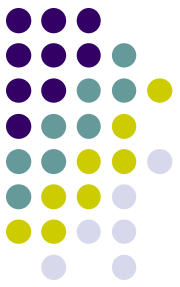


Q

*PAX Ophthalmicana*



?



?

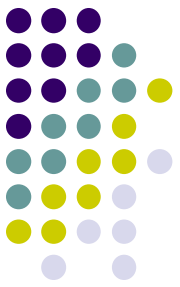


?

*With respect to genetics, to what does the term Central Dogma refer?*

A

*PAX Ophthalmicana*



DNA



RNA



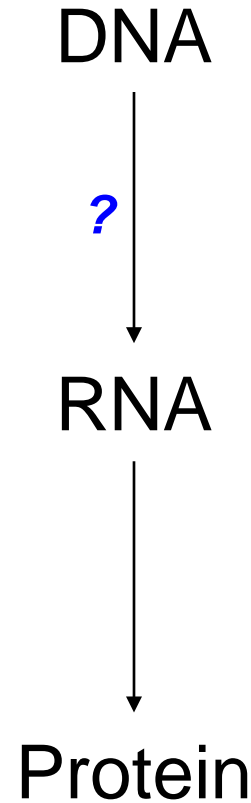
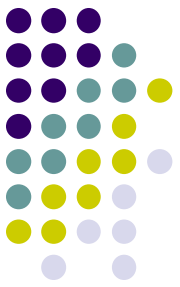
Protein

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It refers to the two steps involved in transforming genetic information into protein

Q

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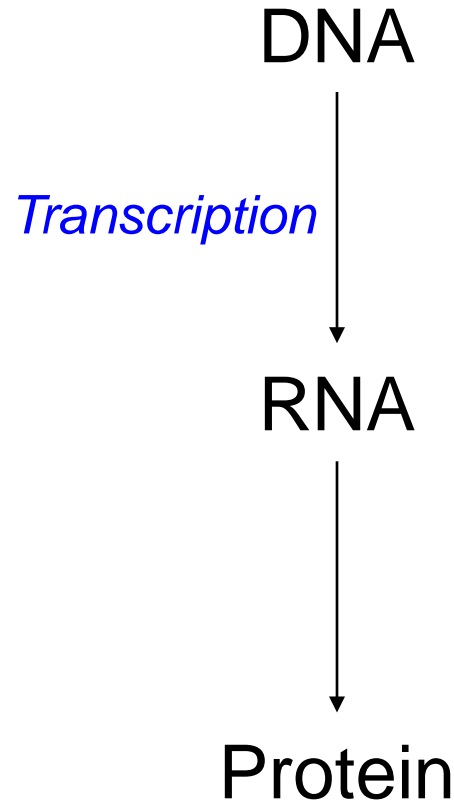
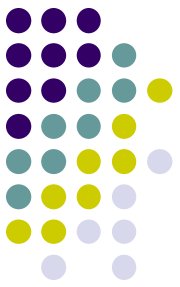
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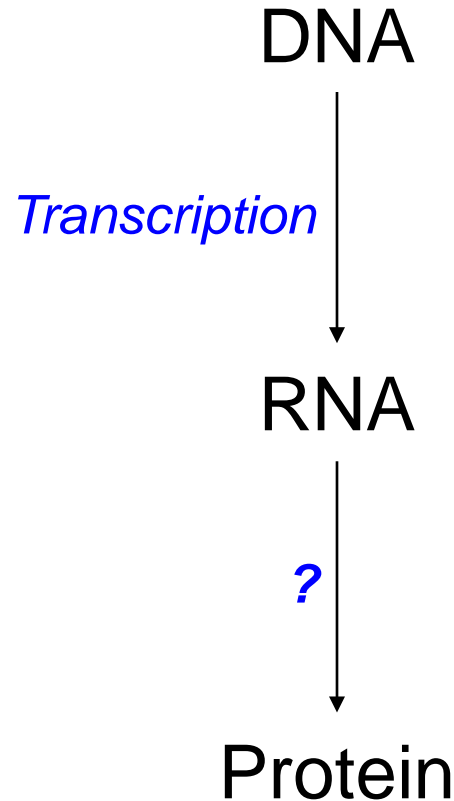
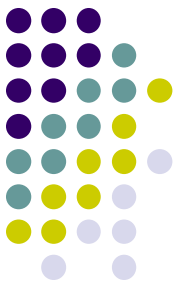
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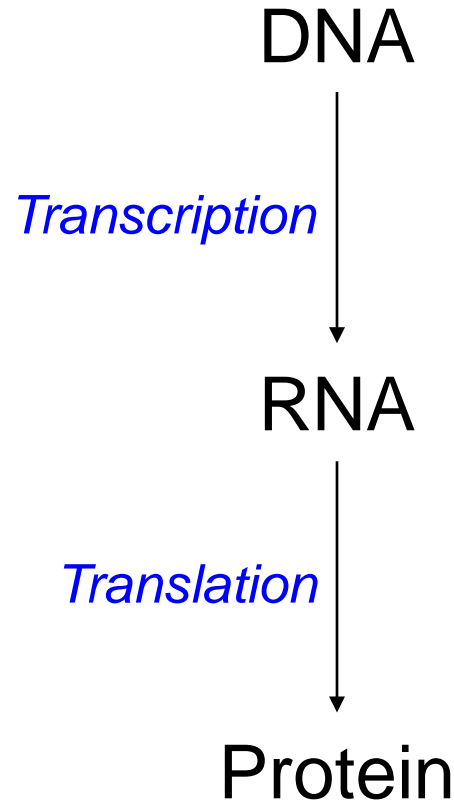
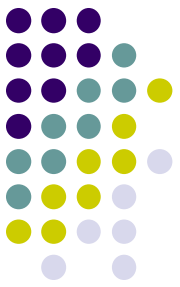
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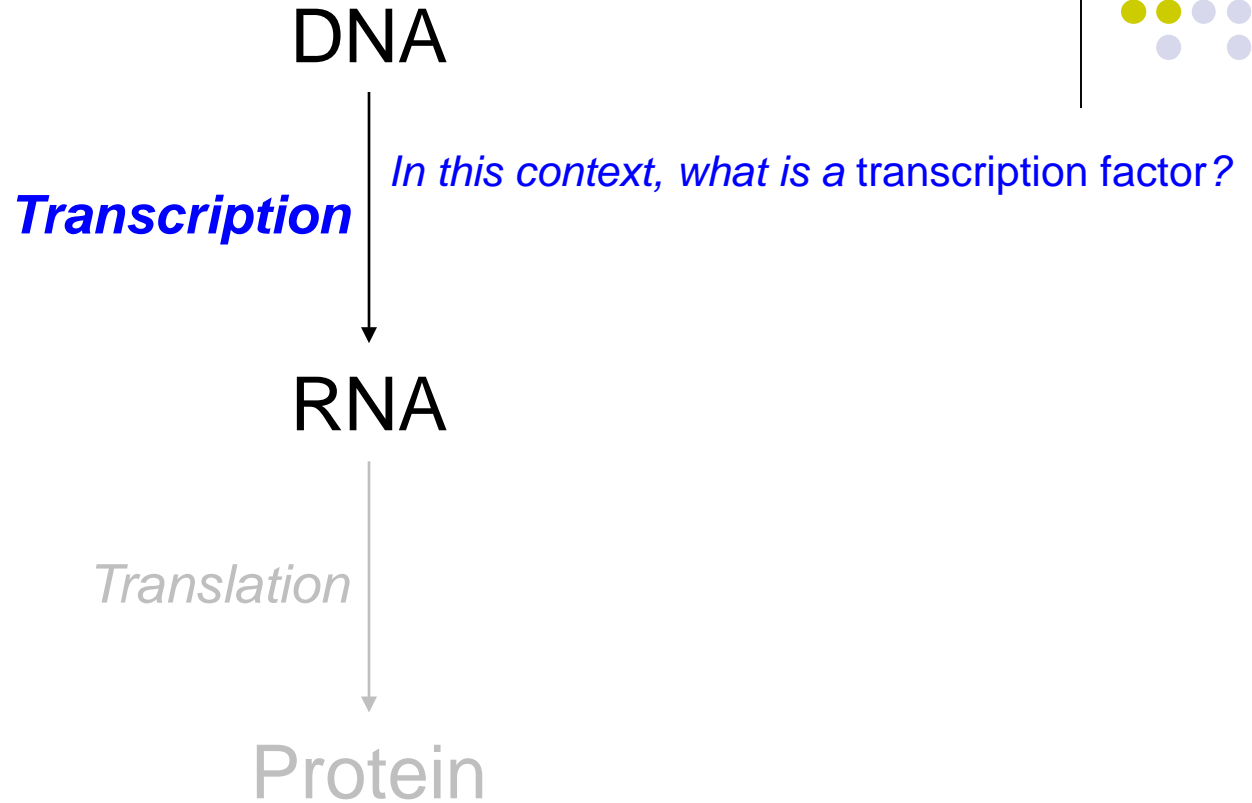
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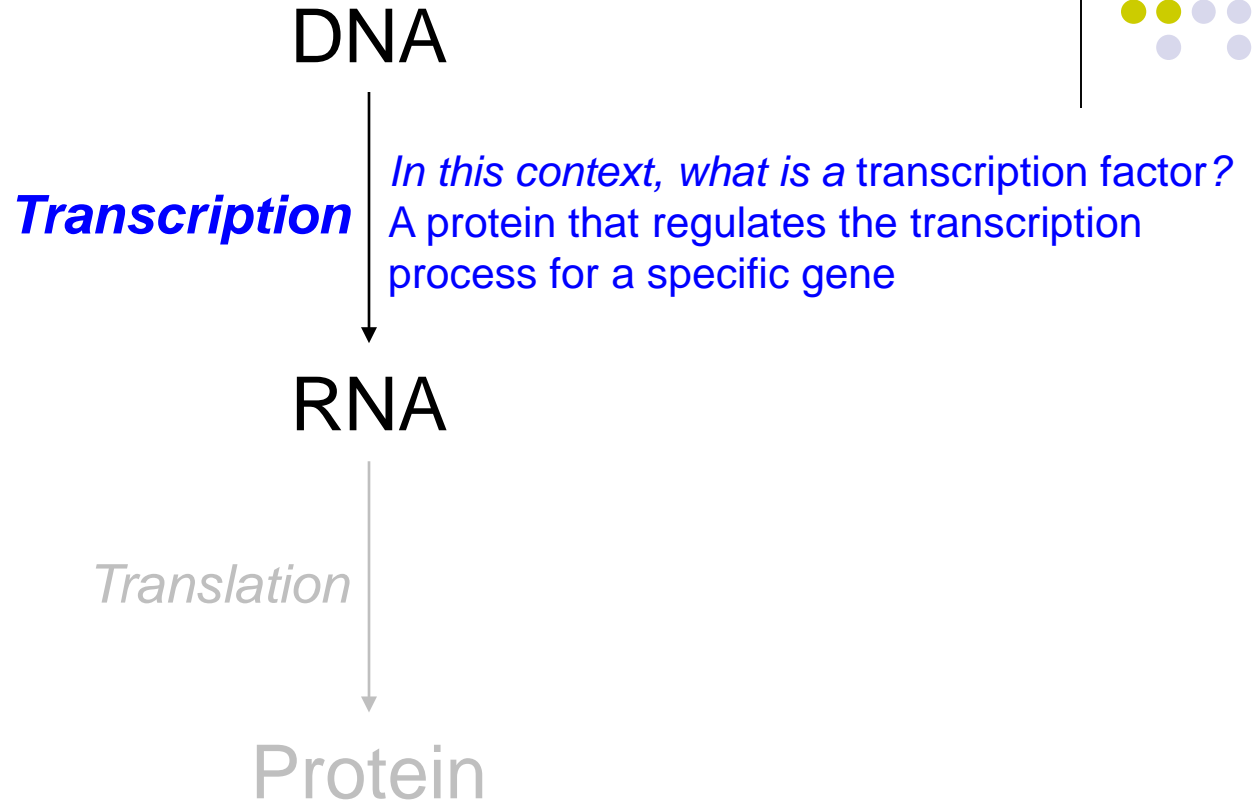
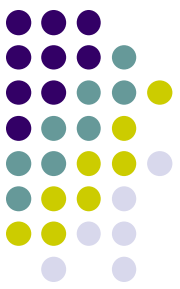


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*PAX Ophthalmicana*



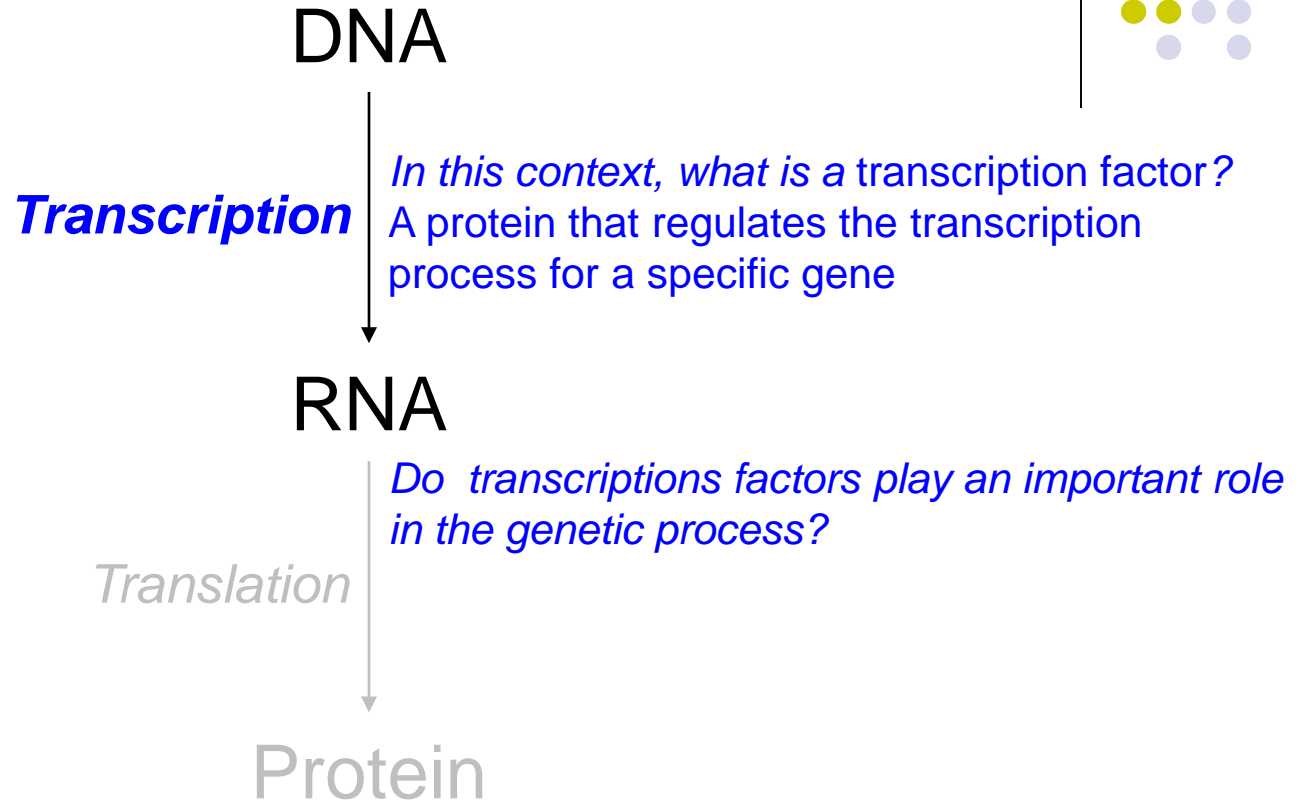
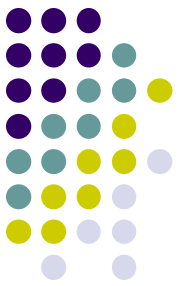
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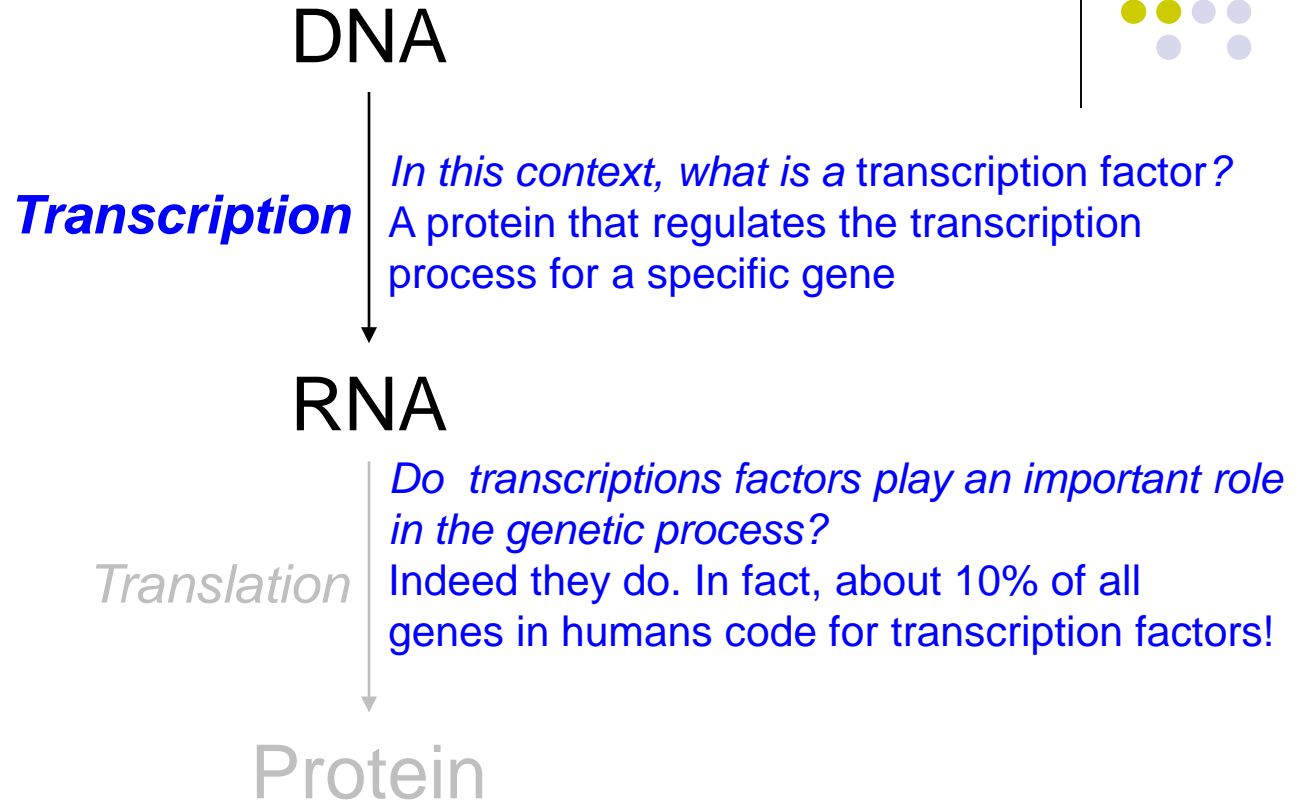


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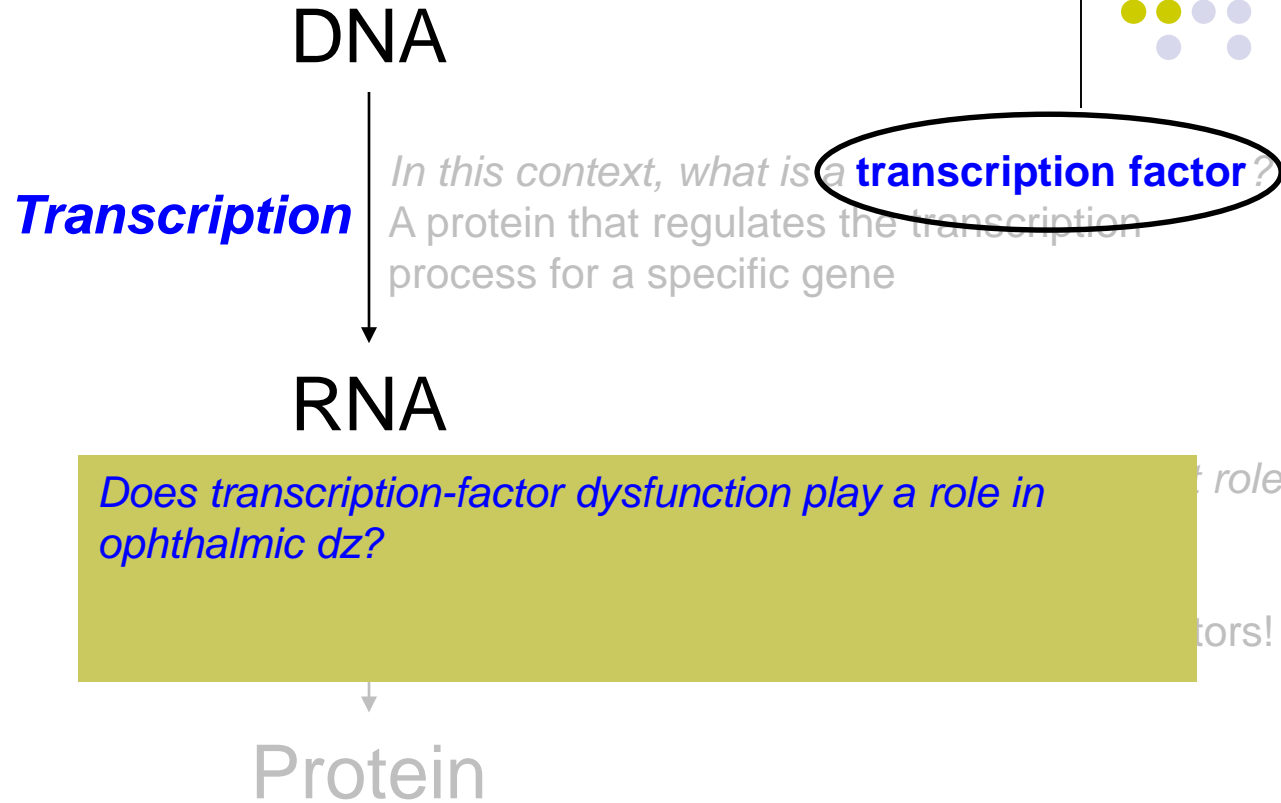
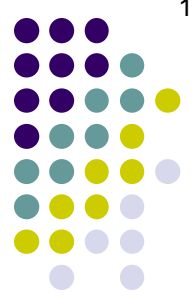
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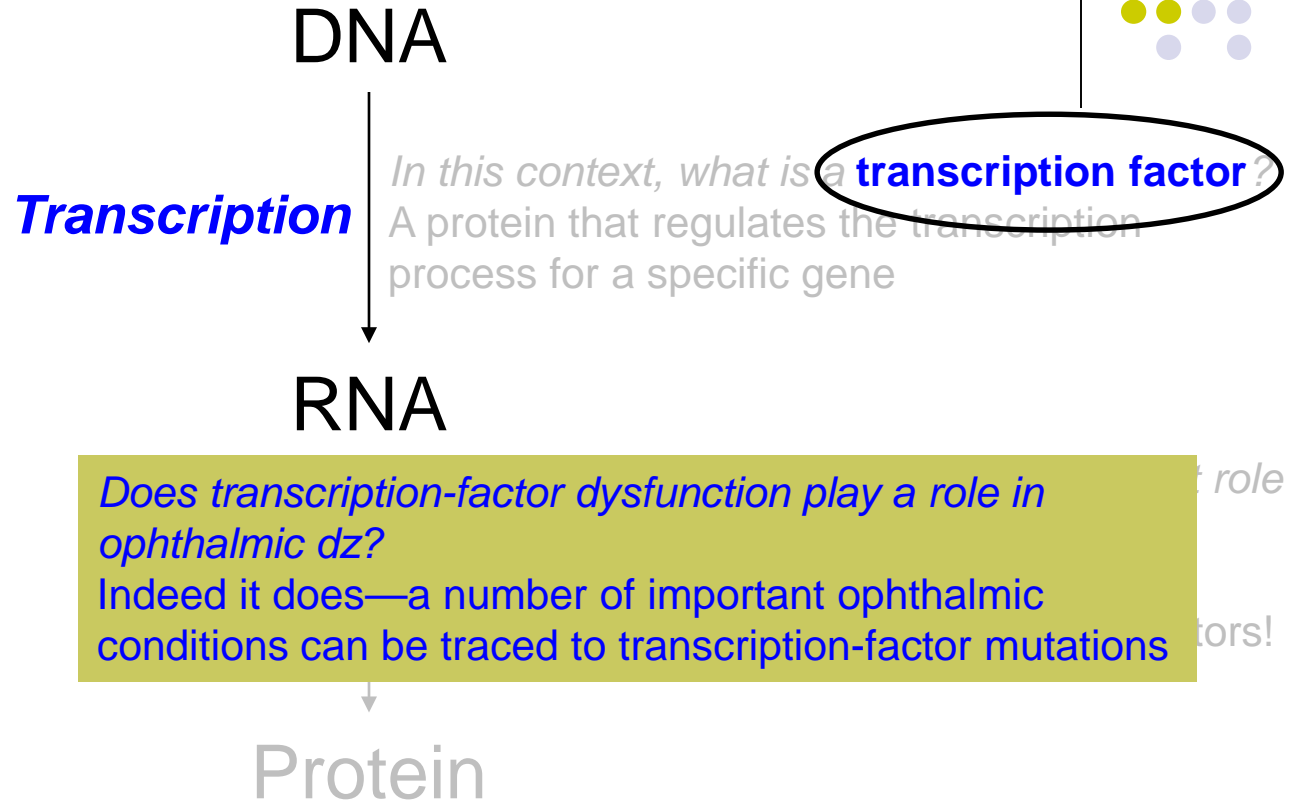
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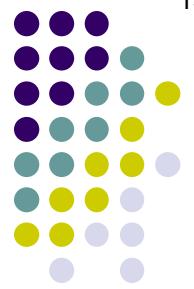
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Q

# PAX Ophthalmicana



DNA

**Transcription**

In this context, what is a **transcription factor**?  
A protein that regulates the transcription process for a specific gene

RNA

Does transcription-factor dysfunction play a role in ophthalmic dz?  
Indeed it does—a number of important ophthalmic conditions can be traced to transcription-factor mutations

Protein

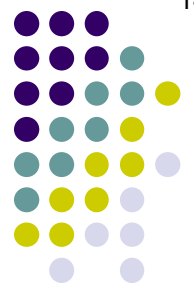
The Fundamentals book lists three transcription-factor (genes) that are especially important for the eye—what are they?

With respect to...  
It refers to the...  
--The first step...  
--the trans...

...into protein:  
...followed by

A

*PAX Ophthalmicana*



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With respect to transcription factors...  
It refers to the...  
--The first step...  
--the trans...  
The Fundamentals book lists three transcription-factor (genes) that are especially important for the eye—what are they?  
--PAX2  
--PAX3  
--PAX6

into protein:  
followed by

Q

## *PAX Ophthalmicana*



*In the present context, what is the origin of the word PAX? Where does it come from?*

*The Fundamentals book lists three transcription-factor (genes) that re especially important for the eye—what are they?*

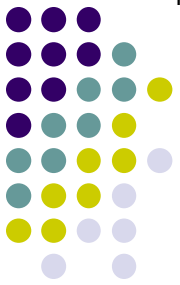
--PAX2

--PAX3

--PAX6

## A

# PAX Ophthalmicana



*In the present context, what is the origin of the word PAX? Where does it come from?*

It is a portmanteau of the term 'PAired (homeo)boX'

*The Fundamentals book refers to PAX genes both as 'paired homeobox' and 'paired box' genes*

*The Fundamentals book lists three transcription-factor (genes) that re especially important for the eye—what are they?*

--PAX2

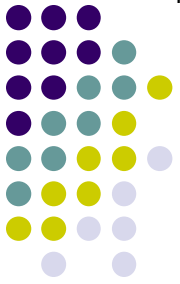
--PAX3

--PAX6



Q

## PAX Ophthalmicana



*In the present context, what is the origin of the word PAX? Where does it come from?  
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*Generally speaking, what are PAX genes involved in?*

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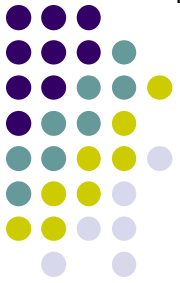
--PAX2

--PAX3

--PAX6

# A

## *PAX Ophthalmicana*



*In the present context, what is the origin of the word PAX? Where does it come from?*  
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Morphogenesis

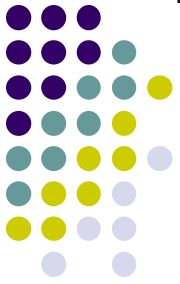
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--PAX2

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## Q

**PAX Ophthalmicana**

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*Of these three PAX genes, which is most important to the development of the eye?*

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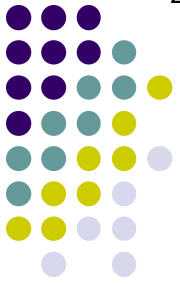
*--PAX2?*

*--PAX3?*

*--PAX6?*

# A

## *PAX Ophthalmicana*



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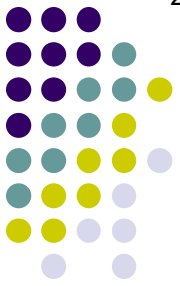
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--PAX2

--PAX3

--**PAX6!**

## *PAX Ophthalmicana*



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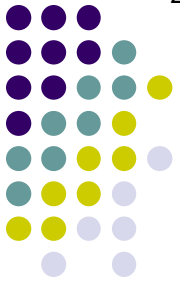
--PAX3

--**PAX6!**

*Next let's take a closer look at PAX6*

# Q

## *PAX Ophthalmicana*

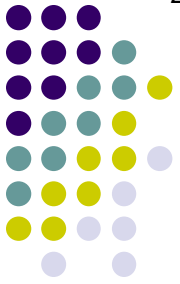


*There are four ocular abnormalities attributed to the PAX6 gene. What are they?*

*The mnemonic is...*

# A

## *PAX Ophthalmicana*



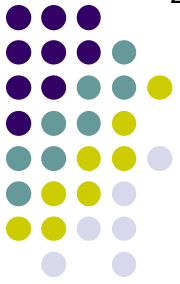
*There are four ocular abnormalities attributed to the PAX6 gene. What are they?*

*P*  
**A**  
**X**  
**6**

*The mnemonic is...PAX6*

Q

## *PAX Ophthalmicana*



*There are four ocular abnormalities attributed to the PAX6 gene. What are they?*

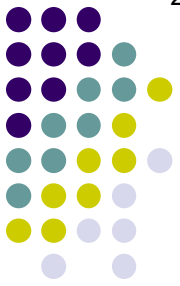
**P**  
**A**  
**X**  
**6**

← Start with the 'P' and work down

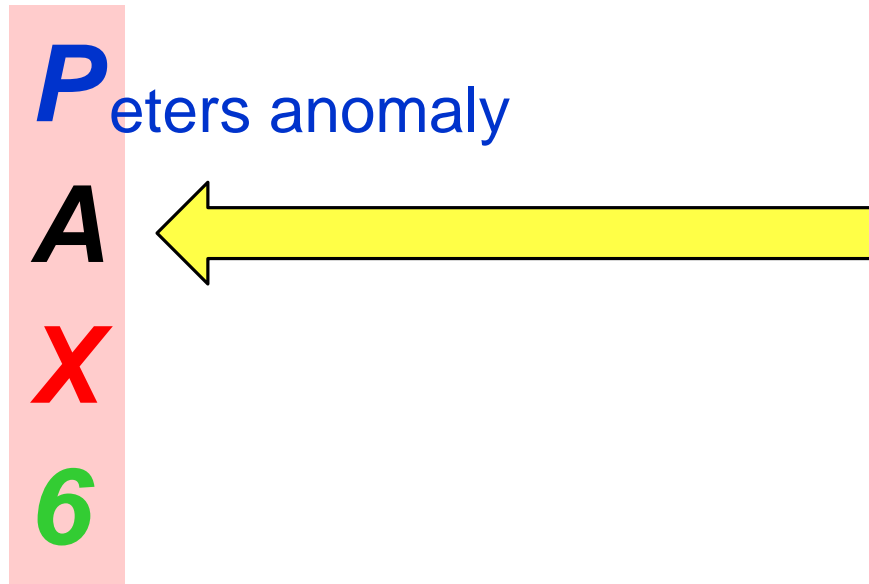


# Q/A

## PAX Ophthalmicana

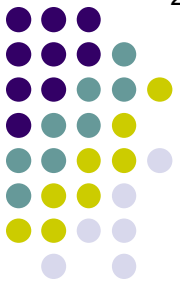


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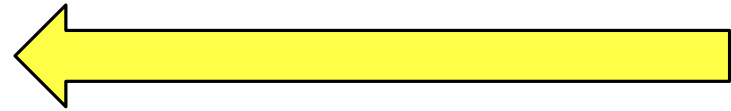
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**P**eters anomaly

**A**niridia

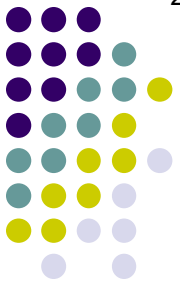
**X**

**6**

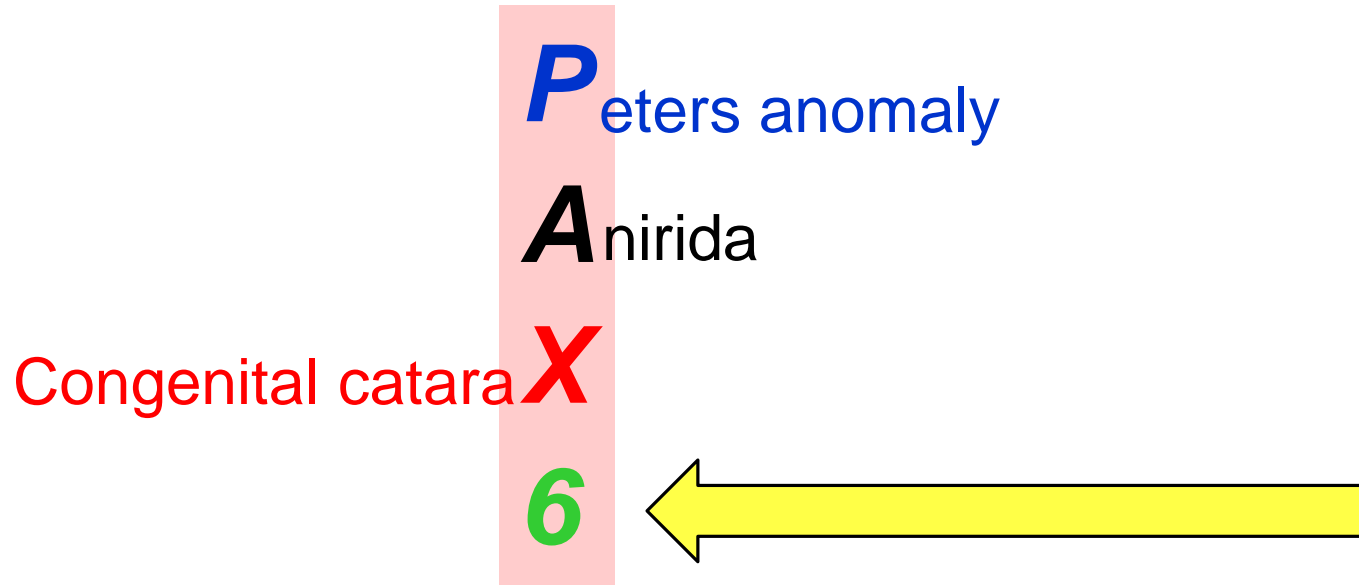


# Q/A

## PAX Ophthalmicana

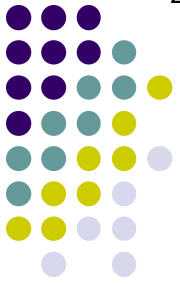


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A

# PAX Ophthalmicana



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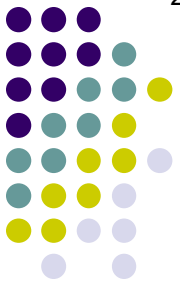
**P**eters anomaly  
**A**niridia  
 Congenital cataract **X**  
 foveal **6** hypoplasia

*If you use your imagination,  
the 6 looks like a lower-case h...*



Q

## PAX Ophthalmicana



*There are four ocular abnormalities attributed to the PAX6 gene. What are they?*

**P**eters anomaly

**A**niridia

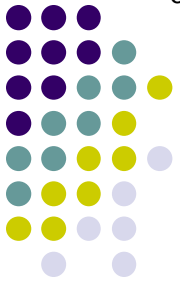
Congenital cataract **X**

foveal **6** hypoplasia

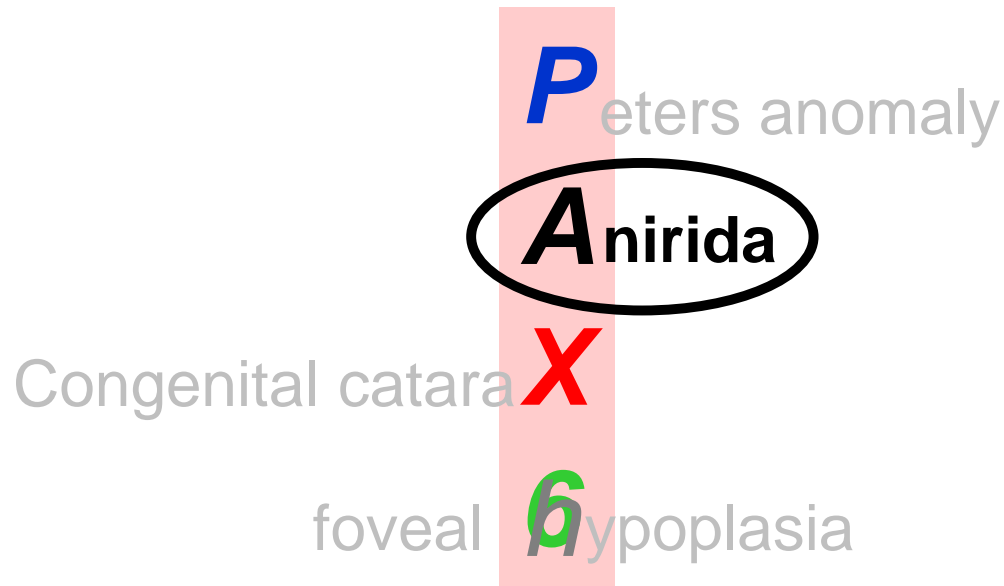
Endeavor to remember all of these. But if you have to pick just one to remember, make it...

# A

## PAX Ophthalmicana

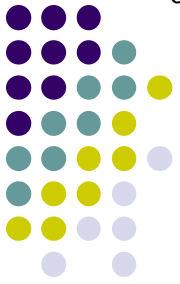


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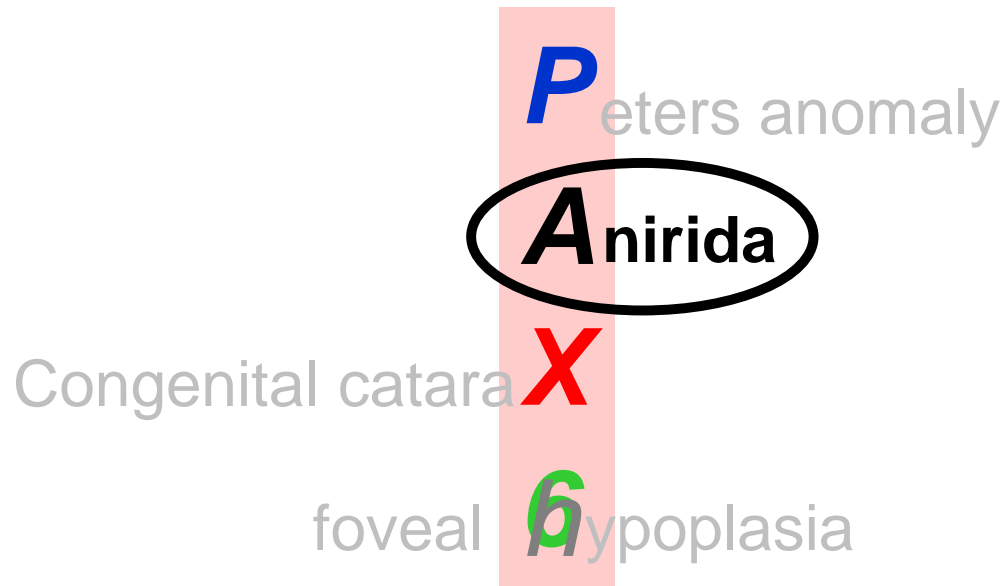


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## PAX Ophthalmicana

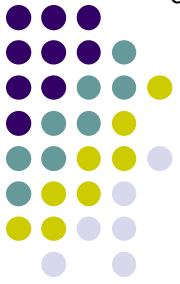


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Endeavor to remember all of these. But if you have to pick just one to remember, make it...**aniridia**. Almost all cases of aniridia are 2ndry to mutations involving PAX6.

## PAX Ophthalmicana



There are four ocular abnormalities attributed to the PAX6 gene. What are they?

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**A**nirida

Congenital cataract **X**

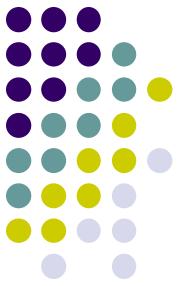
foveal **6** hypoplasia

One final takeaway point  
regarding aniridia...

Endeavor to remember all of these. But if you have to pick just one to remember, make it...**aniridia**. Almost all cases of aniridia are 2ndry to mutations involving PAX6.



## *PAX Ophthalmicana*



*Because all are tied to PAX6,  
you know foveal hypoplasia,  
ON hypoplasia and cataracts  
are associated with aniridia.*

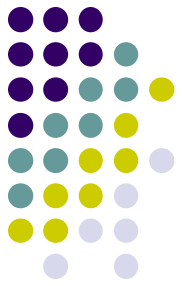
**foveal and optic nerve hypoplasia**

*One final takeaway point  
regarding aniridia...*

**cataracts**

Q

*PAX Ophthalmicana*



?

?

*...But you need to know the three **other** eye findings also associated with aniridia:*

*(Hints forthcoming)*

**foveal and optic nerve hypoplasia**

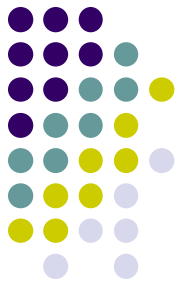
*One final takeaway point regarding aniridia...*

?

**cataracts**

Q

# PAX Ophthalmicana



An eye movement issue

A corneal issue

...But you need to know the three **other** eye findings also associated with aniridia:

(Hints forthcoming)

**foveal and optic nerve hypoplasia**

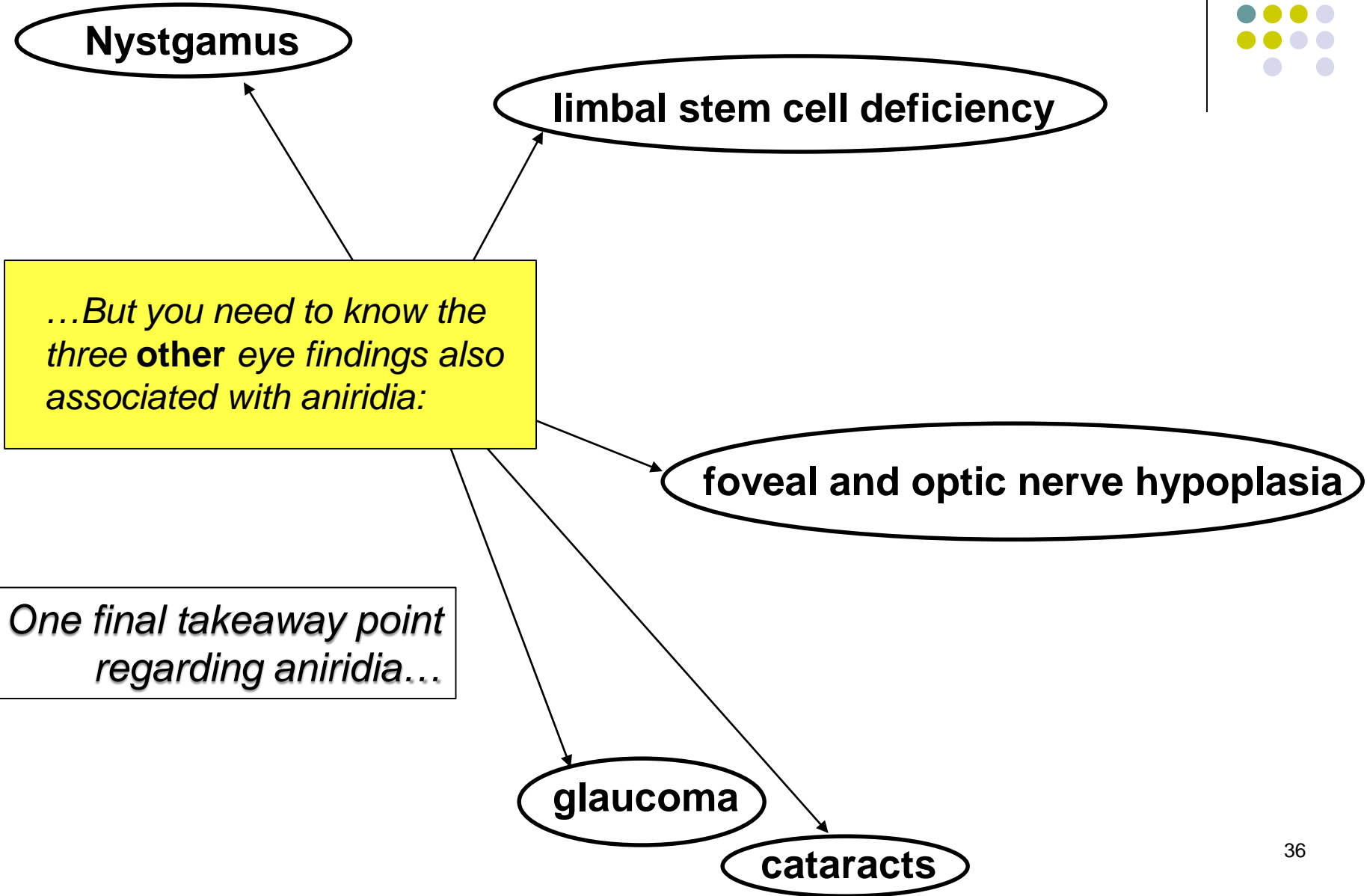
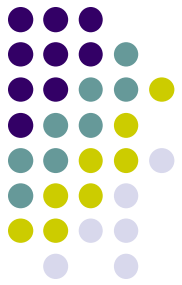
One final takeaway point regarding aniridia...

Angle-related condition

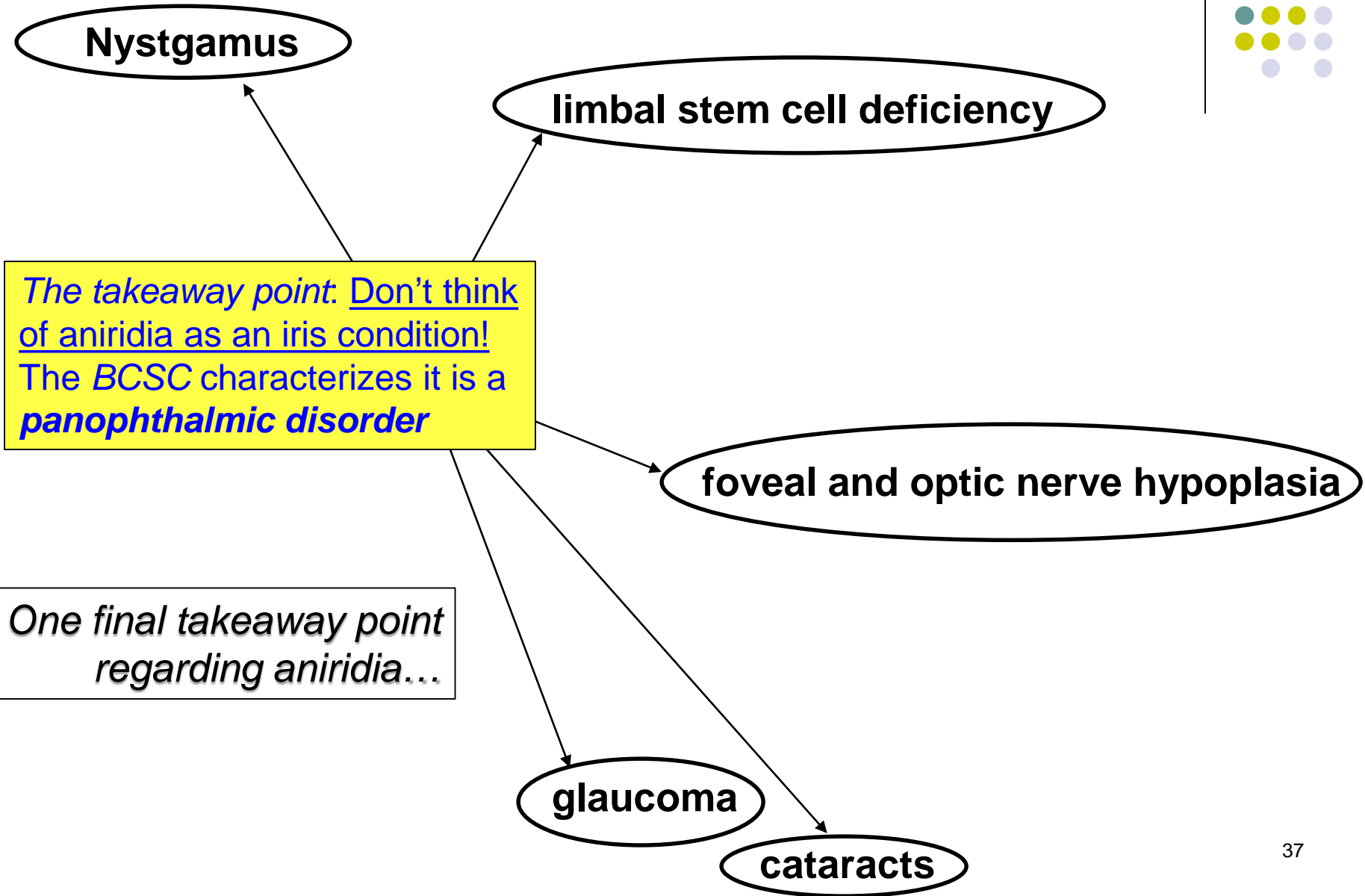
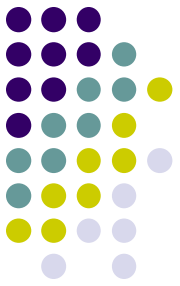
**cataracts**

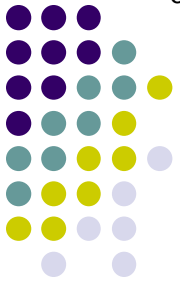
**A**

*PAX Ophthalmicana*



# PAX Ophthalmicana





## *PAX Ophthalmicana*

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*The Fundamentals book lists three transcription-factor (genes) that re especially important for the eye—what are they?*

--PAX2

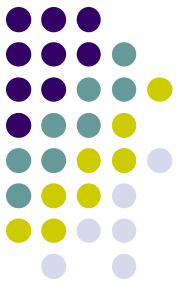
--**PAX3**

--PAX6

*Next, we'll do PAX3*

Q

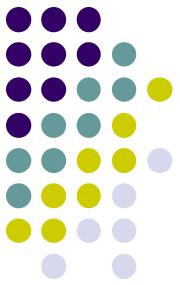
*PAX Ophthalmicana*



*With what eponymous syndrome is PAX3 associated?*

A

*PAX Ophthalmicana*



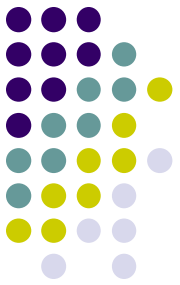
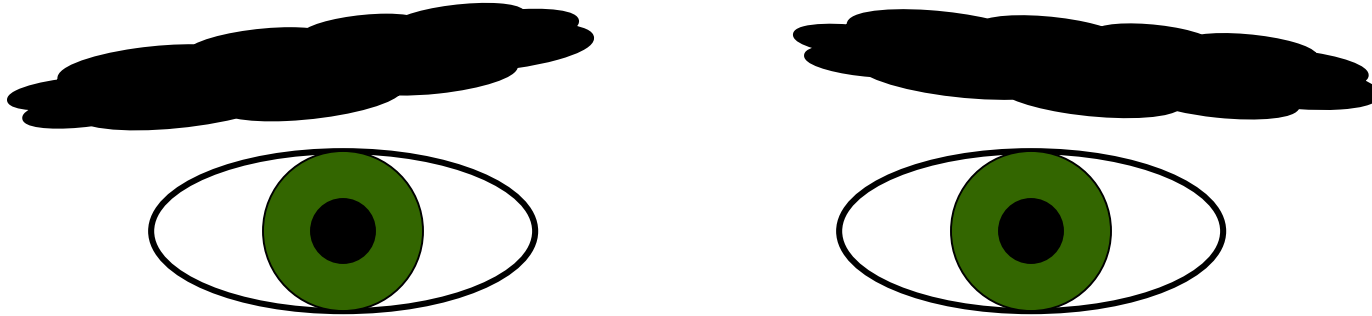
*With what eponymous syndrome is PAX3 associated?*

**Waardenburg Syndrome**



Q

*PAX Ophthalmicana*



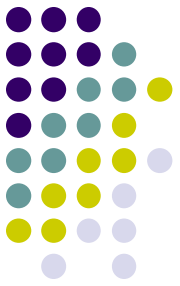
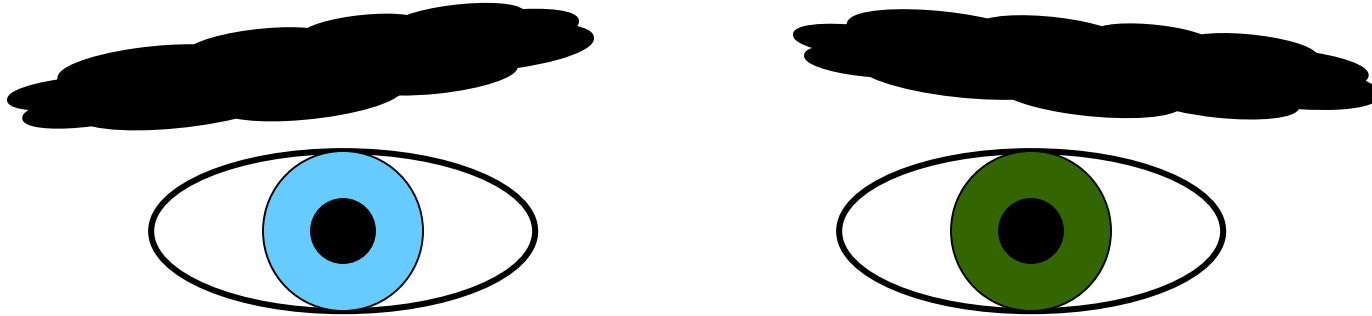
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*What 3 ophthalmic findings are classic for Waardenburg syndrome?*

- 
- 
-

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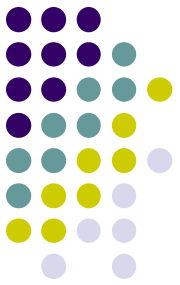


## Waardenburg Syndrome

*What 3 ophthalmic findings are classic for Waardenburg syndrome?*

- Heterochromia iridis**
- Synophrys
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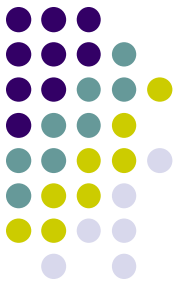
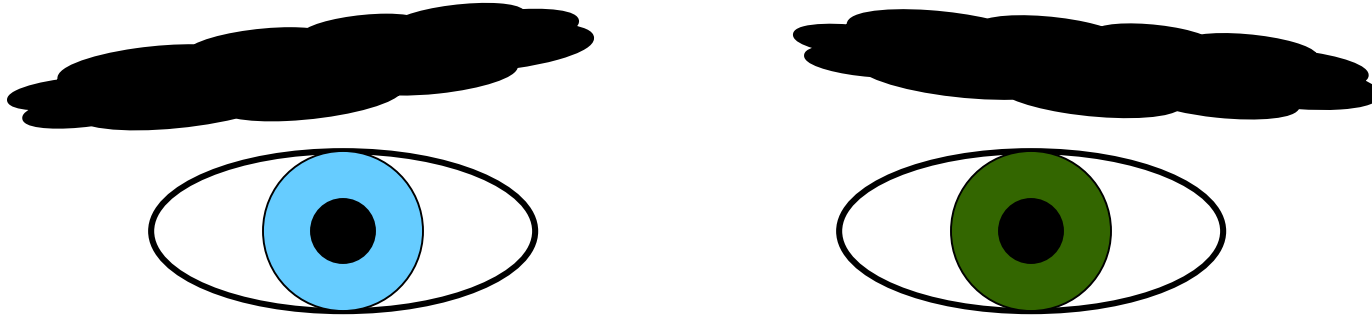
## Congenital lid abnormalities: Matching



Waardenburg syndrome: Heterochromia iridis,  
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(*What the heck is **synophrys**?*)

Q

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## Waardenburg Syndrome

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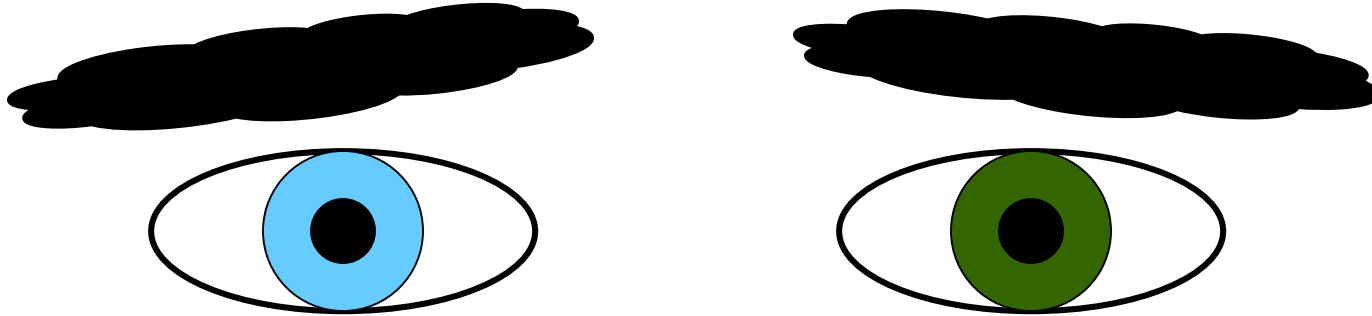
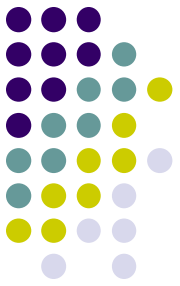
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Q/A

PAX Ophthalmicana



## Waardenburg Syndrome

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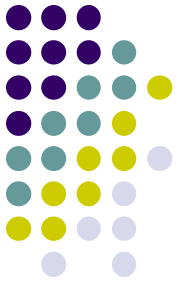
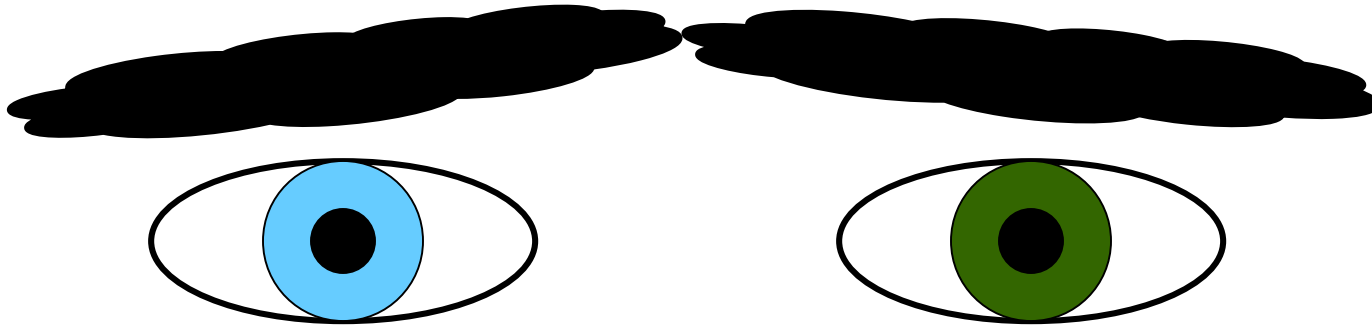
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The formal medical term for a

A

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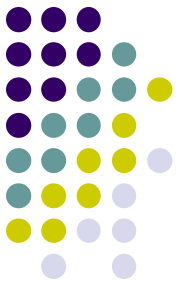
The formal medical term for a **unibrow**

*Congenital lid abnormalities: Matching*



Waardenburg syndrome: Synophrys

## Congenital lid abnormalities: Matching

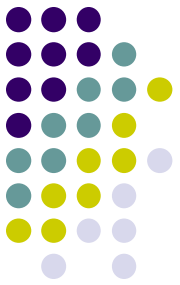
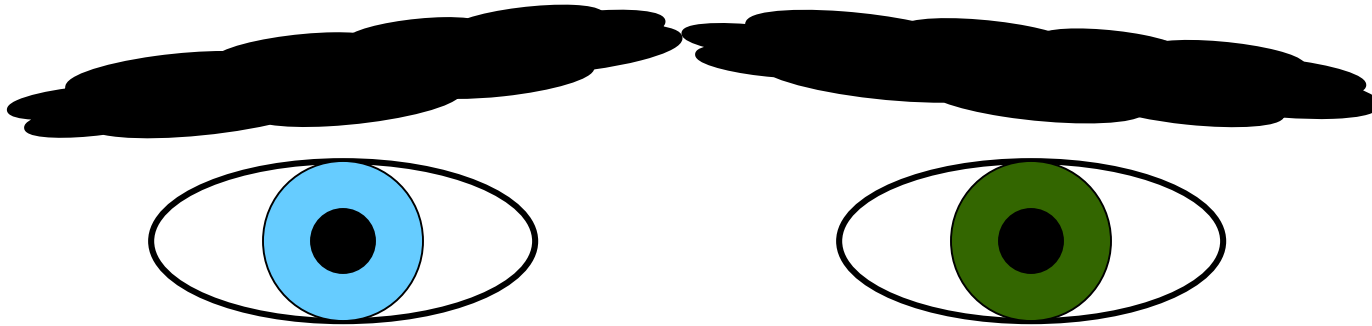


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Q

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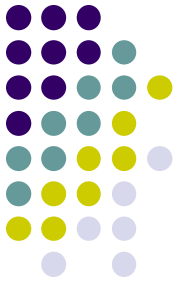
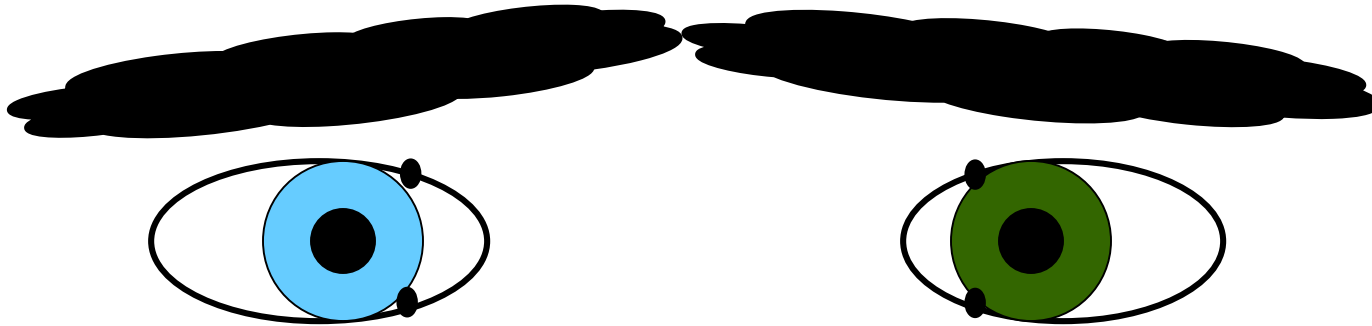
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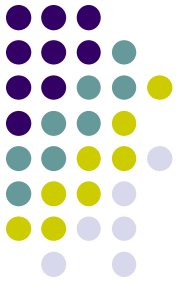
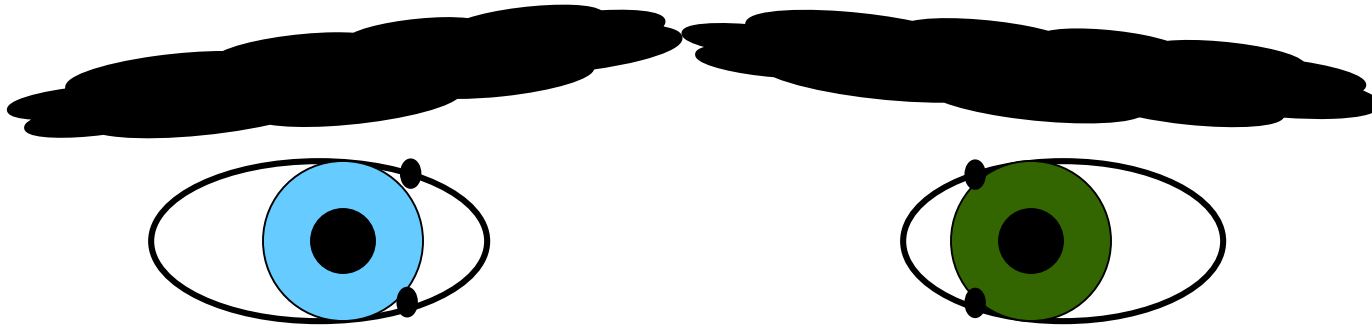
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Lateral displacement of the canthi (ie, telecanthus) **PLUS** laterally displaced lacrimal puncta

Q

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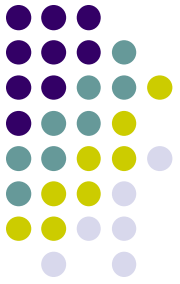
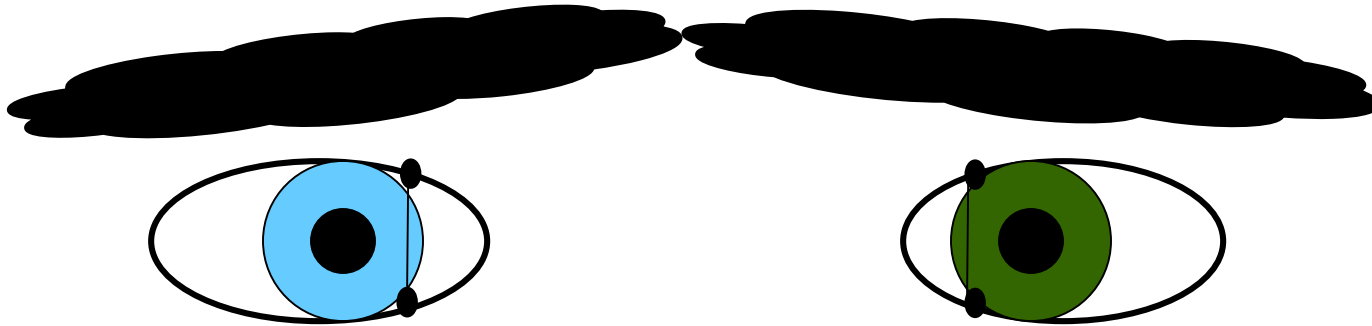
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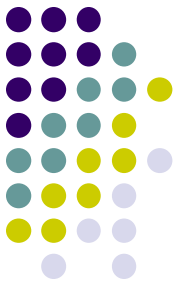
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Draw an imaginary vertical line from the upper to the lower puncta. If this line crosses the cornea, the puncta are displaced. (Next time you examine a pt at the slit-lamp, take note of whether such a line crosses their cornea [it won't].)

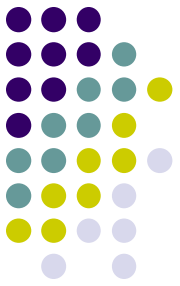
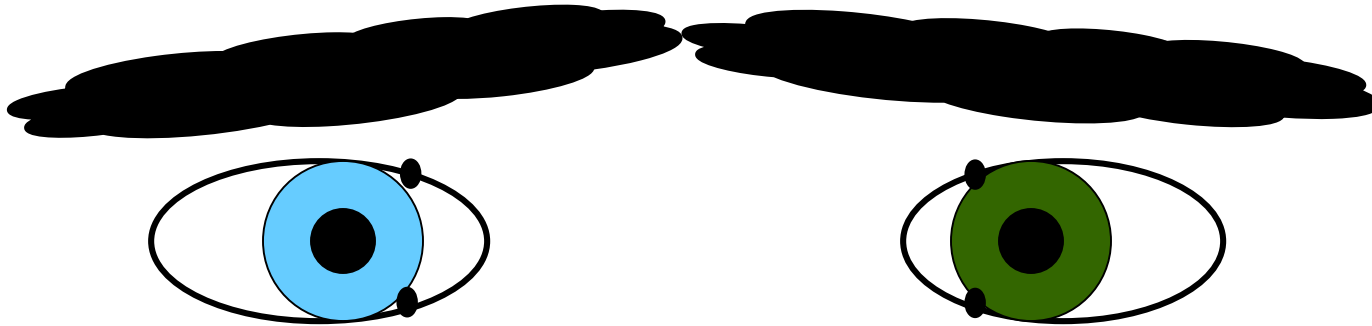
*Congenital lid abnormalities: Matching*



Dystopia canthorum. Note the telecanthus,  
and laterally displaced lacrimal puncta

Q

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## Waardenburg Syndrome

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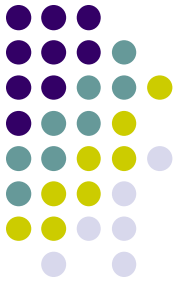
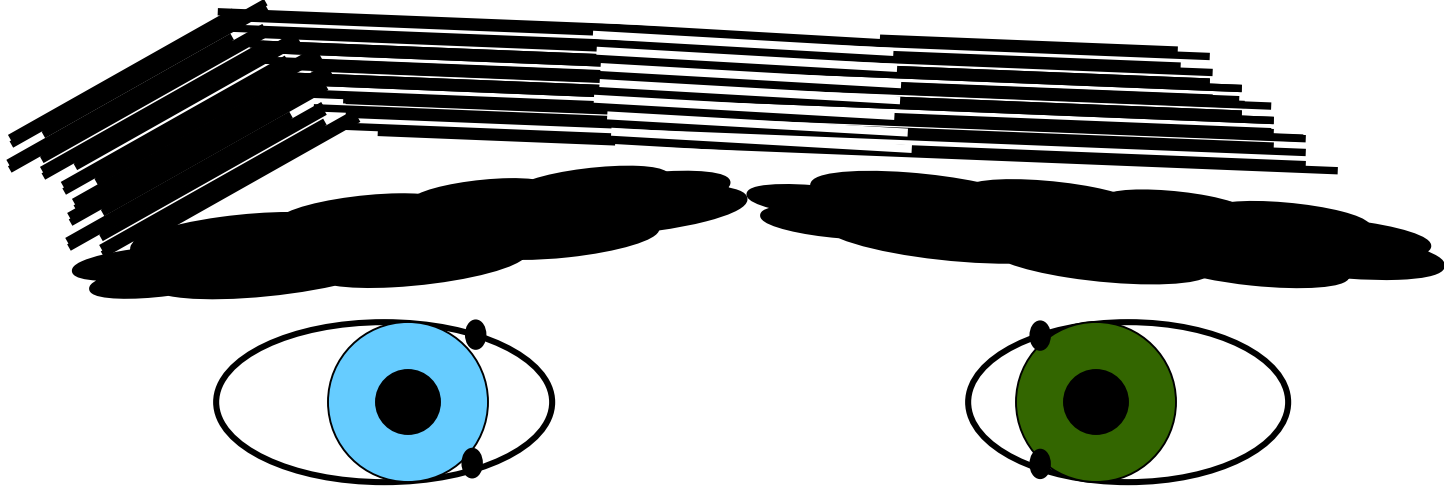
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A



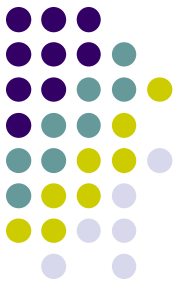
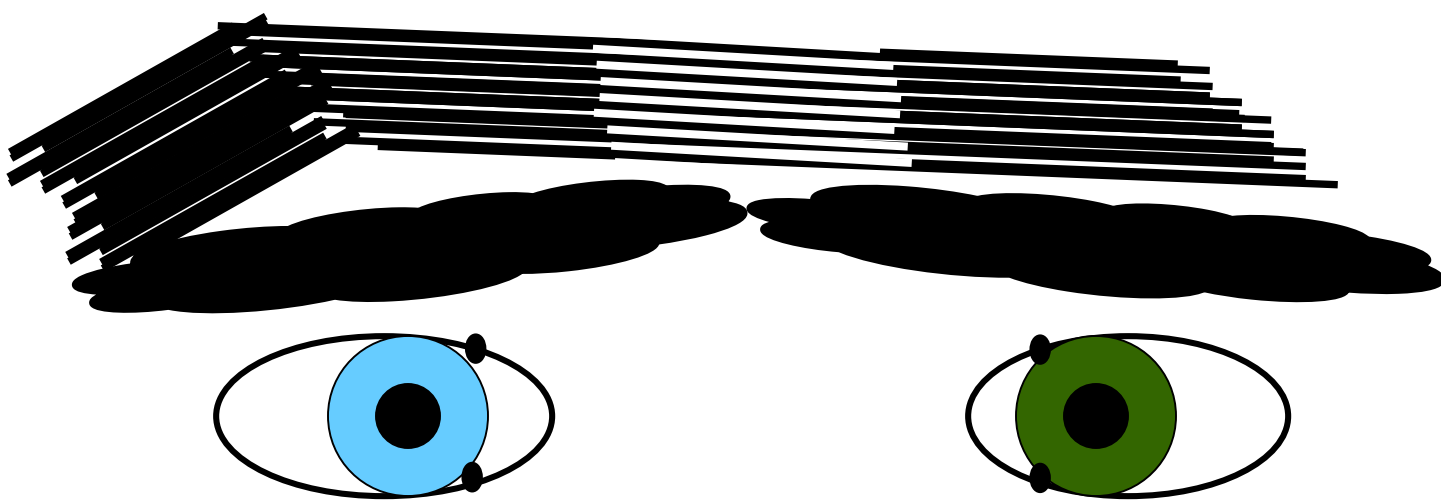
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The presence of a white forelock (ie, an isolated streak of **white hair** in the forehead region)



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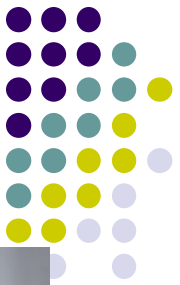
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**W**aardenburg  
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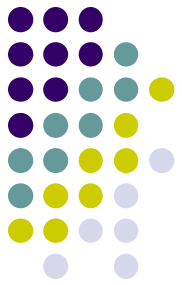


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Waardenburg syndrome: White forelock

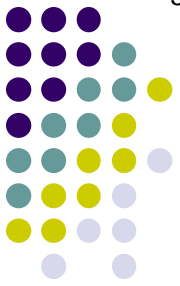
*Congenital lid abnormalities: Matching*



Note that Waardenburg syndrome has forms that do not involve heterochromia

## Q

## PAX Ophthalmicana



*In the present context, what is the origin of the word PAX? Where does it come from?*  
It is a portmanteau of the term 'PAired (homeo)boX'

*Generally speaking, what are PAX genes involved in?*  
Morphogenesis

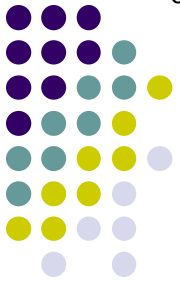
*Of these three PAX genes, which is most important to the development of the eye?*  
**PAX6**. The *Fundamentals* book refers to it as “the master switch for eye development.” The *Peds* book says, “The *PAX6* gene is the master control gene for eye morphogenesis.”

*The Fundamentals book lists three transcription-factor (genes) that re especially important for the eye, what are they?*

- PAX2
- PAX3
- PAX6

Last and most definitely least...PAX2 mutations present with [redacted] of the optic nerve, and [non-eye] hypoplasia

## A

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