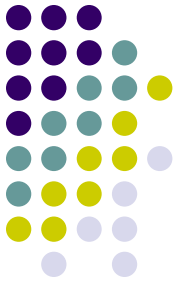


Anterior Segment Dysgenesis



In terms of the fundamental embryological disorder involved, anterior segment dysgenesis is what sort of condition?

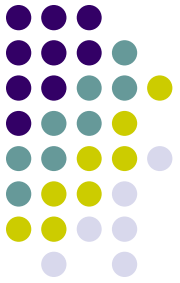
Anterior Segment Dysgenesis



In terms of the fundamental embryological disorder involved, anterior segment dysgenesis is what sort of condition?

A neurocrisstopathy

Anterior Segment Dysgenesis

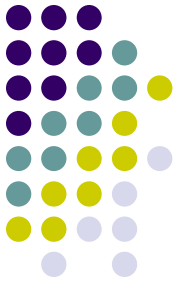


In terms of the fundamental embryological disorder involved, anterior segment dysgenesis is what sort of condition?

A neurocristopathy

What is a neurocristopathy?

Anterior Segment Dysgenesis



In terms of the fundamental embryological disorder involved, anterior segment dysgenesis is what sort of condition?

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What is a neurocristopathy?

A congenital/developmental abnormality owing to flawed neural-crest cell migration or differentiation

Anterior Segment Dysgenesis



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What is/are neural crest cells?

Anterior Segment Dysgenesis



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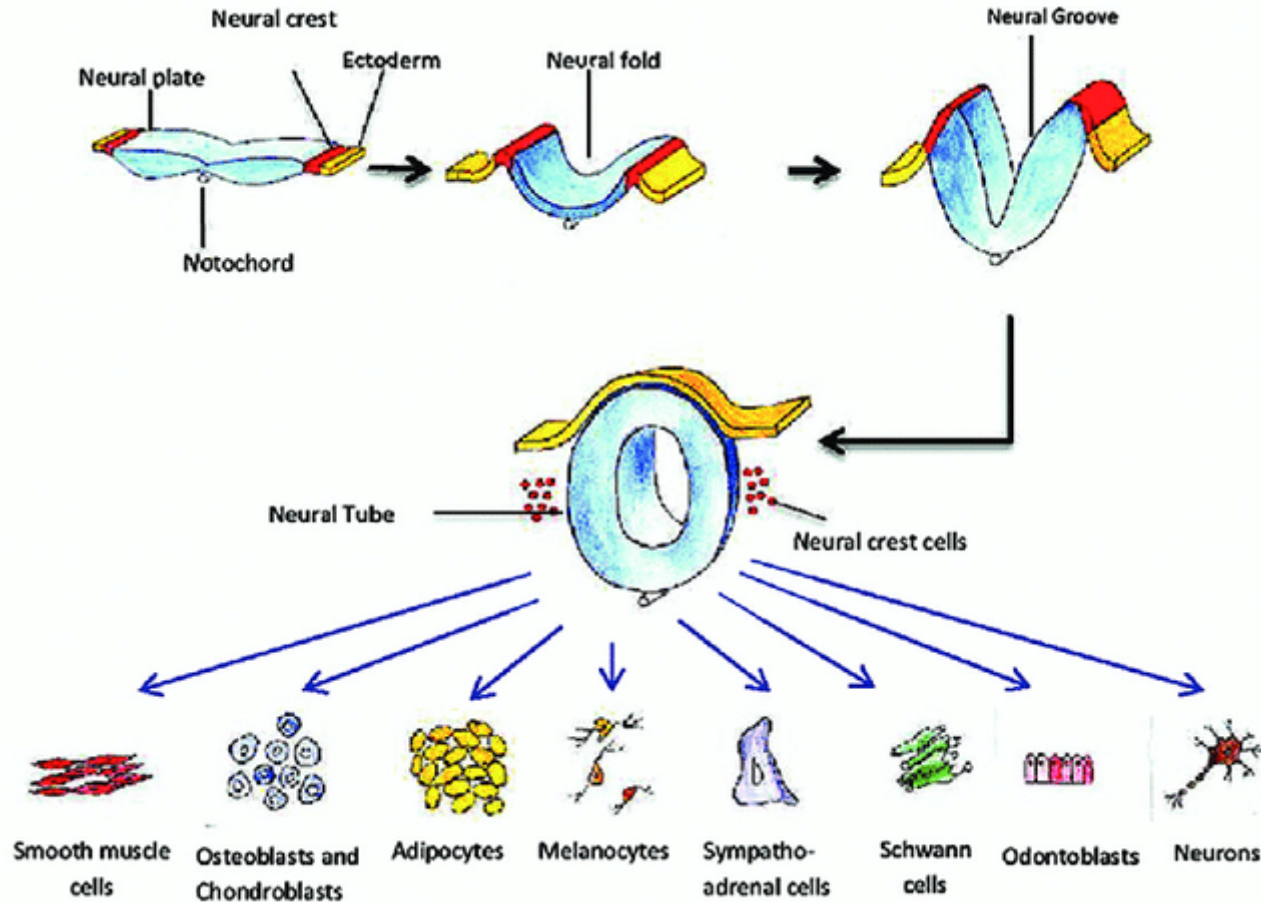
What is a neurocristopathy?

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What is/are neural crest cells?

A special subpopulation of neuroectodermal cells that migrate across the embryo and deposit themselves at a wide variety of locations, eventually differentiating into many distinct tissues

Anterior Segment Dysgenesis



Neural crest cell differentiation (for demo purposes only; don't memorize)

Anterior Segment Dysgenesis



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Neural-crest-cell migration concerning the anterior segment occurs in three ‘waves.’

Which wave involves which future structure?

First wave →?

Second wave →

Third wave →

Anterior Segment Dysgenesis



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Second wave → ?

Third wave →

Anterior Segment Dysgenesis



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Second wave → **Iris stroma**

Third wave → ?

Anterior Segment Dysgenesis



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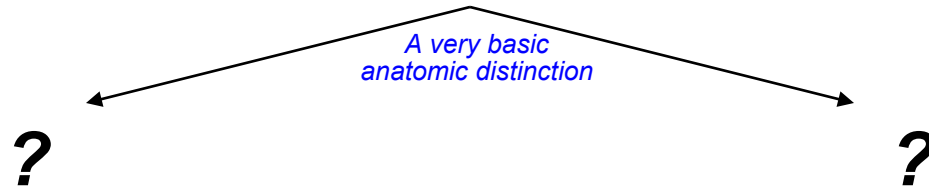
Second wave → Iris stroma

Third wave → **Corneal stroma (keratocytes)**

Anterior Segment Dysgenesis

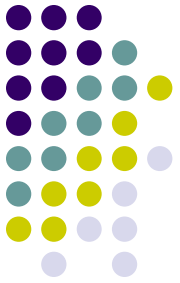


Anterior segment dysgenesis

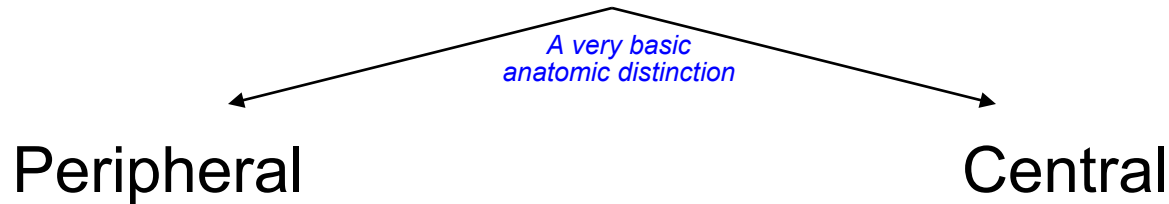


The anterior-segment dysgeneses are divided into two groups on the basis of a very fundamental anatomic distinction among them. What is that distinction?

Anterior Segment Dysgenesis

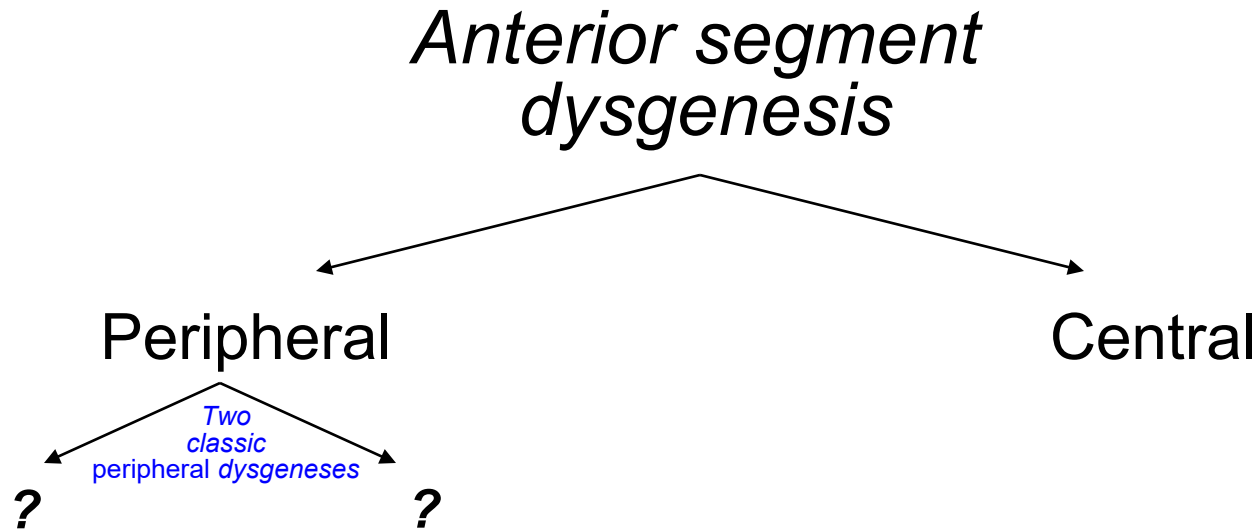
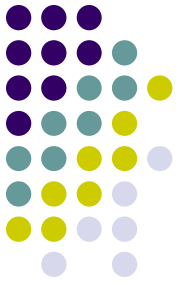


Anterior segment dysgenesis



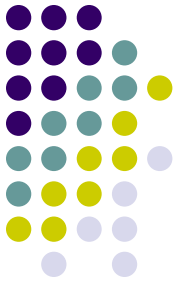
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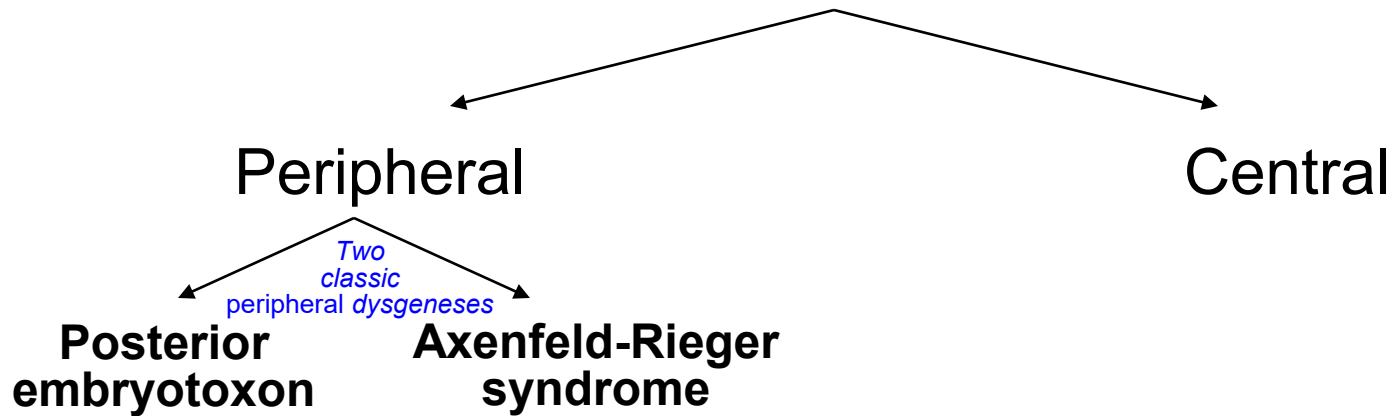


*The BCSC goes into depth on two
peripheral dysgeneses—which two?*

Anterior Segment Dysgenesis



Anterior segment dysgenesis

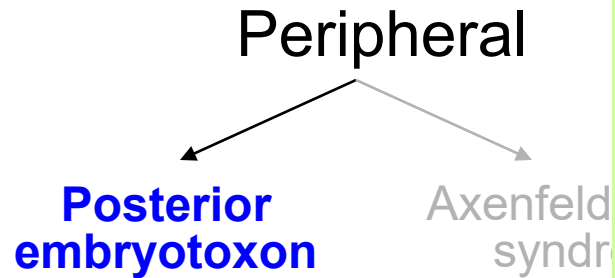


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Anterior Segment Dysgenesis

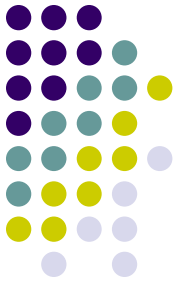


Anterior segment dysgenesis

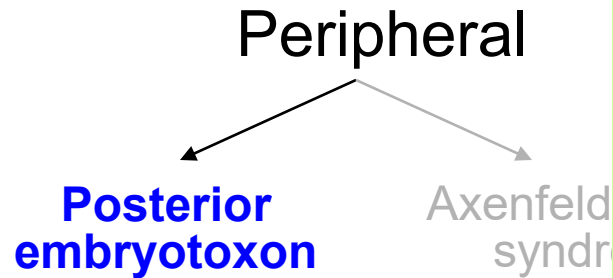


What is a posterior embryotoxon?

Anterior Segment Dysgenesis



Anterior segment dysgenesis



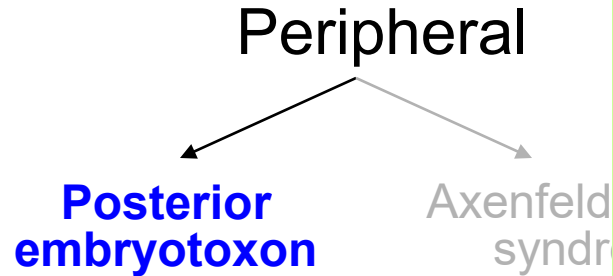
What is a posterior embryotoxon?

An anteriorly displaced and thickened Schwalbe's line/ring

Anterior Segment Dysgenesis



Anterior segment dysgenesis



What is a posterior embryotoxon?

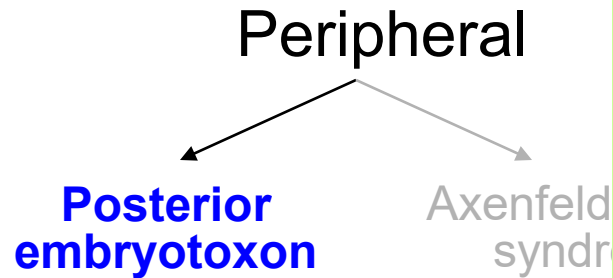
An anteriorly displaced and thickened **Schwalbe's line/ring**

What is Schwalbe's line/ring?

Anterior Segment Dysgenesis



Anterior segment dysgenesis



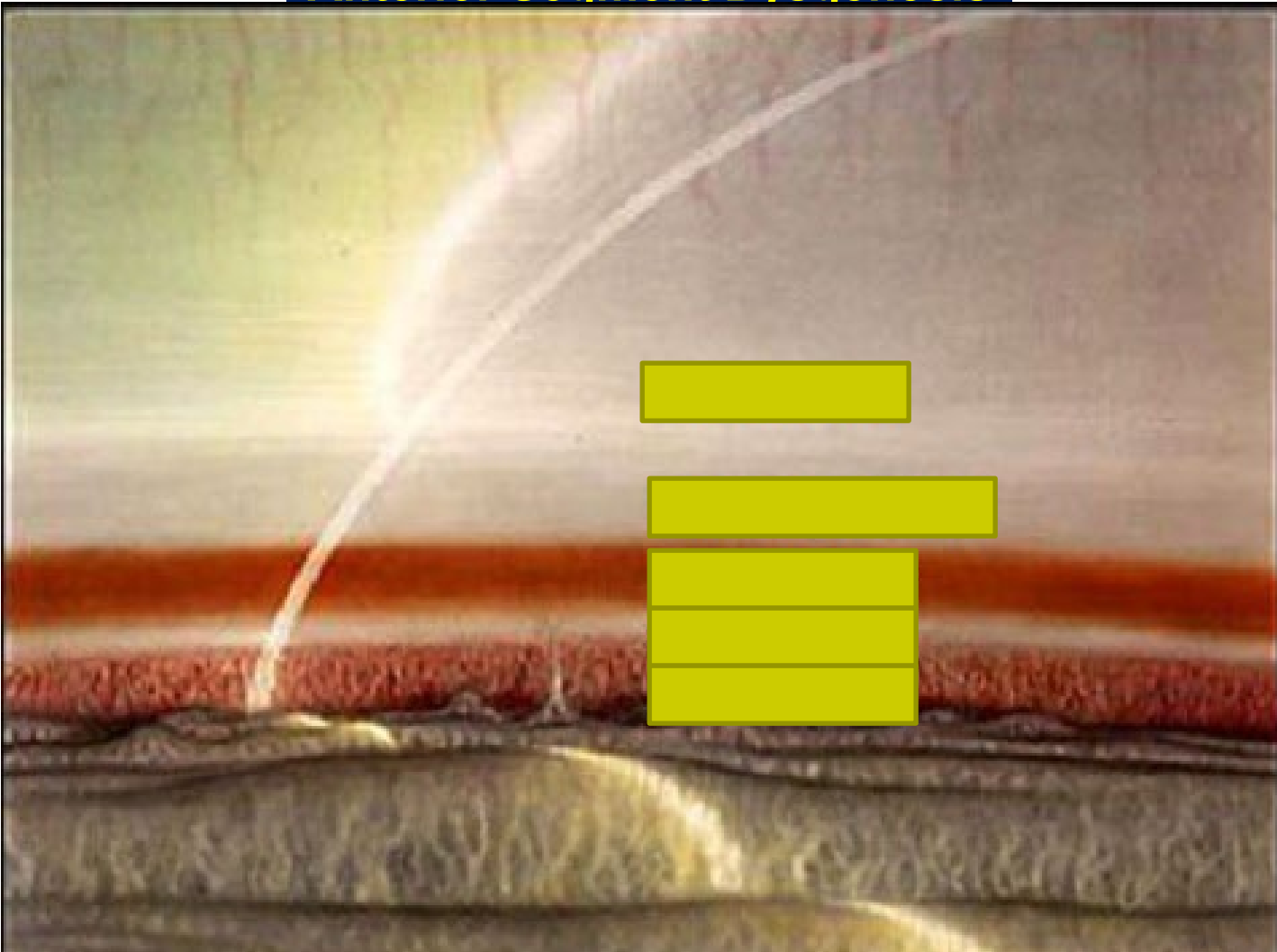
What is a posterior embryotoxon?

An anteriorly displaced and thickened **Schwalbe's line/ring**

What is Schwalbe's line/ring?

The edge or termination of Descemet's layer

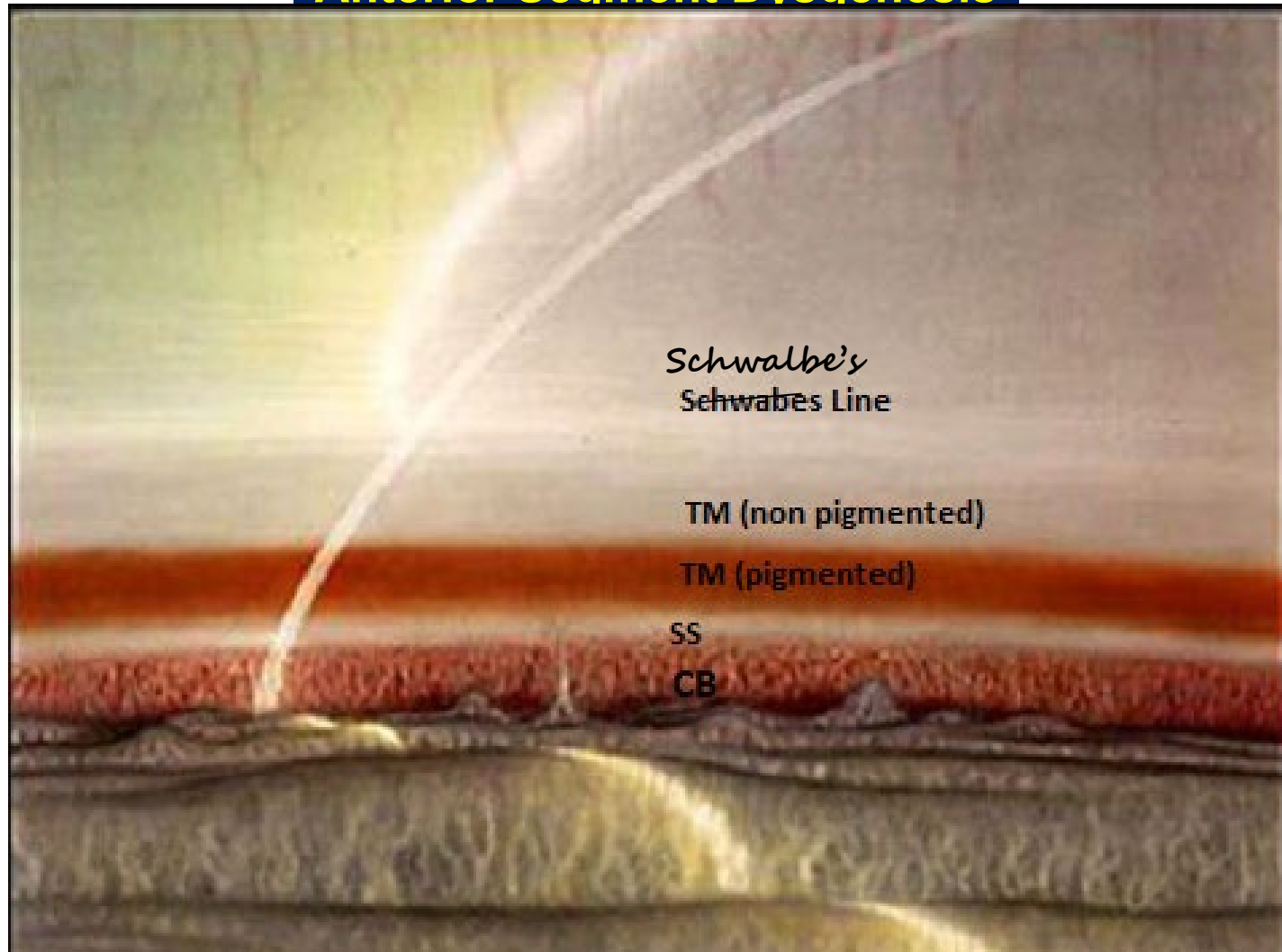
Anterior Segment Dysgenesis



Normal angle anatomy: Identify the structures



Anterior Segment Dysgenesis

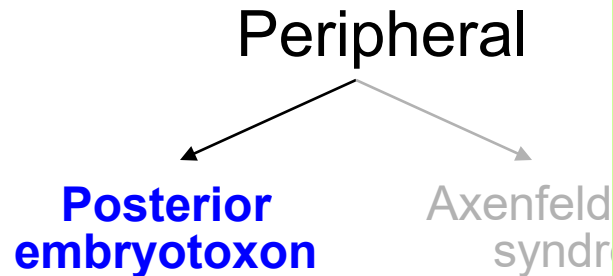


Normal angle anatomy: Identify the structures

Anterior Segment Dysgenesis



Anterior segment dysgenesis



What is a posterior embryotoxon?

An anteriorly displaced and thickened **Schwalbe's line/ring**

What is Schwalbe's line/ring?

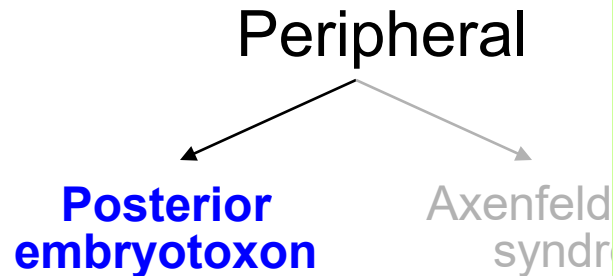
The edge or termination of Descemet's layer

Is it normally apparent during slit-lamp examination?

Anterior Segment Dysgenesis



Anterior segment dysgenesis



What is a posterior embryotoxon?

An anteriorly displaced and thickened **Schwalbe's line/ring**

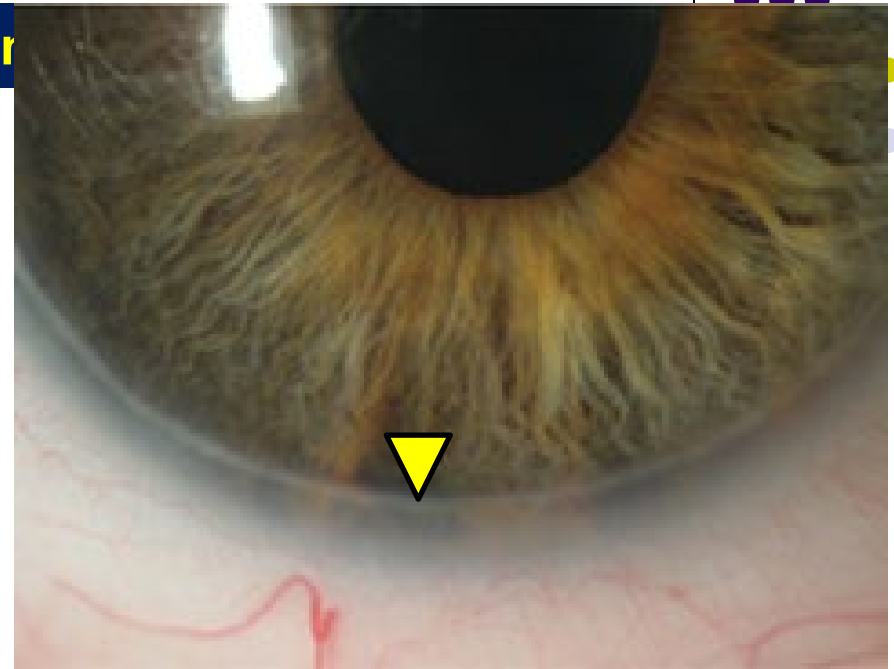
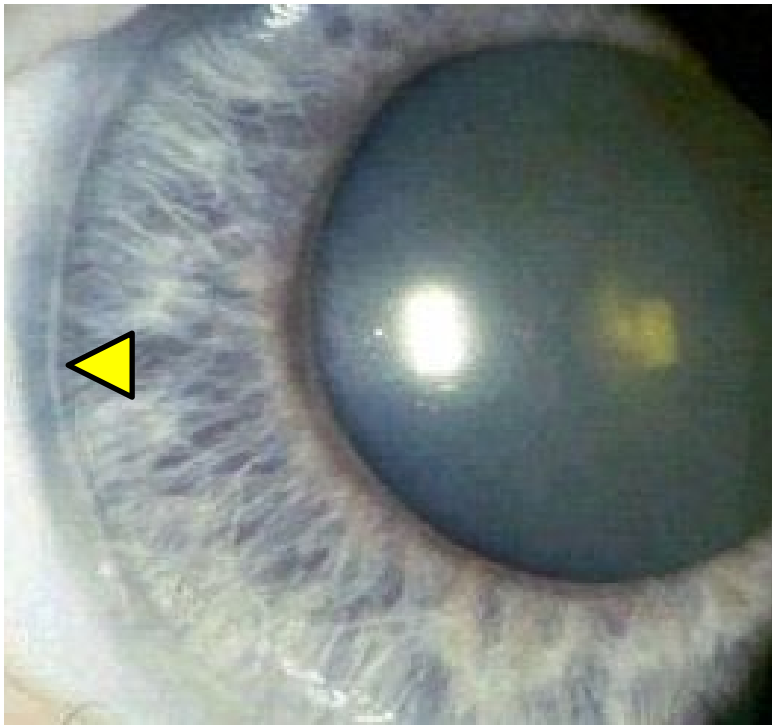
What is Schwalbe's line/ring?

The edge or termination of Descemet's layer

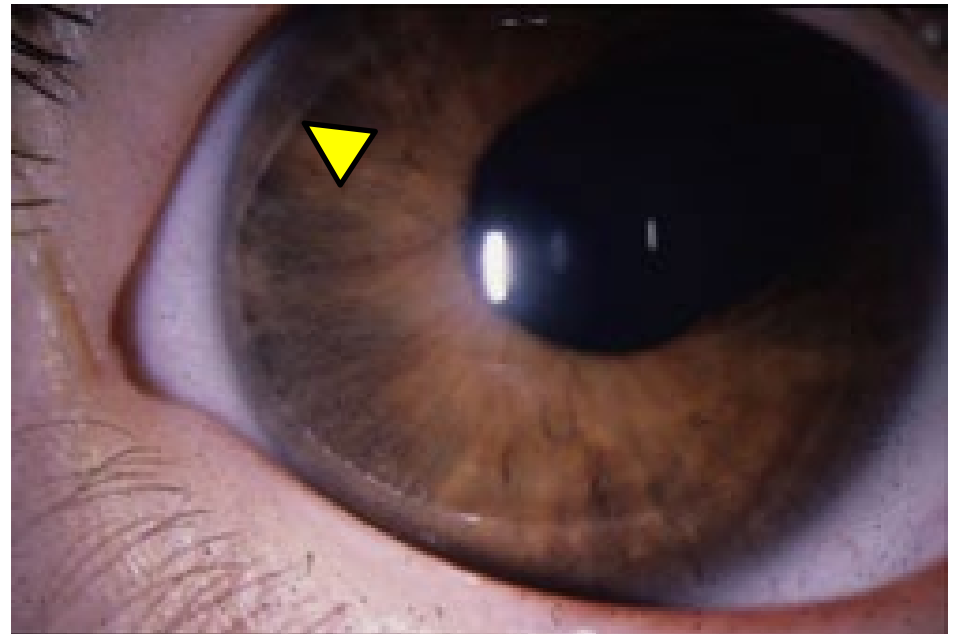
Is it normally apparent during slit-lamp examination?

No—it is usually too thin and posterior to be seen

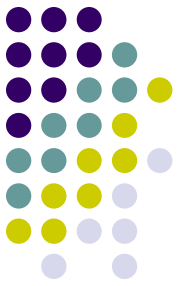
egmer



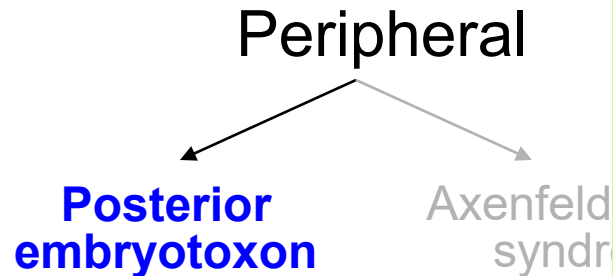
Posterior
embryotoxon



Anterior Segment Dysgenesis



Anterior segment dysgenesis



What is a posterior embryotoxon?

An anteriorly displaced and thickened **Schwalbe's line/ring**

What is Schwalbe's line/ring?

The edge or termination of Descemet's layer

Is it normally apparent during gonioscopy examination?

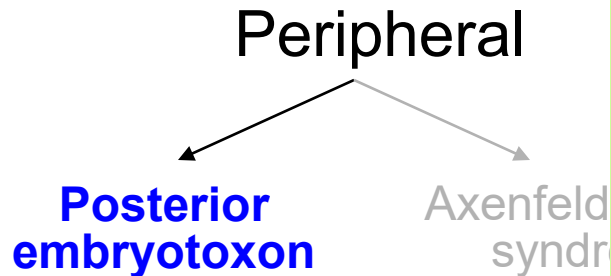
No—it is usually too thin and posterior to be seen

Why the 'line/ring' equivocation?

Anterior Segment Dysgenesis



Anterior segment dysgenesis



What is a posterior embryotoxon?

An anteriorly displaced and thickened **Schwalbe's line/ring**

What is Schwalbe's line/ring?

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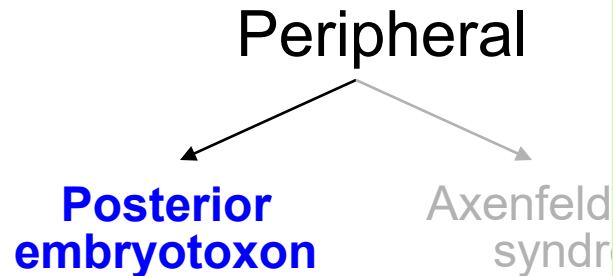
Why the 'line/ring' equivocation?

Most refer to it as Schwalbe's **line**, because that's what it looks like during gonioscopy. However, others point out that because this structure encircles the entire inner aspect of the cornea, it is more properly described as a **ring**.

Anterior Segment Dysgenesis



Anterior segment dysgenesis



What is a posterior embryotoxon?

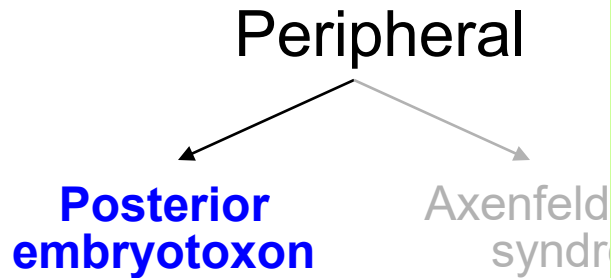
An anteriorly displaced and thickened Schwalbe's line/ring

Is it always a harbinger of significant pathology?

Anterior Segment Dysgenesis



Anterior segment dysgenesis



What is a posterior embryotoxon?

An anteriorly displaced and thickened Schwalbe's line/ring

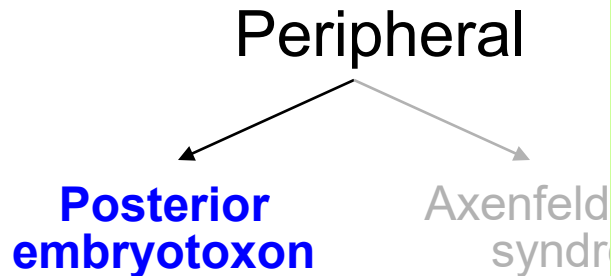
Is it always a harbinger of significant pathology?

No; it is found in about % of otherwise normal eyes

Anterior Segment Dysgenesis



Anterior segment dysgenesis



What is a posterior embryotoxon?

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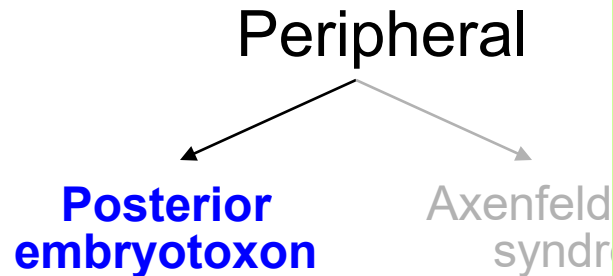
Is it always a harbinger of significant pathology?

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Anterior Segment Dysgenesis



Anterior segment dysgenesis



What is a posterior embryotoxon?

An anteriorly displaced and thickened Schwalbe's line/ring

Is it always a harbinger of significant pathology?

No; it is found in about 15% of otherwise normal eyes

In what three situations is it a significant finding?

1) ?

2) ?

3) ?

Anterior Segment Dysgenesis



Anterior segment dysgenesis

Peripheral

**Posterior
embryotoxon**

Axenfeld
syndr

*Interestingly, all three of these
begin with the letter 'A'*

What is a posterior embryotoxon?

An anteriorly displaced and thickened Schwalbe's line/ring

Is it always a harbinger of significant pathology?

No; it is found in about 15% of otherwise normal eyes

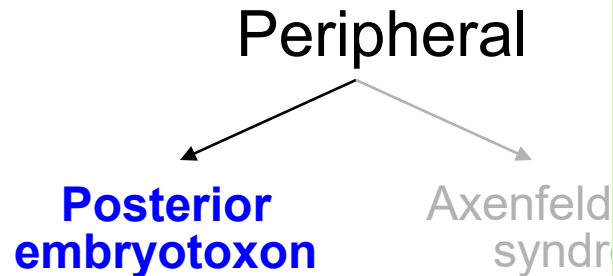
In what three situations is it a significant finding?

- 1) When it is part of the **eponym-eponym** **syndrome**
- 2) When it is associated with **not an eponym**
- 3) When it is associated with **eponym** **syndrome**

Anterior Segment Dysgenesis



Anterior segment dysgenesis



Interestingly, all three of these begin with the letter 'A'

What is a posterior embryotoxon?

An anteriorly displaced and thickened Schwalbe's line/ring

Is it always a harbinger of significant pathology?

No; it is found in about 15% of otherwise normal eyes

In what three situations is it a significant finding?

- 1) When it is part of the **Axenfeld-Rieger syndrome**
- 2) When it is associated with **aniridia**
- 3) When it is associated with **Alagille syndrome**

Anterior Segment Dysgenesis



Anterior segment dysgenesis

Why is the term 'aniridia' technically a misnomer?

embryotoxon?

ced and thickened Schwalbe's line/ring

nger of significant pathology?

out 15% of otherwise normal eyes

ons is it a significant finding?

of the **Axenveld-Rieger syndrome**

ated with **aniridia**

ated with **Alagille syndrome**

Anterior Segment Dysgenesis



Anterior segment dysgenesis

Why is the term 'aniridia' technically a misnomer?
Because a rudimentary iris root is always present

embryotoxon?

ced and thickened Schwalbe's line/ring

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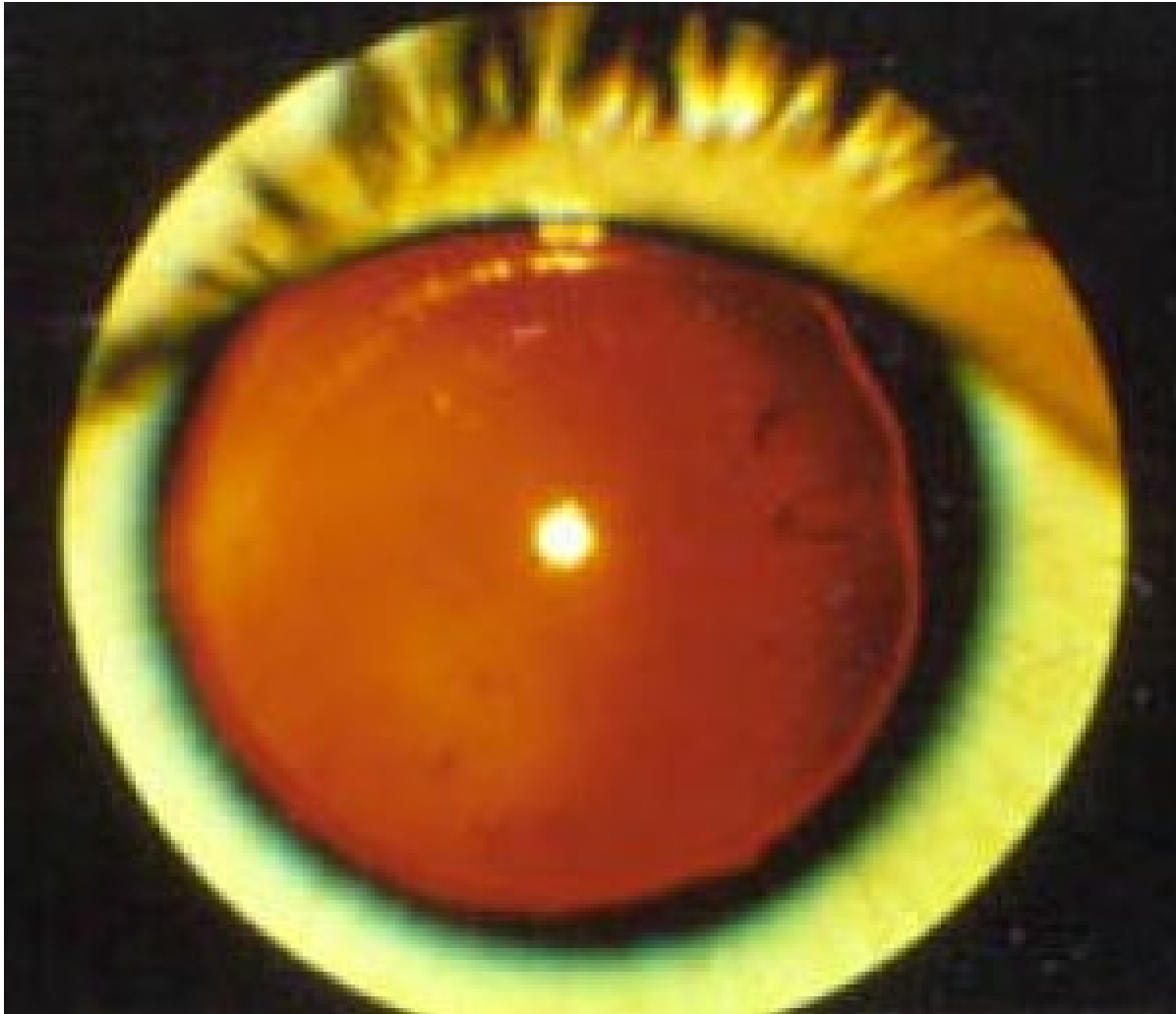
ons is it a significant finding?

of the **Axenfeld-Rieger syndrome**

ated with **aniridia**

ated with **Alagille syndrome**

Anterior Segment Dysgenesis



Aniridia. Note the presence of an iris stub/root

Anterior Segment Dysgenesis



Anterior segment dysgenesis

Why is the term 'aniridia' technically a misnomer?

Because a rudimentary iris root is always present

Is aniridia usually unilateral, or bilateral?

embryotoxon?

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Anterior Segment Dysgenesis



Anterior segment dysgenesis

Why is the term 'aniridia' technically a misnomer?

Because a rudimentary iris root is always present

Is aniridia usually unilateral, or bilateral?

It is almost always bilateral

embryotoxon?

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Is nystagmus commonly associated with aniridia?

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Anterior segment dysgenesis

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It is almost always bilateral

Is nystagmus commonly associated with aniridia?

Yes

***Mental note of aniridia's
ocular associations:***

--Nystagmus (more to follow)

embryotoxon?

ced and thickened Schwalbe's line/ring

nger of significant pathology?

out 15% of otherwise normal eyes

ons is it a significant finding?

of the **Axenfeld-Rieger syndrome**

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Anterior Segment Dysgenesis



Anterior segment dysgenesis

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It is almost always bilateral

*Is **nystagmus** commonly associated with aniridia?*

Is this a sensory or a motor nystagmus?

embryotoxon?

ced and thickened Schwalbe's line/ring

nger of significant pathology?

out 15% of otherwise normal eyes

ons is it a significant finding?

of the **Axenveld-Rieger syndrome**

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Anterior Segment Dysgenesis



Anterior segment dysgenesis

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Sensory

embryotoxon?

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out 15% of otherwise normal eyes

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Anterior Segment Dysgenesis



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Is this a sensory or a motor nystagmus?

Sensory

Is it a jerk, or a pendular nystagmus?

embryotoxon?

ced and thickened Schwalbe's line/ring

nger of significant pathology?

out 15% of otherwise normal eyes

ons is it a significant finding?

of the **Axenveld-Rieger syndrome**

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Anterior Segment Dysgenesis



Anterior segment dysgenesis

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Is this a sensory or a motor nystagmus?

Sensory

Is it a jerk, or a pendular nystagmus?

Pendular

embryotoxon?

ced and thickened Schwalbe's line/ring

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out 15% of otherwise normal eyes

ons is it a significant finding?

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ated with **aniridia**

ated with **Alagille syndrome**

Anterior Segment Dysgenesis



Anterior segment dysgenesis

Why is the term 'aniridia' technically a misnomer?

Because a rudimentary iris root is always present

Is aniridia usually unilateral, or bilateral?

It is almost always bilateral

Is nystagmus commonly associated with aniridia?

Is this a sensory or a motor nystagmus?

Sensory

What anatomic abnormalities are responsible for the poor vision in aniridia?

embryotoxon?

ced and thickened Schwalbe's line/ring

nger of significant pathology?

out 15% of otherwise normal eyes

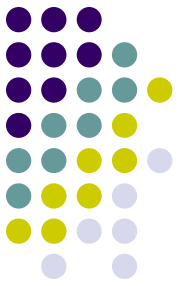
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Anterior Segment Dysgenesis



Anterior segment dysgenesis

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It is almost always bilateral

Is nystagmus commonly associated with aniridia?

Is this a sensory or a motor nystagmus?

Sensory

What anatomic abnormalities are responsible for the poor vision in aniridia?

Foveal and optic nerve hypoplasia

embryotoxon?

ced and thickened Schwalbe's line/ring

nger of significant pathology?

out 15% of otherwise normal eyes

ons is it a significant finding?

of the **Axenfeld-Rieger syndrome**

ated with **aniridia**

ated with **Alagille syndrome**

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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It is almost always bilateral

Is nystagmus commonly associated with aniridia?

Is this a sensory or a motor nystagmus?

Sensory

What anatomic abnormalities are responsible for the poor vision in aniridia?

Foveal and optic nerve hypoplasia

embryotoxon?

and thickened Schwalbe's line/ring

degree of significant pathology?

about 15% of otherwise normal eyes

In other words is it a significant finding?

of the **Axenfeld-Rieger syndrome**

associated with **aniridia**

associated with **Alagille syndrome**

Mental note of aniridia's ocular associations:

- Nystagmus
- Foveal hypoplasia
- ON hypoplasia *(more to follow)*

Anterior Segment Dysgenesis



Anterior segment dysgenesis

Why is the term 'aniridia' technically a misnomer?

Because a rudimentary iris root is always present

Is aniridia usually unilateral, or bilateral?

It is almost always bilateral

Is nystagmus commonly associated with aniridia?

Yes

With what developmental 'complex' is aniridia associated?

embryotoxon?

ced and thickened Schwalbe's line/ring

nger of significant pathology?

out 15% of otherwise normal eyes

ons is it a significant finding?

of the **Axenveld-Rieger syndrome**

ated with **aniridia**

ated with **Alagille syndrome**

Anterior Segment Dysgenesis



Anterior segment dysgenesis

Why is the term 'aniridia' technically a misnomer?

Because a rudimentary iris root is always present

Is aniridia usually unilateral, or bilateral?

It is almost always bilateral

Is nystagmus commonly associated with aniridia?

Yes

With what developmental 'complex' is aniridia associated?

The WAGR complex

embryotoxon?

ced and thickened Schwalbe's line/ring

nger of significant pathology?

out 15% of otherwise normal eyes

ons is it a significant finding?

of the **Axenveld-Rieger syndrome**

ated with **aniridia**

ated with **Alagille syndrome**

Anterior Segment Dysgenesis



Anterior segment dysgenesis

Why is the term 'aniridia' technically a misnomer?

Because a rudimentary iris root is always present

Is aniridia usually unilateral, or bilateral?

It is almost always bilateral

Is nystagmus commonly associated with aniridia?

Yes

With what developmental 'complex' is aniridia associated?

The WAGR complex

WAGR complex consists of:

W

A Aniridia

G

R

embryotoxon?

ced and thickened Schwalbe's line/ring

nger of significant pathology?

out 15% of otherwise normal eyes

ons is it a significant finding?

of the **Axenfled-Rieger syndrome**

ated with **aniridia**

ated with **Alagille syndrome**

Anterior Segment Dysgenesis



Anterior segment dysgenesis

Why is the term 'aniridia' technically a misnomer?

Because a rudimentary iris root is always present

Is aniridia usually unilateral, or bilateral?

It is almost always bilateral

Is nystagmus commonly associated with aniridia?

Yes

With what developmental 'complex' is aniridia associated?

The WAGR complex

WAGR complex consists of:

Wilms tumor

Aniridia

Genitourinary abnormalities

Retardation

embryotoxon?

ced and thickened Schwalbe's line/ring

nger of significant pathology?

out 15% of otherwise normal eyes

ons is it a significant finding?

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What is the noneponymous name for Wilms tumor (ie, what sort of tumor is it)?

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The WAGR complex

WAGR complex consists of:

Wilms tumor

What is the noneponymous name for Wilms tumor (ie, what sort of tumor is it)?

A nephroblastoma

Retardation

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WAGR complex: Wilm's tumor

Anterior Segment Dysgenesis



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familial

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Defects involving what gene are the cause of aniridia?

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The PAX6 gene

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I'm glad you asked...

embryotoxon?

ced and thickened Schwalbe's line/ring

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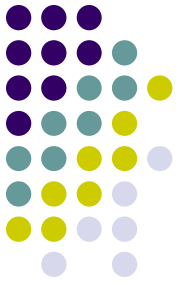
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ated with **Alagille syndrome**

Anterior Segment Dysgenesis



What four ocular abnormalities are attributed to the PAX6 gene?

P
A
X
6

There are four main abnormalities, and the term PAX6 acts as its own mnemonic. Start with the 'P' and make your way down...

Anterior Segment Dysgenesis



What four ocular abnormalities are attributed to the PAX6 gene?

Peters anomaly
A
X
6

Anterior Segment Dysgenesis



What four ocular abnormalities are attributed to the PAX6 gene?

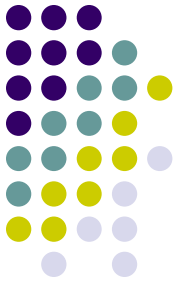
Peters anomaly

Anirida (duh)

X

6

Anterior Segment Dysgenesis



What four ocular abnormalities are attributed to the PAX6 gene?

Peters anomaly

Aniridia (duh)

Congenital cataract **X** (say it out loud)

6

Anterior Segment Dysgenesis



What four ocular abnormalities are attributed to the PAX6 gene?

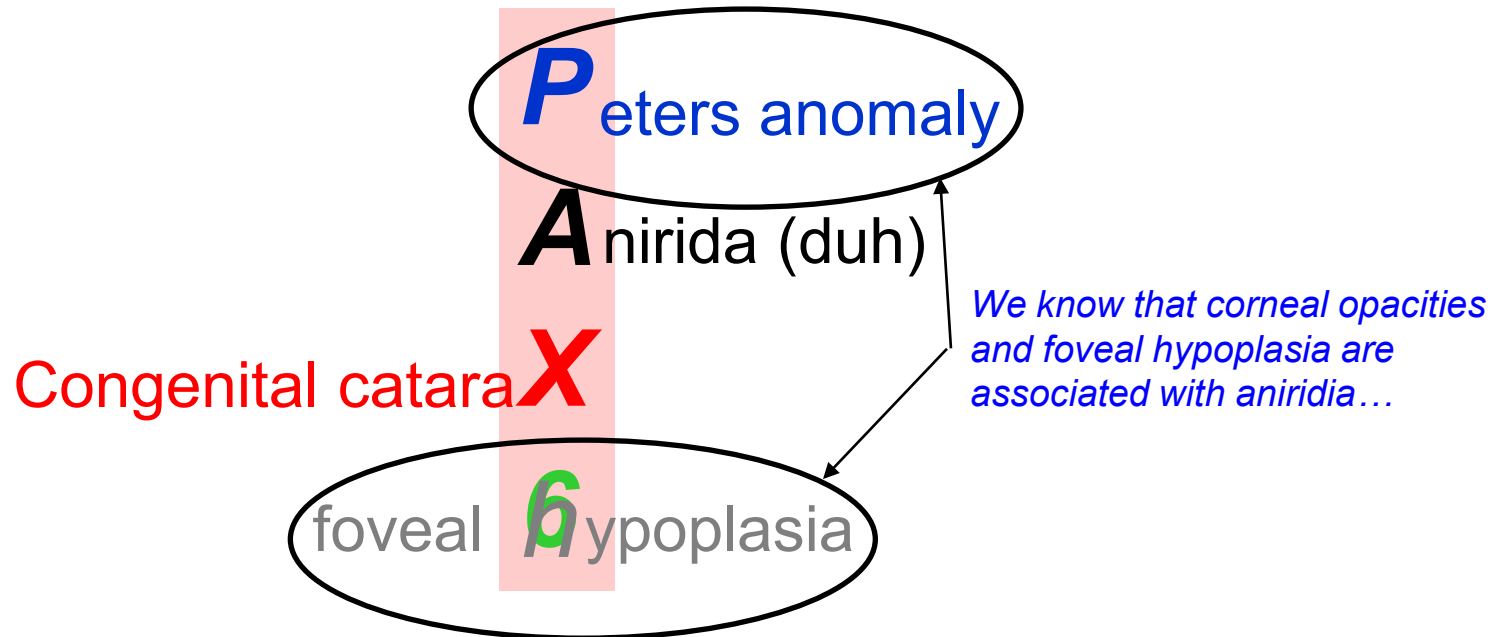
Peters anomaly
Anirida (duh)
Congenital cataract **X**
foveal **6** hypoplasia

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

Anterior Segment Dysgenesis



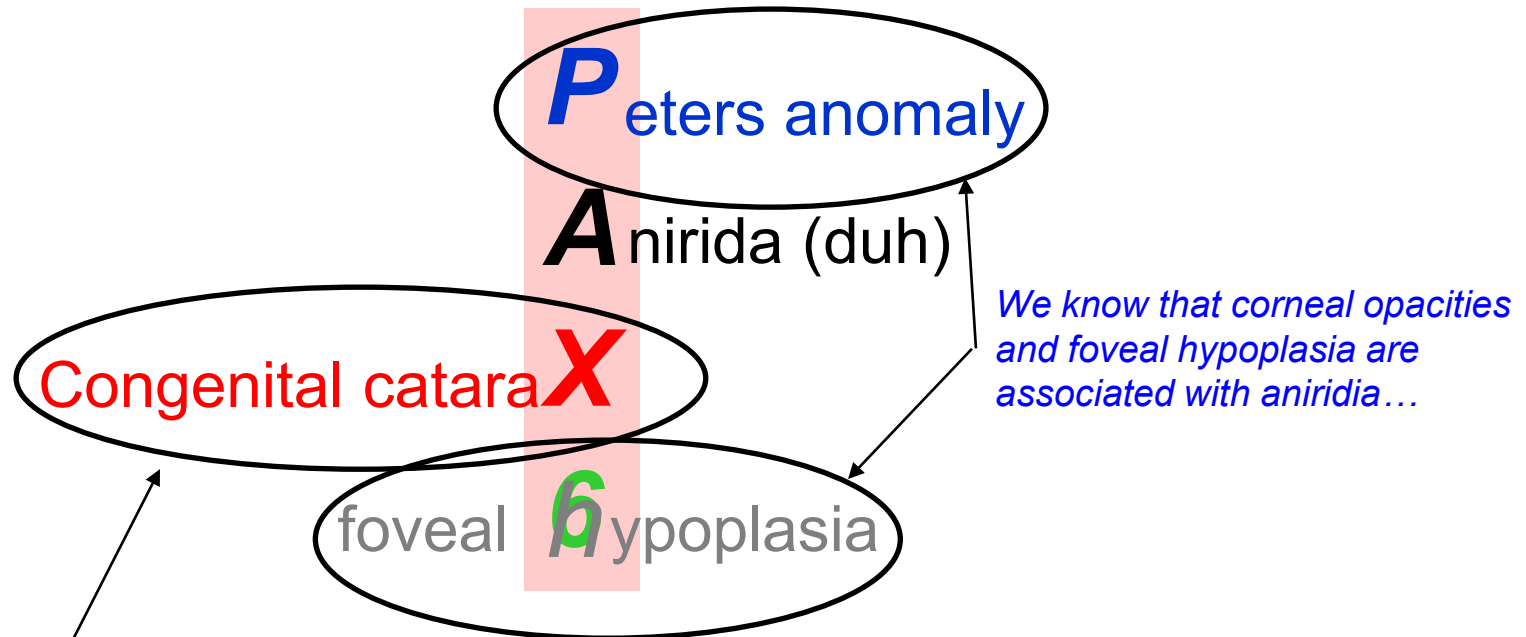
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Anterior Segment Dysgenesis



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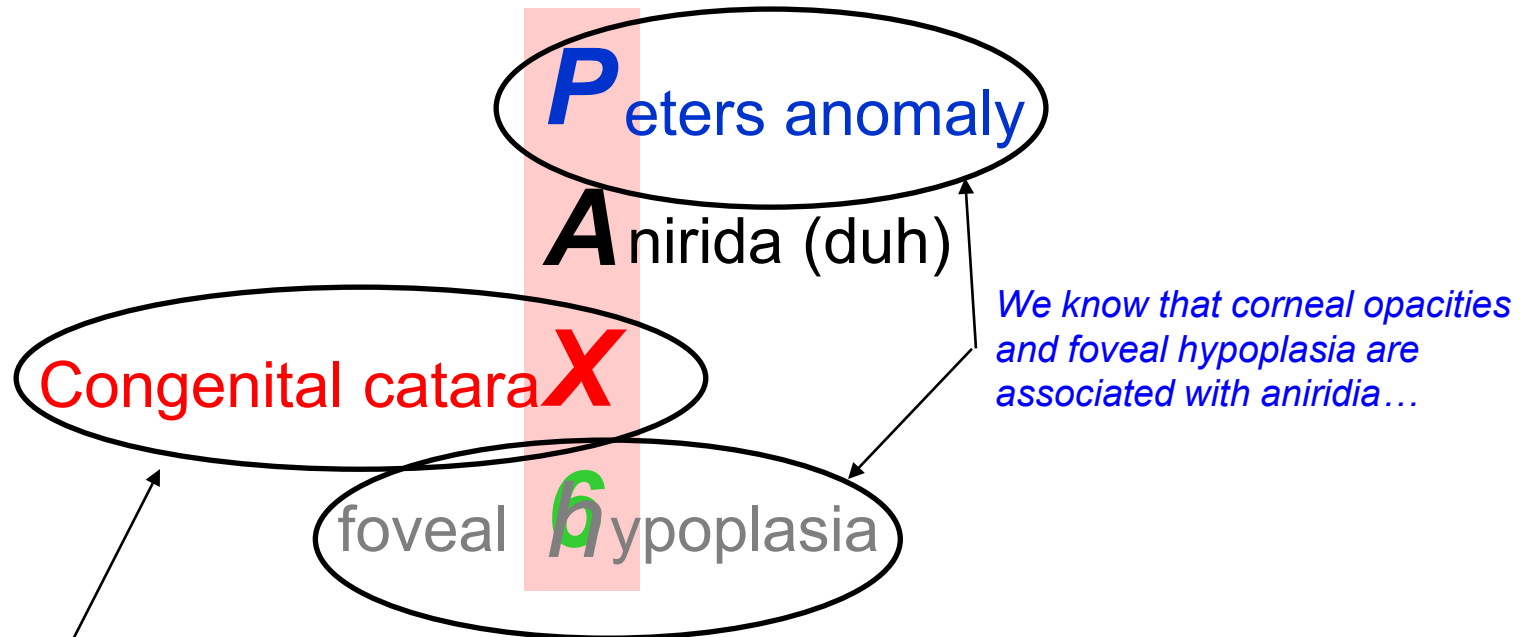


...but are cataracts associated with it as well?

Anterior Segment Dysgenesis



What four ocular abnormalities are attributed to the PAX6 gene?



*...but are cataracts associated with it as well?
Indeed they are*

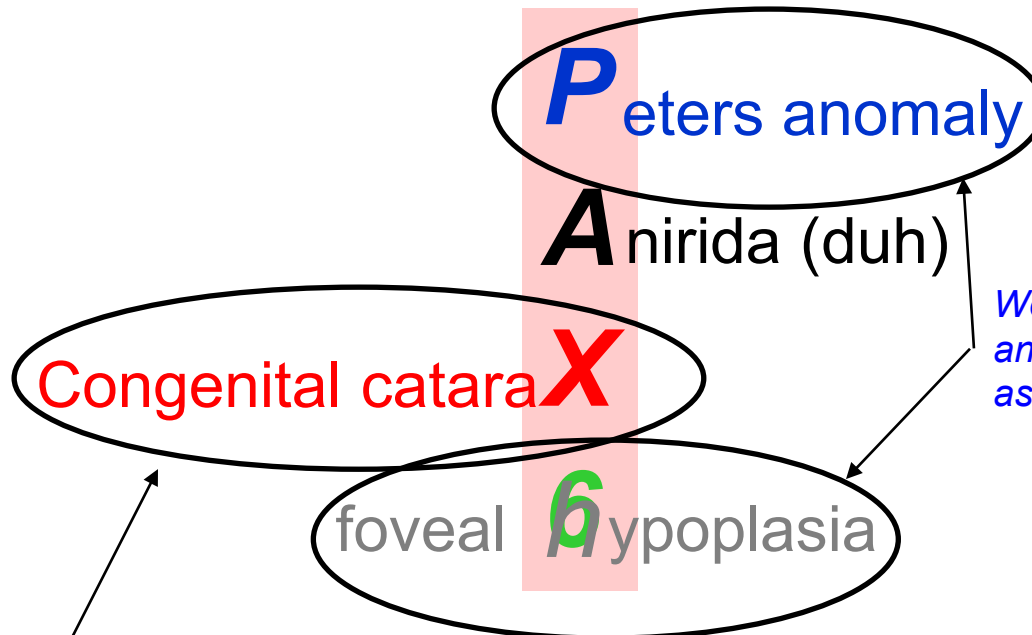
Anterior Segment Dysgenesis

*Mental note of aniridia's
ocular associations:*

- Nystagmus
- Foveal hypoplasia
- ON hypoplasia *(more to follow)*
- Peters anomaly
- Congenital cataracts

What four ocular ab

the PAX6 gene?



*We know that corneal opacities
and foveal hypoplasia are
associated with aniridia...*

*...but are **cataracts** associated with it as well?
Indeed they are*



What sort of gene is PAX6 anyway?

sis



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

Peters anomaly

Aniridia (duh)

Congenital cataract **X**

foveal **6** hypoplasia

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What sort of gene is PAX6 anyway?
A homeobox gene

sis



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

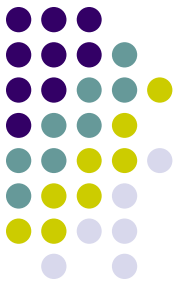
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What is a homeobox gene?



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Peters anomaly
Aniridia (duh)
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A homeobox gene

sis

What is a homeobox gene?
One that regulates morphogenesis



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

Peters anomaly
Aniridia (duh)
Congenital cataract **X**
foveal **6** hypoplasia

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

What sort of gene is PAX6 anyway?
A homeobox gene

sis



As the BCSC *Peds* book puts it, “The PAX6 gene is the master control gene for eye morphogenesis.”

One that regulates morphogenesis

What four ocular abnormalities are attributed to the PAX6 gene?

Peters anomaly
Aniridia (duh)
Congenital cataract
foveal hypoplasia

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

Anterior Segment Dysgenesis



*Why is **sporadic** aniridia associated with Wilms tumor, but not **familial** aniridia?*

Are all aniridia cases at risk for WAGR complex?

No, only those in which the genetic mutation is **sporadic**

Anterior Segment Dysgenesis



*Why is **sporadic** aniridia associated with Wilms tumor, but not **familial** aniridia?*

The PAX6 gene and the Wilms tumor gene (called) are adjacent to one another on chromosome .

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Anterior Segment Dysgenesis



*Why is **sporadic** aniridia associated with Wilms tumor, but not **familial** aniridia?*

The PAX6 gene and the Wilms tumor gene (called *WT1*) are adjacent to one another on chromosome 11p.

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Because of the PAX6/*WT1* spatial relationship, all infants presenting with sporadic aniridia **must** undergo genetic screening for the Wilms tumor defect.

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If a child tests positive for the Wilms tumor defect, how should they be screened for Wilms tumor?

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If a child tests positive for the Wilms tumor defect, how should they be screened for Wilms tumor?

Via periodic renal ultrasound

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If a child tests positive for the Wilms tumor defect, how should they be screened for Wilms tumor?

Via periodic renal ultrasound

How often, and for how long?

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If a child tests positive for the Wilms tumor defect, how should they be screened for Wilms tumor?

Via periodic renal ultrasound

How often, and for how long?

Every 3 months until age 7 years

Are all aniridia cases at risk for WAGR complex?

No, only those in which the genetic mutation is **sporadic**

Anterior Segment Dysgenesis



Nystagmus

***Mental note of aniridia's
ocular associations:***

- Nystagmus
- Foveal hypoplasia
- ON hypoplasia
- Peters anomaly
- Congenital cataracts
- ? *Two final aniridia associations*
- ? *we have yet to mention*

A corneal issue

Peters anomaly

foveal and optic nerve hypoplasia

*Angle-related
condition*

cataracts

Anterior Segment Dysgenesis



Nystagmus

*Mental note of aniridia's
ocular associations:*

- Nystagmus
- Foveal hypoplasia
- ON hypoplasia
- Peters anomaly
- Congenital cataracts
- Limbal stem-cell deficiency
- Glaucoma

limbal stem cell deficiency

Peters anomaly

foveal and optic nerve hypoplasia

glaucoma

cataracts

Anterior Segment Dysgenesis



Nystagmus

limbal stem cell deficiency

Peters anomaly

One final point regarding aniridia:

foveal and optic nerve hypoplasia

glaucoma

cataracts

Mental note of aniridia's ocular associations:

- Nystagmus
- Foveal hypoplasia
- ON hypoplasia
- Peters anomaly
- Congenital cataracts
- Limbal stem-cell deficiency
- Glaucoma

Anterior Segment Dysgenesis



Nystagmus

limbal stem cell deficiency

Peters anomaly

*Don't think of aniridia as just an iris condition! The BCSC refers to it as a **panophthalmic disorder***

One final point regarding aniridia:

foveal and optic nerve hypoplasia

glaucoma

cataracts

Anterior Segment Dysgenesis



Anterior segment dysgenesis

What is the noneponymous name of Alagille syndrome?

oxon?

thickened Schwalbe's line/ring

significant pathology?

of otherwise normal eyes

a significant finding?

enfeld-Rieger syndrome

n aniridia

n Alagille syndrome

Anterior Segment Dysgenesis



Anterior segment dysgenesis

What is the noneponymous name of Alagille syndrome?
Arterohepatic dysplasia

oxon?

thickened Schwalbe's line/ring

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h Alagille syndrome

Anterior Segment Dysgenesis



Anterior segment dysgenesis

What is the noneponymous name of Alagille syndrome?
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Anterior Segment Dysgenesis



Anterior segment dysgenesis

What is the noneponymous name of Alagille syndrome?
Arterohepatic dysplasia

How is it inherited?
Autosomal dominant, but the expressivity varies widely

oxon?
thickened Schwalbe's line/ring

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enfeld-Rieger syndrome
aniridia
Alagille syndrome

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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What is the classic presentation vis a vis us eye dentists?

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Alagille pts have a characteristic facial appearance—in a word, what is it?

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Alagille pts have a characteristic facial appearance—in a word, what is it?
'Triangular.' They have a broad forehead, and their face tapers to a pointy chin.

oxon?
thickened Schwalbe's line/ring

significant pathology?
of otherwise normal eyes

a significant finding?
enfeld-Rieger syndrome
aniridia

Alagille syndrome

Anterior Segment Dysgenesis



Alagille syndrome: Facies

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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In addition to liver, eye and face findings, what other organs are commonly affected?

--?

--?

oxon?

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enfeld-Rieger syndrome

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In addition to liver, eye and face findings, what other organs are commonly affected?

- The heart
- The skeleton

oxon?

thickened Schwalbe's line/ring

significant pathology?

of otherwise normal eyes

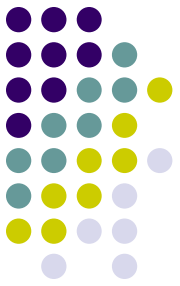
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enfeld-Rieger syndrome

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Anterior Segment Dysgenesis



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--The heart: ?

--The skeleton

oxon?

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In addition to liver, eye and face findings, what other organs are commonly affected? How are they affected?

--The heart: Septal defects, PDA, and tetralogy of Fallot are common

--The skeleton: ?

oxon?

thickened Schwalbe's line/ring

significant pathology?

of otherwise normal eyes

a significant finding?

enfeld-Rieger syndrome

aniridia

Alagille syndrome

Anterior Segment Dysgenesis



Anterior segment dysgenesis

What is the noneponymous name of Alagille syndrome?
Arterohepatic dysplasia

How is it inherited?
Autosomal dominant, but the expressivity varies widely

What is the classic presentation vis a vis us eye dentists?
An infant with jaundice who presents to the eye service as a 'rule out Alagille syndrome' consult

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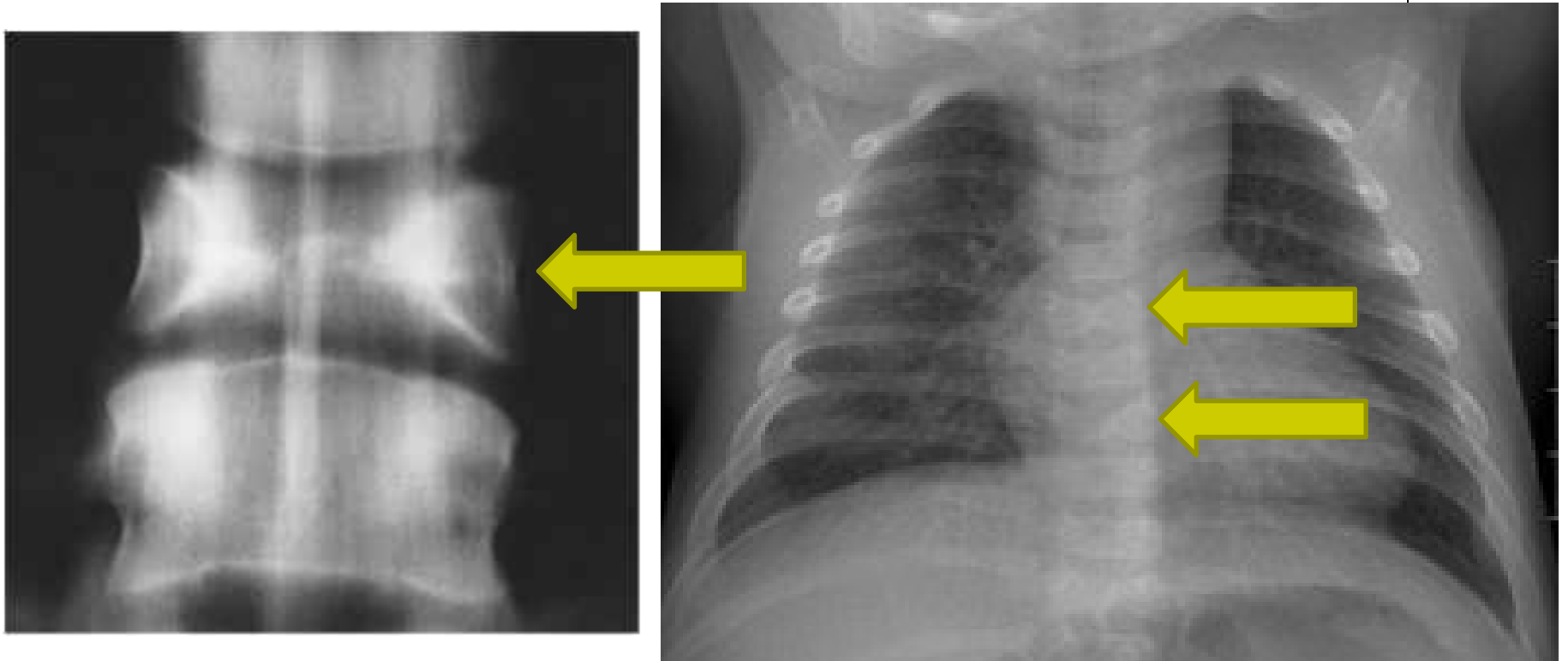
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Alagille syndrome

Anterior Segment Dysgenesis



Alagille syndrome: Butterfly vertebrae

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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Anterior Segment Dysgenesis



Anterior segment dysgenesis

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Proximal?

thickened Schwalbe's line/ring

What is the most common ocular pathology?

Otherwise normal eyes

What is the most significant finding?

Rieger syndrome

Sturge-Weber syndrome

Alagille syndrome

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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Another syndrome of ophthalmic concern includes butterfly vertebrae as a finding. What is it?
Goldenhar syndrome

In two words, what sort of condition is Goldenhar?
A

What is the classic ophthalmic finding?
Thickened Schwalbe's line/ring

What is the classic systemic pathology?
Otherwise normal eyes

What is the classic systemic finding?
Rieger syndrome
Goldenhar syndrome

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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Stemoxon?

thickened Schwalbe's line/ring

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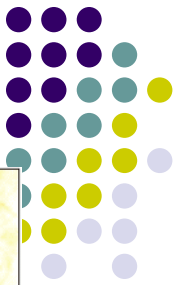
What is the noneponymous name for Goldenhar syndrome?
Oculo-auricular-**vertebral** (OAV) syndrome

What is the classic ophthalmic finding?
Thickened Schwalbe's line/ring

What is the classic systemic pathology?
Otherwise normal eyes

What is the classic finding?
Rieger syndrome
Chandrasekhar syndrome
Alagille syndrome

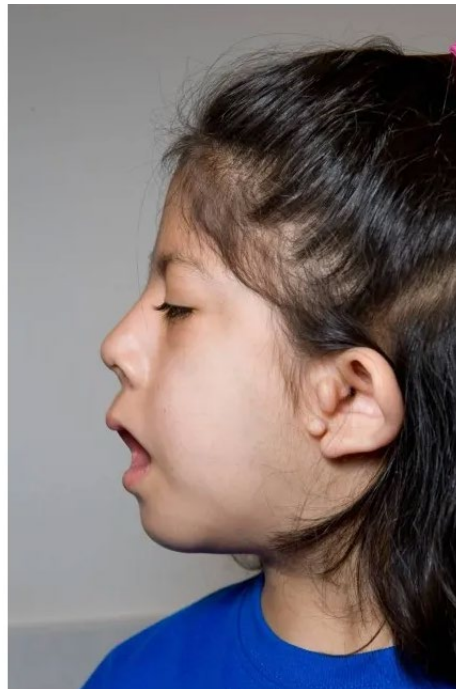
Anterior Segment Dysgenesis



Goldenhar: Limbal (epibulbar) dermoids; lid coloboma (OCULO-auriculo-vertebral syndrome)

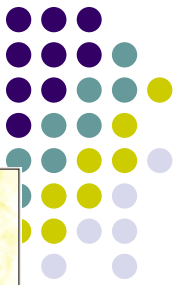


Goldenhar: Ear abnormalities (Oculo-AURICULO-vertebral syndrome)



Goldenhar syndrome: Hemifacial microsomia

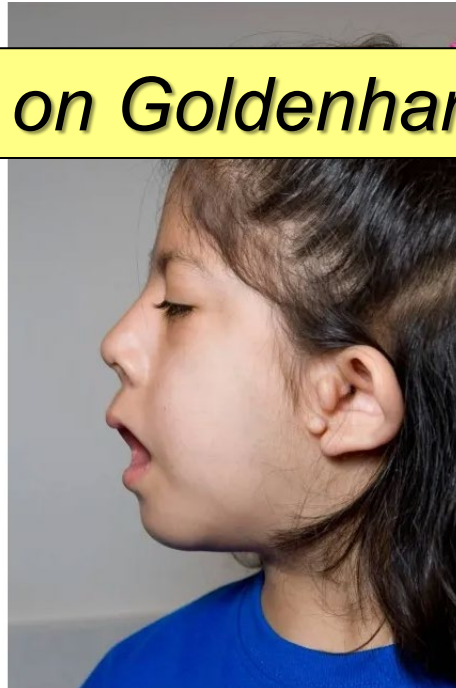
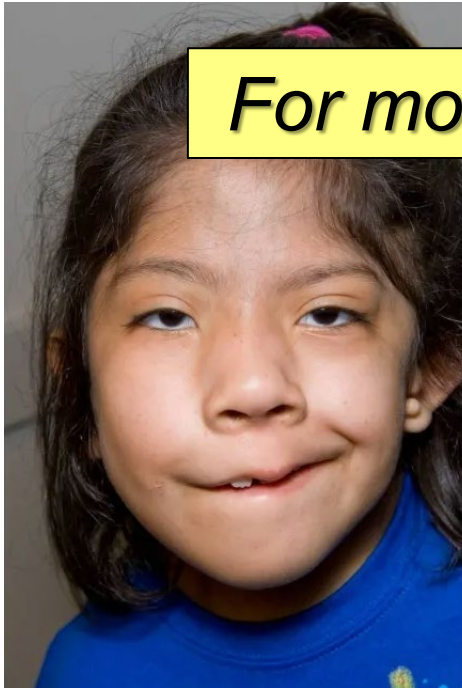
Anterior Segment Dysgenesis



Goldenhar: Limbal (epibulbar) dermoids; lid coloboma (OCULO-auriculo-vertebral syndrome)



For more on Goldenhar, see slide-set P22



Goldenhar syndrome: Hemifacial microsomia

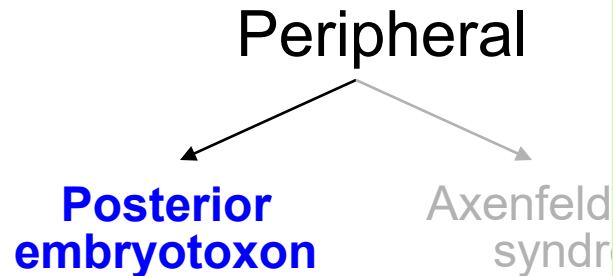


Goldenhar: Ear abnormalities (Oculo-AURICULO-vertebral syndrome)

Anterior Segment Dysgenesis



Anterior segment dysgenesis



What is a posterior embryotoxon?

An anteriorly displaced and thickened Schwalbe's line/ring

Is it always a harbinger of significant pathology?

No; it is found in about 15% of otherwise normal eyes

In what three situations is it a significant finding?

- 1) When it is part of the **Axenfeld-Rieger syndrome**
- 2) When it is associated with **aniridia**
- 3) When it is associated with **Alagille syndrome**

*Is there such a thing as an **anterior** embryotoxon?*

*anterior
embryotoxon*

Anterior Segment Dysgenesis



Anterior segment dysgenesis

Peripheral

Posterior
embryotoxon

Axenfeld
syndr

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Yes--it is another name for arcus

Arcus juvenilis
aka
*anterior
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Anterior Segment Dysgenesis



Anterior segment dysgenesis

Peripheral

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Anterior Segment Dysgenesis



Anterior segment dysgenesis

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Arcus juvenilis
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What is arcus juvenilis?

Anterior Segment Dysgenesis



Anterior segment dysgenesis

Peripheral

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Axenfeld
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Corneal arcus

Arcus juvenilis
aka
anterior
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Arcus senilis

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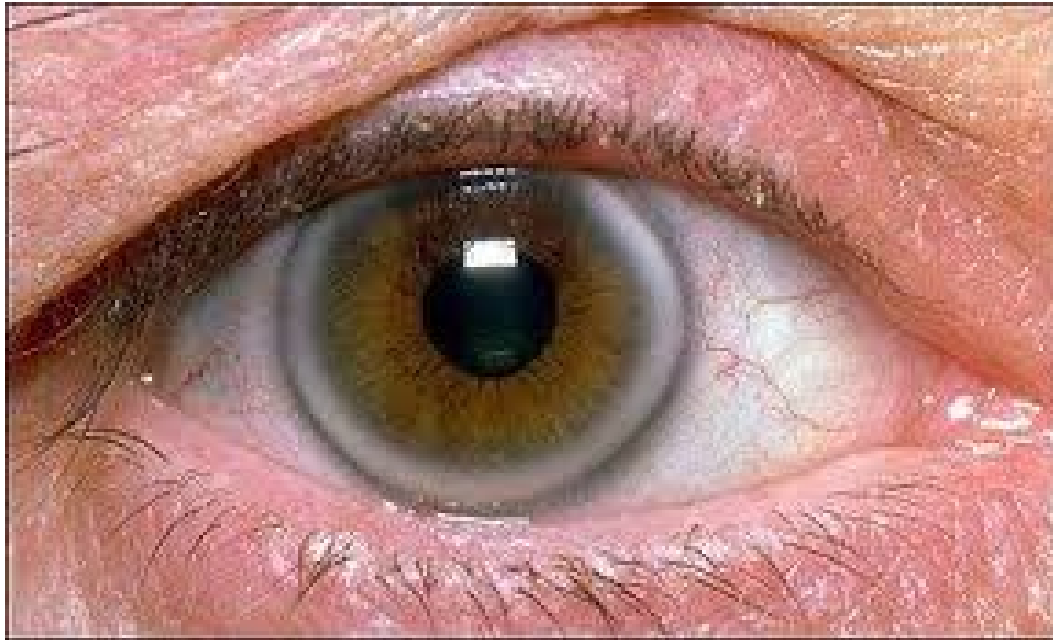
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Yes--it is another name for **arcus juvenilis**

What is arcus juvenilis?

It is the congenital version of **arcus senilis**

Anterior Segment Dysgenesis



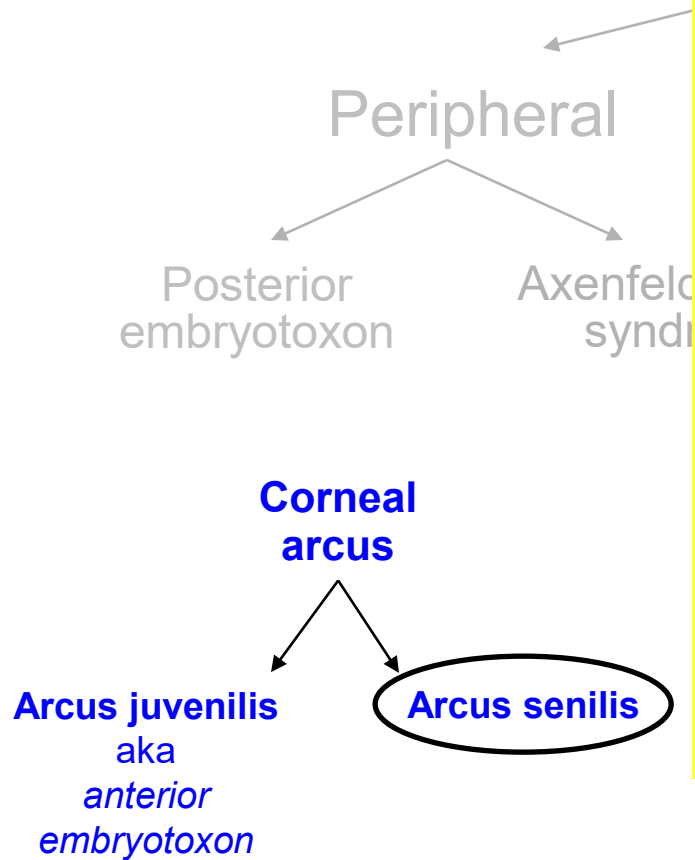
Arcus senilis

Anterior Segment Dysgenesis



Regarding arcus senilis...

What is its main chemical component?



What is arcus juvenilis?

It is the congenital version of **arcus senilis**

Anterior Segment Dysgenesis



Regarding arcus senilis...

What is its main chemical component?
Cholesterol

Peripheral

Posterior
embryotoxon

Axenfeld
syndrome

Corneal
arcus

Arcus juvenilis
aka
anterior
embryotoxon

Arcus senilis

What is arcus juvenilis?

It is the congenital version of **arcus senilis**

Anterior Segment Dysgenesis



A

Regarding arcus senilis...

What is its main chemical component?

Cholesterol

Is it usually unilateral, or bilateral?

Peripheral

Posterior
embryotoxon

Axenfeld
syndrome

Corneal
arcus

Arcus juvenilis
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Anterior Segment Dysgenesis



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Arcus senilis

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Anterior Segment Dysgenesis



A

Regarding arcus senilis...

What is its main chemical component?

Cholesterol

Is it usually **unilateral**, or bilateral?

Bilateral

Periph

What condition should be suspected if a pt has unilateral arcus?

Posterior
embryotoxon

Axenfeld
syndr

Corneal
arcus

Arcus juvenilis
aka
anterior
embryotoxon

Arcus senilis

What is arcus juvenilis?

It is the congenital version of **arcus senilis**

Anterior Segment Dysgenesis



A Regarding arcus senilis...

What is its main chemical component?
Cholesterol

Is it usually **unilateral**, or bilateral?
Bilateral

Periph What condition should be suspected if a pt has **unilateral arcus**?
Carotid occlusive dz, or ocular ischemic syndrome (OIS)

Posterior
embryotoxon

Axenfeld
syndr

**Corneal
arcus**

Arcus juvenilis
aka
anterior
embryotoxon

Arcus senilis

What is arcus juvenilis?
It is the congenital version of **arcus senilis**

Anterior Segment Dysgenesis



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What is its main chemical component?
Cholesterol

Is it usually **unilateral**, or bilateral?
Bilateral

Periph

What condition should be suspected if a pt has unilateral arcus?
Carotid occlusive dz, or ocular ischemic syndrome (OIS)

Posterior
embryotoxon

Axial
s

If arcus is a sign of carotid occlusion or OIS, which
side is occluded/ischemic--the side **with** the arcus,
or the side **without** the arcus?

**Corneal
arcus**



Arcus juvenilis
aka
anterior
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Arcus senilis

What is arcus juvenilis?

It is the congenital version of **arcus senilis**

Anterior Segment Dysgenesis



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What is its main chemical component?
Cholesterol

Is it usually **unilateral**, or bilateral?
Bilateral

Periph

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**Corneal
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Anterior Segment Dysgenesis



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Is it usually unilateral, or bilateral?

Bilateral

Is it a dystrophy?

Peripheral

Posterior
embryotoxon

Axenfeld
syndrome

Corneal
arcus

Arcus juvenilis
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Arcus senilis

What is arcus juvenilis?

It is the congenital version of **arcus senilis**

Anterior Segment Dysgenesis



A

Regarding arcus senilis...

What is its main chemical component?

Cholesterol

Is it usually unilateral, or bilateral?

Bilateral

Is it a dystrophy?

No; per the *Cornea* book, it is an “involutional change”

Peripheral

Posterior
embryotoxon

Axenfeld
syndrome

**Corneal
arcus**

Arcus juvenilis
aka
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Arcus senilis

What is arcus juvenilis?

It is the congenital version of **arcus senilis**

Anterior Segment Dysgenesis



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Regarding arcus senilis...

What is its main chemical component?

Cholesterol

Is it usually unilateral, or bilateral?

Bilateral

Is it a dystrophy?

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In what pattern does it typically declare itself?

Peripheral

Posterior
embryotoxon

Axenfeld
syndrome

Corneal
arcus

Arcus juvenilis
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Arcus senilis

What is arcus juvenilis?

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Anterior Segment Dysgenesis



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What is its main chemical component?

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In what pattern does it typically declare itself?

It starts at the poles, then spreads circumferentially

Peripheral

Posterior
embryotoxon

Axenfeld
syndrome

Corneal
arcus

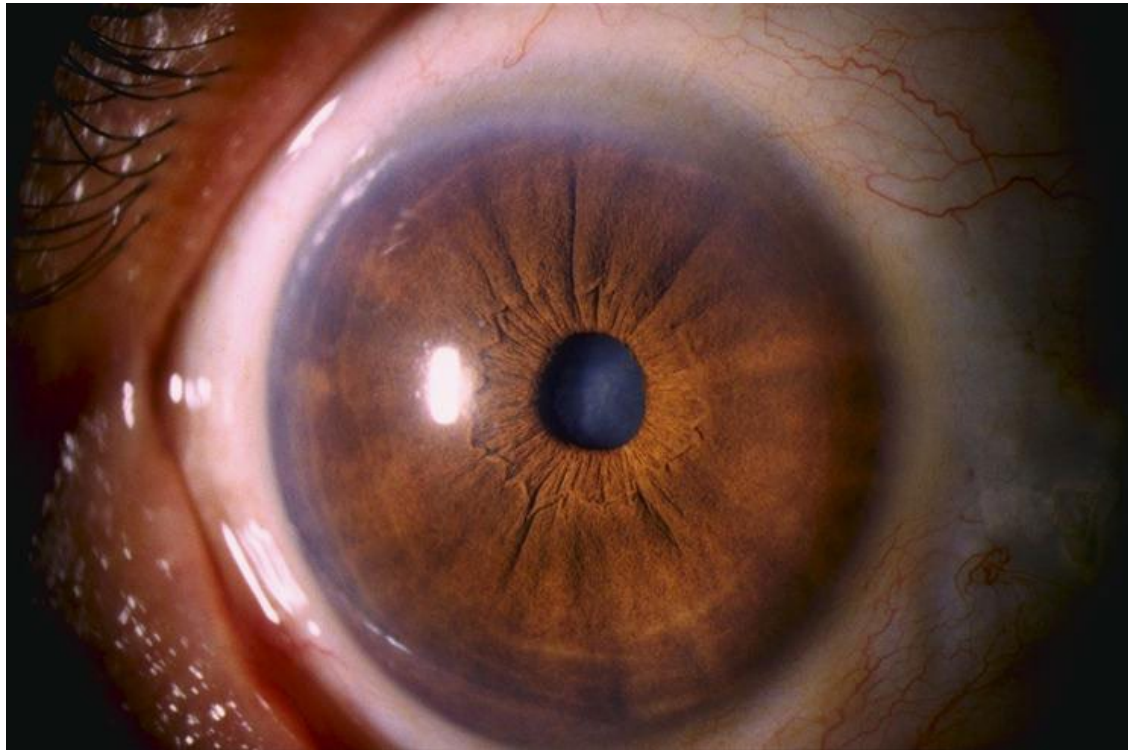
Arcus juvenilis
aka
anterior
embryotoxon

Arcus senilis

What is arcus juvenilis?

It is the congenital version of **arcus senilis**

Anterior Segment Dysgenesis



Early arcus senilis

Anterior Segment Dysgenesis



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Cholesterol

Is it usually unilateral, or bilateral?

Bilateral

Is it a dystrophy?

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In what pattern does it typically declare itself?

It starts at the poles, then spreads circumferentially

Does it exhibit a gender predilection?

Peripheral

Posterior
embryotoxon

Axenfeld
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Corneal
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What is arcus juvenilis?

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Anterior Segment Dysgenesis



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In what pattern does it typically declare itself?

It starts at the poles, then spreads circumferentially

Does it exhibit a gender predilection?

Yes, are more likely to develop it

Peripheral

Posterior
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Axenfeld
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Corneal
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In what pattern does it typically declare itself?

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Does it exhibit a gender predilection?

Yes, men are more likely to develop it

Peripheral

Posterior
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Axenfeld
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Anterior Segment Dysgenesis



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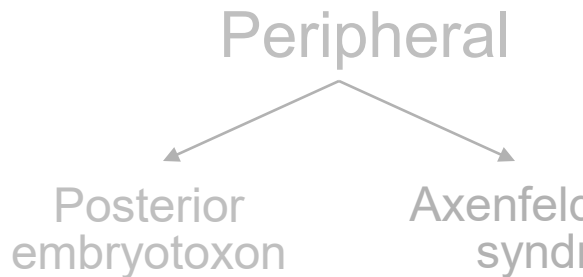
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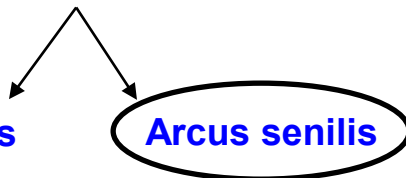
Does it exhibit a gender predilection?

Yes, men are more likely to develop it

Does it exhibit a racial predilection?



Corneal arcus



Arcus juvenilis
aka
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Arcus senilis

What is arcus juvenilis?

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Anterior Segment Dysgenesis



A

Regarding arcus senilis...

What is its main chemical component?

Cholesterol

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Is it a dystrophy?

No; per the *Cornea* book, it is an “involutional change”

In what pattern does it typically declare itself?

It starts at the poles, then spreads circumferentially

Does it exhibit a gender predilection?

Yes, men are more likely to develop it

Does it exhibit a racial predilection?

Yes, it is more common in [redacted]

Peripheral

Posterior
embryotoxon

Axenfeld
syndrome

Corneal
arcus

Arcus juvenilis
aka
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Arcus senilis

What is arcus juvenilis?

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In what pattern does it typically declare itself?

It starts at the poles, then spreads circumferentially

Does it exhibit a gender predilection?

Yes, men are more likely to develop it

Does it exhibit a racial predilection?

Yes, it is more common in AAs

Peripheral

Posterior
embryotoxon

Axenfeld
syndrome

Corneal
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Arcus juvenilis
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Arcus senilis

What is arcus juvenilis?

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Anterior Segment Dysgenesis



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Regarding arcus senilis...

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Is it usually unilateral, or bilateral?
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In what pattern does it typically declare itself?
It starts at the poles, then spreads circumferentially

Does it exhibit a gender predilection?
Yes, men are more likely to develop it

Does it exhibit a racial predilection?
Yes, it is more common in AAs

Does its prevalence increase with age?

Peripheral

Posterior
embryotoxon

Axenfeld
syndrome

Corneal
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In what pattern does it typically declare itself?

It starts at the poles, then spreads circumferentially

Does it exhibit a gender predilection?

Yes, men are more likely to develop it

Does it exhibit a racial predilection?

Yes, it is more common in AAs

Does its prevalence increase with age?

Yes; after age 50, the prevalence is ~100%

What is arcus juvenilis?

It is the congenital version of **arcus senilis**

Peripheral

Posterior
embryotoxon

Axenfeld
syndrome

**Corneal
arcus**

Arcus juvenilis
aka
anterior
embryotoxon

Arcus senilis

Anterior Segment Dysgenesis



A

Regarding arcus senilis...

What is its main chemical component?

Cholesterol

Is it usually unilateral, or bilateral?

Bilateral

Is it a dystrophy?

No; per the *Cornea* book, it is an “involutional change”

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aka
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Arcus senilis

's line/ring

eyes

ome

n?

Anterior Segment Dysgenesis



Arcus senilis in older AAM

Anterior Segment Dysgenesis



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No, it is a benign finding

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Post
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Necrobiotic xanthogranuloma

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Necrobiotic xanthogranuloma



Adult-onset asthma with periocular xanthogranuloma

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The 'adult xanthogranulomatous diseases'

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What is the fourth adult xanthogranulomatous disease? (with periorcular xanthogranuloma process)? Erdheim-Chester dz

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It is called simply *adult-onset xanthogranuloma*, and presents like the 'asthma' type (but without the asthma, duh)

with periocular xanthogranuloma (process)? Erdheim-Chester dz

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The 'adult xanthogranulomatous diseases'

In three words, how would you characterize these conditions?

They are non-Langerhans cell histiocytoses

Are they composed of lipid?

Sort of, but more specifically, they are composed of lipid-filled macrophages

Are they always a harbinger of elevated serum lipids?

No, they can (and often do) appear in individuals with normal lipid panels

Can they be congenital?

Yes, and when they are, they usually are a sign of lipid derangement

often present in such pts. What is it?

Xanthelasma

Does its prevalence increase with age?

Yes; after age 80, the prevalence is ~100%
before age 40

What is arcus juvenilis?

It is the congenital version of **arcus senilis**

Arcus juvenilis
aka
anterior
embryotoxon

Arcus senilis

Speaking of 'xanthelasmas'...What dz comes to mind if an adult presents with what appear to be xanthelasmas, but:

The xanthelasmas are ulcerated? Necrobiotic xanthogranuloma

The pt c/o adult-onset asthma? Adult-onset asthma with periorcular xanthogranuloma

The pt is severely systemically ill (by the same dz process)? Erdheim-Chester dz

The abd

The 'ad

**For more on the adult-onset
xanthogranulomas, see slide-set K20**

In three words, how would you characterize these conditions?

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Anterior Segment Dysgenesis



A Regarding arcus senilis...

What are xanthelasmas, that is, what is their clinical appearance?

They are yellowish plaques located in the medial canthal region, usually on the upper lids

Do they present unilaterally, or bilaterally?

Bilaterally

Are they composed of lipid?

Sort of, but more specifically, they are composed of **lipid-filled macrophages**

Are they always a harbinger of elevated serum lipids?

Speaking of 'foamy macrophages' part deux...

What does come to mind if, instead of an adult

with xanthelasmas, the pt in question was a

very young child with unilateral pigmented iris nodules? ← First clue...

Arcus juven
aka
anterior
embryotoxon

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Anterior Segment Dysgenesis



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Speaking of 'foamy macrophages' part deux...

What does come to mind if, instead of an adult with xanthelasmas, the pt in question was a very young child with unilateral pigmented iris nodules?

And heterochromia iridis secondary to those nodules? ← Second clue

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aka
anterior
embryotoxon

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Anterior Segment Dysgenesis



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And heterochromia iridis secondary to those nodules?

Along with a nontraumatic hyphema in the affected eye? ← Clue #3

Arcus juvenilis
aka
anterior
embryotoxon

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Anterior Segment Dysgenesis



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And heterochromia iridis secondary to those nodules?

Along with a nontraumatic hyphema in the affected eye?

Associated with orangish skin papules? ← Last clue--the answer is next

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Arcus juvenilis
aka
anterior
embryotoxon

Anterior Segment Dysgenesis



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Along with a nontraumatic hyphema in the affected eye?

Associated with orangish skin papules?

Juvenile xanthogranuloma (JXG)

What is arcus juvenilis?

It is the congenital version of **arcus senilis**

Arcus juve
aka
anterior
embryotoxon

Anterior Segment Dysgenesis

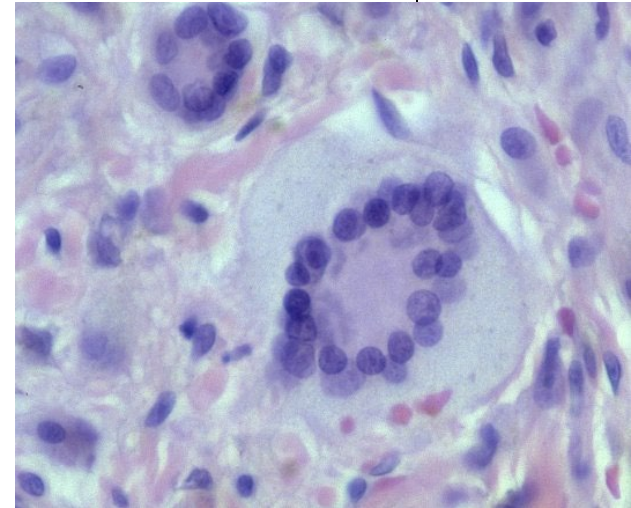


Skin papules. The orange color is classic

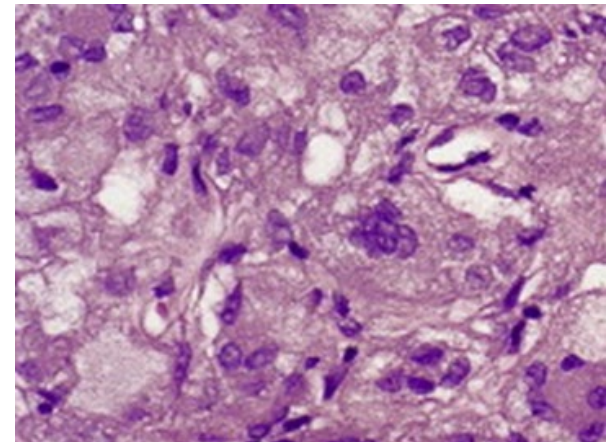


Spontaneous hyphema

JXG

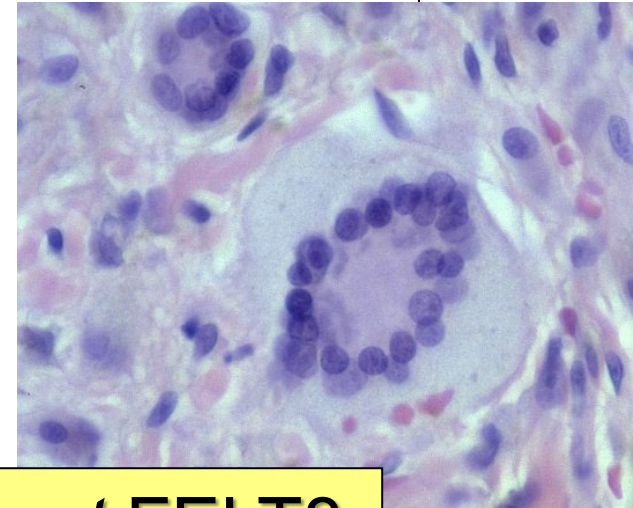
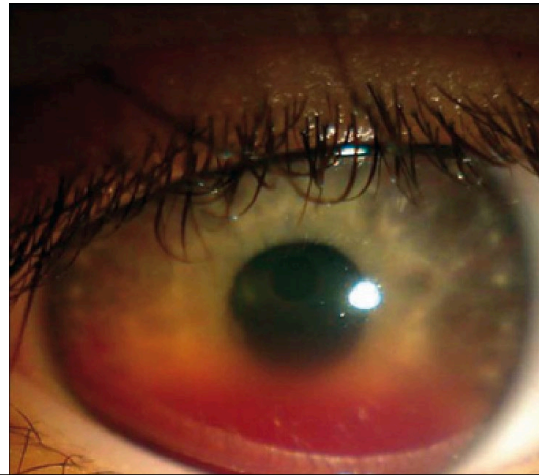


Touton giant cells



Foamy macrophages

Anterior Segment Dysgenesis

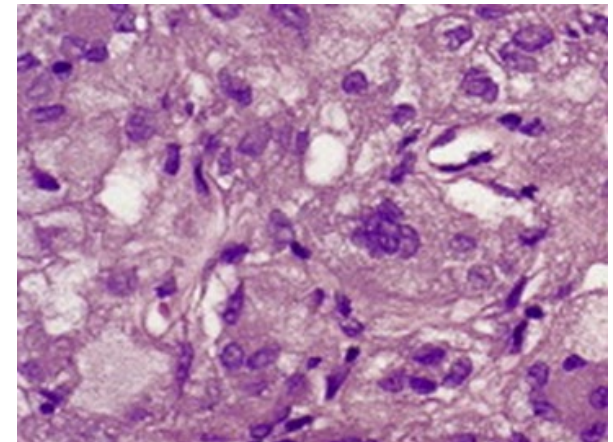


For more on JXG, see slide-set FELT8

cells

Spontaneous hyphema

JXG



Foamy macrophages



Skin papules. The orange color is classic

Anterior Segment Dysgenesis



Anterior segment dysgenesis

Peripheral

Posterior
embryotoxon

Axenfeld
syndr

Corneal arcus

Arcus juvenilis
aka
anterior
embryotoxon

Arcus senilis
aka
?

What is a posterior embryotoxon?

An anteriorly displaced and thickened Schwalbe's line/ring

Is it always a harbinger of significant pathology?

No; it is found in about 15% of otherwise normal eyes

In what three situations is it a significant finding?

- 1) When it is part of the **Axenfeld-Rieger syndrome**
- 2) When it is associated with **aniridia**
- 3) When it is associated with **Alagille syndrome**

*Finally: Anterior embryotoxon is another name for arcus juvenilis.
Is there an equivalent 'another name' for arcus senilis?*

Anterior Segment Dysgenesis



Anterior segment dysgenesis

Peripheral

Posterior
embryotoxon

Axenfeld
syndr

Corneal arcus

Arcus juvenilis
aka
anterior
embryotoxon

Arcus senilis
aka
anterior
gerontoxon

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Yes—anterior **gerontoxon**

Anterior Segment Dysgenesis



Anterior segment dysgenesis

Peripheral

Posterior
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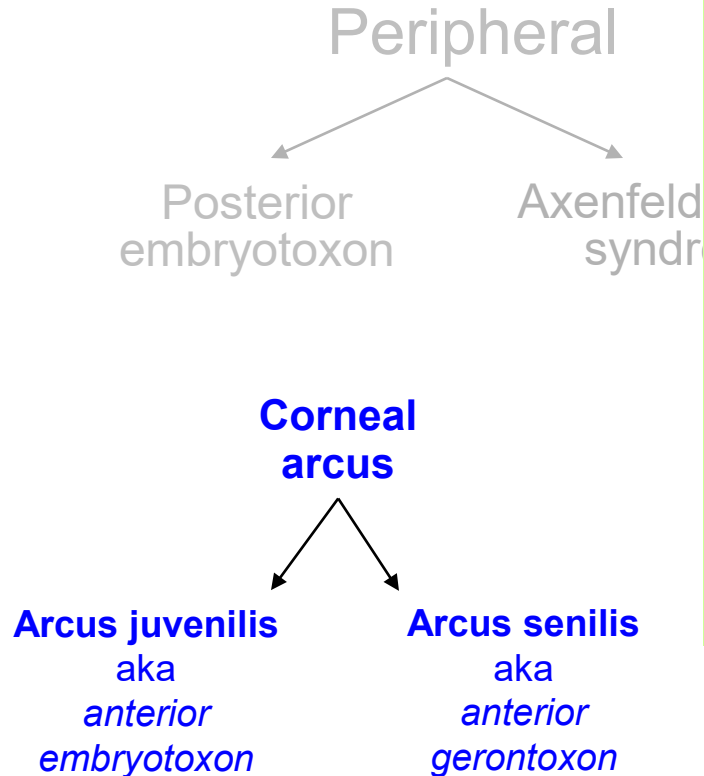
Yes—**anterior gerontoxon**

Gerontoxon?

Anterior Segment Dysgenesis



Anterior segment dysgenesis



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Finally: Anterior embryotoxon is another name for arcus juvenilis.

Is there an equivalent 'another name' for arcus senilis?

Yes—**anterior gerontoxon**

Gerontoxon?

Yeah, 'geron-' as in *gerontology*, as in old folks

Anterior Segment Dysgenesis



Anterior segment dysgenesis

	Embryotoxon	Gerontoxon
Anterior	Arcus juvenilis	Arcus senilis
Posterior	Posterior embryotoxon	?

To complete the matrix...There's an anterior embryotoxon,
a posterior embryotoxon, and an anterior gerontoxon.
That just leaves posterior gerontoxon. Is it a thing?

Arcus juvenilis
aka
anterior
embryotoxon

gerontoxon

Gerontoxon?

Yeah, 'geron-' as in *gerontology*, as in old folks

Anterior Segment Dysgenesis



Anterior segment dysgenesis

	Embryotoxon	Gerontoxon
Anterior	Arcus juvenilis	Arcus senilis
Posterior	Posterior embryotoxon	Nope

To complete the matrix... There's an anterior embryotoxon, a posterior embryotoxon, and an anterior gerontoxon. That just leaves posterior gerontoxon. Is it a thing? Not that I'm aware of (hit me up if you know different)

Arcus juvenilis
aka
anterior
embryotoxon

gerontoxon

Gerontoxon?

Yeah, 'geron-' as in gerontology, as in old folks

be's line/ring

y?
nal eyes

g?
drome

me

ous juvenilis.
s?

Anterior Segment Dysgenesis



*Anterior segment
dysgenesis*

What features define Axenfeld-Rieger syndrome?

Peripheral

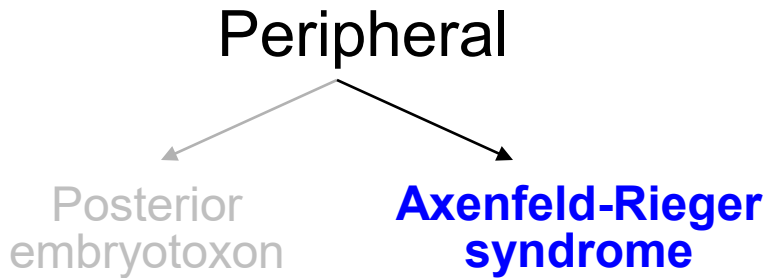
Posterior
embryotoxon

**Axenfeld-Rieger
syndrome**

Anterior Segment Dysgenesis

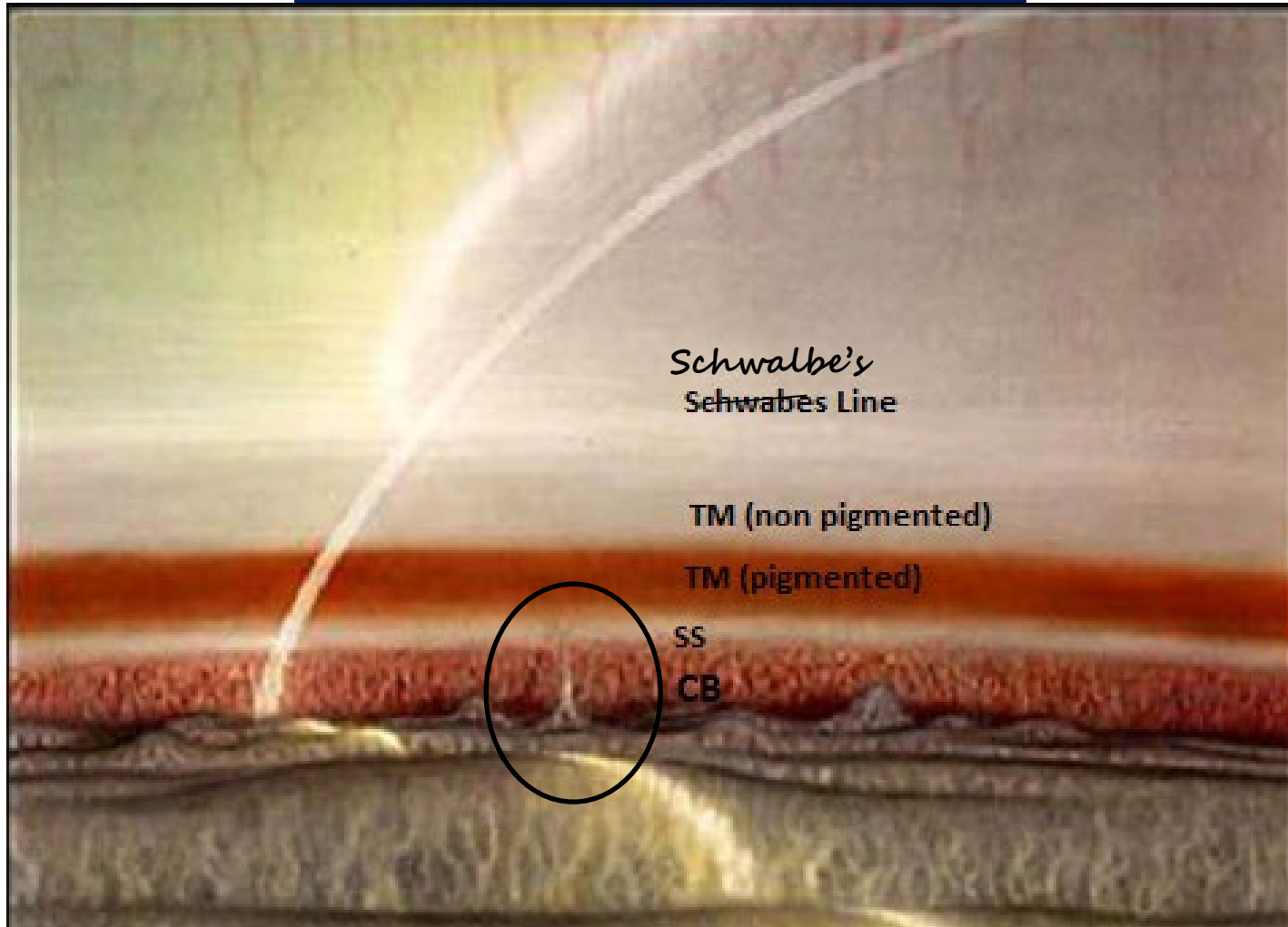


Anterior segment dysgenesis



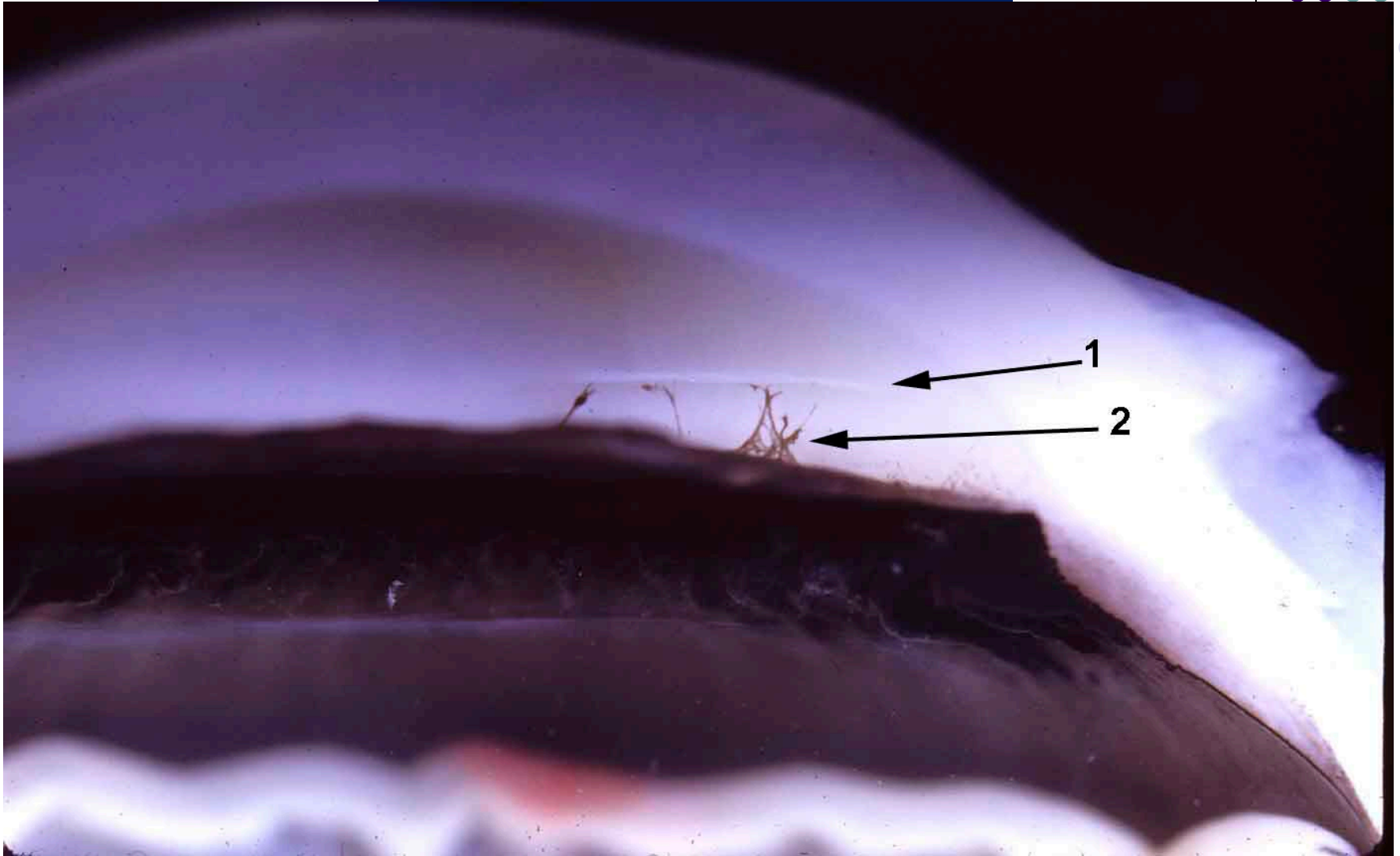
What features define Axenfeld-Rieger syndrome?
Posterior embryotoxon with attached iris strands +
iris hypoplasia + angle abnormalities

Anterior Segment Dysgenesis



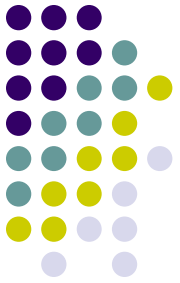
Normal iris strands attached to SS

Anterior Segment Dysgenesis

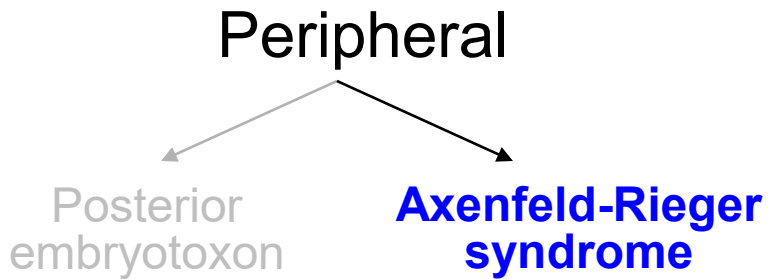


Abnormal iris strands attached to posterior embryotoxon in A-R

Anterior Segment Dysgenesis



Anterior segment dysgenesis



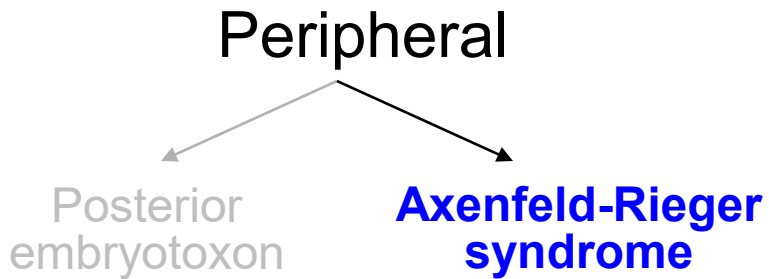
What features define Axenfled-Rieger syndrome?
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Where does ARS rank as a cause of iris hypoplasia?

Anterior Segment Dysgenesis



Anterior segment dysgenesis



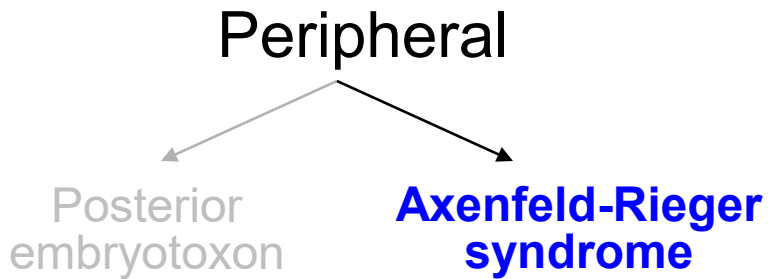
What features define Axenfeld-Rieger syndrome?
Posterior embryotoxon with attached iris strands + **iris hypoplasia** + angle abnormalities

Where does ARS rank as a cause of iris hypoplasia?
It is the most common

Anterior Segment Dysgenesis



Anterior segment dysgenesis



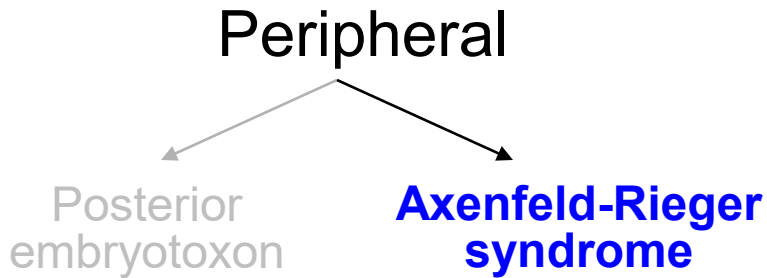
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'Angle abnormalities' suggests an increased risk of glaucoma. Does ARS in fact convey such a risk?

Anterior Segment Dysgenesis



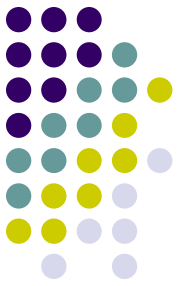
Anterior segment dysgenesis



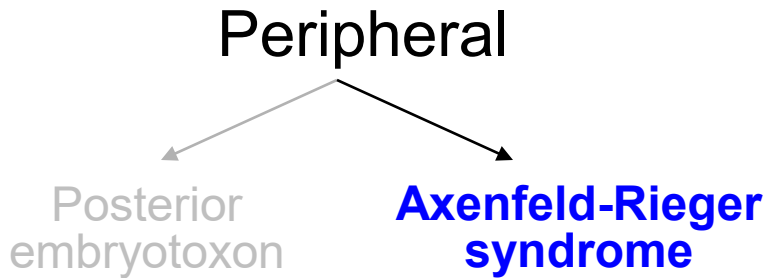
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It does indeed*

Anterior Segment Dysgenesis



Anterior segment dysgenesis



What features define Axenfeld-Rieger syndrome?
Posterior embryotoxon with attached iris strands +
iris hypoplasia + **angle abnormalities**

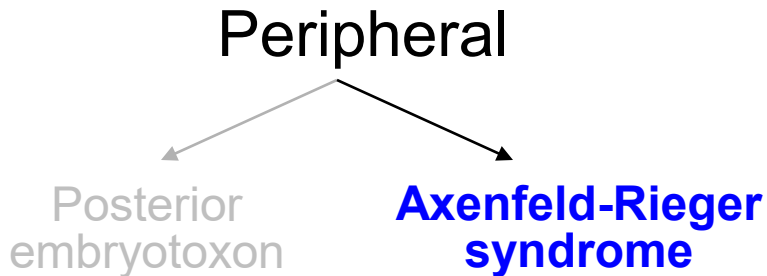
'Angle abnormalities' suggests an increased risk of glaucoma. Does ARS in fact convey such a risk? It does indeed

What is the lifetime risk of developing glaucoma?

Anterior Segment Dysgenesis



Anterior segment dysgenesis



What features define Axenfeld-Rieger syndrome?
Posterior embryotoxon with attached iris strands +
iris hypoplasia + **angle abnormalities**

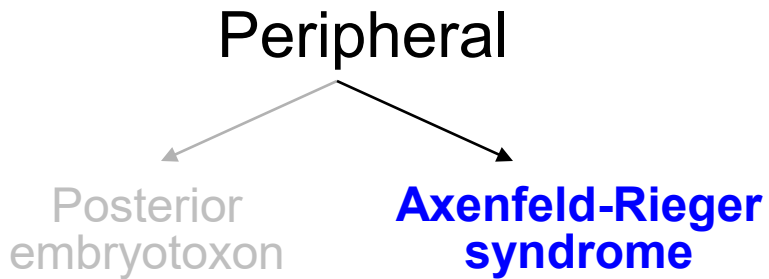
'Angle abnormalities' suggests an increased risk of glaucoma. Does ARS in fact convey such a risk? It does indeed

*What is the lifetime risk of developing glaucoma?
50%*

Anterior Segment Dysgenesis



Anterior segment dysgenesis

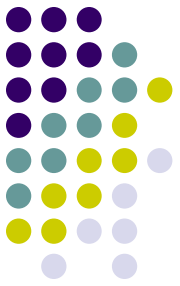


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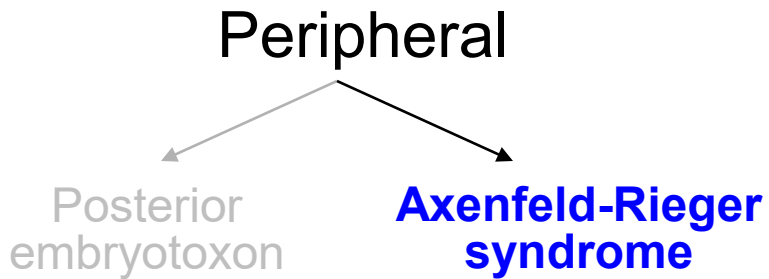
What other iris abnormalities may be present?

- 1) ?
- 2) ?
- 3) ?

Anterior Segment Dysgenesis



Anterior segment dysgenesis

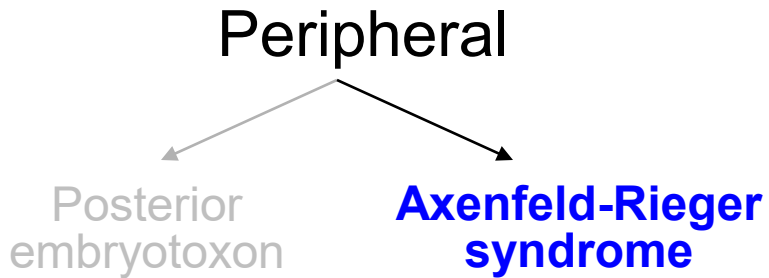


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Posterior embryotoxon with attached iris strands + iris hypoplasia + angle abnormalities

What other iris abnormalities may be present?
1) Corectopia
2) Ectropion uveae
3) Cryptless, glassy surface

Anterior Segment Dysgenesis

A What is corectopia?

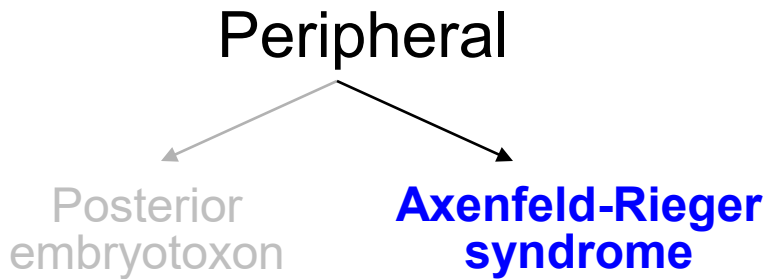


What other iris abnormalities may be present?

- 1) **Corectopia**
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Anterior Segment Dysgenesis

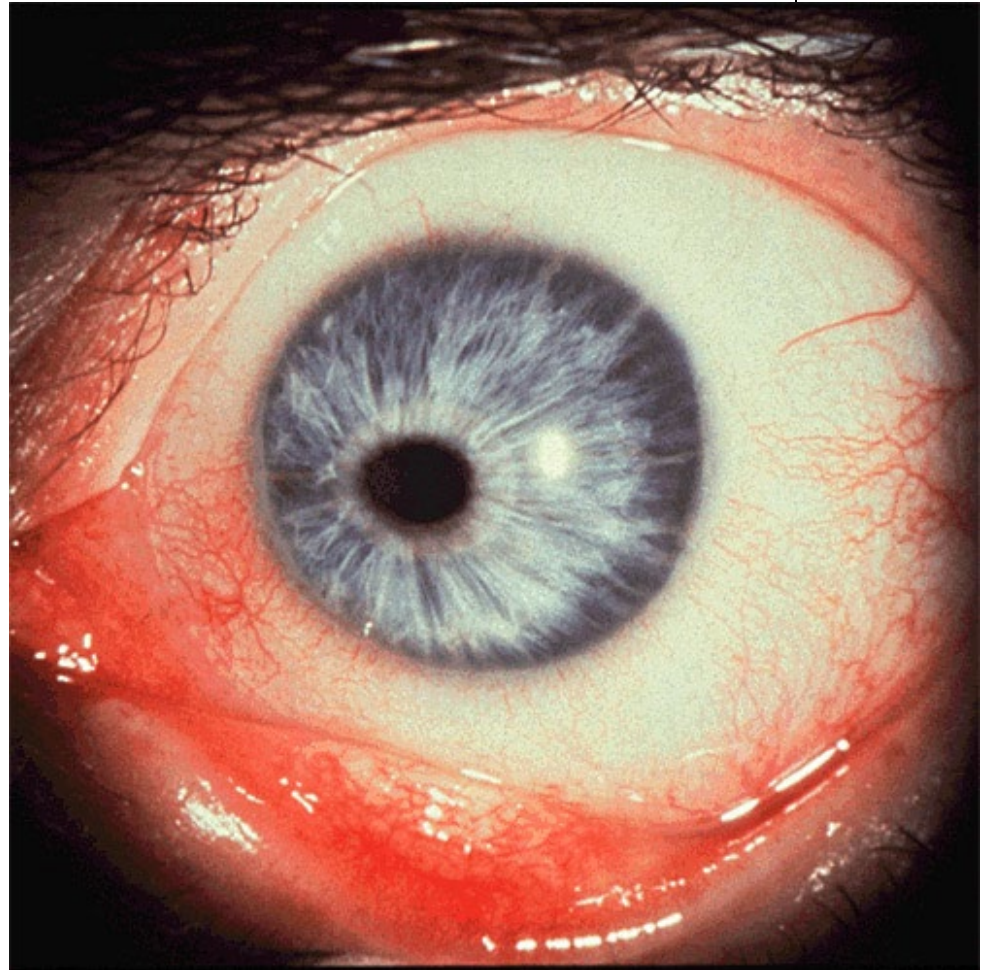
A *What is **corectopia**?*
The displacement of the pupil from its normal central-ish location



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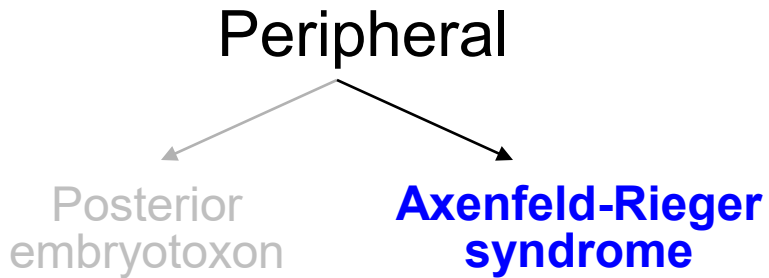
Anterior Segment Dysgenesis



Corectopia

Anterior Segment Dysgenesis

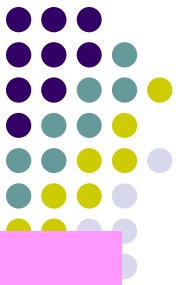
A What is **corectopia**?
The displacement of the pupil from its normal central-ish location
Why central-ish?



What other iris abnormalities may be present?

- 1) **Corectopia**
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Anterior Segment Dysgenesis



A What is **corectopia**?
The displacement of the pupil from its normal central-ish location

Why central-ish?

Deviation from centrality of 1/2 mm is common, and up to 1 mm is considered normal

Peripheral

Posterior
embryotoxon

**Axenfeld-Rieger
syndrome**

What other iris abnormalities may be present?

- 1) **Corectopia**
- 2) Estropion uveae
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Anterior Segment Dysgenesis



Anterior segment dysgenesis

What features define Axenfeld-Rieger syndrome?
Posterior embryotoxon with attached iris strands +
iris hypoplasia + angle abnormalities

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- 2) **Ectropion uveae**
- 3) Cryptose, glassy surface

Peripheral

Posterior
embryotoxon

*What does the term **ectropion uveae** refer to?*

Anterior Segment Dysgenesis



Anterior segment dysgenesis

Peripheral

Posterior
embryotoxon

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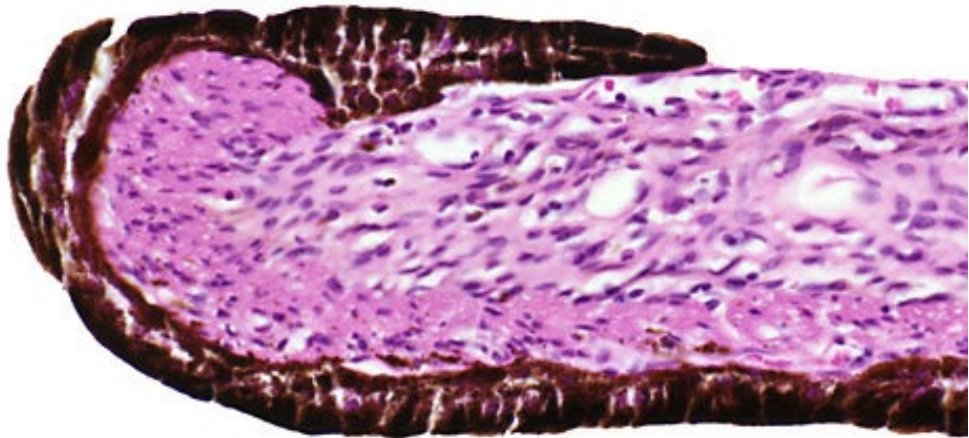
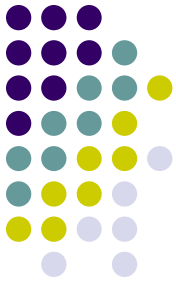
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- 2) **Ectropion uveae**
- 3) Cryptose, glassy surface

*What does the term **ectropion uveae** refer to?*

The presence of posterior pigmented iris epithelium on the anterior surface of the iris

Anterior Segment Dysgenesis



Ectropion uveae

Anterior Segment Dysgenesis



Anterior segment dysgenesis

Peripheral

Posterior
embryotoxon

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The presence of posterior pigmented iris epithelium on the anterior surface of the iris

Technically speaking, the term a misnomer. Why?

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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Peripheral

Posterior
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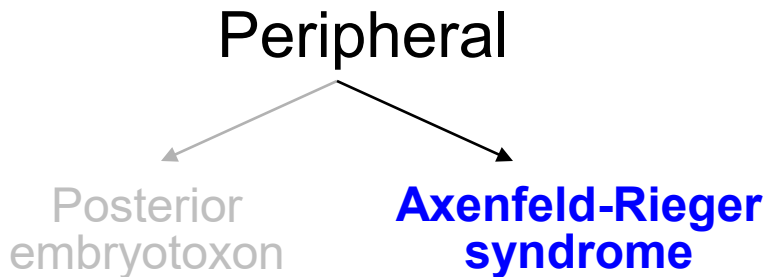
Technically speaking, the term a misnomer. Why?

Because the posterior pigmented epithelium derives from *neuroectoderm*, not uvea

Anterior Segment Dysgenesis



Anterior segment dysgenesis



What features define Axenfeld-Rieger syndrome?
Posterior embryotoxon with attached iris strands +
iris hypoplasia + angle abnormalities

What other iris abnormalities may be present?

- 1) Corectopia
- 2) Ectropion uveae
- 3) Cryptless, glassy surface

What corneal abnormalities may be present?

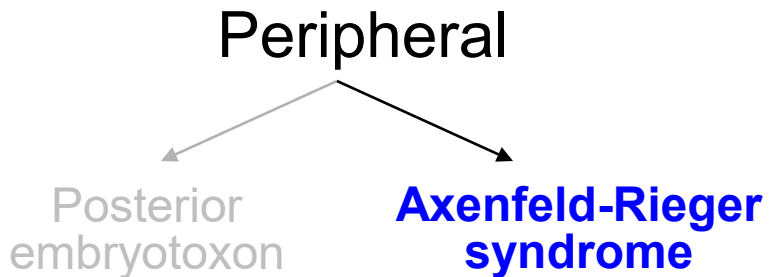
- 1) ?
- 2) ?

} Not simultaneously, obviously

Anterior Segment Dysgenesis



Anterior segment dysgenesis



What features define Axenfeld-Rieger syndrome?
Posterior embryotoxon with attached iris strands + iris hypoplasia + angle abnormalities

What other iris abnormalities may be present?

- 1) Corectopia
- 2) Ectropion uveae
- 3) Cryptless, glassy surface

What corneal abnormalities may be present?

- 1) Megalocornea
 - 2) Microcornea
- } Not simultaneously, obviously

Anterior Segment Dysgenesis

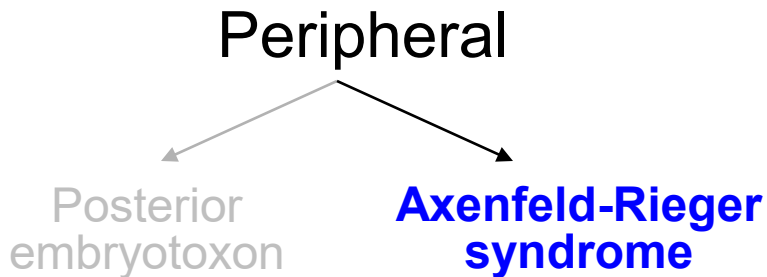


Megalocornea in a 2 y.o. with Axenfeld-Reiger

Anterior Segment Dysgenesis



Anterior segment dysgenesis



What features define Axenfled-Rieger syndrome?
 Posterior embryotoxon with attached iris strands +
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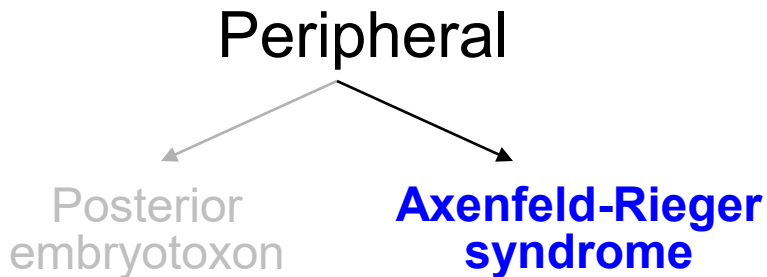
- 1) **Megalocornea**
- 2) **Microcornea**

	<i>Horizontal diameter at birth (mm)</i>	<i>Horizontal diameter at ≥2 years (mm)</i>
Megalocornea	>?	
Microcornea		

Anterior Segment Dysgenesis



Anterior segment dysgenesis



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Posterior embryotoxon with attached iris strands + iris hypoplasia + angle abnormalities

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- 3) Cryptless, glassy surface

What corneal abnormalities may be present?

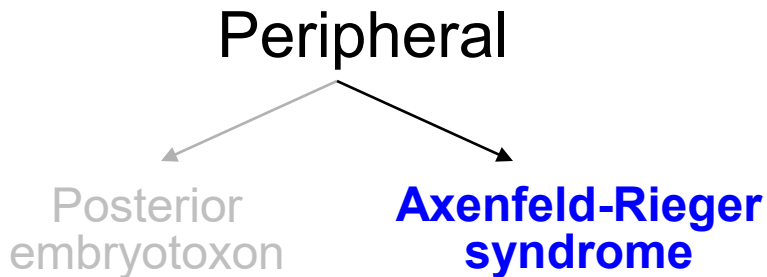
- 1) **Megalocornea**
- 2) **Microcornea**

	Horizontal diameter at birth (mm)	Horizontal diameter at ≥ 2 years (mm)
Megalocornea	>12	
Microcornea		

Anterior Segment Dysgenesis



Anterior segment dysgenesis



What features define Axenfeld-Rieger syndrome?
Posterior embryotoxon with attached iris strands + iris hypoplasia + angle abnormalities

What other iris abnormalities may be present?

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- 2) Ectropion uveae
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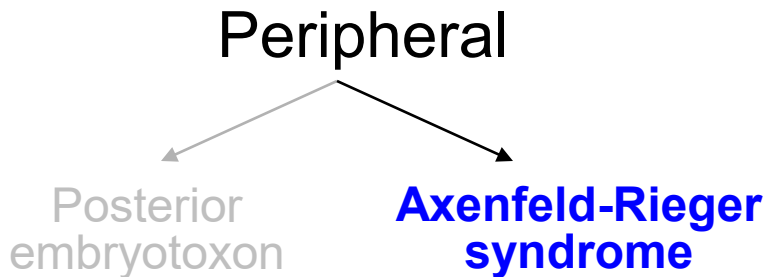
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Anterior Segment Dysgenesis



Anterior segment dysgenesis



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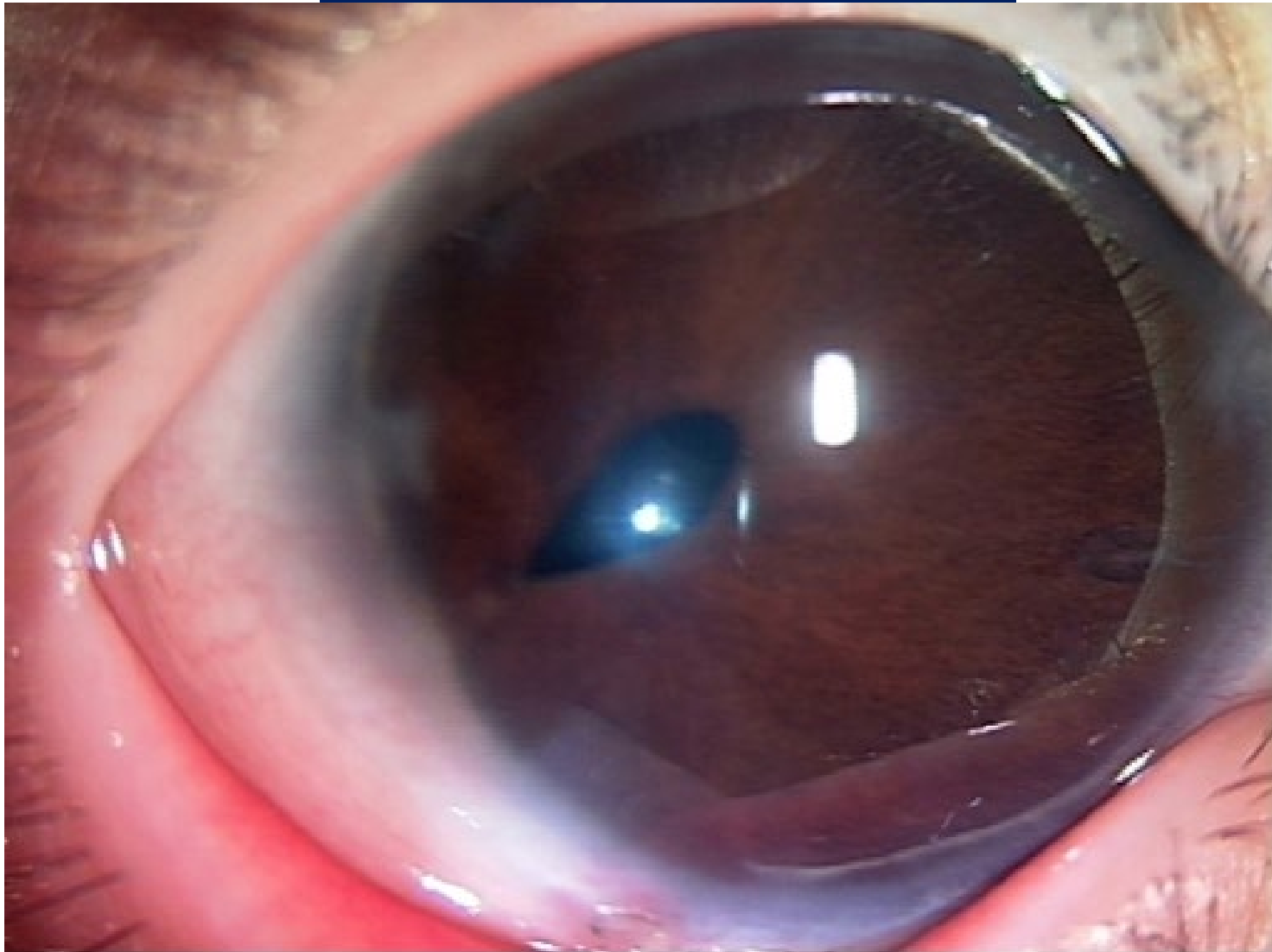
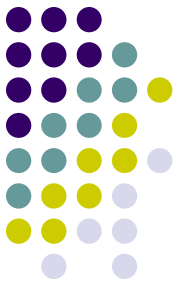
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Microcornea		

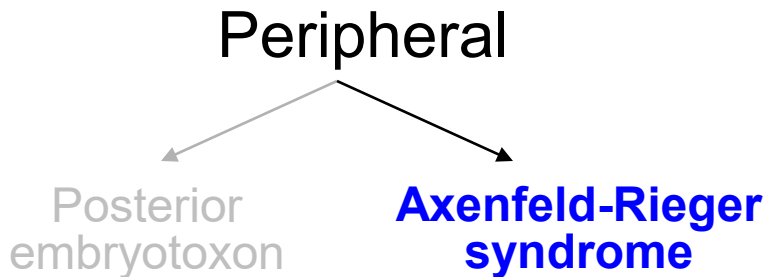


3 y.o. girl who presented at three months of age with hazy megalocornea, posterior embryotoxon, iris hypoplasia, corectopia with early onset severe glaucoma. The horizontal/vertical corneal diameters were 13.0/12.5 mm.

Anterior Segment Dysgenesis



Anterior segment dysgenesis



What features define Axenfled-Rieger syndrome?
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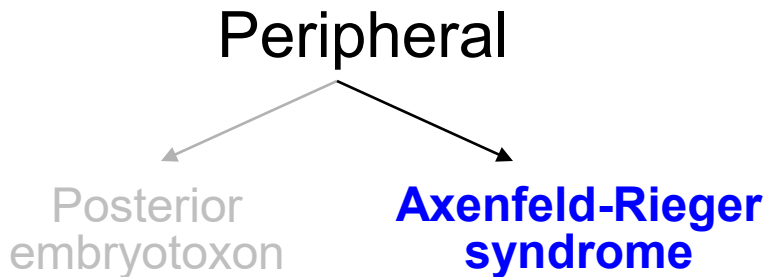
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Microcornea	<?	

Anterior Segment Dysgenesis



Anterior segment dysgenesis



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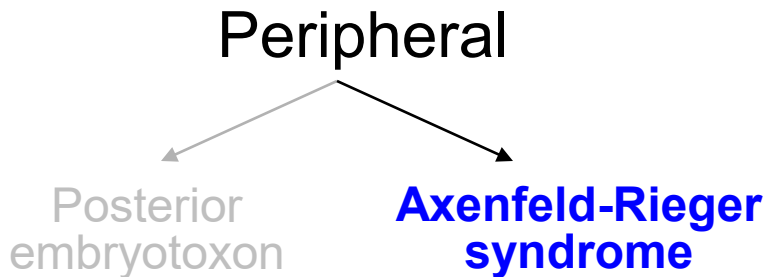
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Megalocornea	>12	>13
Microcornea	<9	

Anterior Segment Dysgenesis



Anterior segment dysgenesis



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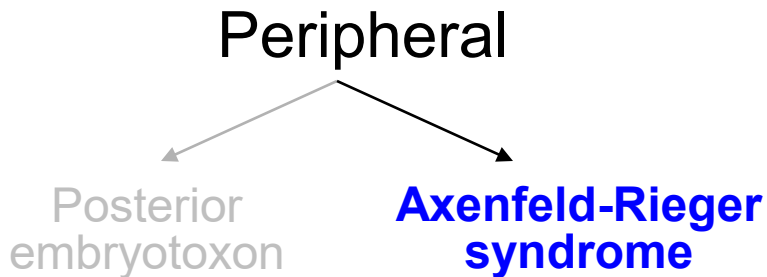
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Anterior Segment Dysgenesis



Anterior segment dysgenesis



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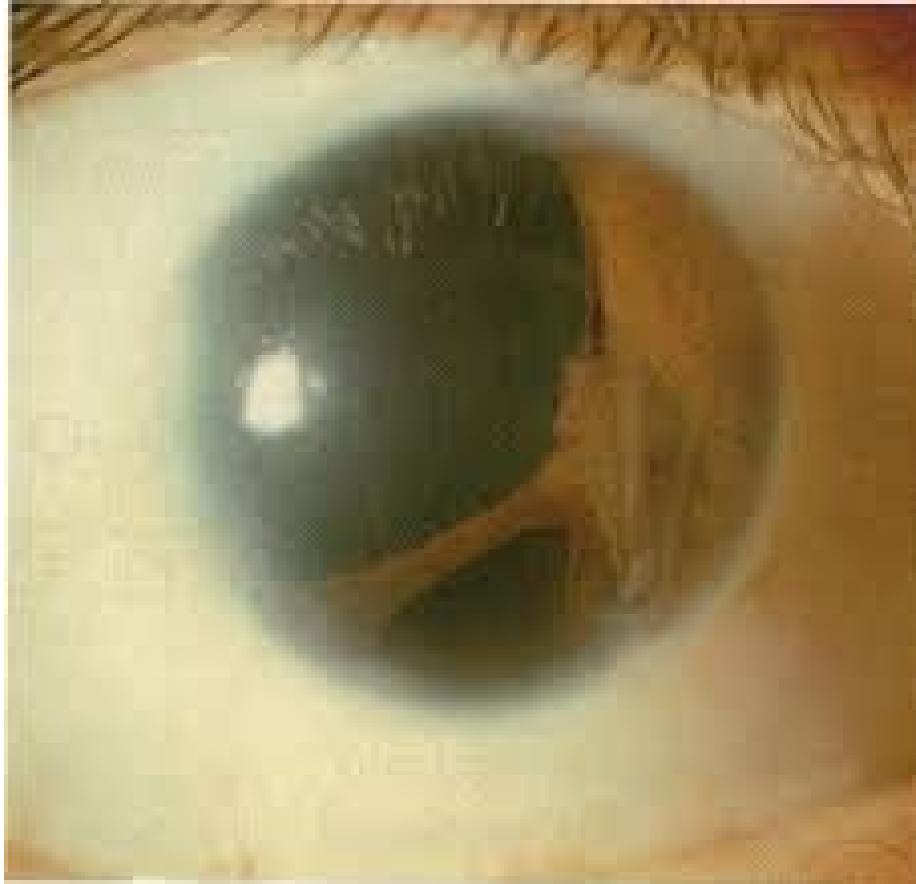
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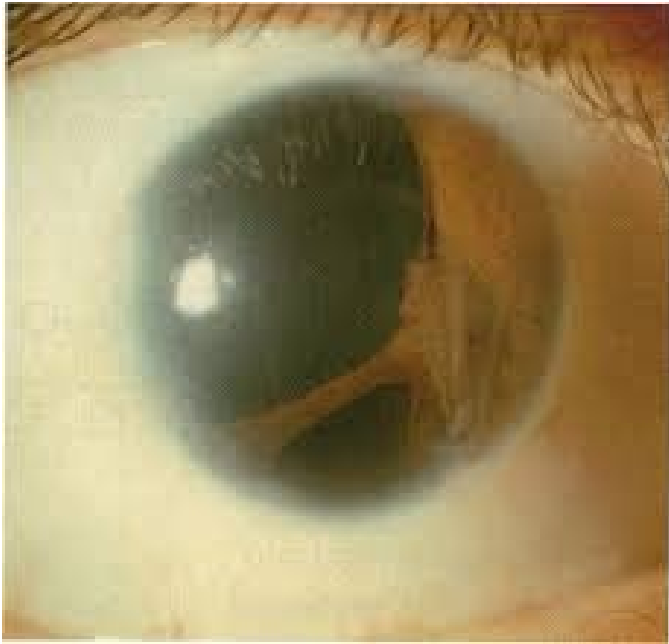
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Megalocornea	>12	>13
Microcornea	<9	<10

Anterior Segment Dysgenesis

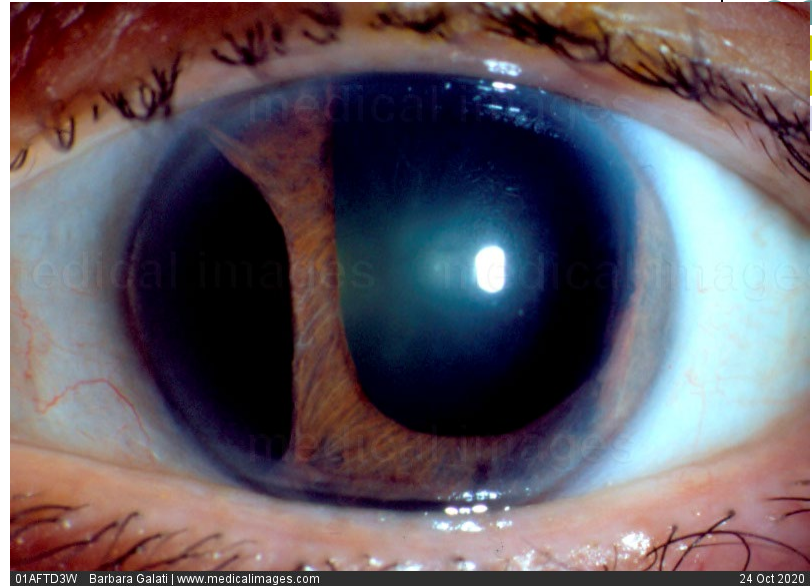


Axenfeld-Reiger with microcornea (8.5 mm)

Anterior Segment Dysgenesis



Axenfeld-Reiger with microcornea (8.5 mm)

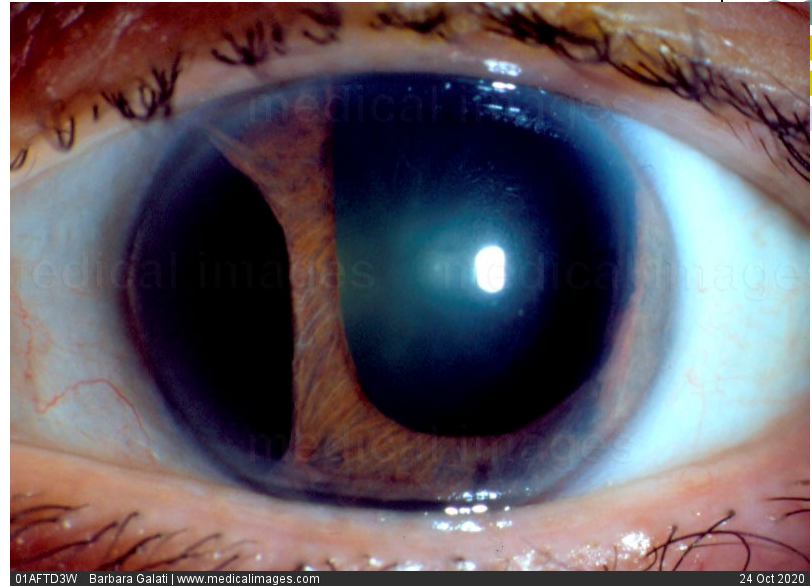


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Anterior Segment Dysgenesis



Axenfeld-Reiger with microcornea (8.5 mm)

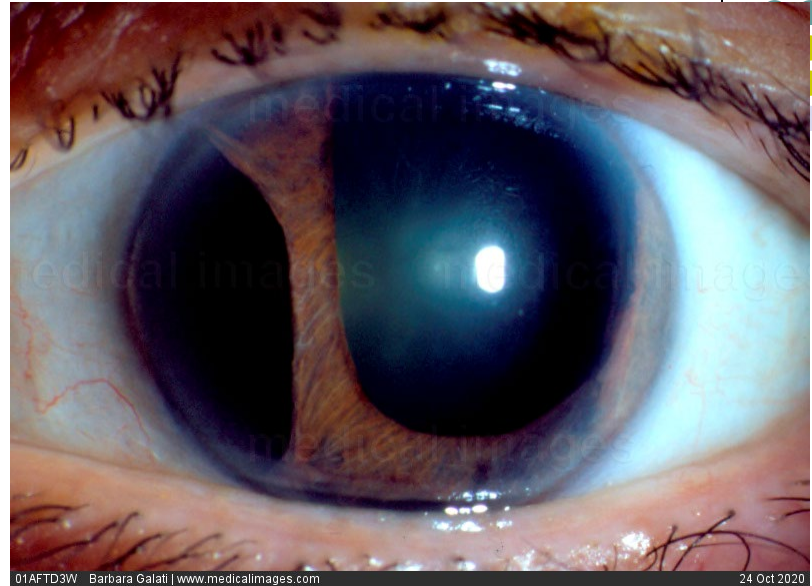


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Essential iris atrophy

Anterior Segment Dysgenesis



Axenfeld-Reiger with microcornea (8.5 mm)



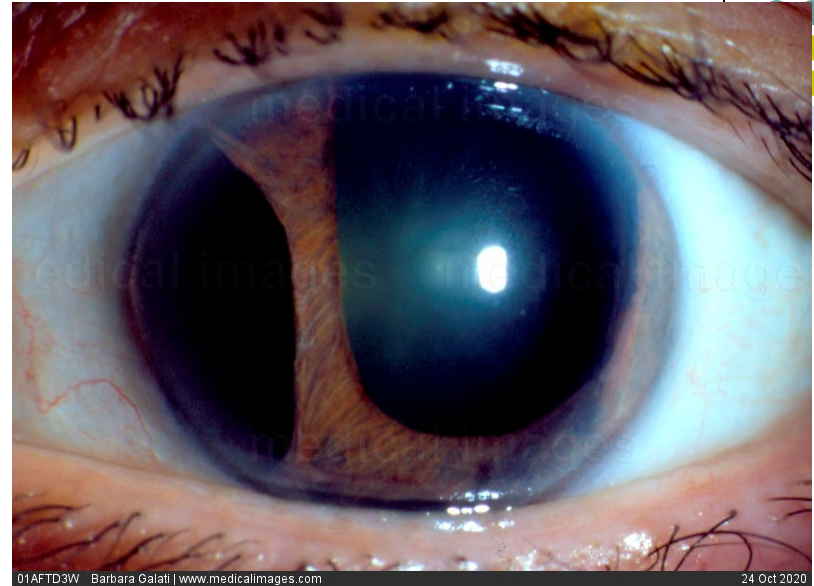
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Essential iris atrophy is a variant/form of what condition?

Anterior Segment Dysgenesis



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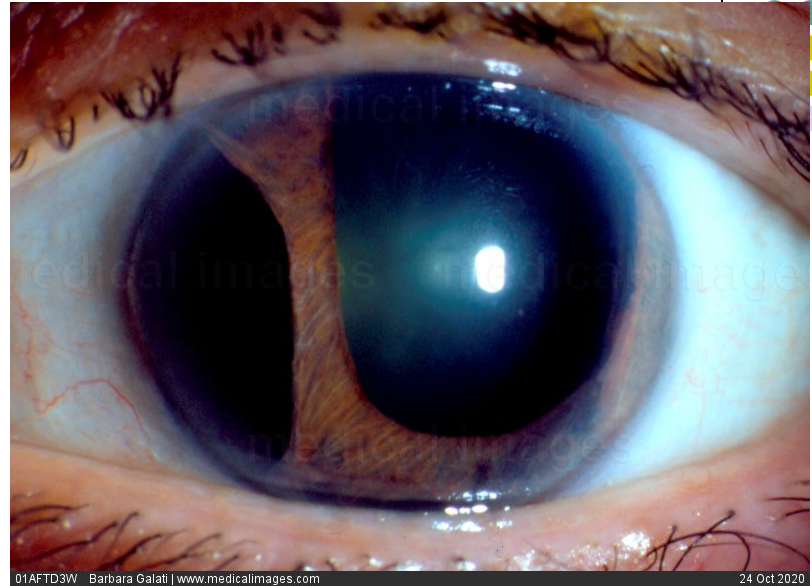
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Iridocorneal endothelial (ICE) syndrome

Anterior Segment Dysgenesis



Axenfeld-Reiger with microcornea (8.5 mm)



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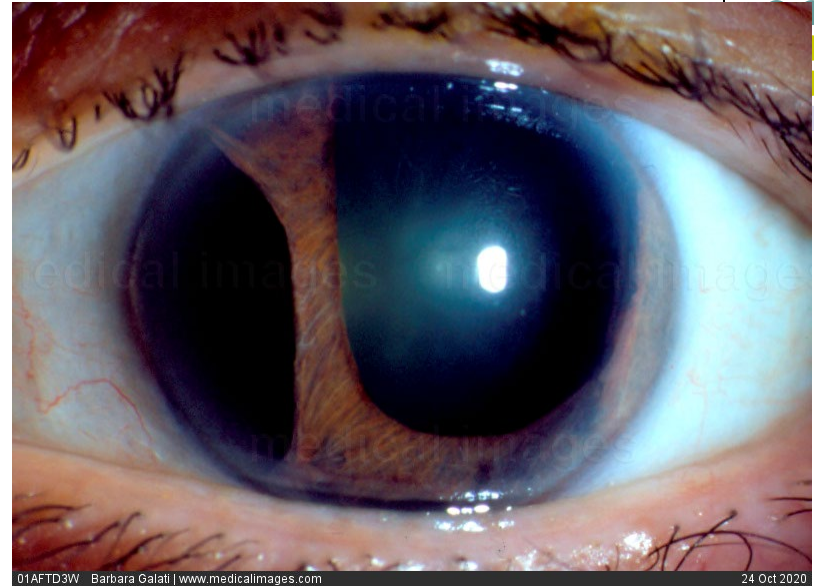
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How do we know that the ARS pic isn't actually ICE?

Anterior Segment Dysgenesis



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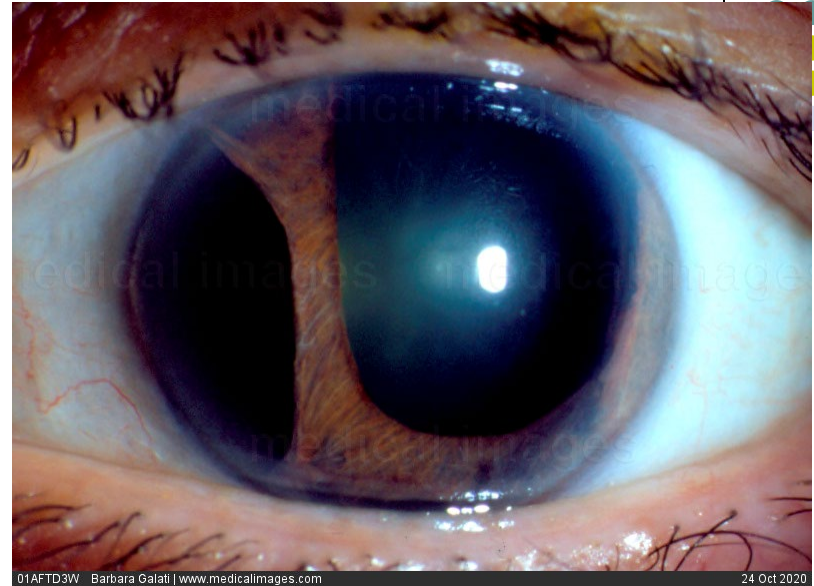
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Anterior Segment Dysgenesis



Axenfeld

Is ICE sporadic, or inherited?



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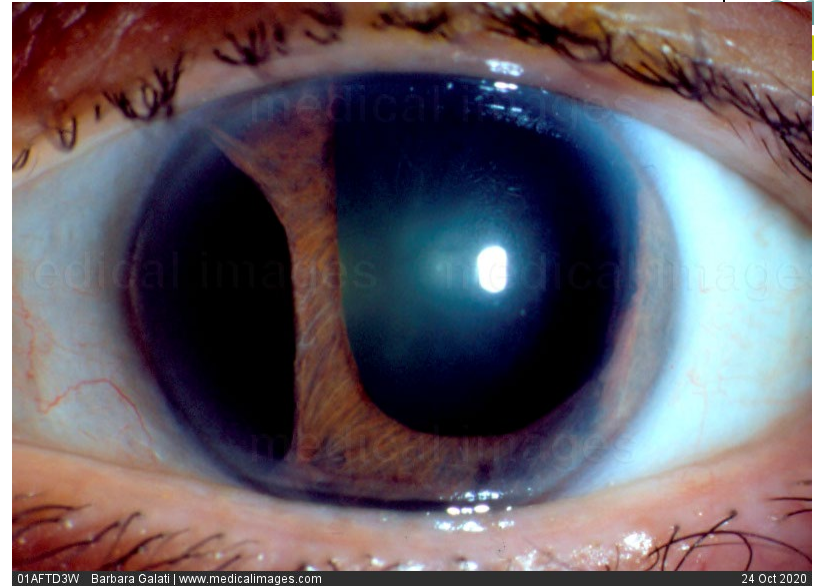
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Anterior Segment Dysgenesis



Axenfeld

Is ICE sporadic, or inherited?
Sporadic



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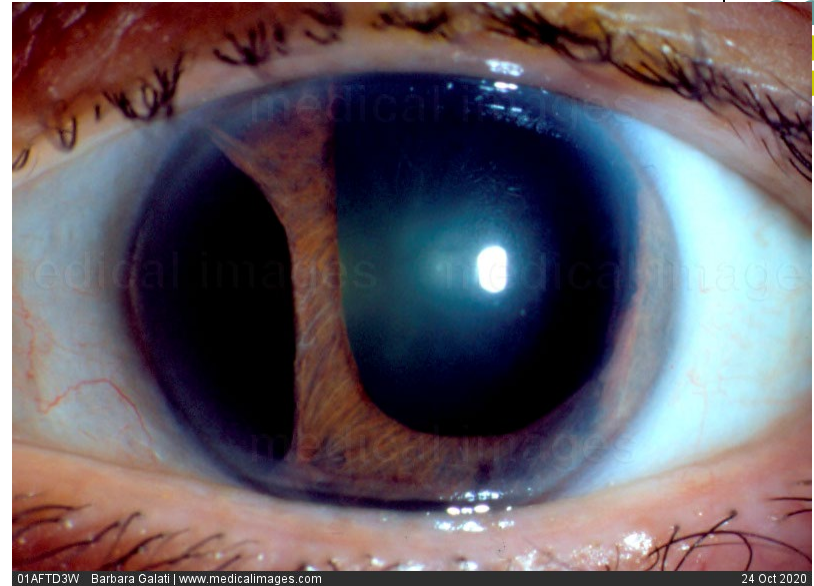


Axenfeld

Is ICE sporadic, or inherited?

Sporadic

Is it unilateral, or bilateral?



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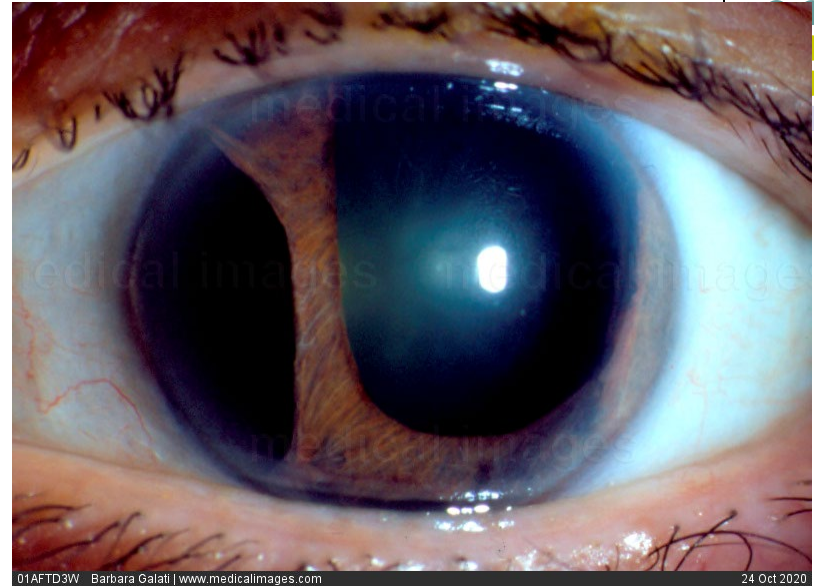
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Anterior Segment Dysgenesis



Axenfeld

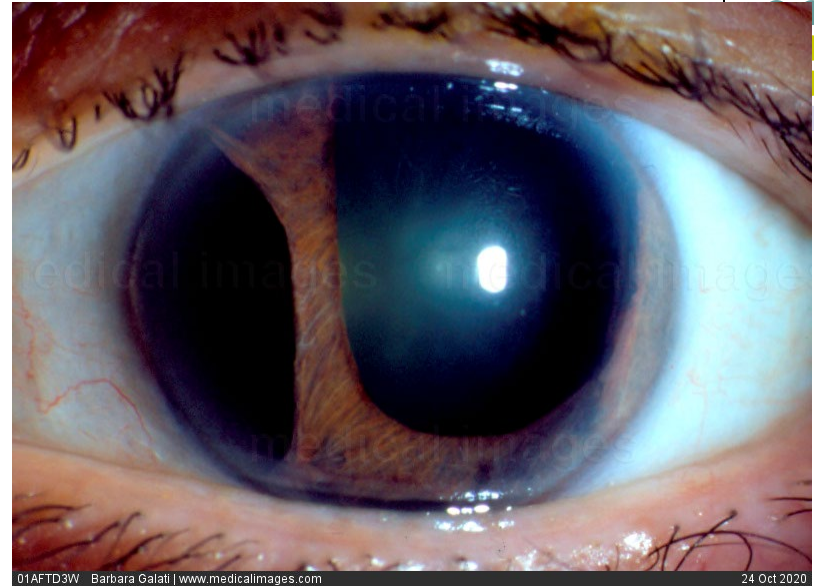
Is ICE sporadic, or inherited?

Sporadic

Is it unilateral, or bilateral?

Unilateral

Does it tend to affect males, or females?



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Essential iris atrophy is a variant/form of what condition?

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Anterior Segment Dysgenesis



Axenfeld

Is ICE sporadic, or inherited?

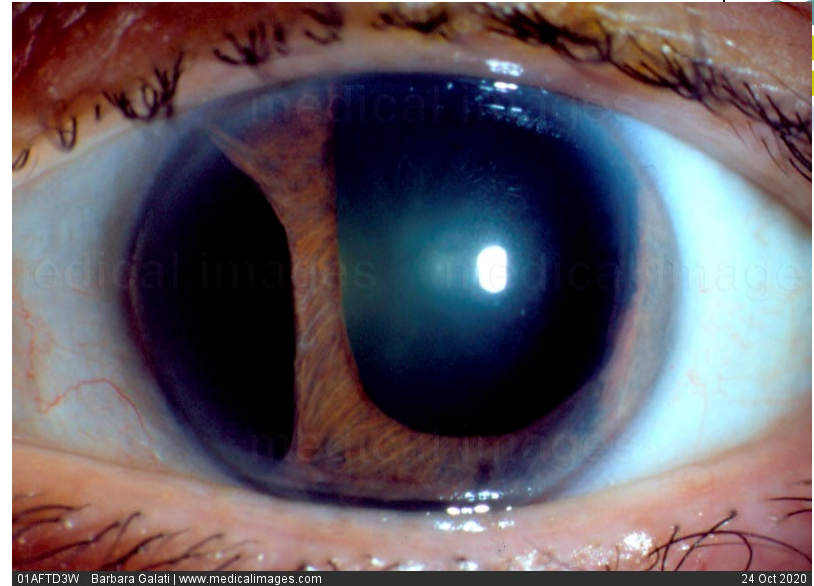
Sporadic

Is it unilateral, or bilateral?

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Does it tend to affect males, or females?

Females



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Essential iris atrophy

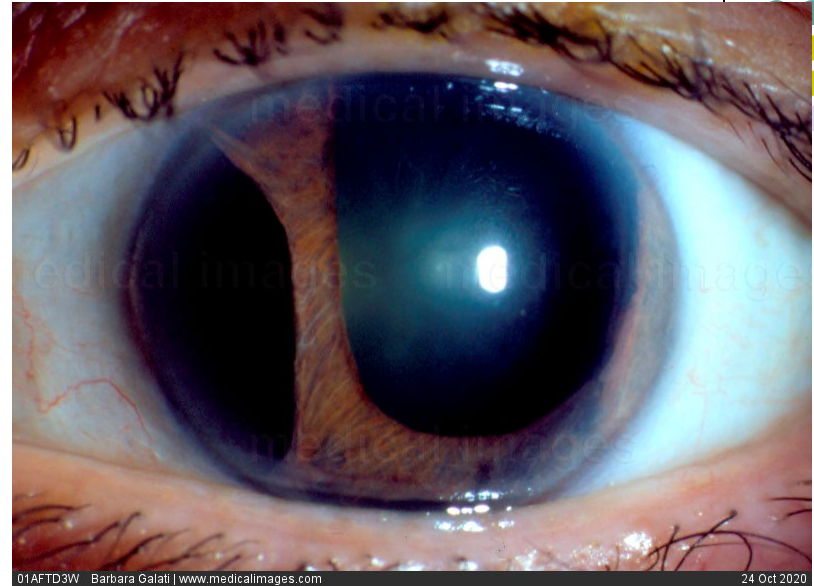
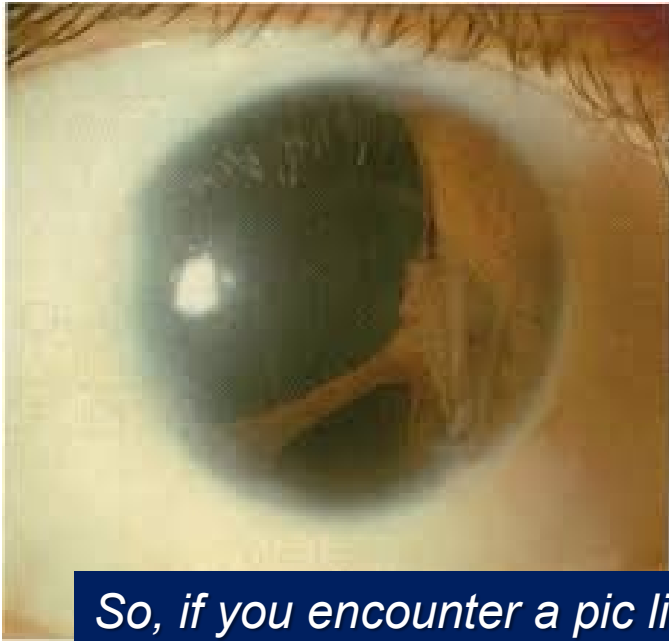
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Anterior Segment Dysgenesis



←That pic should have immediately reminded you of another condition (pictured above)—what is it?

Essential iris atrophy

So, if you encounter a pic like these on the OKAP/Boards:

--If the answer is ICE, the pt will be an adult female with one wonky eye, and there will be no family hx of similar eye issues

--If the answer is ARS, the pt will be a child, the cornea may be too small (or large), and s/he will have other stigmata of ARS (we are continuing to unpack these)

Axenf

dition?

E?

Anterior Segment Dysgenesis



For more on ICE, see slide-set K26

←That pic should have immediately reminded you of another condition (pictured above)—what is it?

Essential iris atrophy

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Axenf

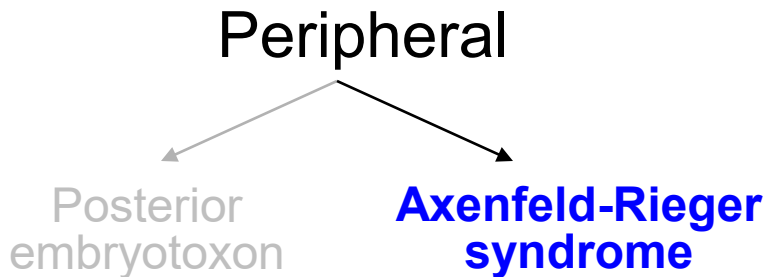
dition?

E?

Anterior Segment Dysgenesis



Anterior segment dysgenesis



What features define Axenfled-Rieger syndrome?
Posterior embryotoxon with attached iris strands + iris hypoplasia + angle abnormalities

What other iris abnormalities may be present?

- 1) Corectopia
- 2) Ectropion uveae
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What corneal abnormalities may be present?

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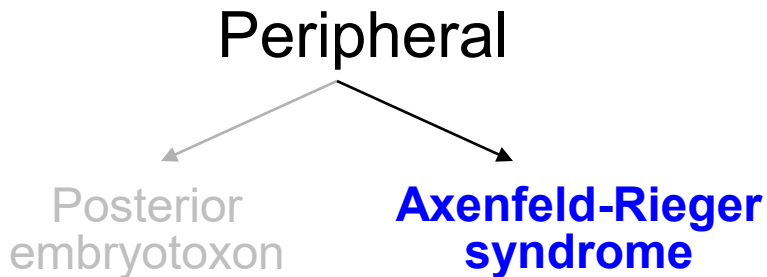
What nonocular abnormalities may be present?

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

Anterior Segment Dysgenesis



Anterior segment dysgenesis



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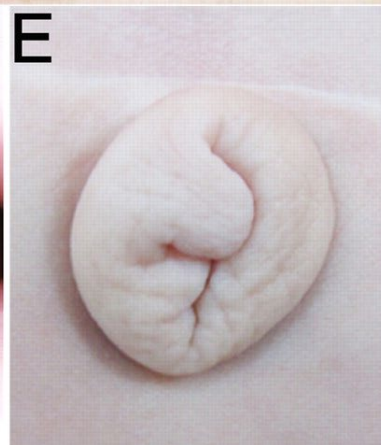
- 1) Corectopia
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What corneal abnormalities may be present?

- 1) Megalocornea
- 2) Microcornea

What nonocular abnormalities may be present?

- 1) Abnormal dentition
- 2) Characteristic facies
- 3) Periumbilical skin folds
- 4) Cardiac valve problems



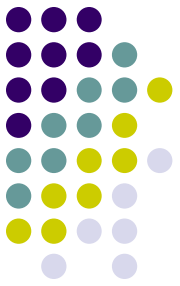
esis



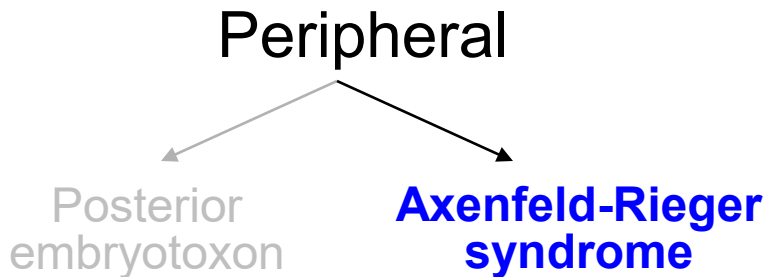
- (A) Facial photograph showing maxillary hypoplasia, thin upper lip, and broad nasal bridge.
- (B) Left eye with corectopia.
- (C) Right eye with posterior embryotoxon.
- (D) Dental anomalies, including maxillary hypodontia.
- (E) Redundant periumbilical skin.

Axenfeld-Reiger *syndrome*

Anterior Segment Dysgenesis



Anterior segment dysgenesis



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What nonocular abnormalities may be present?

- 1) **Abnormal dentition**
- 2) Characteristic facies

Speaking of eye dentistry: When you hear that a pt has teephus issues, four conditions should spring immediately to mind. One is Axenfled-Reiger; what are the other three?

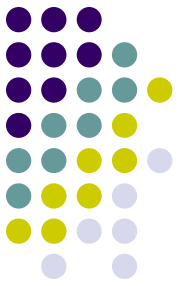
--Axenfled-Reiger

--?

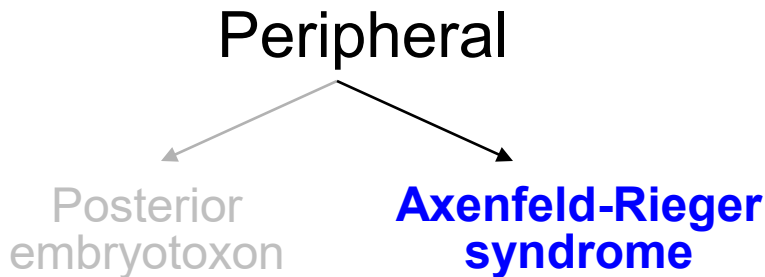
--?

--?

Anterior Segment Dysgenesis



Anterior segment dysgenesis



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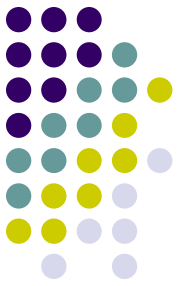
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- Axenfled-Reiger
- Gardner syndrome
- Congenital syphilis
- Incontinentia pigmenti

Anterior Segment Dysgenesis



Anterior segment dysgenesis

What features define Axenfeld-Rieger syndrome?
Posterior embryotoxon with attached iris strands +
iris hypoplasia + angle abnormalities

What is the noneponymous name of this syndrome?

What abnormalities may be present?

ectopic
fuzzy surface

What abnormalities may be present?

ectopic

What abnormalities may be present?

ectopic

hear that a pt has teephus issues,
ately to mind. One is Axenfeld-Reiger;

Abnormal dentition

What are the other three?

- Axenfeld-Reiger
- Gardner syndrome**
- Congenital syphilis
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Anterior Segment Dysgenesis



Anterior segment dysgenesis

What features define Axenfeld-Rieger syndrome?
Posterior embryotoxon with attached iris strands +
iris hypoplasia + angle abnormalities

What is the noneponymous name of this syndrome?
Familial adenomatous polyposis

What other abnormalities may be present?

ectopic cilia
fuzzy surface

What other abnormalities may be present?

ectopic cilia

What other abnormalities may be present?

ectopic cilia

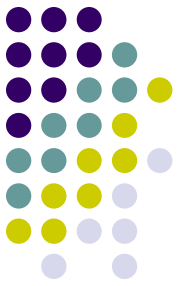
When you hear that a pt has teething issues, immediately to mind. One is Axenfeld-Reiger;

Abnormal dentition

What are the other three?

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- Gardner syndrome**
- Congenital syphilis
- Incontinentia pigmenti

Anterior Segment Dysgenesis



Anterior segment dysgenesis

What features define Axenfeld-Rieger syndrome?
Posterior embryotoxon with attached iris strands +
iris hypoplasia + angle abnormalities

What is the noneponymous name of this syndrome?
Familial adenomatous polyposis

Is it common, or rare?

What other abnormalities may be present?

ectopic
fleshy surface

What other abnormalities may be present?

ectopic

What other abnormalities may be present?

ectopic

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Anterior Segment Dysgenesis



Anterior segment dysgenesis

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ectopic cilia
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What other abnormalities may be present?

ectopic cilia

What other abnormalities may be present?

ectopic cilia

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Anterior Segment Dysgenesis



Anterior segment dysgenesis

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What is the main issue facing these pts? (It's not ophthalmic.)

Abnormal dentition

Gardner syndrome

What are the other three?
--Axenfeld-Rieger
--Gardner syndrome
--Congenital syphilis
--Incontinentia pigmenti

What other abnormalities may be present?

Teeth
Glossy surface

What other abnormalities may be present?

What other abnormalities may be present?

When you hear that a pt has teething issues, immediately to mind. One is Axenfeld-Reiger;

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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Is it common, or rare?
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What is the main issue facing these pts? (It's not ophthalmic.)
They develop innumerable two words at a young age

Abnormal dentition

Gardner syndrome

What are the other three?
--Axenfeld-Rieger
--Gardner syndrome
--Congenital syphilis
--Incontinentia pigmenti

What other abnormalities may be present?

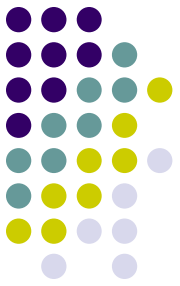
hearing
fatty surface

What other abnormalities may be present?

What other abnormalities may be present?

When you hear that a pt has teething issues, immediately to mind. One is Axenfeld-Reiger;

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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Rare

What is the main issue facing these pts? (It's not ophthalmic.)
They develop innumerable colonic polyps at a young age

What other abnormalities may be present?

Macular degeneration
Leaky surface

What other abnormalities may be present?

What other abnormalities may be present?

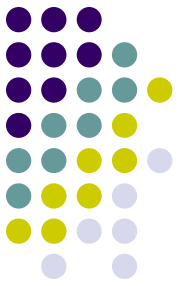
Abnormal dentition

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Anterior Segment Dysgenesis



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at extremely high risk of developing colon cancer by age 50 or so

Abnormal dentition

Gardner syndrome

What are the other three?
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--Gardner syndrome
--Congenital syphilis
--Incontinentia pigmenti

What other abnormalities may be present?

ear
fleshy surface

What other abnormalities may be present?

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When you hear that a pt has teething issues, immediately to mind. One is Axenfeld-Reiger;

Anterior Segment Dysgenesis



Anterior segment dysgenesis

What features define Axenfeld-Rieger syndrome?
Posterior embryotoxon with attached iris strands +
iris hypoplasia + angle abnormalities

What is the noneponymous name of this syndrome?
Familial adenomatous polyposis

Is it common, or rare?
Rare

What is the main issue facing these pts? (It's not ophthalmic.)
They develop innumerable colonic polyps at a young age, and are
at extremely high risk of developing colon cancer by age 40 or so

Abnormal dentition

Gardner syndrome

What are the other three?
--Axenfeld-Rieger
--Gardner syndrome
--Congenital syphilis
--Incontinentia pigmenti

What other abnormalities may be present?

ear
fleshy surface

What other abnormalities may be present?

What other abnormalities may be present?

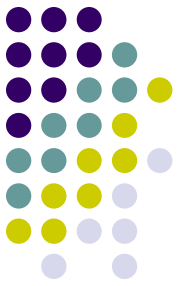
When you hear that a pt has teething issues, immediately to mind. One is Axenfeld-Reiger;

Anterior Segment Dysgenesis



Gardner syndrome: Colonic polyps

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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Why are we talking about it, ie, what is its ocular involvement?

What are the other three?
--Axenfeld-Rieger
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What abnormalities may be present?

tear
fleshy surface

What abnormalities may be present?

What abnormalities may be present?

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Anterior Segment Dysgenesis



Anterior segment dysgenesis

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Abnormal dentition

Why are we talking about it, ie, what is its ocular involvement?
Pts have something-like lesions in their retina

Gardner syndrome

What are the other three?
--Axenfeld-Rieger
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--Incontinentia pigmenti

abnormalities may be present?

eeae
assy surface

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Anterior Segment Dysgenesis



Anterior segment dysgenesis

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They develop innumerable colonic polyps at a young age, and are
at extremely high risk of developing colon cancer by age 40 or so

Abnormal dentition

Why are we talking about it, ie, what is its ocular involvement?
Pts have CHRPE-like lesions in their retina

Gardner syndrome

--Axenfeld-Rieger
--Congenital syphilis
--Incontinentia pigmenti

abnormalities may be present?

eeae
assy surface

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Anterior Segment Dysgenesis



CHRPE



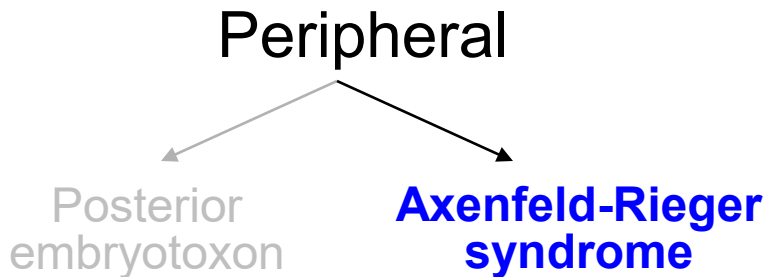
CHRPE-like lesions of Gardner syndrome

For more on Gardners, see slide-set P3

Anterior Segment Dysgenesis



Anterior segment dysgenesis



What features define Axenveld-Rieger syndrome?
Posterior embryotoxon with attached iris strands + iris hypoplasia + angle abnormalities

What other iris abnormalities may be present?

- 1) Corectopia
- 2) Ectropion uveae
- 3) Cryptless, glassy surface

What corneal abnormalities may be present?

- 1) Megalocornea
- 2) Microcornea

What nonocular abnormalities may be present?

- 1) **Abnormal dentition**
- 2) Characteristic facies

What is the eponymous name for the abnormal dentition of congenital syphilis?

eye dentistry: When you hear that a pt has teeptus issues, what should spring immediately to mind. One is Axenveld-Reiger; what other three?

Reiger syndrome

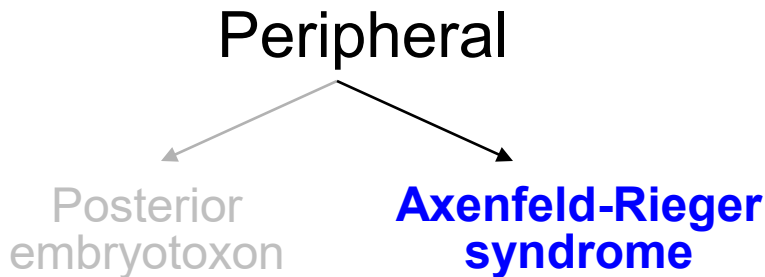
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Anterior Segment Dysgenesis



Anterior segment dysgenesis



What features define Axenfled-Rieger syndrome?
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Hutchinson teeth

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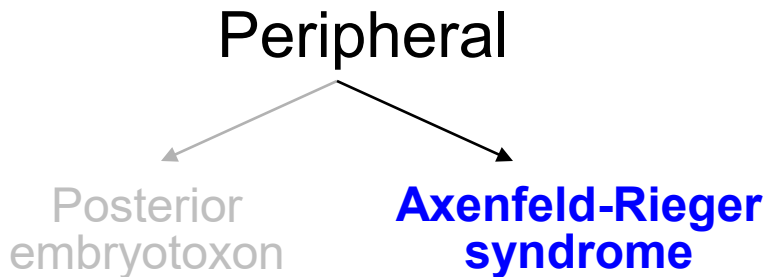
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Anterior Segment Dysgenesis



Anterior segment dysgenesis



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What description is commonly applied to the appearance of Hutchinson teeth?

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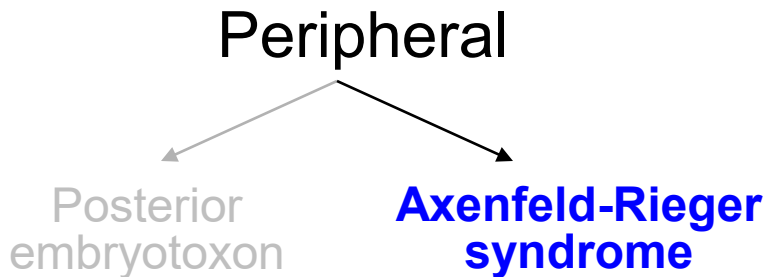
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Anterior Segment Dysgenesis



Anterior segment dysgenesis



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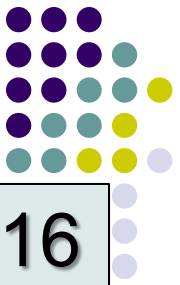
What description is commonly applied to the appearance of Hutchinson teeth?
'Peg shaped'

Eye dentistry: When you hear that a pt has teeptus issues, what should spring immediately to mind. One is Axenfled-Reiger; what other three?

-- **Congenital syphilis**

-- Incontinentia pigmenti

Anterior Segment Dysgenesis



For more on congenital syphilis, see slide-set U16

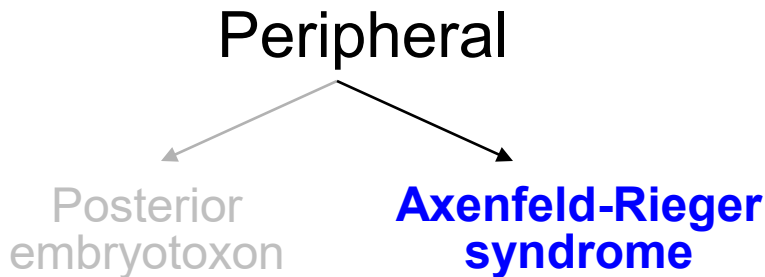


Congenital syphilis: Hutchinson teeth

Anterior Segment Dysgenesis



Anterior segment dysgenesis



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In one word, what sort of condition is IP?

of eye dentistry: When you hear that a pt has teeptus issues, conditions should spring immediately to mind. One is Axenfeld-Reiger; the other three?

*d-Reiger
r syndrome*

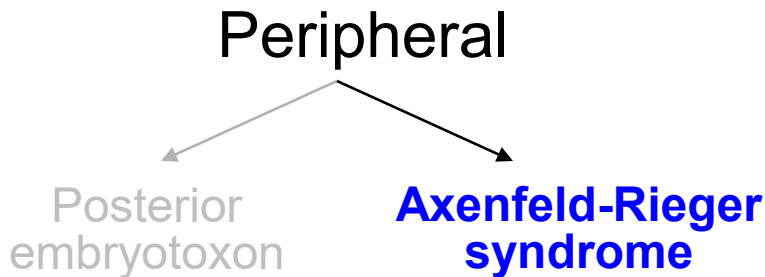
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Anterior Segment Dysgenesis



Anterior segment dysgenesis



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In one word, what sort of condition is IP?
A phakomatosis

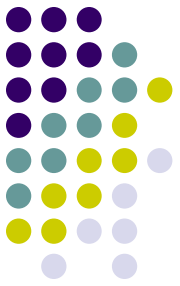
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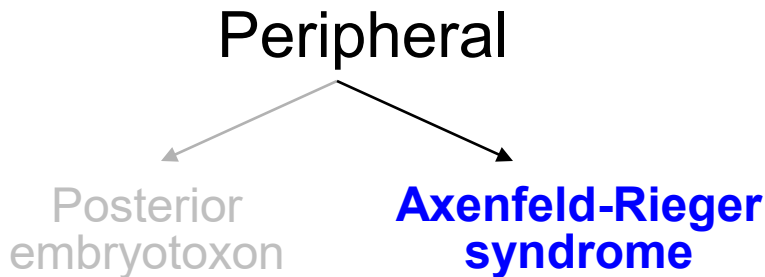
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Incontinentia pigmenti

Anterior Segment Dysgenesis



Anterior segment dysgenesis



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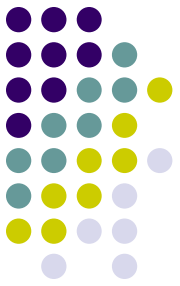
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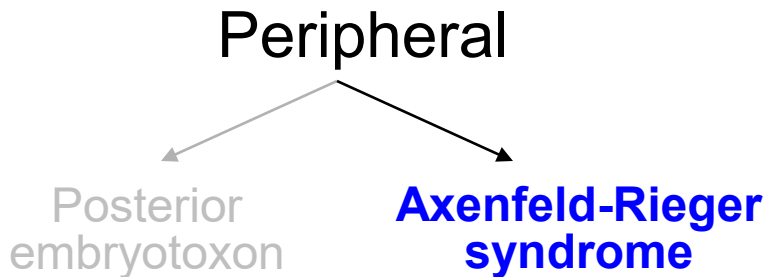
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Anterior Segment Dysgenesis



Anterior segment dysgenesis



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What is the eponymous name for IP?
Bloch-Sulzberger syndrome

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Incontinentia pigmenti

Anterior Segment Dysgenesis



Anterior segment dysgenesis

Peripheral

Very broadly, what is a phakomatosis?

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Anterior Segment Dysgenesis



Anterior segment dysgenesis

Peripheral

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Microcornea

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Incontinentia pigmenti

Anterior Segment Dysgenesis



Anterior segment dysgenesis

Peripheral

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Megalocornea
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Abnormal dentition

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phakomatosis

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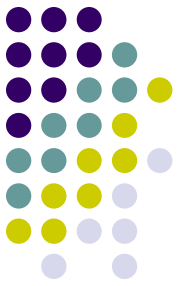
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d-Reiger
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Incontinentia pigmenti

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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Anterior Segment Dysgenesis



Anterior segment dysgenesis

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'Splashed paint' appearance

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d-Reiger
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--Congenital syphilis

Incontinentia pigmenti

Anterior Segment Dysgenesis

For more on IP, see slide-set P10

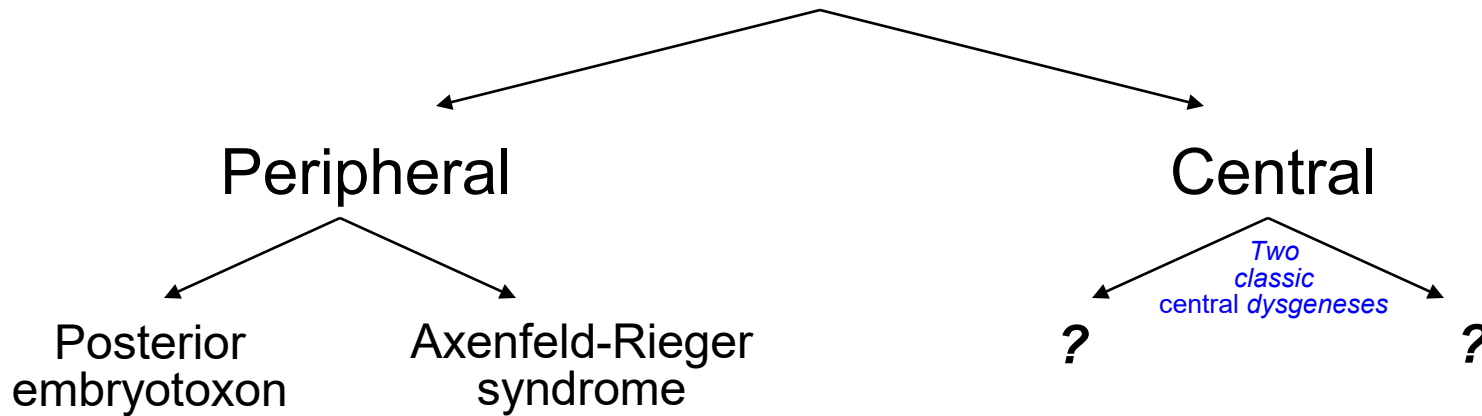


Incontinentia pigmenti: Splashed-paint appearance

Anterior Segment Dysgenesis



Anterior segment dysgenesis

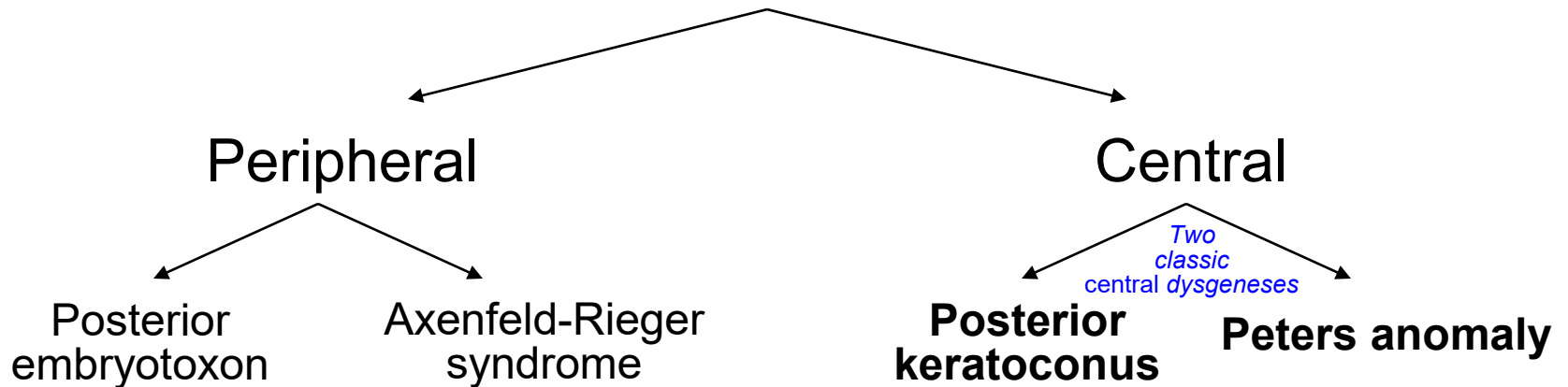


The BCSC goes into depth on two central dysgeneses—which two?

Anterior Segment Dysgenesis



Anterior segment dysgenesis



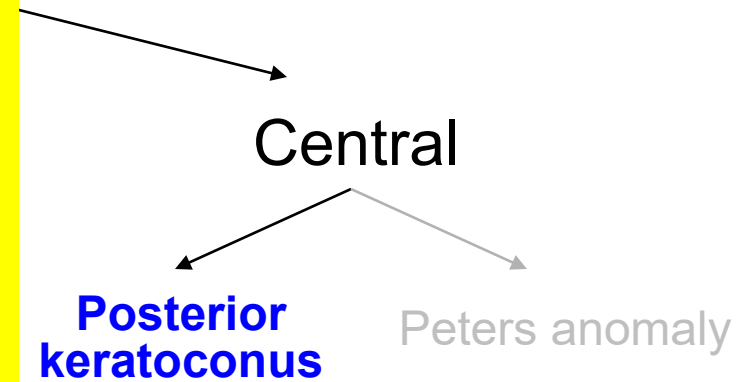
*The BCSC goes into depth on two
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Anterior Segment Dysgenesis



What is posterior keratoconus?

Anterior Segment
Dysgenesis



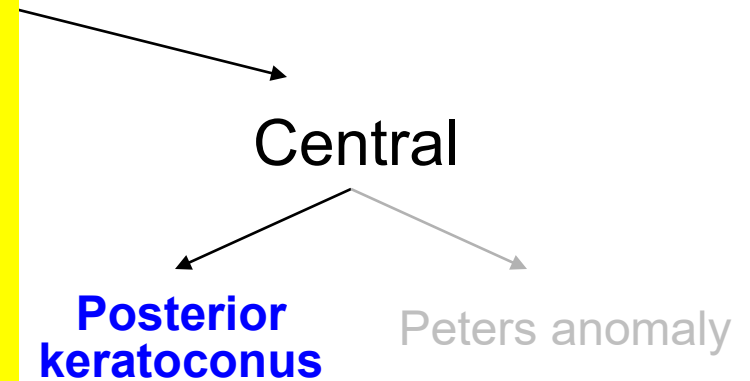
Anterior Segment Dysgenesis



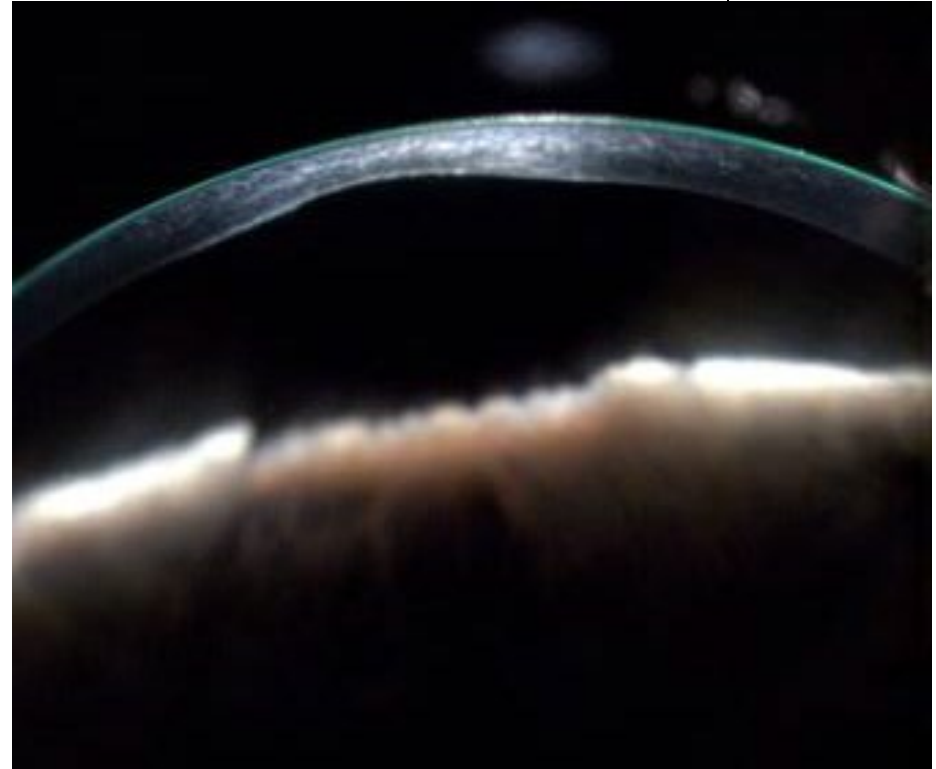
What is posterior keratoconus?

A focal and discrete indentation of the posterior corneal surface

Anterior Segment
Dysgenesis



Anterior Segment Dysgenesis



Posterior keratoconus

Anterior Segment Dysgenesis

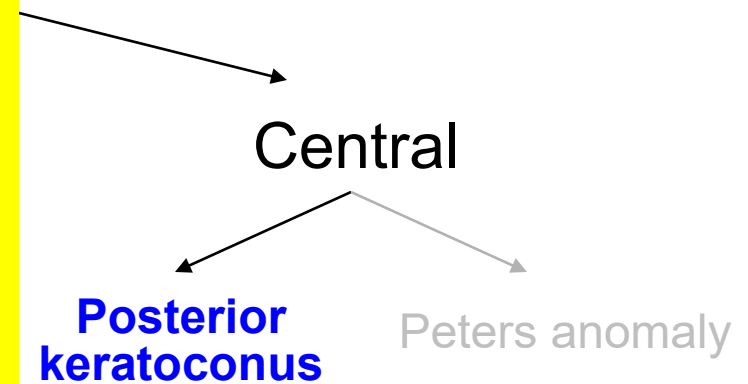


What is posterior keratoconus?

A focal and discrete indentation of the posterior corneal surface

Is the indentation secondary to a defect in the endothelium and/or Descemet's?

ment
sis



Anterior Segment Dysgenesis



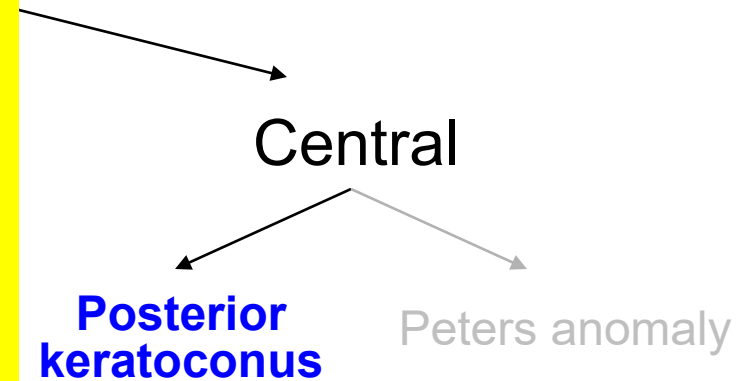
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No, both are usually present and intact

ment
sis



Anterior Segment Dysgenesis



What is posterior keratoconus?

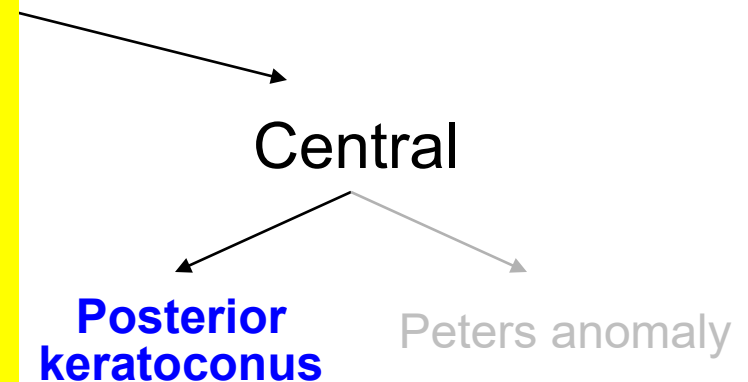
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ment
sis



Anterior Segment Dysgenesis



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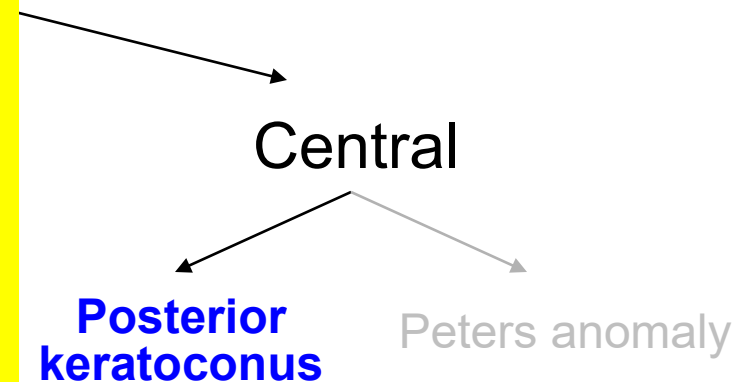
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Is it common, like regular keratoconus?

No, it is rare

ment
sis



Anterior Segment Dysgenesis



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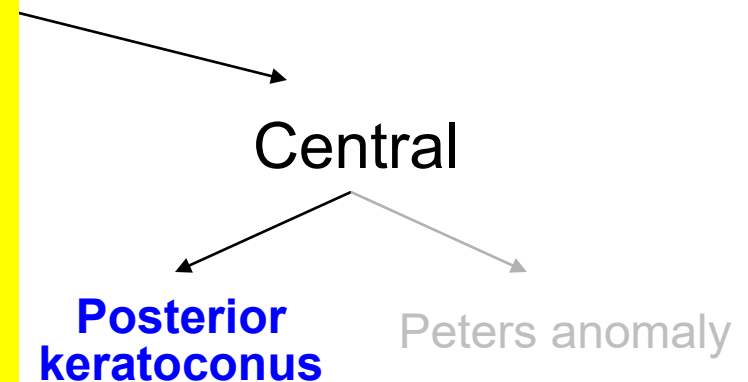
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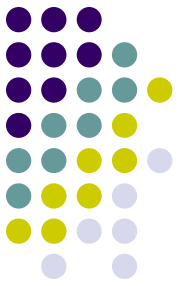
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Does it affect vision?

ment
sis



Anterior Segment Dysgenesis



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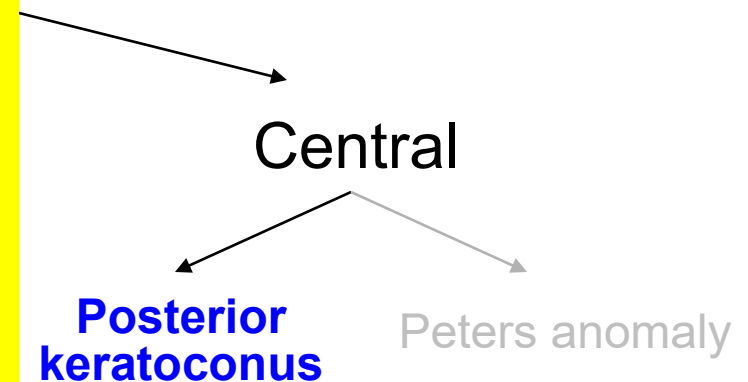
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Does it affect vision?

Yes, it causes irregular astigmatism, which can be severe enough to result in amblyopia

ment
sis



Anterior Segment Dysgenesis



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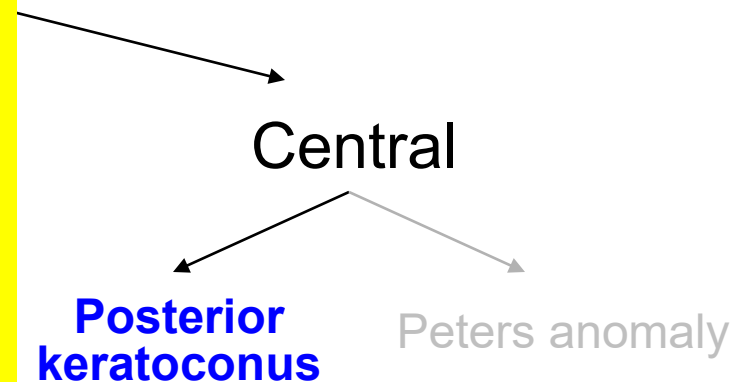
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Are most cases unilateral, or bilateral?

ment
sis



Anterior Segment Dysgenesis



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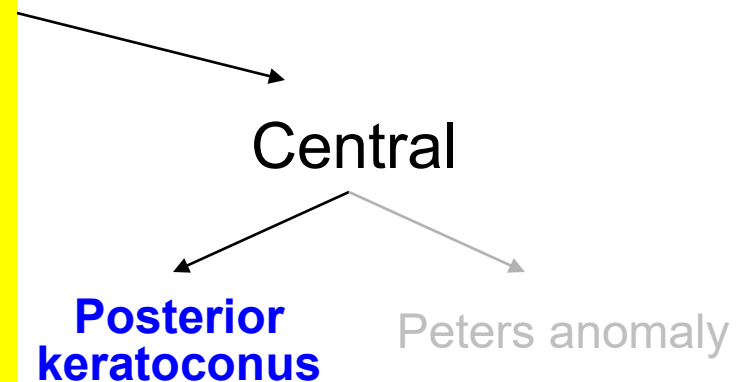
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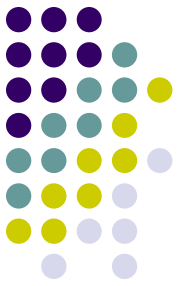
Are most cases unilateral, or bilateral?

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ment
sis



Anterior Segment Dysgenesis



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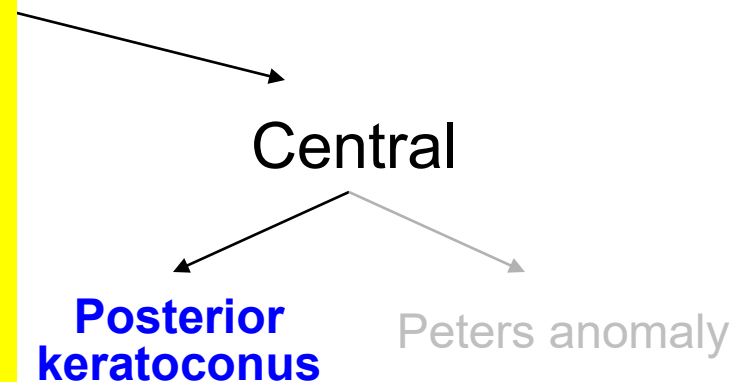
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Are most cases familial, or sporadic?

Anterior Segment
Dysgenesis



Anterior Segment Dysgenesis



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Does it affect vision?

Yes, it causes irregular astigmatism, which can be severe enough to result in amblyopia

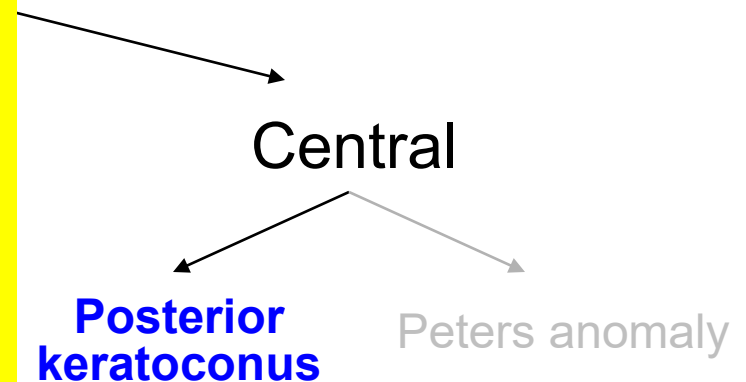
Are most cases unilateral, or bilateral?

Unilateral

Are most cases familial, or sporadic?

Sporadic

ment
sis



Anterior Segment Dysgenesis



Anterior segment dysgenesis

What is Peters anomaly?

Central

Peters anomaly

Anterior Segment Dysgenesis



Anterior segment dysgenesis

What is Peters anomaly?

A defect of the posterior central cornea, including the absence of Descemet's and subadjacent endothelium. Adhesions extending from the iris to the posterior corneal defect are often present.

Central

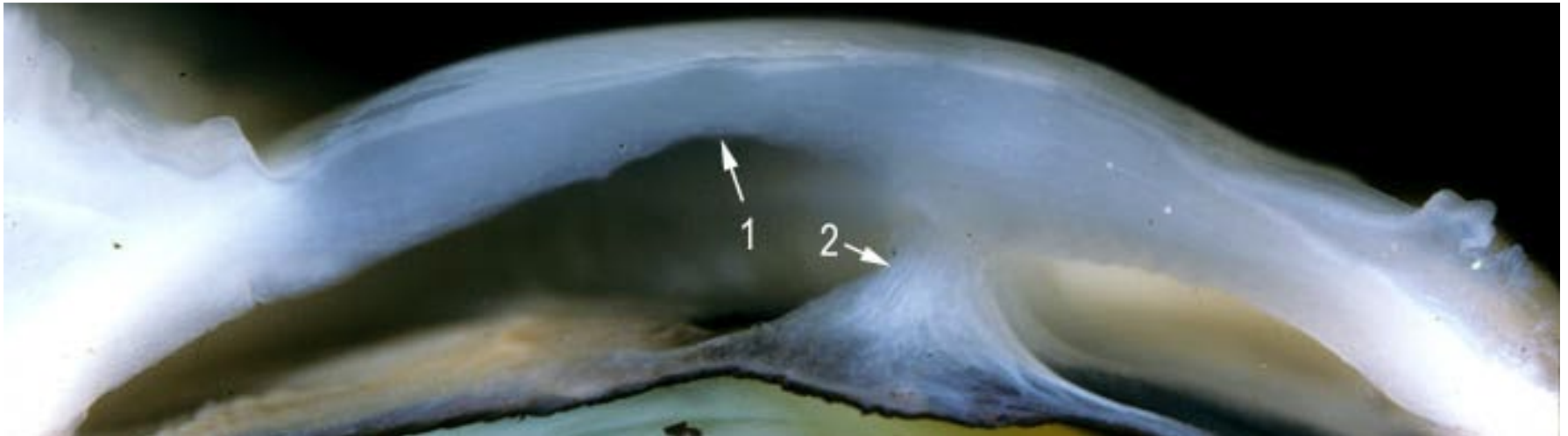
Peters anomaly

Anterior Segment Dysgenesis



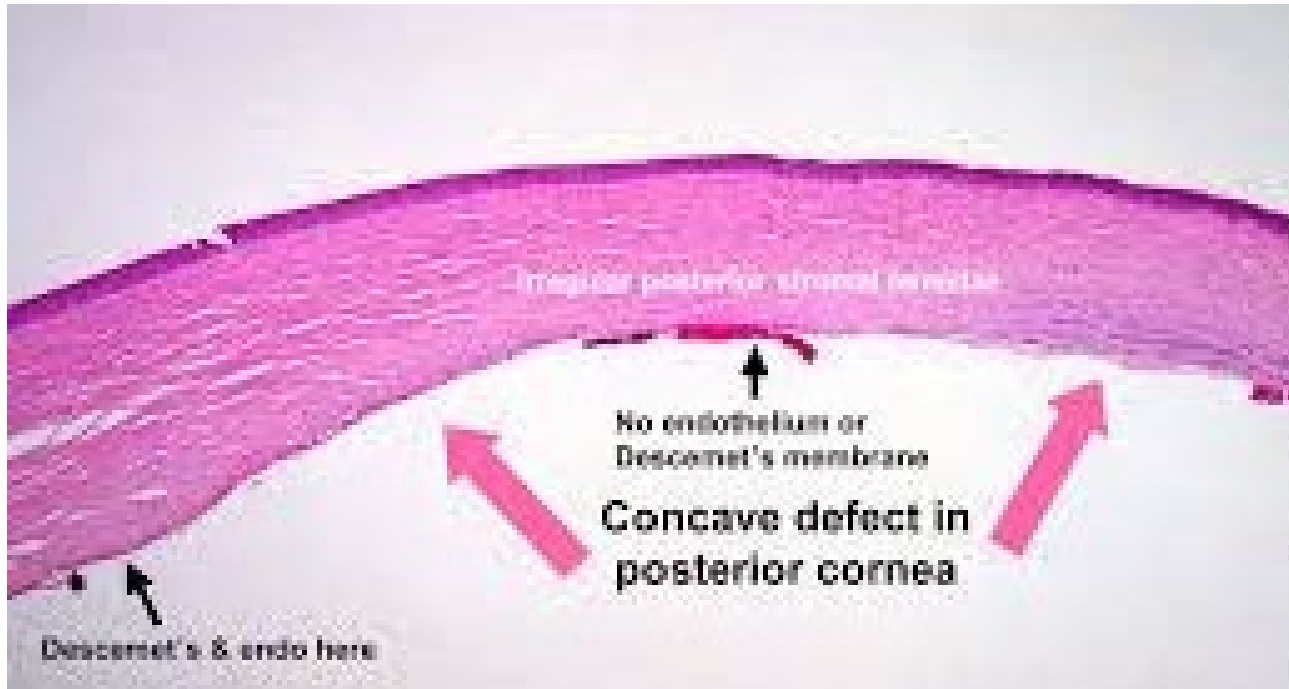
1. Defect of the posterior central cornea, including the absence of Descemet's and subjacent endothelium

2. Adhesions extending from the iris to the posterior corneal defect



Peters anomaly

Anterior Segment Dysgenesis



Peters anomaly

Anterior Segment Dysgenesis



Anterior segment dysgenesis

What is Peters anomaly?

A defect of the posterior central cornea, including the absence of Descemet's and subjacent endothelium. Adhesions extending from the iris to the posterior corneal defect are often present.

How does it present?

Central

Peters anomaly

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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How does it 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.

Central

Peters anomaly

Anterior Segment Dysgenesis



Peters anomaly: Hazy cornea

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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The opacity ranges in severity from a faint haze to an opaque, elevated and vascularized mass.

Central

Peters anomaly

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

What is the STUMPED mnemonic for a cloudy cornea in an infant?

S
T
U
M
Peters anomaly
E
D

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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The opacity ranges in severity from a faint haze to an opaque, elevated and vascularized mass.

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Peters anomaly

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

What is the STUMPED mnemonic for a cloudy cornea in an infant?

Sclerocornea; **S**tomal dystrophy (CHSD)

Trauma (eg, forcep injury)

Ulcer

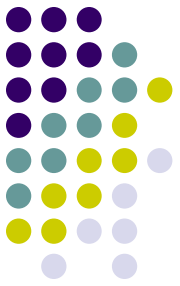
Mucopolysaccharidosis

Peters anomaly

Endothelial dystrophy (CHED); **E**levated IOP (congenital glaucoma)

Dermoid of the cornea

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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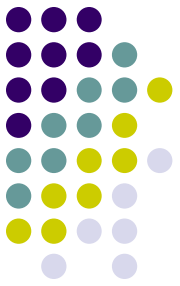
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How might the lens be involved?

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Peters anomaly

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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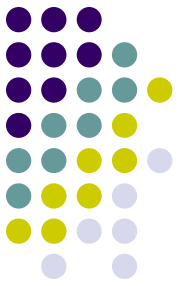
How might the lens be involved?

--It is often.. one word
--It may be.. one word to the three word
--Occasionally it is.. size, shape, and location (four words)

Central

Peters anomaly

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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How might the lens be involved?

- It is often...cataractous
- It may be...adherent to the posterior corneal defect
- Occasionally it is...small, misshapen, and displaced into the AC

Central

Peters anomaly

Anterior Segment Dysgenesis



Peters anomaly: Cataractous lens

Anterior Segment Dysgenesis



Anterior segment dysgenesis

What is Peters anomaly?

A d
Des
the

Hmm...The notion of a 'small, misshapen' lens in this context should bring to mind particular condition. What is it?

How
As
opa
and

How
--It
--It

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Anterior Segment Dysgenesis



Anterior segment dysgenesis

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Microspherophakia

How

As

opa

and

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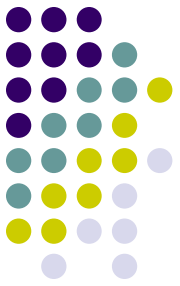
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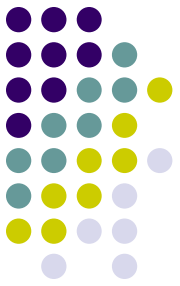
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--It
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How *What common slit-lamp observation owes to the lens' small size?*

--It *Typically, the entirety of the lens equator can be seen in the*

--It *pupillary aperture when the pt is widely dilated*

--Occasionally it is...**small** , misshapen , and displaced into the AC

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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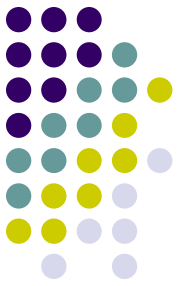
How *How does refractive status manifest the lens' spherical shape?*

--It

--It

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Anterior Segment Dysgenesis



Anterior segment dysgenesis

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and *The name says it all: the lens is small ('micro') and **round ('sphero')***

How *How does refractive status manifest the lens' spherical shape?*
--It *Pts are usually highly myopic*
--It

--Occasionally it is...small , **misshapen** , and displaced into the AC

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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How *If zonular laxity allows the lens to drift forward, what clinical condition may result?*

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Anterior Segment Dysgenesis



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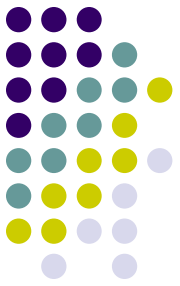
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--It The lens may 'clog' the pupillary opening, resulting in pupillary block angle-closure glaucoma

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Anterior Segment Dysgenesis



Anterior segment dysgenesis

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Instead, what condition should come immediately to mind?

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Ho **Weill-Marchesani syndrome**

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--It The lens may 'clog' the pupillary opening, resulting in pupillary block angle-closure glaucoma

--Occasionally it is...small , misshapen , and displaced into the AC

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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an *Instead, what condition should you consider?*

Weill-Marchesani syndrome

He *If zonular laxity allows the lens to dislocate*

--It *The lens may 'clog' the pupillary opening*

--It *resulting in angle closure glaucoma*

--Occasionally it is...small, misshapen

What is the classic stature of a W-M pt? *lens?*

result?

closure glaucoma

Anterior Segment Dysgenesis



Anterior segment dysgenesis

What is Peters anomaly?

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He *If zonular laxity allows the lens to dislocate*

--It *The lens may 'clog' the pupillary opening*

--It *resulting in angle closure glaucoma*

--Occasionally it is...small, misshapen

What is the classic stature of a W-M pt? *lens?*
Quite short

result?

closure glaucoma

Anterior Segment Dysgenesis



Weill-Marchesani syndrome: Short stature

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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What is the classic stature of a W-M pt?

Quite short

What is notable about their digits?

lens?

result?

closure glaucoma

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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What is the classic stature of a W-M pt?

Quite short

What is notable about their digits?

They are short as well

lens?

result?

closure glaucoma

Anterior Segment Dysgenesis



Weill-Marchesani syndrome: Short fingers

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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an *Instead, what condition should you consider?*

Weill-Marchesani syndrome

He *If zonular laxity allows the lens to dislocate*
--It *The lens may 'clog' the pupillary opening*
--It *resulting in secondary angle closure glaucoma*

--Occasionally it is...small, misshapen

What is the classic stature of a W-M pt?
Quite short

What is notable about their digits?
They are short as well

What is notable about their joints?

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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Ho *If zonular laxity allows the lens to dislocate*
--It *The lens may 'clog' the pupillary opening*
--It *resulting in*

--Occasionally it is...small, misshapen

What is the classic stature of a W-M pt?
Quite short

What is notable about their digits?
They are short as well

What is notable about their joints?
They are stiff

result?
closure glaucoma

Anterior Segment Dysgenesis



Anterior segment dysgenesis

What is Peters anomaly?

A d Hmm...The notion of a 'small, misshapen' lens in this context should bring to mind particular
Des condition. What is it?
the **Microspherophakia**

How Is microspherophakia associated with Peters anomaly?

As Yes, a "microspherophakia" is a "Peters anomaly" (Peters anomaly is a form of microspherophakia)

For more on W-M, see slide-set FELT14

When

Instead, what condition should you consider? Quite short

Weill-Marchesani syndrome

What is notable about their digits?

They are short as well

result? closure glaucoma

What is notable about their joints?

They are stiff

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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How does it 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.

How might the lens be involved?

- It is often...cataractous
- It may be...adherent to the posterior corneal defect
- Occasionally it is...small, misshapen, and displaced into the AC

Central

Peters anomaly

Anterior Segment Dysgenesis



Anterior segment dysgenesis

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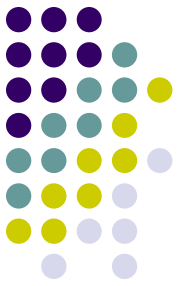
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Does Peters anomaly require a workup?

Central

Peters anomaly

Anterior Segment Dysgenesis



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Does Peters anomaly require a workup?

No if it's...

Yes if it's...

Central

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Anterior Segment Dysgenesis



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- It may be...adherent to the posterior corneal defect
- Occasionally it is...small, misshapen, and displaced into the AC

Does Peters anomaly require a workup?

No if it's...**unilateral** (usually sporadic)

Yes if it's...**bilateral** (do a complete genetic and systemic workup)

Central

Peters anomaly