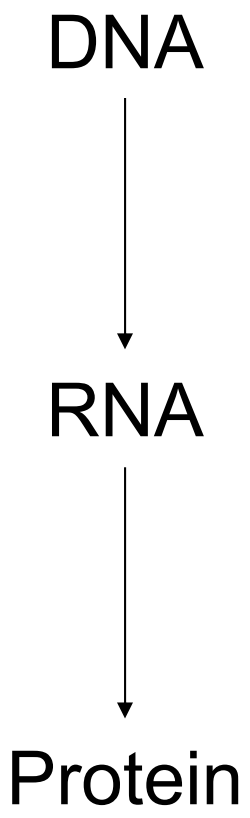
Q

PAX Ophthalmicana

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

A

PAX Ophthalmicana



With respect to genetics, to what does the term Central Dogma *refer?*
It refers to the two steps involved in transforming genetic information into protein

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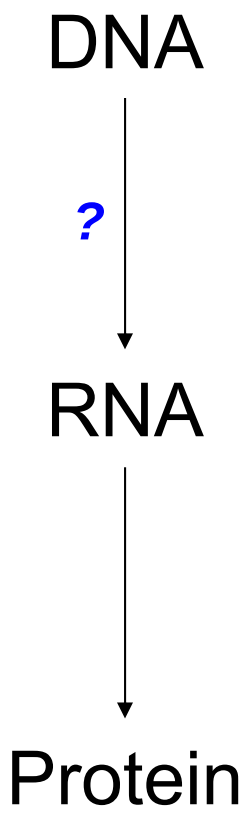

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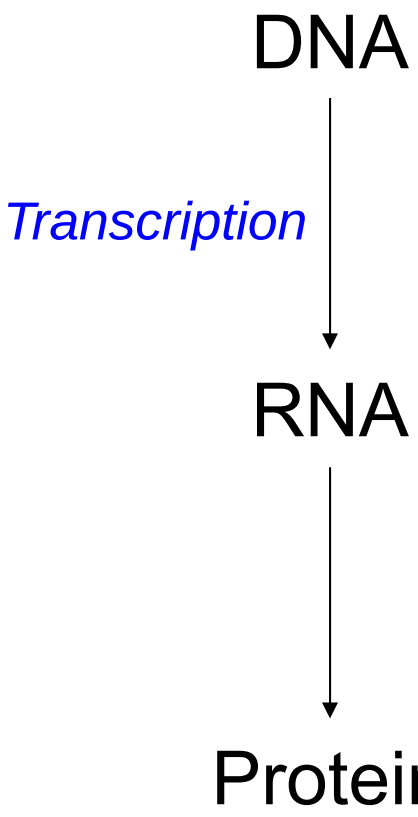

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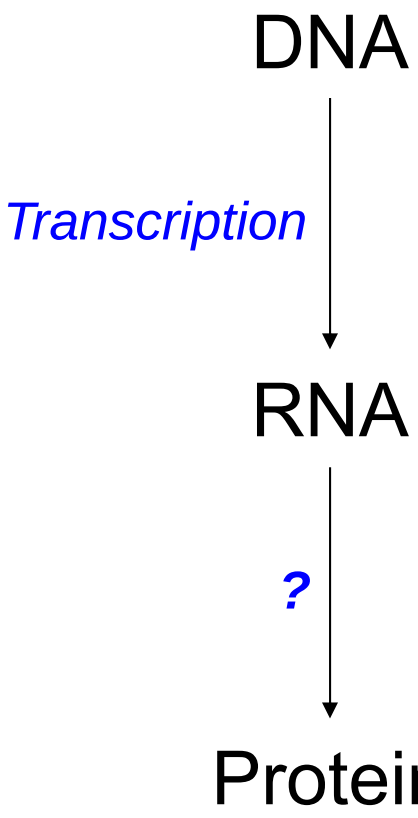

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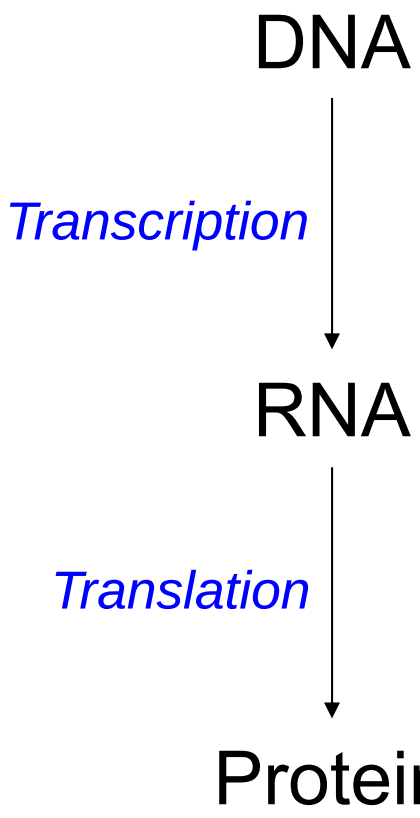

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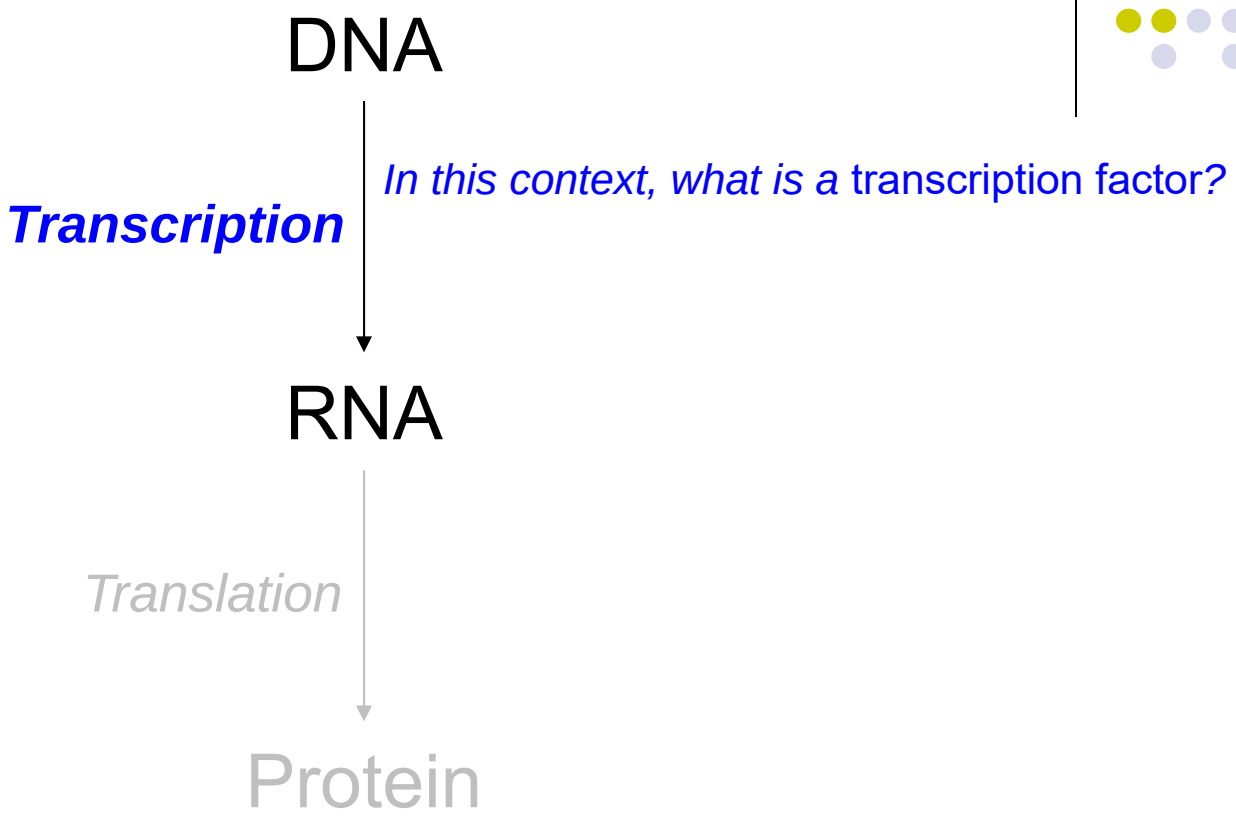

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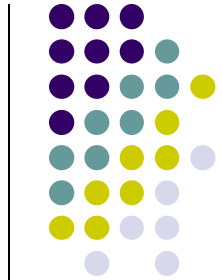

DNA

Transcription

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

RNA

Translation

Protein

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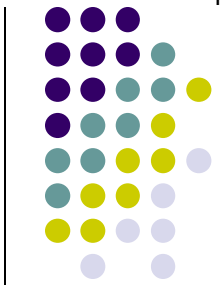

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With respec

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--The first st owed by

--the **transl**

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--

--

--

A

PAX Ophthalmicana

DNA

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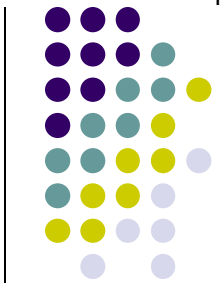
--PAX3
--PAX6

nto protein:
owed by

Q

PAX Ophthalmicana



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--**PAX2**
--**PAX3**
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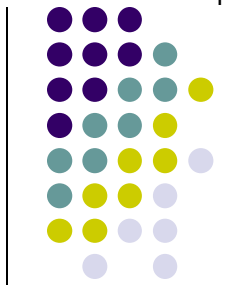
A

PAX Ophthalmicana



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It is a portmanteau of the term '**PA**ired (homeo)bo**X**'

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

The Fundamentals *book lists three transcription-factor genes that are especially important for the eye—what are they?*
--**PAX2**
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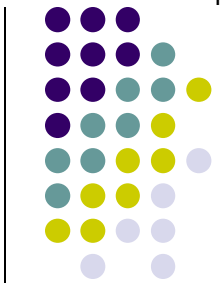
--**PAX6**

Q

PAX Ophthalmicana



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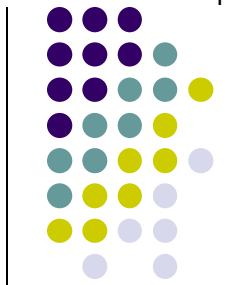
PAX Ophthalmicana



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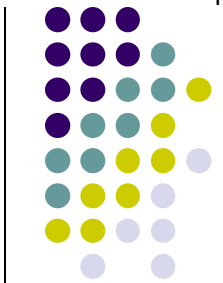

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--***PAX2?***
--***PAX3?***
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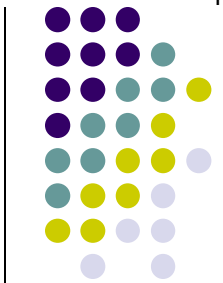
A

PAX Ophthalmicana



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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."

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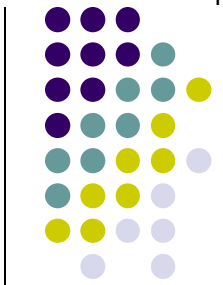
--PAX2
--PAX3
--**PAX6!**

PAX Ophthalmicana



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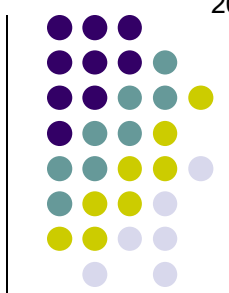
Next let's take a closer look at PAX6

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

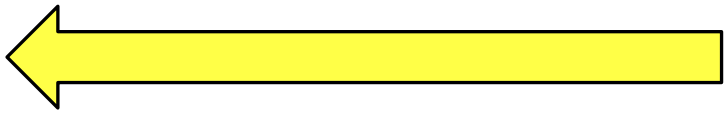
Start with the 'P' and work down

Q/A

PAX Ophthalmicana

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

P eters anomaly

A 

X

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Q/A

PAX Ophthalmicana

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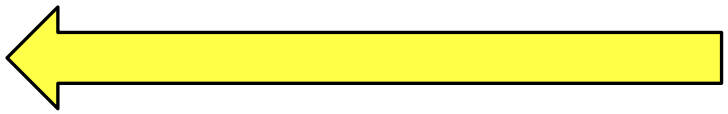

Q/A

PAX Ophthalmicana

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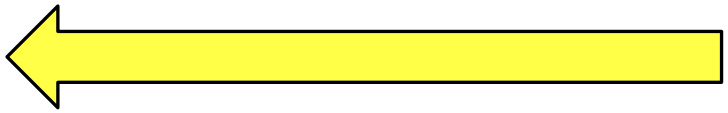
Congenital catara***X***

6



A

PAX Ophthalmicana

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

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Congenital catara***X***

Fovea and optic nerve ***6*** ypoplasia

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

Q

PAX Ophthalmicana

In three words, what sort of condition is Peters anomaly?

An three words

P eters anomaly

A nirida

Congenital catara *X*

Fovea and optic nerve *6* ypoplasia

A

PAX Ophthalmicana

In three words, what sort of condition is Peters anomaly?
An anterior segment dysgenesis

Peters anomaly

Anirida

Congenital catara**X**

Fovea and optic nerve **6**ypoplasia

Q

Anterior segment dysgenesis

? **?**

In thr
An a

Peters anomaly

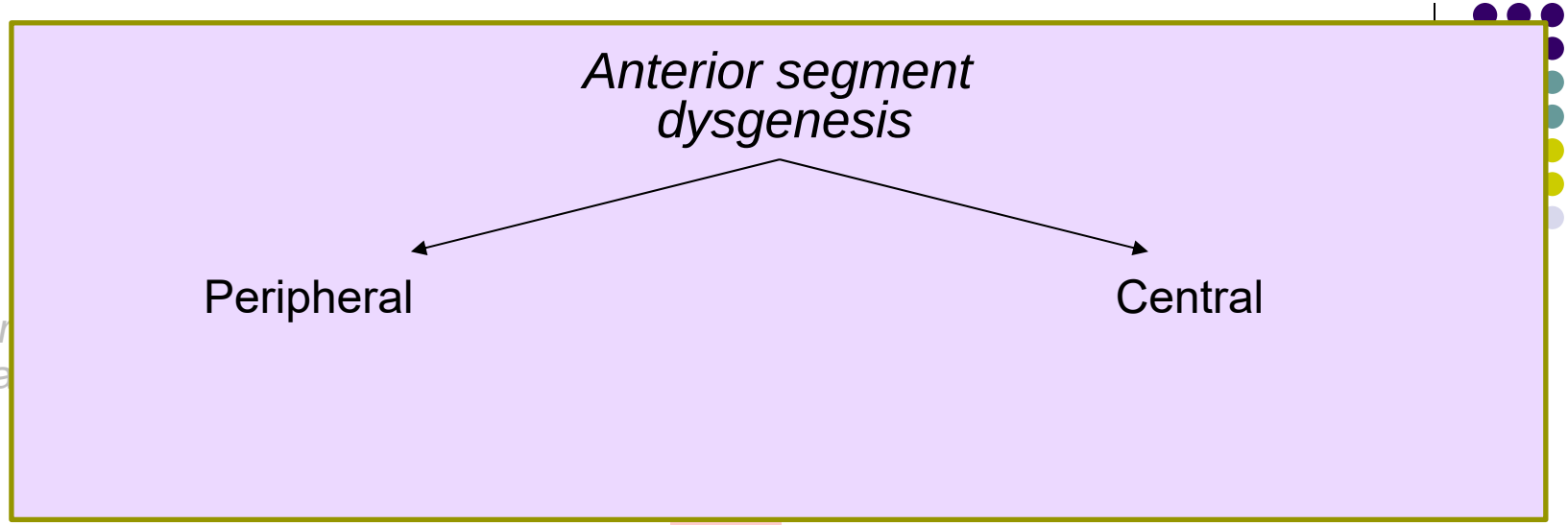
The Q

The BCSC *divides anterior segment dysgeneses into two broad categories based on a fundamental anatomic distinction. What is it?*

Fo

A

In thr
An a



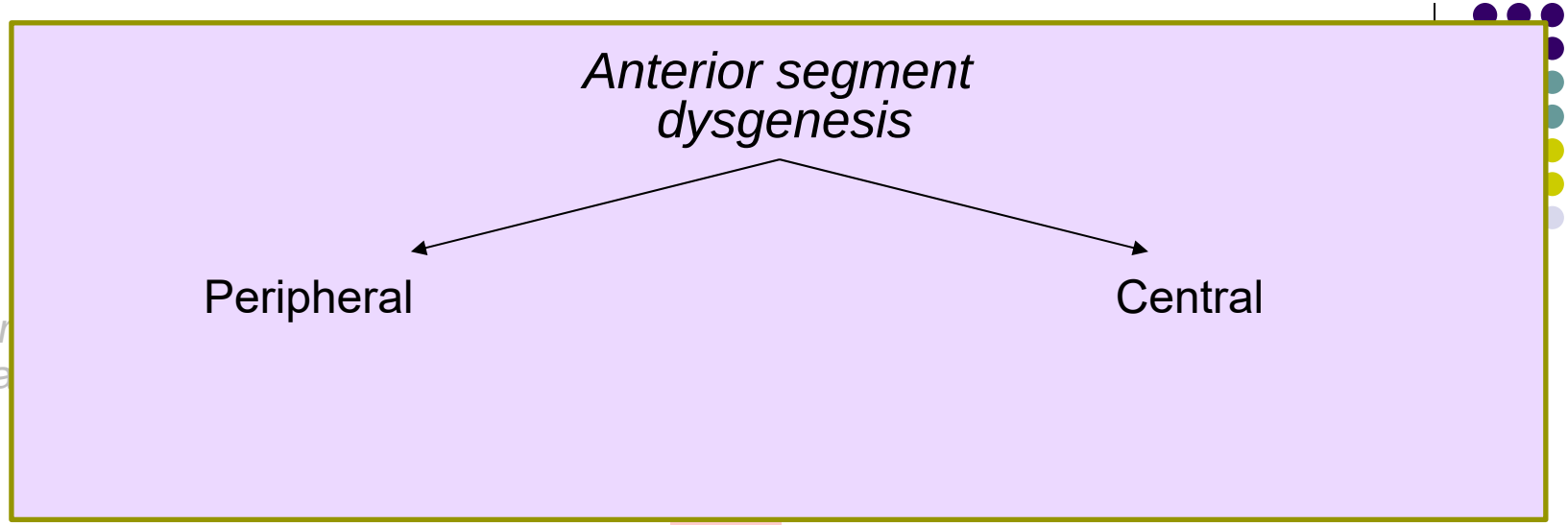
***P*eters anomaly**

The Q

The BCSC *divides anterior segment dysgeneses into two broad categories based on a fundamental anatomic distinction. What is it?*
It's whether the dysgenesis involves the *central* vs *peripheral* anterior segment

Fo

Q



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Is Peters a peripheral, or central dysgenesis?

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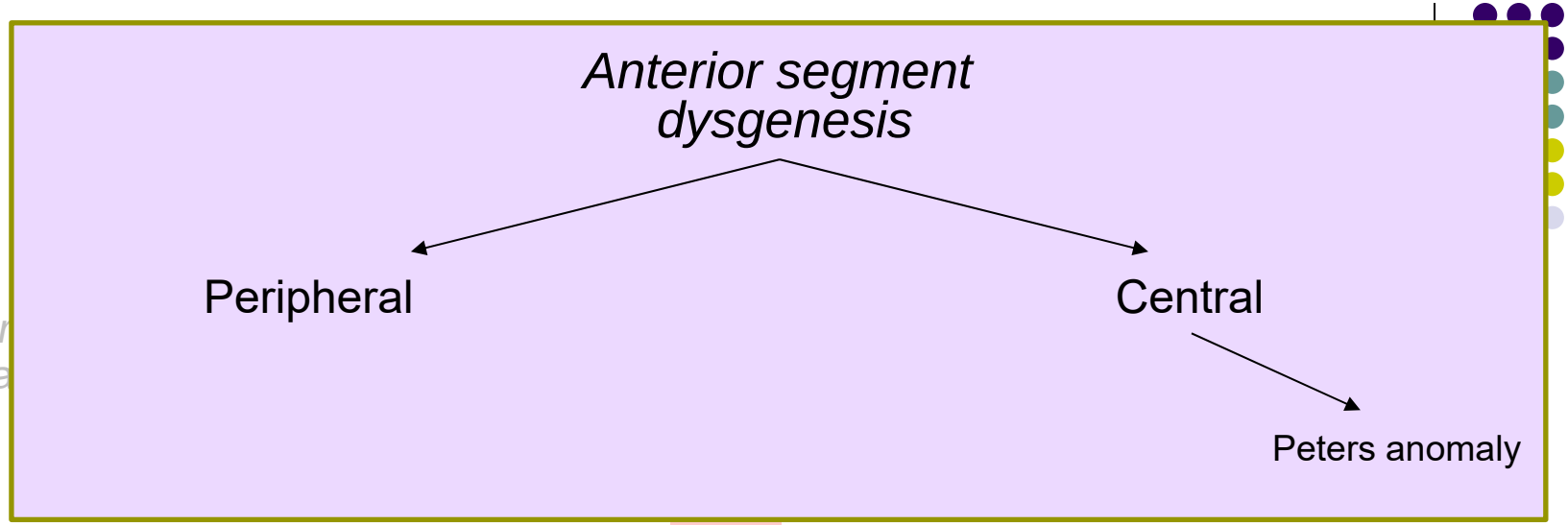

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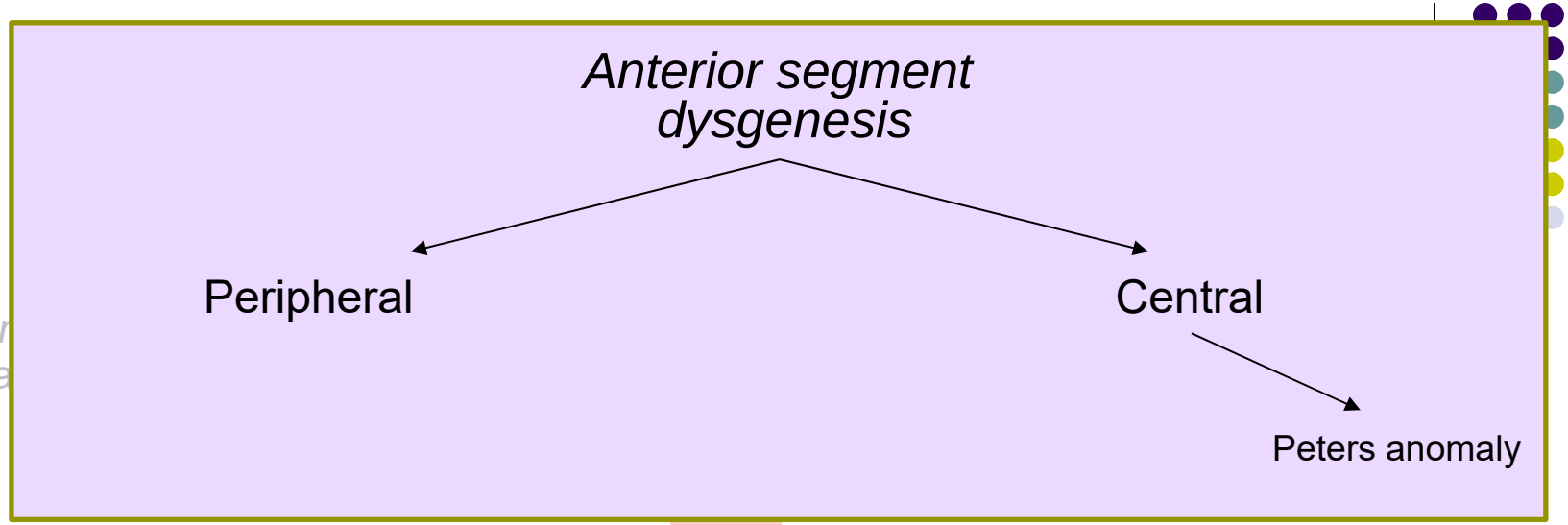
Central

Fo

Q



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How does Peters anomaly present?

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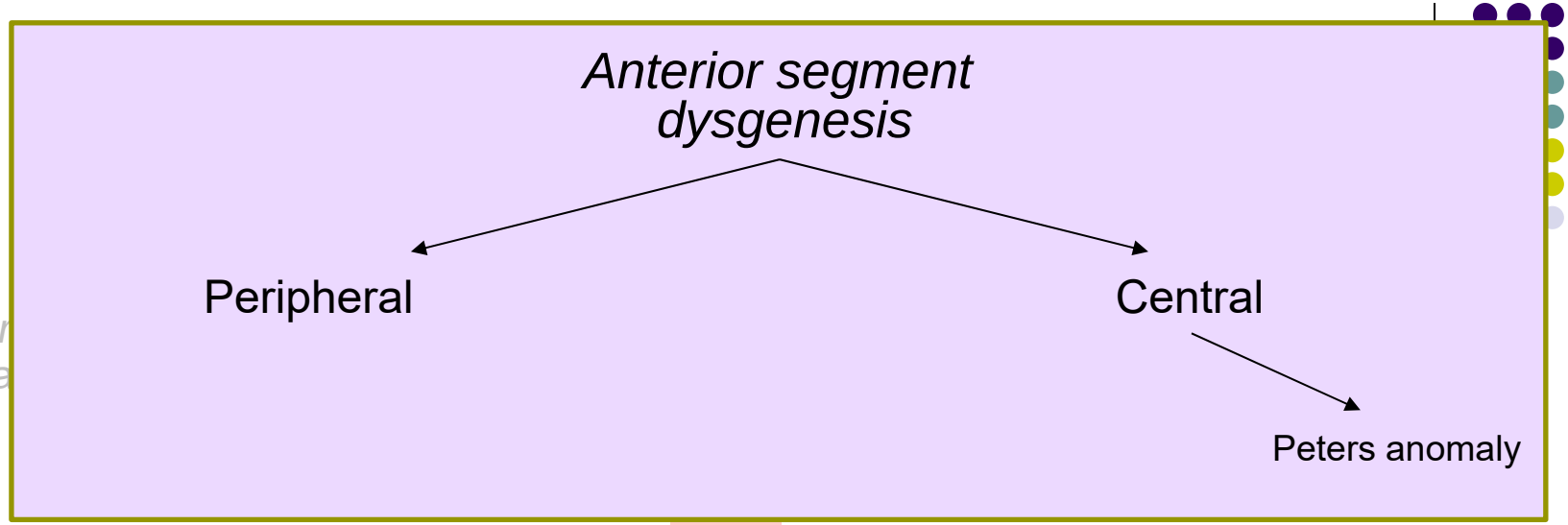

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How does Peters anomaly present?
As a corneal opacity at birth (it's in the STUMPED mnemonic).

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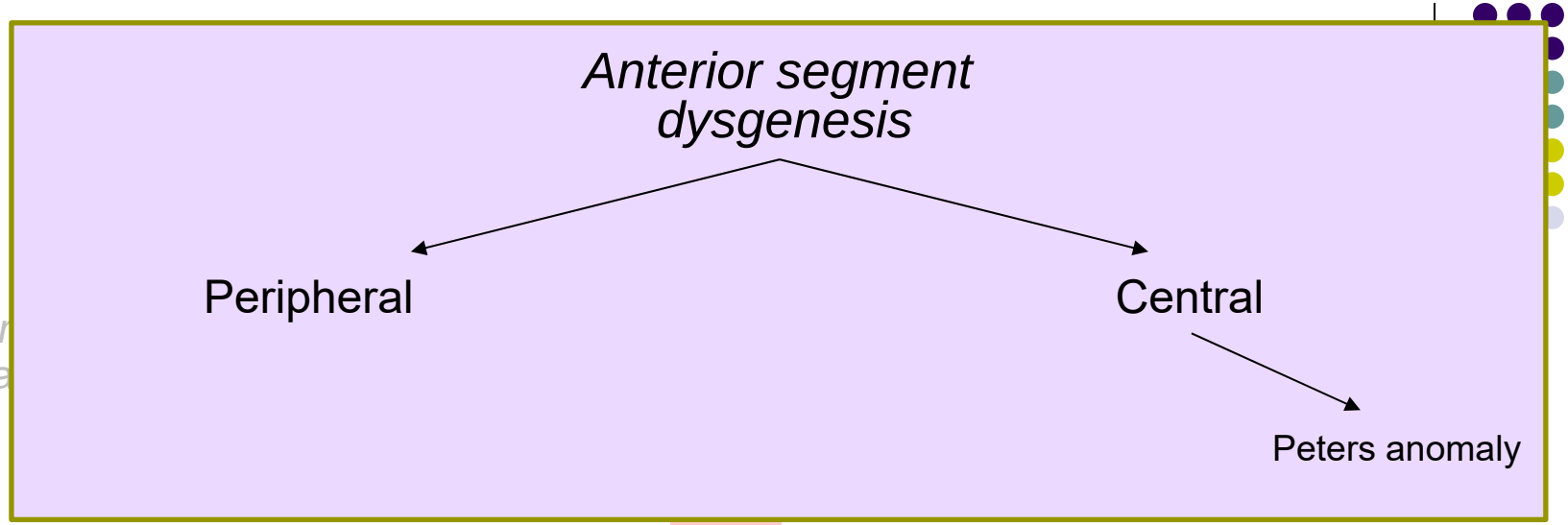

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Fo

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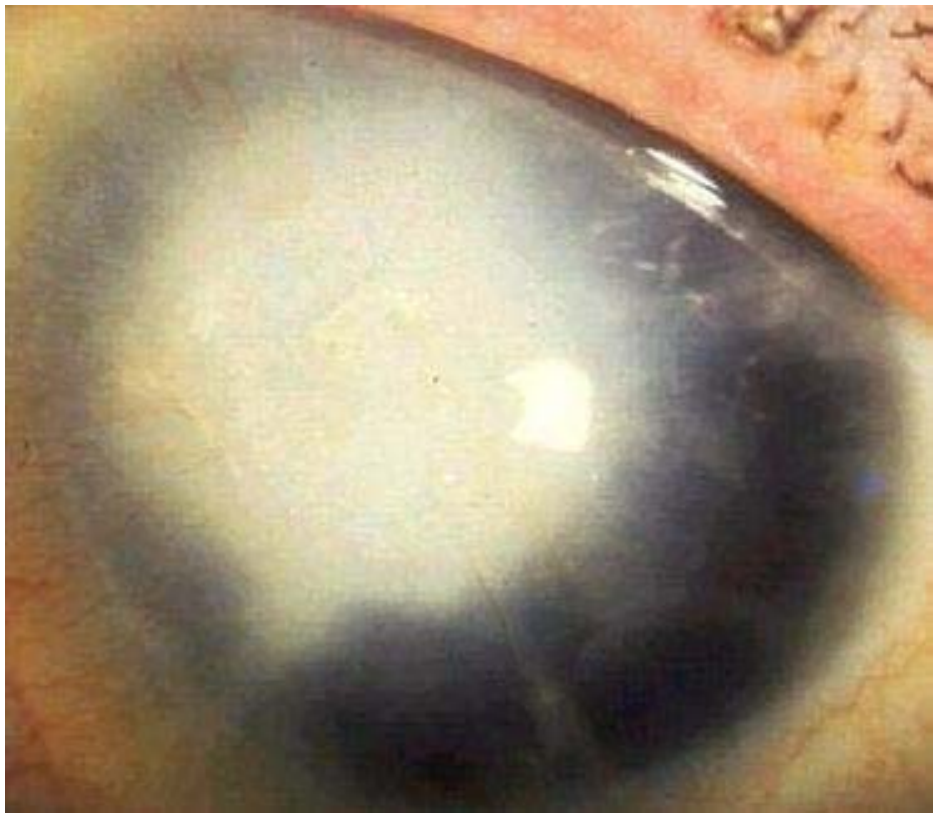
As a corneal opacity at birth (it's in the STUMPED mnemonic). The opacity ranges in severity from a faint haze to an opaque, elevated and vascularized mess.

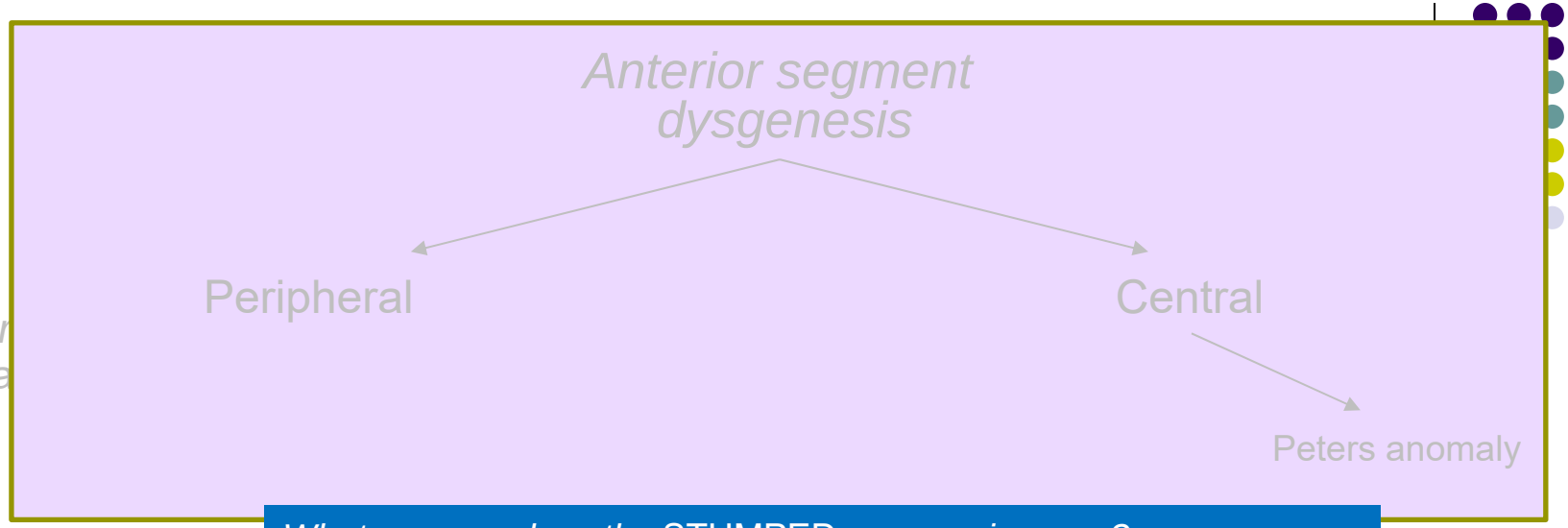
PAX Ophthalmicana



Peters anomaly: Hazy cornea

Q

In thr
An a



What purpose does the STUMPED *mnemonic serve?*

The BCS
based or
It's wheth

Is Peters
Central

Fo *How does*

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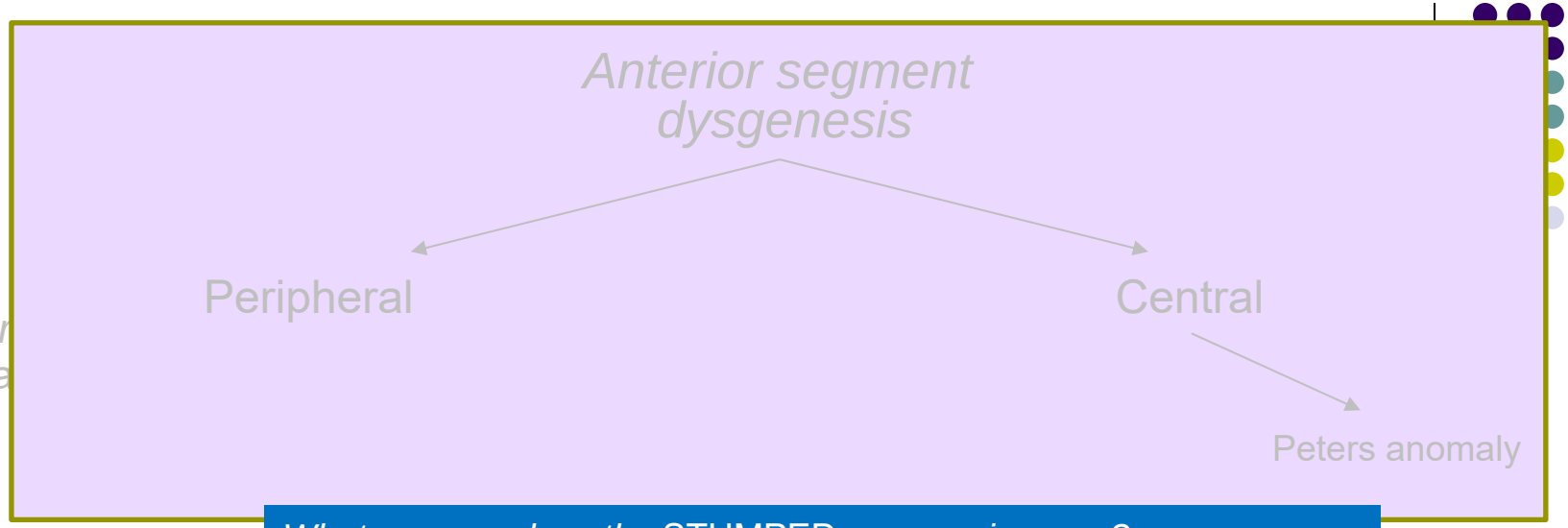

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What purpose does the STUMPED *mnemonic serve?*
Remembering the DDx for a cloudy cornea in an infant

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Q

Anterior segment dysgenesis

Peripheral

Central

Peters anomaly

In thr
An a

What purpose does the STUMPED *mnemonic serve?*
Remembering the DDx for a cloudy cornea in an infant

What are the other conditions in the mnemonic?
S
T
U
M
Peters anomaly
E
D

Note: There are two S's and two E's

The BCS
Central

Fo

How does Peters anomaly present?
As a corneal opacity at birth (it's in the **STUMPED mnemonic**). The opacity ranges in severity from a faint haze to an opaque, elevated and vascularized mess.

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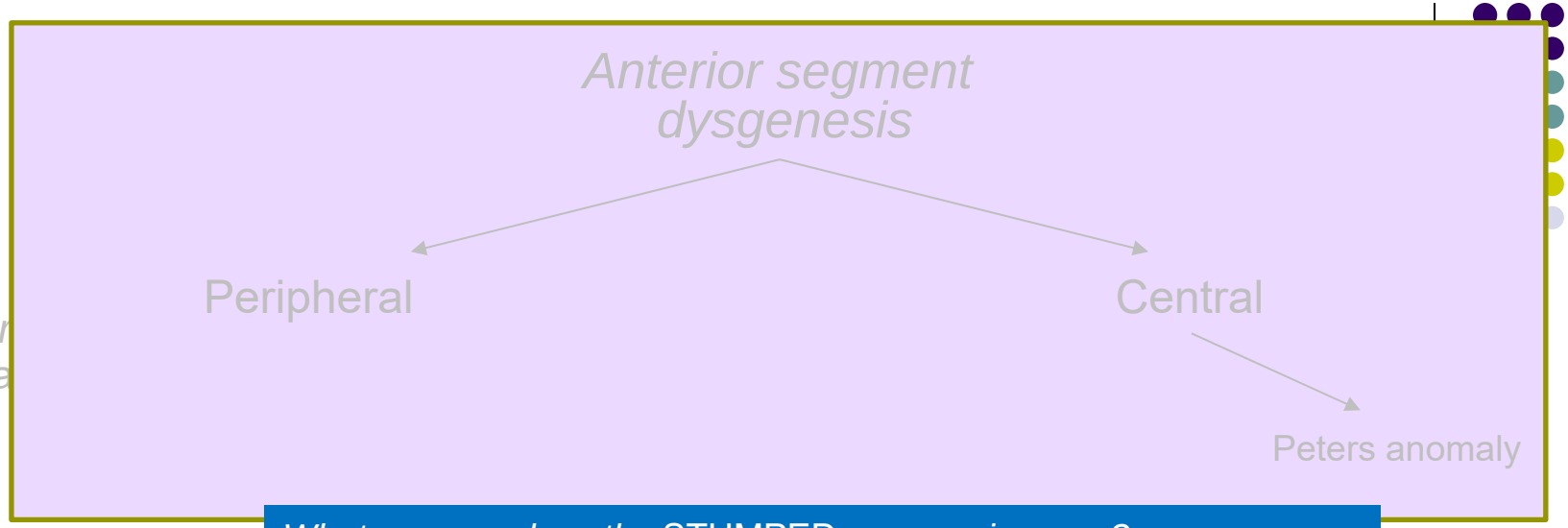


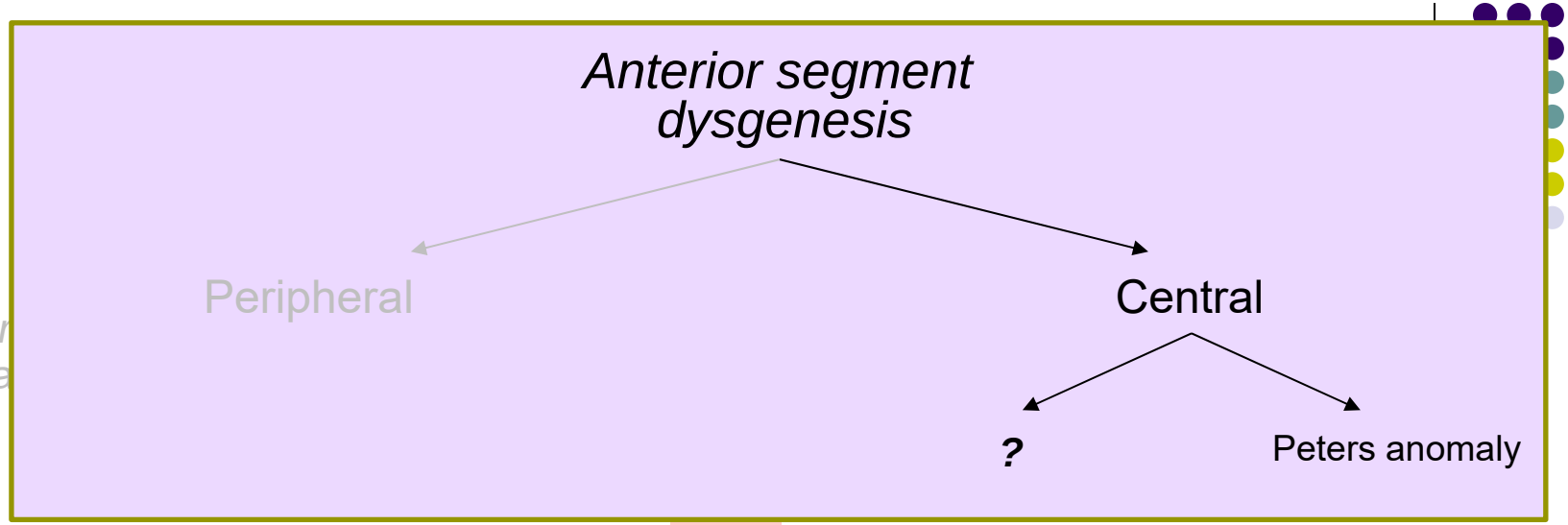
What purpose does the STUMPED *mnemonic serve?*
Remembering the DDx for a cloudy cornea in an infant

What are the other conditions in the mnemonic?
Sclerocornea; **S**tromal dystrophy (CHSD)
Trauma (eg, forcep injury)
Ulcer
Mucopolysaccharidosis
Peters anomaly
Endothelial dystrophy (CHED); **E**levated IOP (congenital glaucoma)
Dermoid of the cornea

Note: There are two S's and two E's

STUMPED mnemonic

Q



In thr
An a

P ete

The BCSC emphasizes one other central anterior-segment dysgenesis—what is it?

The BCSC divides anterior segment dysgeneses into two broad categories based on a fundamental anatomic distinction. What is it?
It's whether the dysgenesis involves the *central* vs *peripheral* anterior segment

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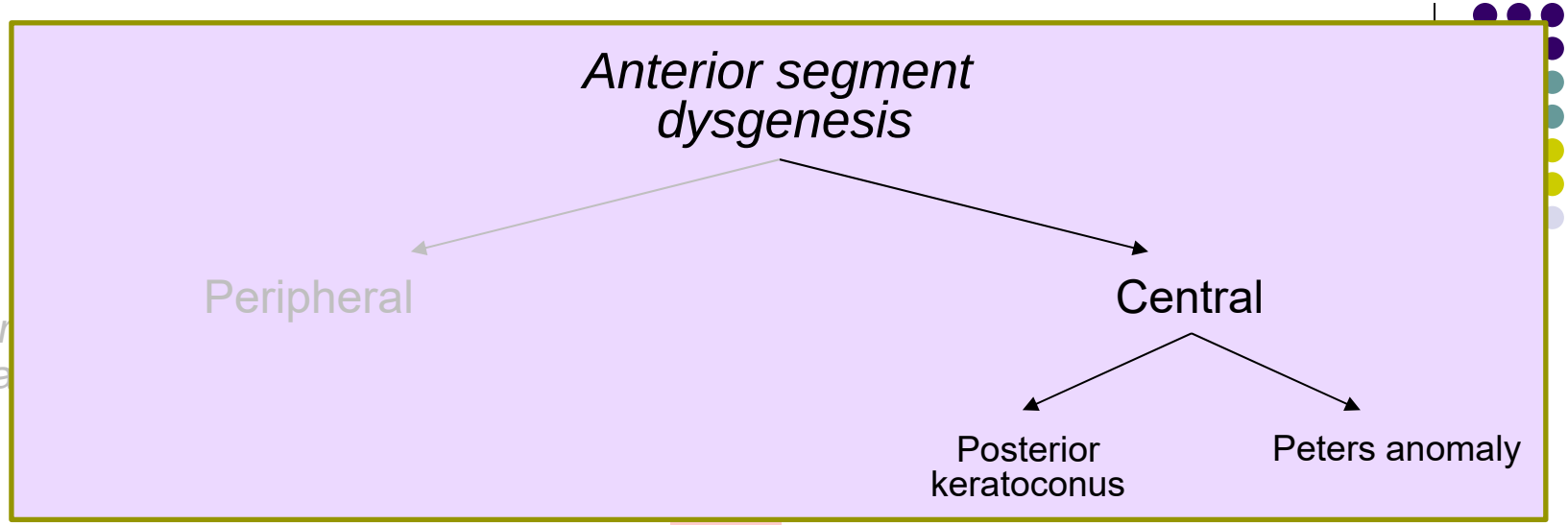


In thr
An a

P ete

The BCSC emphasizes one other central anterior-segment dysgenesis—what is it?
Posterior keratoconus

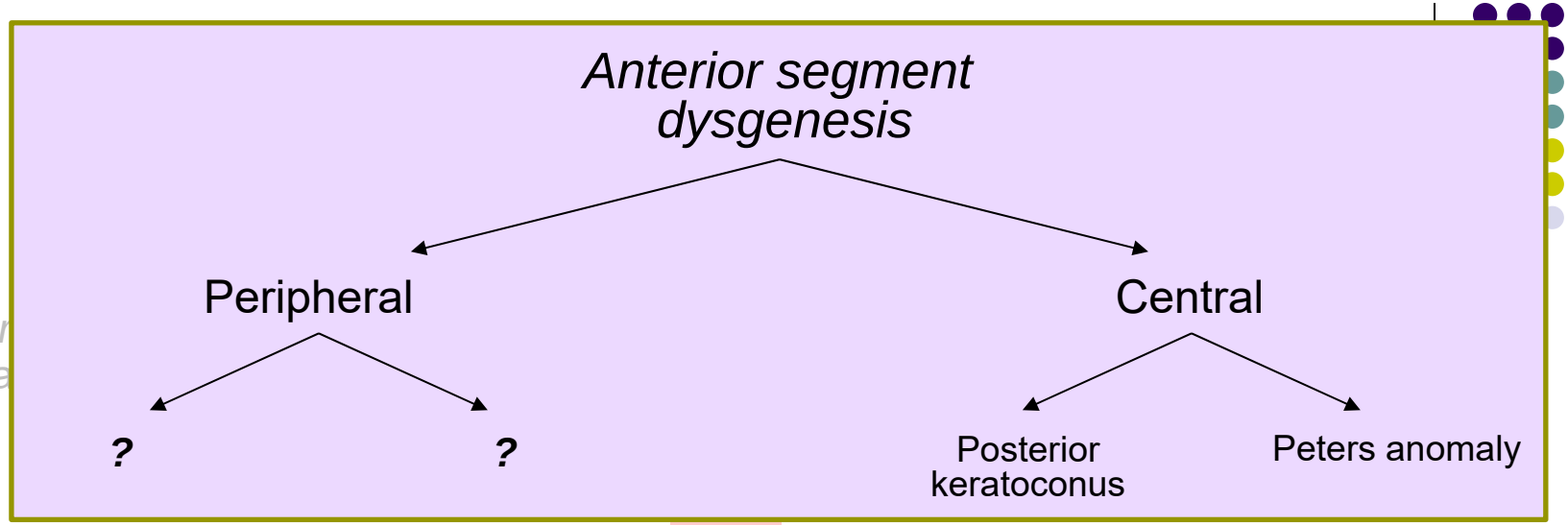
The BCSC divides anterior segment dysgeneses into two broad categories based on a fundamental anatomic distinction. What is it?
It's whether the dysgenesis involves the *central* vs *peripheral* anterior segment

Is Peters a peripheral, or central dysgenesis?
Central

Fo

How does Peters anomaly present?
As a corneal opacity at birth (it's in the STUMPED mnemonic). The opacity ranges in severity from a faint haze to an opaque, elevated and vascularized mess.

Q



In thr
An a

The BCSC *emphasizes two* peripheral *dysgeneses—what are they?*

P ete

The BCSC *emphasizes one other central anterior-segment dysgenesis—what is it?*
Posterior keratoconus

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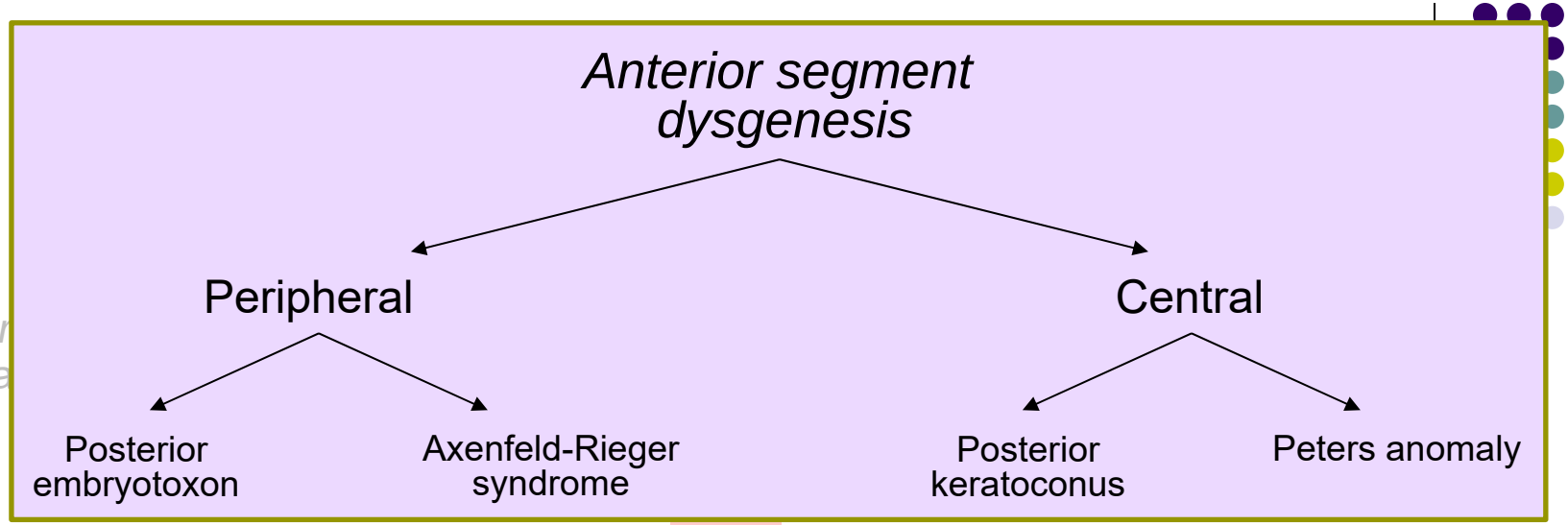

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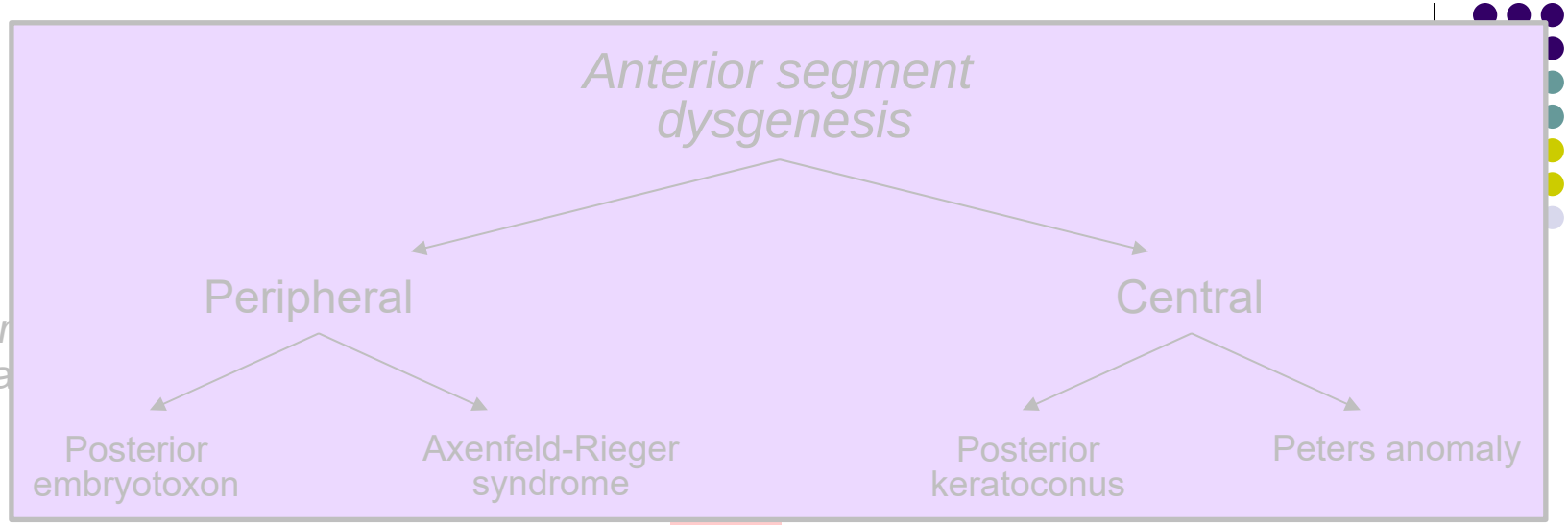

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For more on the anterior segment dysgeneses, see slide-set FELT7

Posterior embryotoxon, and Axenfeld-Reiger syndrome

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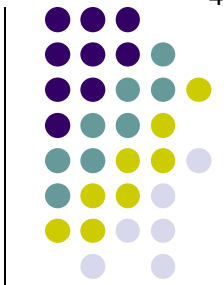
PAX Ophthalmicana



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

P eters anomaly

A nirida

Congenital catara***X***

Fovea and optic nerve ***6*** ypoplasia

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

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PAX Ophthalmicana

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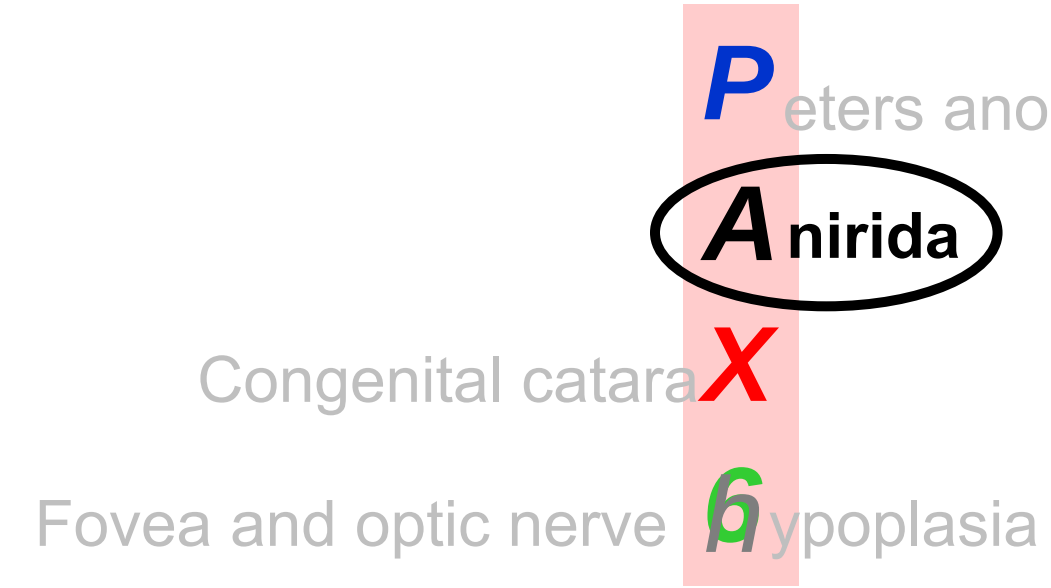

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PAX Ophthalmicana

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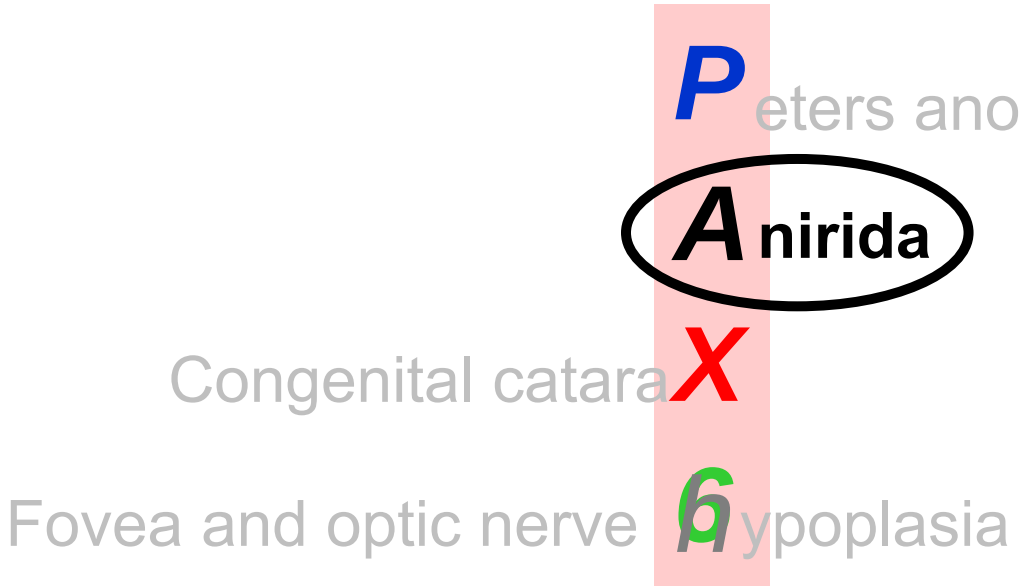

Endeavor to remember all of these. But if you have to pick just one to remember, make it…**aniridia**. *Almost all cases of aniridia are 2ndry to problems involving* PAX6.

PAX Ophthalmicana

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

A final takeaway point regarding aniridia…

P eters anomaly

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Congenital catara **X**

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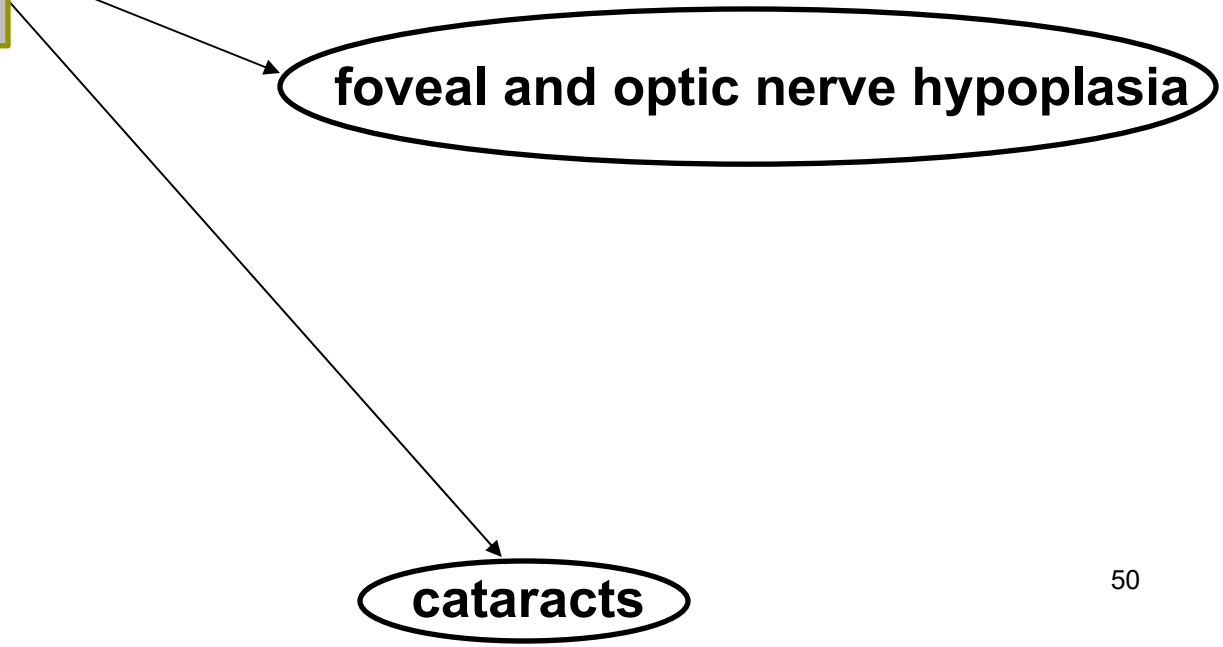
Endeavor to remember all of these. But if you have to pick just one to remember, make it…**aniridia**. *Almost all cases of aniridia are 2ndry to problems involving* PAX6.

PAX Ophthalmicana

Because all are tied to PAX6*, it shouldn't surprise you to hear that foveal hypoplasia, ON hypoplasia and cataracts are associated with it.*



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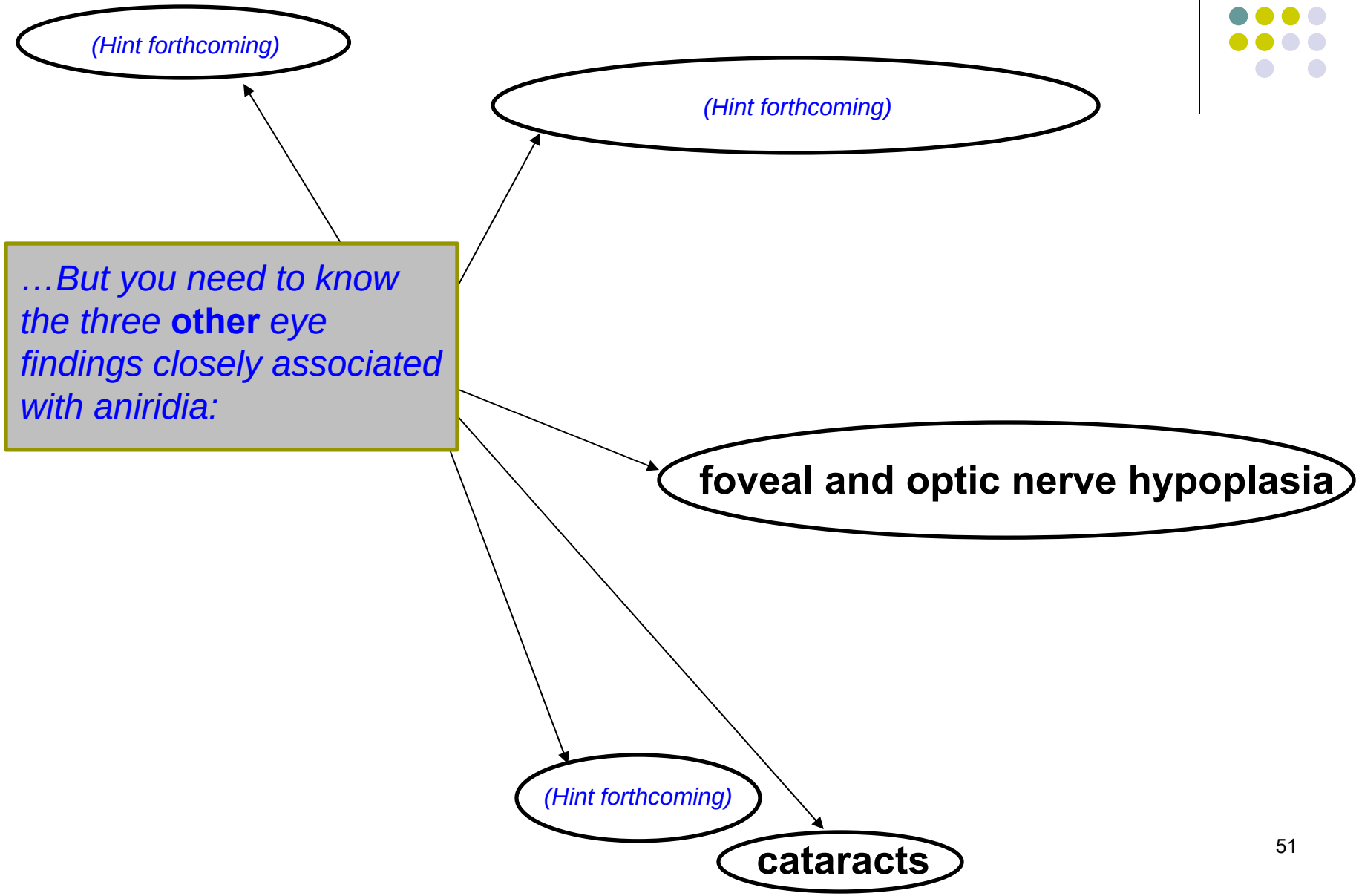
PAX Ophthalmicana



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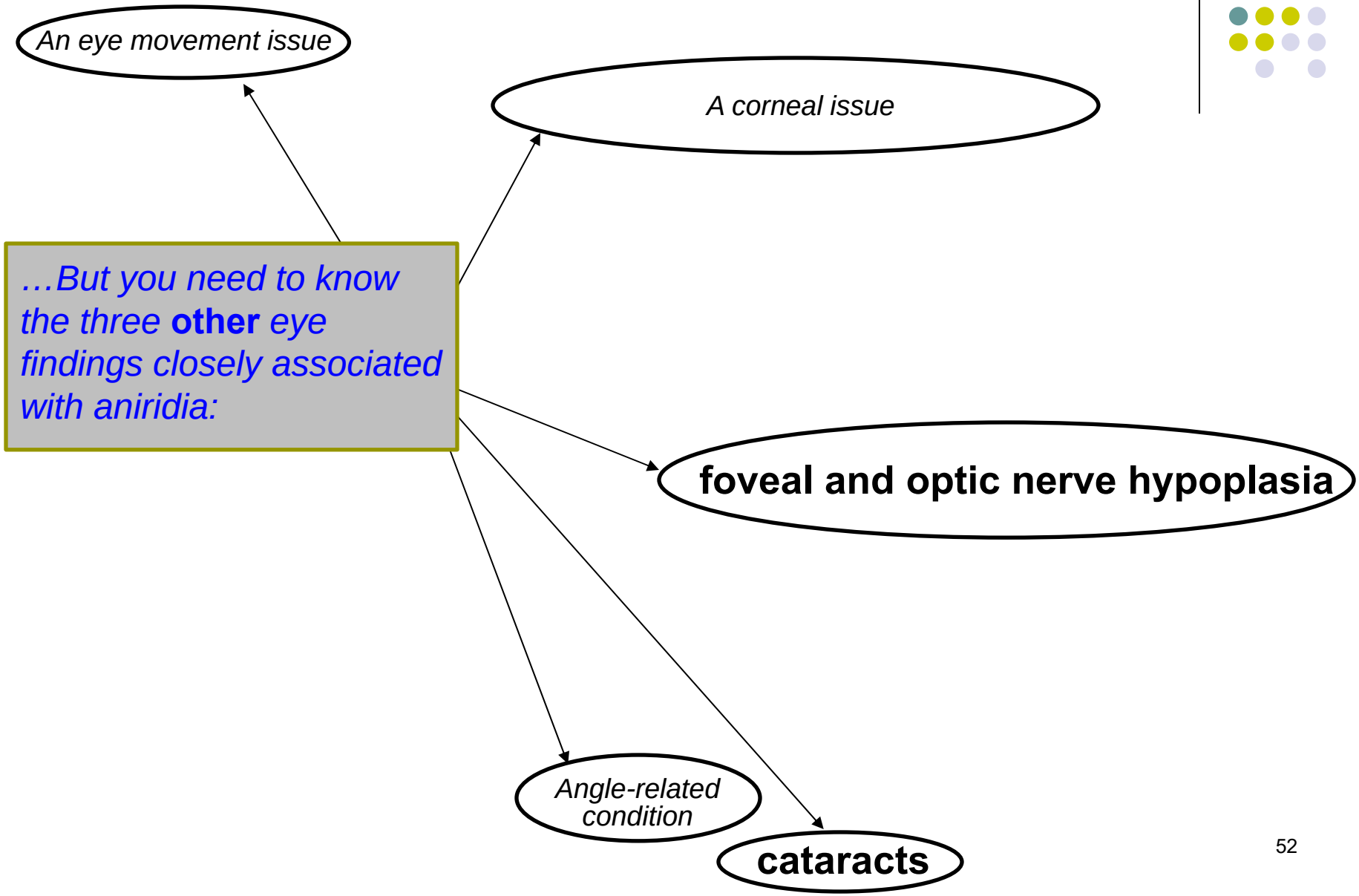

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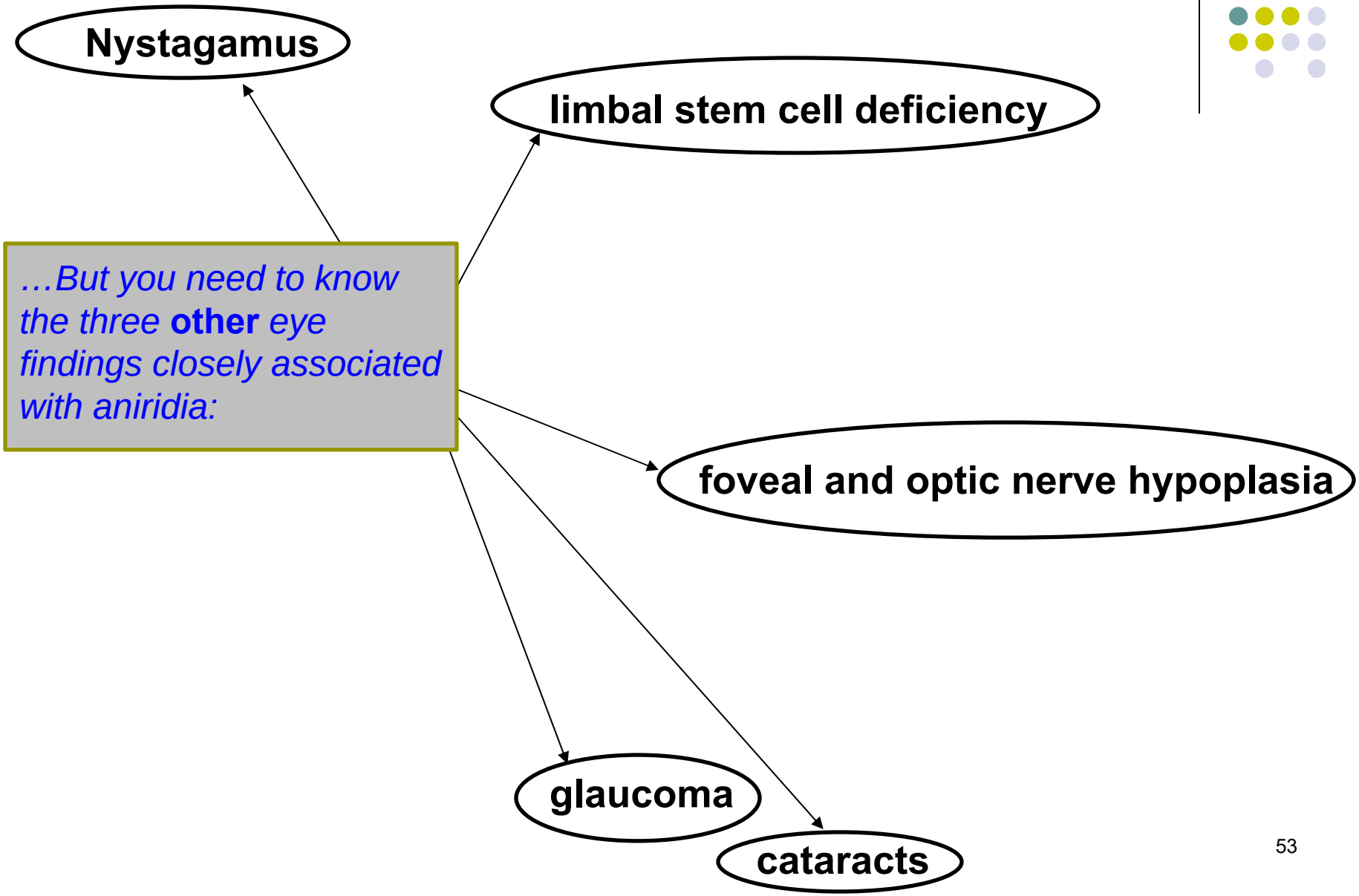
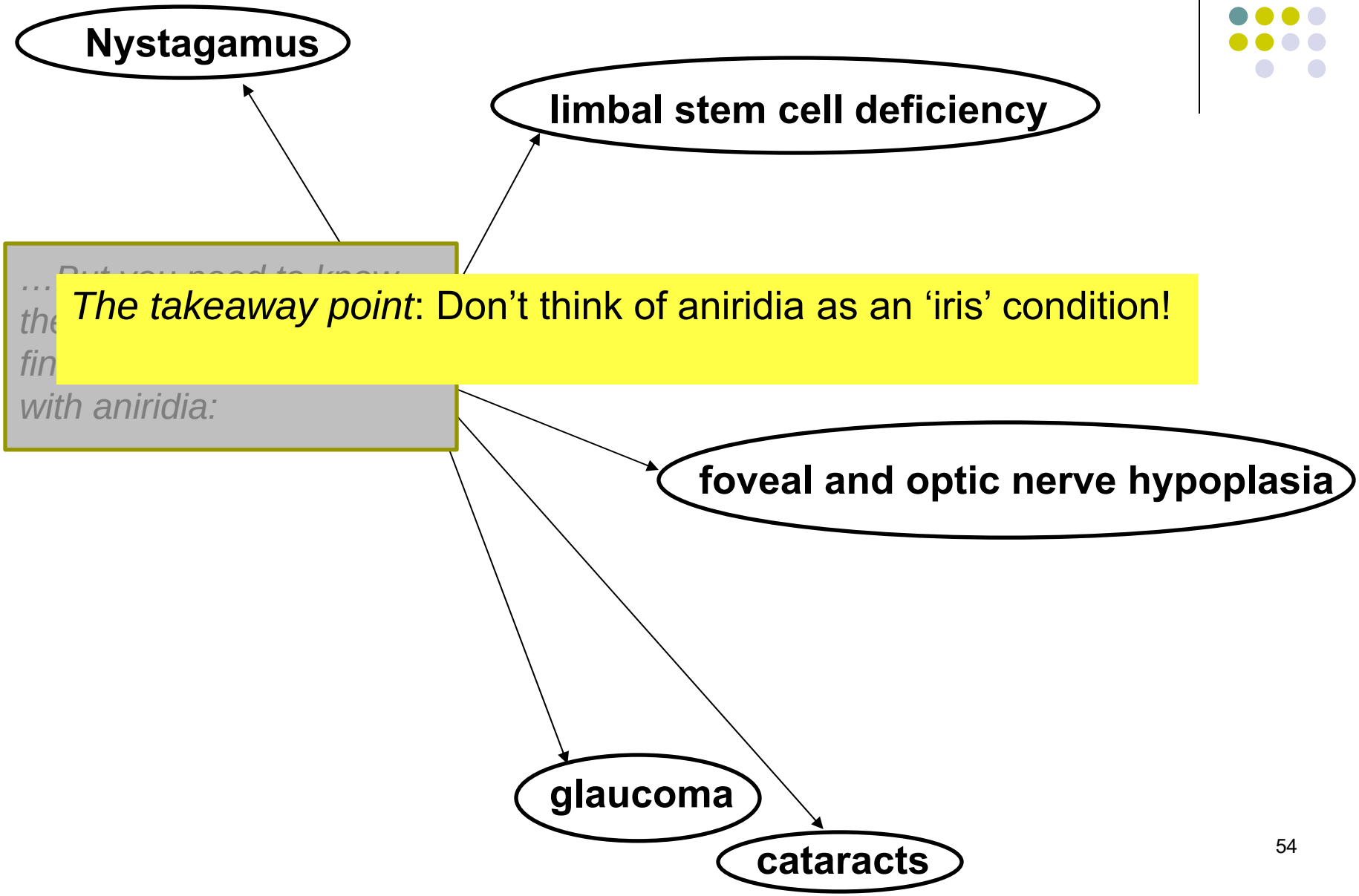
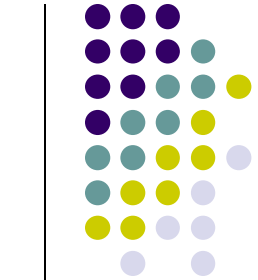


PAX Ophthalmicana



PAX Ophthalmicana



Nystagamus

limbal stem cell deficiency

… But you need to know the fin… with aniridia:

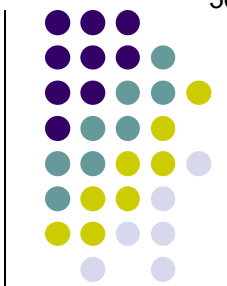
The takeaway point: Don't think of aniridia as an 'iris' condition!
The *BCSC* characterizes it is a ***panophthalmic disorder***

foveal and optic nerve hypoplasia

glaucoma

cataracts

PAX Ophthalmicana



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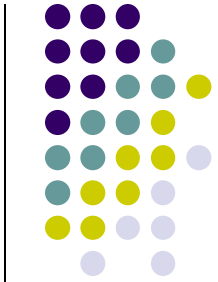
The Fundamentals *book lists three transcription-factor genes that are especially important for the eye—what are they?*
--PAX2
--**PAX3**
--PAX6

Next, we'll do PAX3

Q

PAX Ophthalmicana



With what eponymous syndrome is PAX3 *associated?*

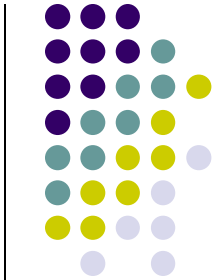
A

PAX Ophthalmicana

With what eponymous syndrome is PAX3 *associated?*

Waardenburg Syndrome

PAX Ophthalmicana

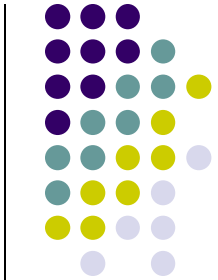


With what eponymous syndrome is PAX**3** *associated?*

W**aa**rdenburg Syndrome

Sidebar: Isn't it frustrating that, with its two AAs, Waardenburg syndrome is not associated with *PAX***2**? I mean, seriously: Would it have been **that** big a deal to name Waardenburg's gene *PAX2*, and the **other** one *PAX3*?

PAX Ophthalmicana



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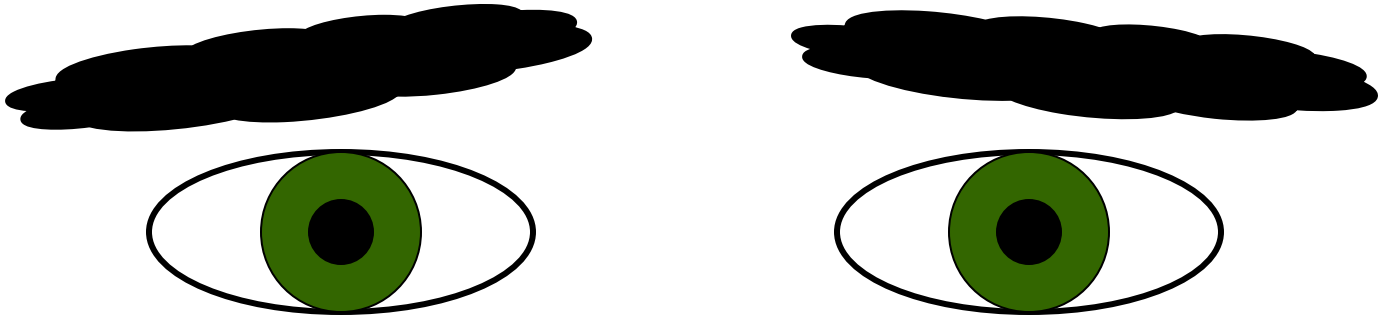
For me, this is **so** annoying that the annoyance itself serves as a memory aid; ie, when trying to recall whether Waardenburg is *PAX2* vs *PAX3*, in my head pops 'Oh yeah, that's the condition that missed out on the perfect mnemonic,' and so I know its *PAX3*.

Q

PAX Ophthalmicana



Waardenburg Syndrome

What 3 ophthalmic findings are classic for Waardenburg syndrome?

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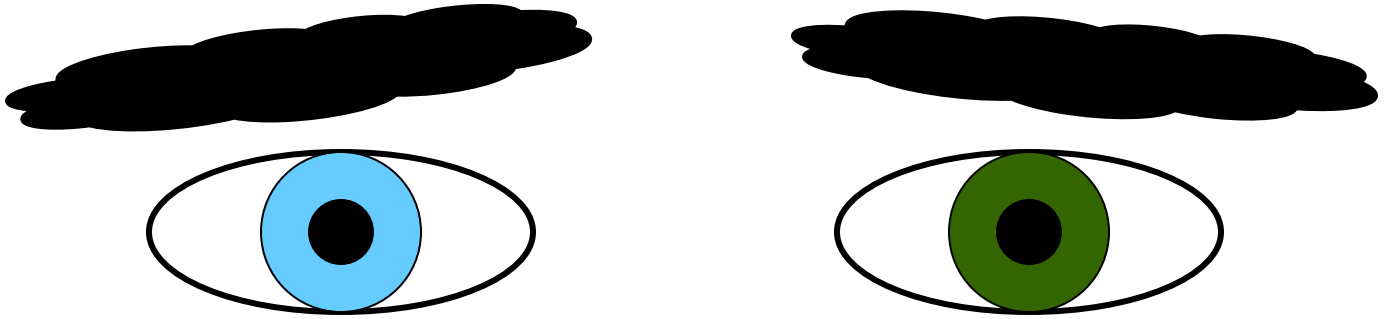
A

PAX Ophthalmicana



Waardenburg Syndrome

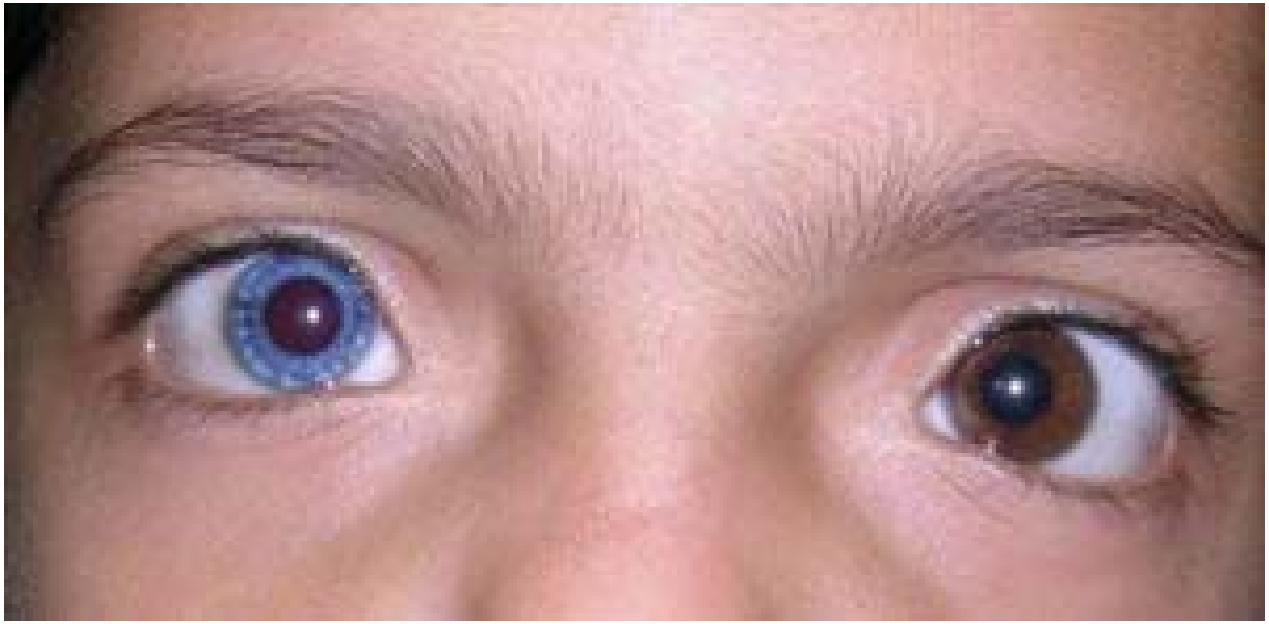
What 3 ophthalmic findings are classic for Waardenburg syndrome?

--Heterochromia iridis

--Synophrys

--Dystopia canthorum

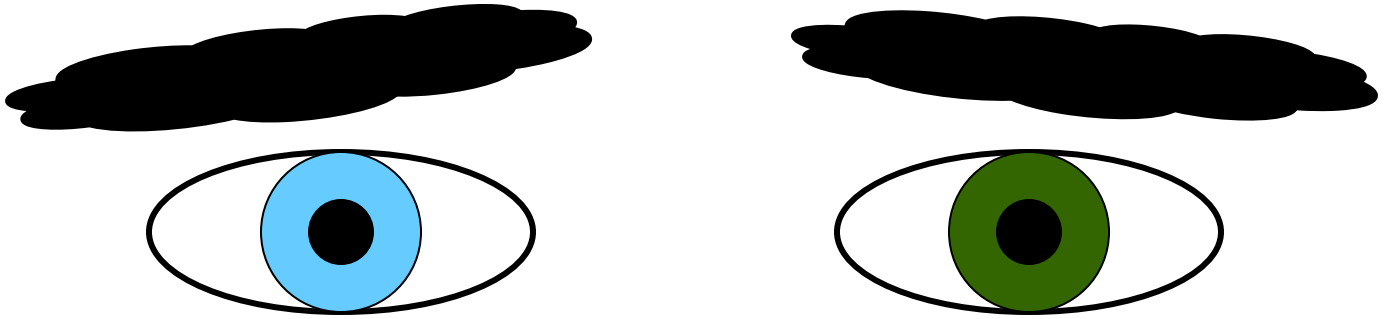
PAX Ophthalmicana



Waardenburg syndrome: Heterochromia iridis, dystopia canthorum, and mild synophrys
(What the heck is **synophrys***?)*

Q

PAX Ophthalmicana



Waardenburg Syndrome

What 3 ophthalmic findings are classic for Waardenburg syndrome?

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What the heck is synophrys*?*

Q/A

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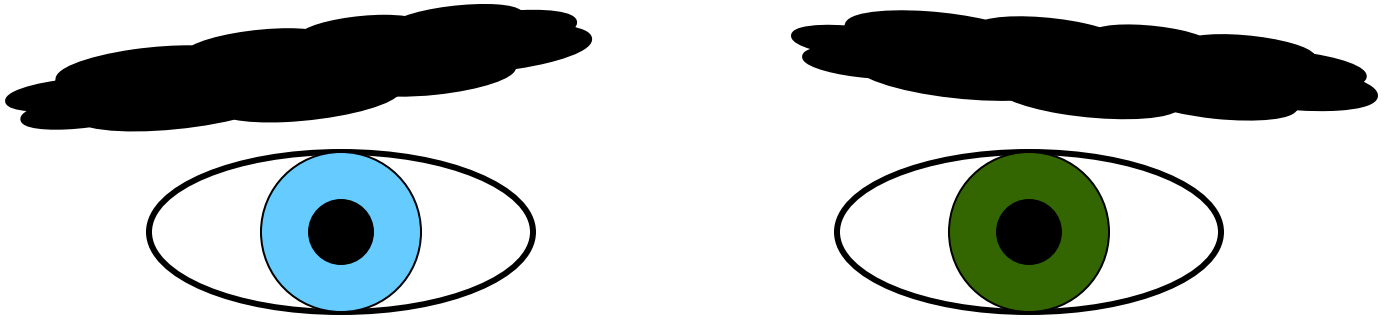

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What the heck is synophrys*?*

The formal medical term for a

A

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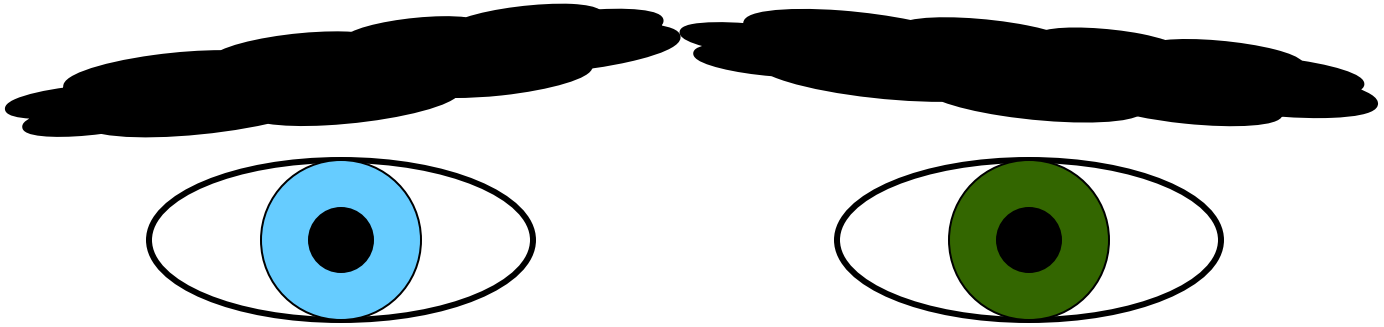

Waardenburg Syndrome

What 3 ophthalmic findings are classic for Waardenburg syndrome?

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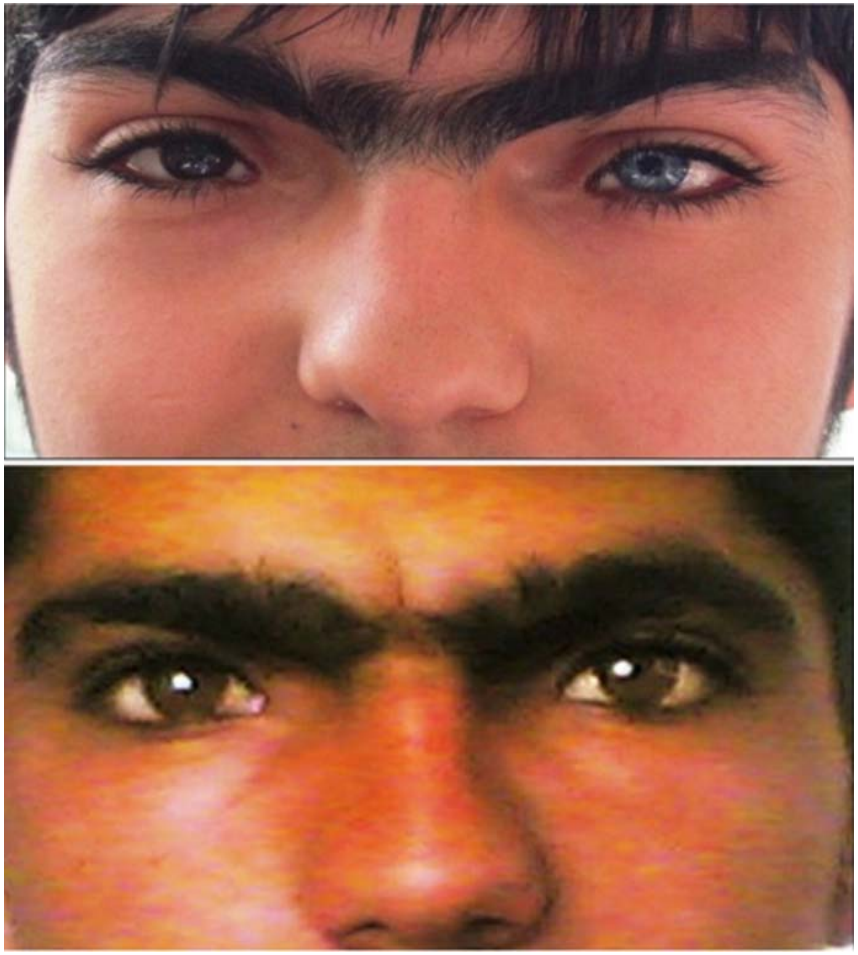
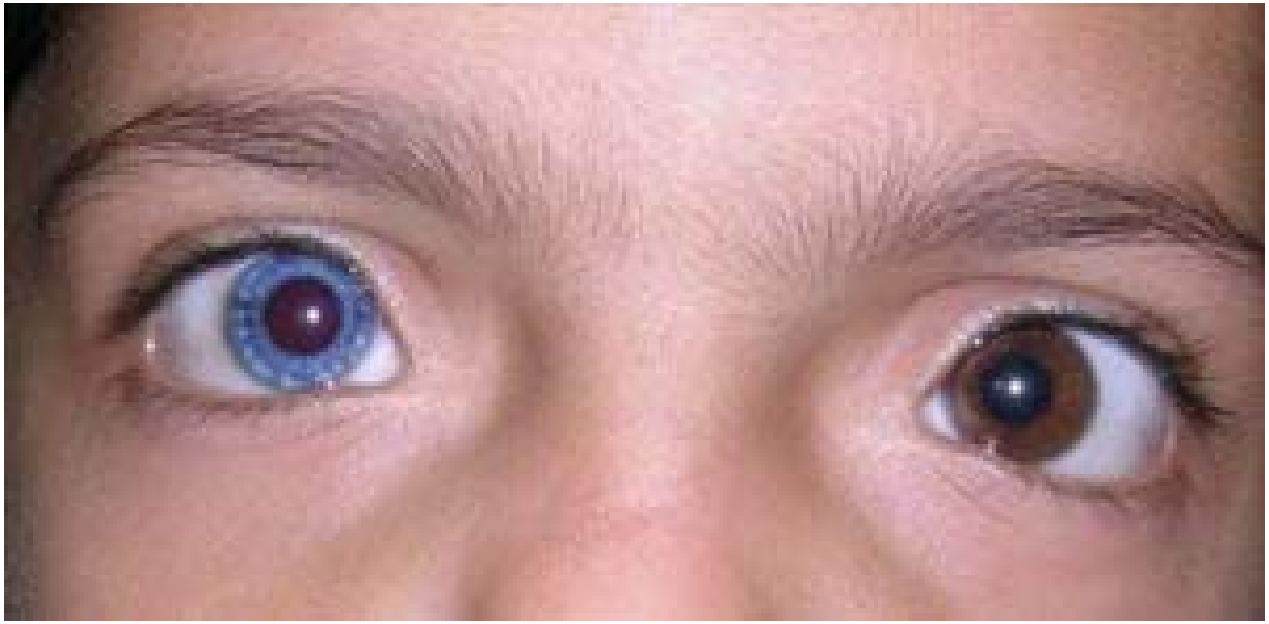
What the heck is synophrys*?*
The formal medical term for a **unibrow**

PAX Ophthalmicana



Waardenburg syndrome: Synophrys

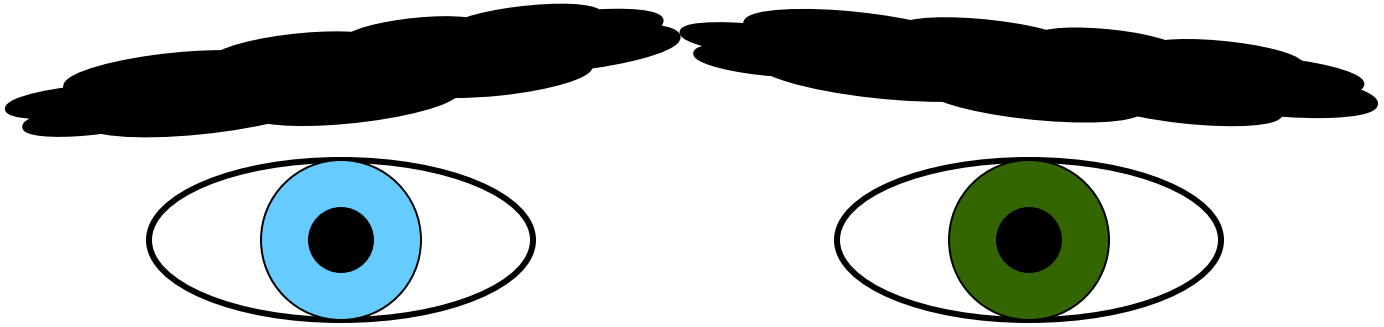
PAX Ophthalmicana



Waardenburg syndrome: Heterochromia iridis, dystopia canthorum, and mild synophrys
(What the heck is **dystopia canthorum***?)*

Q

PAX Ophthalmicana



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What the heck is dystopia canthorum?

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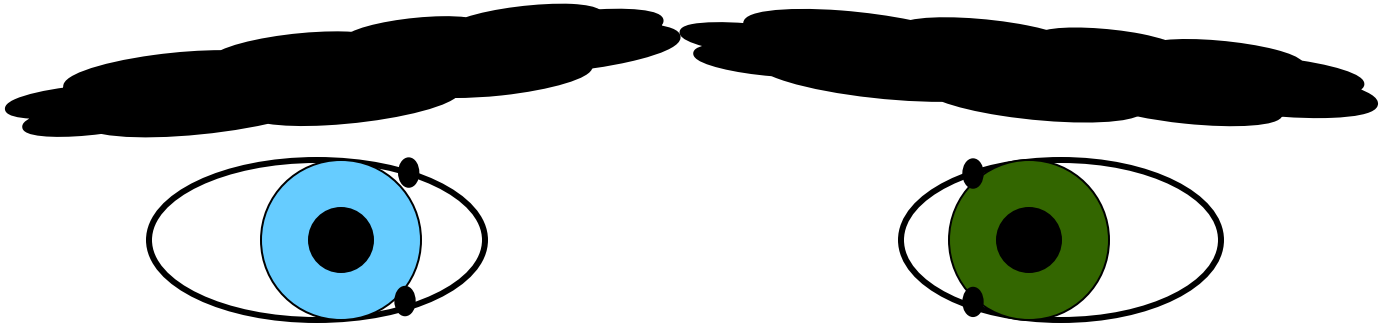

Waardenburg Syndrome

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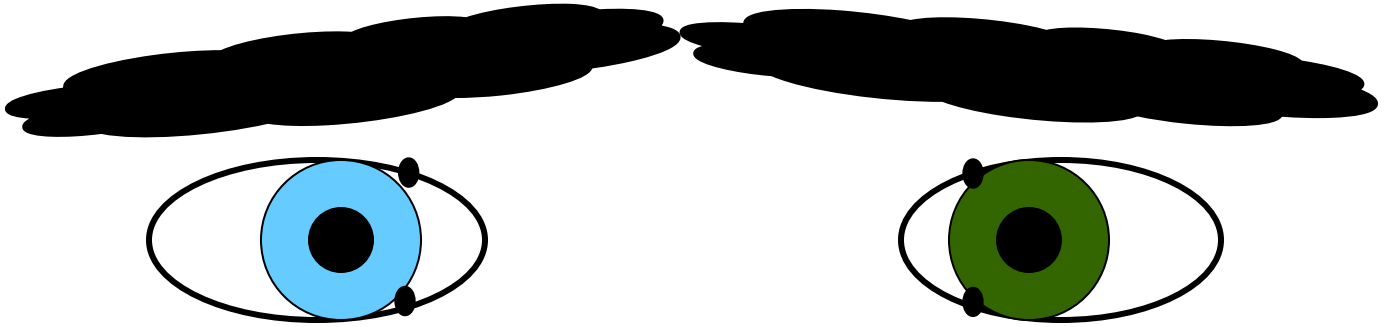
--**Dystopia canthorum**

What the heck is dystopia canthorum?
Lateral displacement of the canthi (ie, telecanthus) ***PLUS*** laterally displaced lacrimal puncta

Q

PAX Ophthalmicana



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How on earth are you supposed to recognize that the puncta are too lateral?

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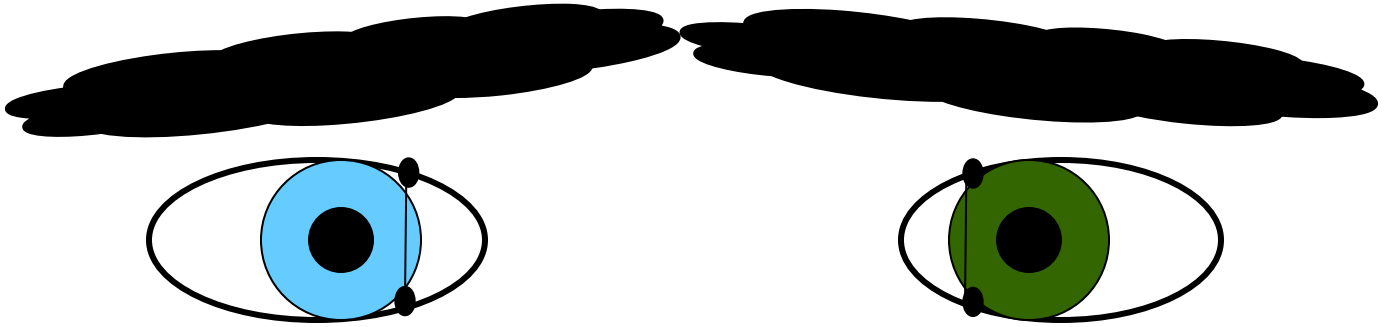
PAX Ophthalmicana



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laterally displaced lacrimal puncta

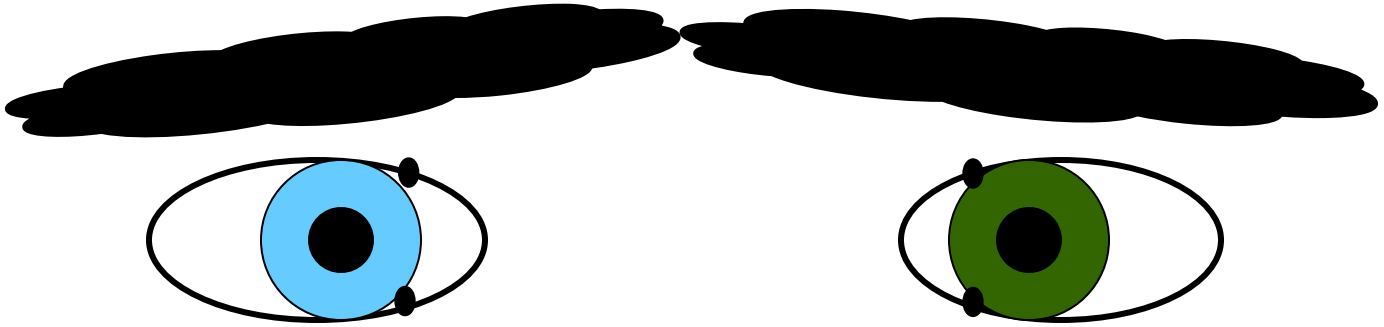
How on earth are you supposed to recognize that the puncta are too lateral?
Draw an imaginary vertical line from the upper to the lower puncta. If this line crosses the cornea, the puncta are displaced. (Next time you examine a pt at the slit-lamp, take note of whether such a line crosses their cornea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

PAX Ophthalmicana

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

Q

PAX Ophthalmicana



Waardenburg Syndrome

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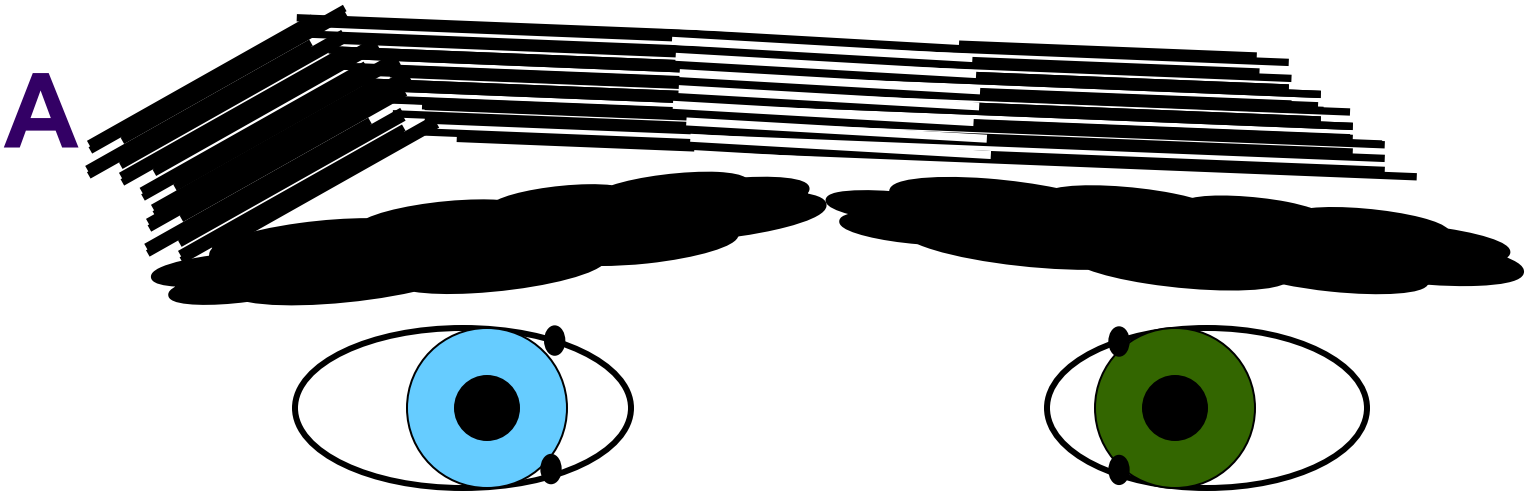
What **non-ophthalmic** *finding is classic for Waardenburg syndrome?*



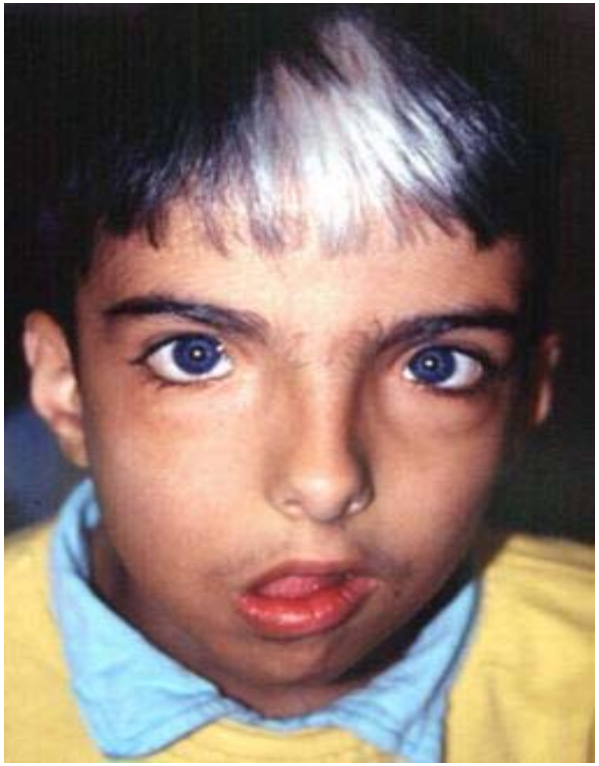
Waardenburg Syndrome

What 3 ophthalmic findings are classic for Waardenburg syndrome?
-- **Heterochromia iridis**
--Synophrys
--Dystopia canthorum

What **non-ophthalmic** *finding is classic for Waardenburg syndrome?*
The presence of a white forelock (ie, an isolated streak of **white hair** in the forehead region)

Waardenburg
White forelock

PAX Ophthalmicana



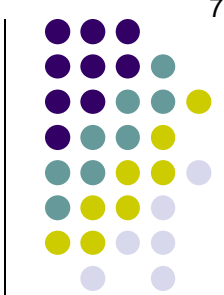
Waardenburg syndrome: White forelock

PAX Ophthalmicana

Note that Waardenburg syndrome has forms that do not involve heterochromia

Q

PAX Ophthalmicana



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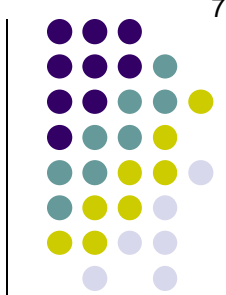
--PAX3
--PAX6

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

A

PAX Ophthalmicana



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