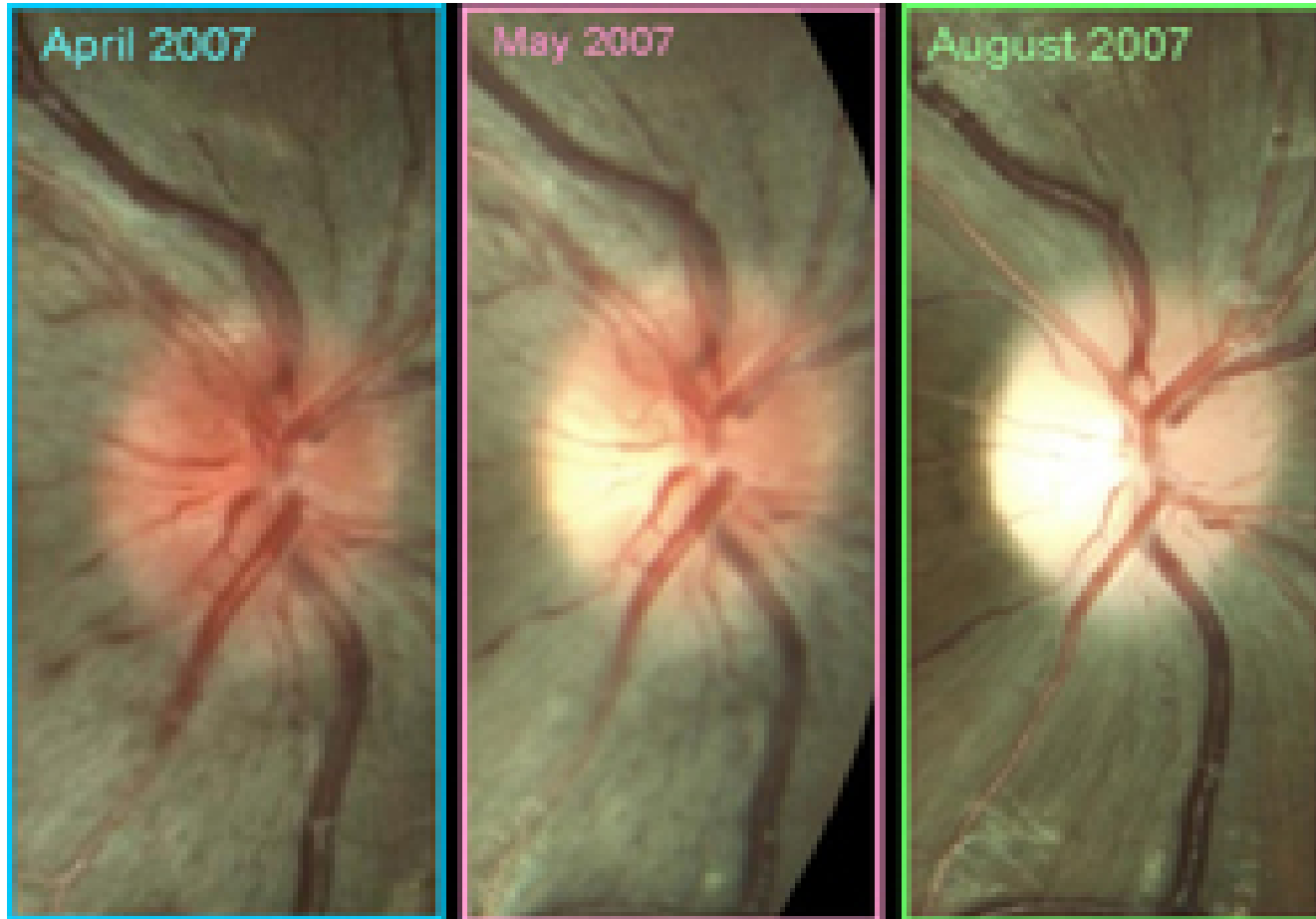


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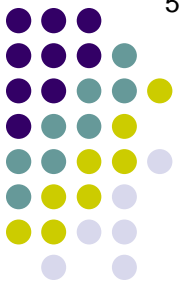


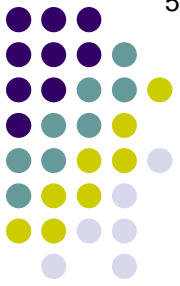
6. A careful comparison of the discs revealed subtle findings—disc hyperemia, relative opacity of the retinal nerve fiber layer, and mild telangiectatic (corkscrew) vessels—that were more marked in the right eye.

On LHON



LHON: Progression of ONH atrophy



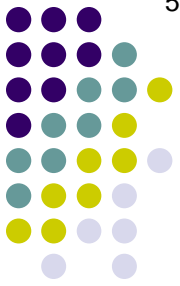


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- Cardiac co-morbidity:

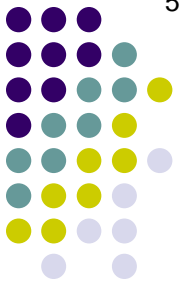
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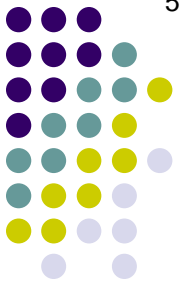


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What is WPW?



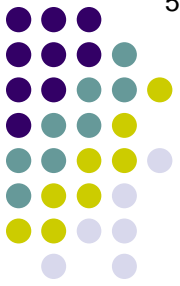
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What is WPW?

An abnormality of cardiac **conduction**



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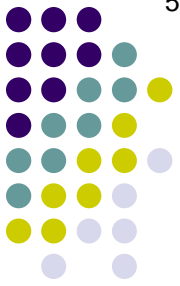
An abnormality of cardiac **conduction**

What are the classic EKG findings in WPW?

--The PR interval is abnormally...[long vs short]

--

--



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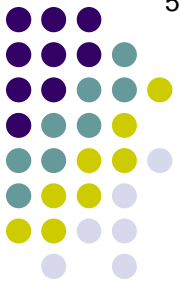
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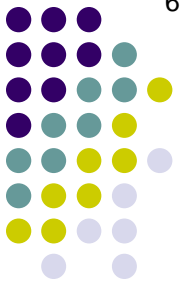
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--The QRS complex is abnormally...*[wide vs narrow]*



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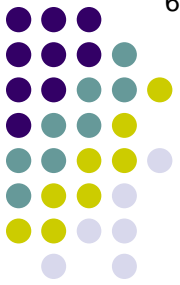
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--The QRS complex is abnormally...**wide**



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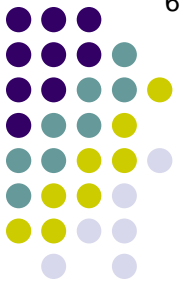
An abnormality of cardiac **conduction**

What are the classic EKG findings in WPW?

--The PR interval is abnormally...**short**

--The QRS complex is abnormally...**wide**

--The QRS complex onset is...*[classic descriptor]*



A

On LHON

- Classic DFE findings:
 - ONH...telangiectasias
 - ONH...pseudoedema
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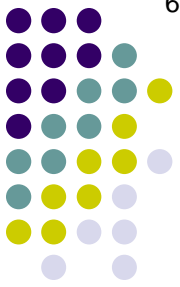
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What are the classic EKG findings in WPW?

--The PR interval is abnormally...**short**

--The QRS complex is abnormally...**wide**

--The QRS complex onset is...**'slurred'**



WPW: Slurred onset of the QRS complex



Q

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WPW renders pts prone to what abnormal rhythm?



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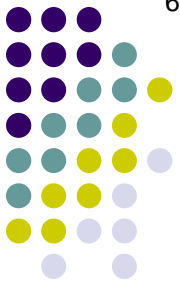
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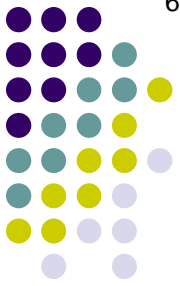
Supraventricular tachycardia (SVT)



Q

On LHON

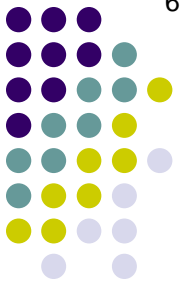
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- Diagnosis: Blood assay for abb + word



A

On LHON

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What are the genetic positions for the three most common mutations?

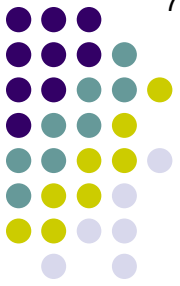


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*What are the genetic positions for the three most common mutations?
11778, 3460 and 14484*



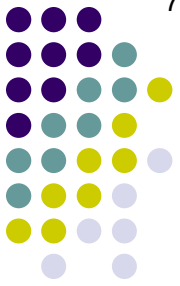
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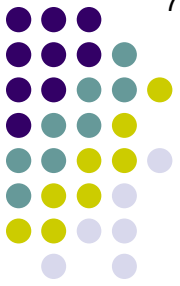
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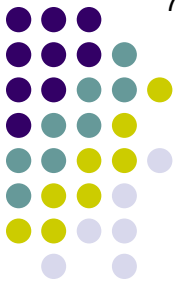
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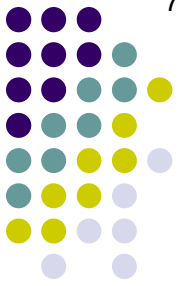
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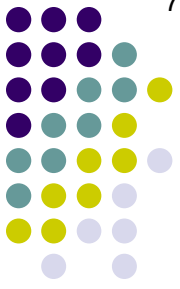
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A

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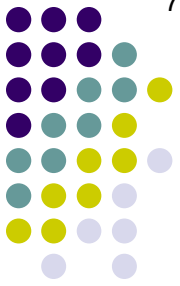
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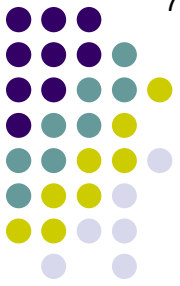
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Q

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- Treatment:



A

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