In terms of the fundamental embryological disorder involved, anterior segment dysgenesis is what sort of condition?
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A neurocristopathy
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What is a neurocristopathy?
In terms of the fundamental embryological disorder involved, anterior segment dysgenesis is a condition.
A neurocristopathy

What is a neurocristopathy?
A congenital/developmental abnormality owing to flawed neural-crest cell migration or differentiation
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What is/are neural crest cells?
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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)
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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 ➔
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First wave → Corneal endothelium
Second wave → Iris stroma
Third wave → Corneal stroma (keratocytes)
Anterior segment dysgenesis

A very basic anatomic distinction

?   ?
Anterior segment dysgenesis

A very basic anatomic distinction

Peripheral  Central
Anterior Segment Dysgenesis

Anterior segment dysgenesis

Peripheral
- Two classic peripheral dysgeneses

Central

?
Anterior segment dysgenesis

Peripheral
- Posterior embryotoxon
- Two classic peripheral dysgeneses

Central
- Axenfeld-Rieger syndrome
Anterior segment dysgenesis

Peripheral

Posterior embryotoxon

Axenfeld-Rieger syndrome

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

Peripheral

Posterior embryotoxon

Axenfeld-Rieger syndrome
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
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

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Normal angle anatomy: Identify the structures
Anterior Segment Dysgenesis

Normal angle anatomy: Identify the structures
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Is it always a harbinger of significant pathology?

Axenfeld-Rieger syndrome

Posterior embryotoxon
Anterior Segment Dysgenesis

Anterior segment dysgenesis

Peripheral

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Axenfeld-Rieger syndrome

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
Anterior segment dysgenesis

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Interestingly, all three of these begin with the letter ‘A’
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Is aniridia usually unilateral, or bilateral?

Is nystagmus commonly associated with aniridia?

With what developmental ‘complex’ is aniridia associated?

Are all aniridia cases at risk for WAGR complex?
Anterior segment dysgenesis

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- **Why is the term ‘aniridia’ technically a misnomer?**
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  - It is almost always bilateral

- **Is nystagmus commonly associated with aniridia?**
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- **With what developmental ‘complex’ is aniridia associated?**
  - The WAGR complex

- **Are all aniridia cases at risk for WAGR complex?**
  - No, only those in which the genetic mutation is sporadic
Aniridia. Note the presence of an iris stub/root
**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
Anterior segment dysgenesis

**Anterior segment dysgenesis**

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What anatomic abnormalities are responsible for the poor vision in aniridia?
Foveal and optic nerve hypoplasia
**Anterior Segment Dysgenesis**

### Anterior segment dysgenesis

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**Mental note of aniridia’s ocular associations:**
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WAGR complex consists of:
- W (Wilms tumor)
- Aniridia
- G (Genitourinary abnormalities)
- R (Retardation)

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

- **Anterior 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.

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- **What is the noneponymous name for Wilms tumor (ie, what sort of tumor is it)?**
  - A nephroblastoma.

- **WAGR complex consists of:**
  - Wilms tumor
  - Aniridia
  - Genitourinary abnormalities
  - Retardation
Anterior Segment Dysgenesis

WAGR complex: Wilm’s tumor
Anterior Segment Dysgenesis

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Are all aniridia cases at risk for WAGR complex?
No, only those in which the genetic mutation is sporadic
**Anterior Segment Dysgenesis**

Anterior segment dysgenesis

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Are defects involving what gene the cause of aniridia?
No

---

**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

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**Defects involving what gene are the cause of aniridia?**
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### Anterior Segment Dysgenesis

#### Anterior segment dysgenesis

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### Notes
- **Axenfeld-Rieger syndrome**
- **aniridia**
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Anterior Segment Dysgenesis

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I’m glad you asked…
What four ocular abnormalities are attributed to the PAX6 gene?

There are four main abnormalities, and the term PAX6 acts as its own mnemonic. Start with the ‘P’ and make your way down…
What four ocular abnormalities are attributed to the PAX6 gene?

- Anterior Segment Dysgenesis
- Peters anomaly
What four ocular abnormalities are attributed to the **PAX6** gene?

- **Peters anomaly**
- **Anirida (duh)**
- **Anterior Segment Dysgenesis**
What four ocular abnormalities are attributed to the PAX6 gene?

- Peters anomaly
- Anirida (duh)
- Congenital cataract (say it out loud)
- Anterior Segment Dysgenesis
What four ocular abnormalities are attributed to the PAX6 gene?

- Peters anomaly
- Anirida (duh)
- Congenital cataract
- Foveal hypoplasia

*If you use your imagination, the 6 looks like a lower-case h...*
What four ocular abnormalities are attributed to the PAX6 gene?

- Peters anomaly
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We know that corneal opacities and foveal hypoplasia are associated with aniridia...
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...but are **cataracts** associated with it as well?

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Mental note of aniridia’s ocular associations:
--Nystagmus
--Foveal hypoplasia
--ON hypoplasia
--Peters anomaly
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We know that corneal opacities and foveal hypoplasia are associated with aniridia…

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What sort of gene is PAX6 anyway?

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If you use your imagination, the 6 looks like a lower-case h…
What sort of gene is PAX6 anyway?
A homeobox gene

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What sort of gene is PAX6 anyway?
A homeobox gene

What is a homeobox gene?
One that regulates morphogenesis
What sort of gene is PAX6 anyway?
A homeobox gene

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

One that regulates morphogenesis

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An anteriorly displaced and thickened Schwalbe’s line/ring

Is it always a harbinger of significant pathology?

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In what three situations is it a significant finding?

1) When it is part of the Axenfeld-Rieger syndrome
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Yes; arcus senilis is the anterior embryotoxon

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Is aniridia usually unilateral, or bilateral?

It is almost always bilateral

Is nystagmus commonly associated with aniridia?

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With what developmental ‘complex’ is aniridia associated?

The WAGR complex

Are all aniridia cases at risk for WAGR complex?

No, only those in which the genetic mutation is sporadic

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. Inherited genetic abnormalities leading to familial aniridia are located within the PAX6 gene itself, and thus do not affect the viability of the nearby WT1. In contrast, sporadic cases of aniridia are usually the result of the wholesale deletion of a chunk of genetic material in the PAX6 ‘neighborhood.’ And since WT1 is its next-door neighbor, it is often affected as well by these deletions. Because of the PAX6/WT1 spatial relationship, all infants presenting with sporadic aniridia must undergo genetic screening for the Wilms tumor defect.

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
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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.

Are all aniridia cases at risk for WAGR complex? No, only those in which the genetic mutation is **sporadic**.
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Are all aniridia cases at risk for WAGR complex?

No, only those in which the genetic mutation is **sporadic**.
**Anterior Segment Dysgenesis**

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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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*How often, and for how long?*

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Every 3 months until age 7 years

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Nystagmus

Mental note of aniridia’s ocular associations:
--Nystagmus
--Foveal hypoplasia
--ON hypoplasia
--Peters anomaly
--Congenital cataracts
--? Two more aniridia associations
--? we have yet to mention

Anterior Segment Dysgenesis

A corneal issue

Peters anomaly

foveal and optic nerve hypoplasia

Angle-related condition

cataracts
Nystagmus

Mental note of aniridia’s ocular associations:
-- Nystagmus
-- Foveal hypoplasia
-- ON hypoplasia
-- Peters anomaly
-- Congenital cataracts
-- Limbal stem-cell deficiency
-- Glaucoma

Anterior Segment Dysgenesis

limbal stem cell deficiency

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One final point regarding aniridia:

Peters anomaly
foveal and optic nerve hypoplasia

Anterior Segment Dysgenesis

limbal stem cell deficiency

glaucoma
cataracts
Nystagmus

limbal stem cell deficiency

Peters anomaly

foveal and optic nerve hypoplasia

glaucoma

cataracts

**Anterior Segment Dysgenesis**

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

One final point regarding aniridia:
Anterior segment dysgenesis

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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?
An infant with jaundice who presents to the eye service as a 'rule out Alagille syndrome' consult
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.
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Renal, neurological and vascular abnormalities are common as well.
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
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Is there such a thing as an anterior embryotoxon?
Yes; arcus senilis is the anterior embryotoxon
What is the noneponymous name of Alagille syndrome?
Arterohepatic dysplasia
How is it inherited?
Autosomal dominant, but the expressivity varies widely
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Alagille syndrome: Facies
Anterior Segment Dysgenesis

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Anterior Segment Dysgenesis

Alagille syndrome: Butterfly vertebrae
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 craniofacial malformation

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

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Anterior segment dysgenesis

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

Goldenhar syndrome: Hemifacial microsomia

Goldenhar: Ear abnormalities (Oculo-AURICULO-vertebral syndrome)
Anterior Segment Dysgenesis

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

For more on Goldenhar, see slide-set P22

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Yes--it is another name for arcus juvenilis

Arcus juvenilis
aka anterior embryotoxon
Anterior Segment Dysgenesis

**Anterior segment dysgenesis**

- **Peripheral**
  - Posterior embryotoxon
  - Axenfeld-Rieger syndrome

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

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.”

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 80, the prevalence is ~100%.
Anterior Segment Dysgenesis

**Regarding arcus senilis...**

*What is its main chemical component?*
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**Arcus senilis**

- It is the congenital version of arcus senilis

**Arcus juvenilis**
aka
*anterior embryotoxon*

**Corneal arcus**

**Axenfeld-Rieger syndrome**

**Posterior embryotoxon**

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Arcus juvenilis
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Arcus senilis

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Arcus juvenilis aka anterior embryotoxon

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Regarding arcus senilis...

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Bilateral.

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

Carotid occlusive dz, or ocular ischemic syndrome (OIS).

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What is arcus senilis?

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Is it always a harbinger of significant pathology?

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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?

Yes--it is another name for arcus juvenilis

What is arcus juvenilis?

It is the congenital version of arcus senilis

Regarding arcus senilis…

What is its main chemical component?

Cholesterol

Is it usually unilateral, or bilateral?

Bilateral

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

Carotid occlusive dz, or ocular ischemic syndrome (OIS)

What is arcus senilis?

It is the congenital version of arcus senilis
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?
Yes--it is another name for arcus juvenilis.

 Regarding arcus senilis...

What is its main chemical component?
Cholesterol.

Is it usually unilateral, or bilateral?
Bilateral.

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

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?
The side without the arcus.

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
What condition should be suspected if a pt has unilateral arcus?
Carotid occlusive dz, or ocular ischemic syndrome (OIS)
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? The side without the arcus

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

Corneal arcus
Arcus juvenilis aka anterior embryotoxon
Arcus senilis
Axenfeld-Rieger syndrome
Posterior embryotoxon
Peripheral
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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?

What is arcus juvenilis?
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Anterior Segment Dysgenesis

Regarding arcus senilis...

What is its main chemical component?
Cholesterol

Is it usually unilateral, or bilateral?
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Is it a dystrophy?
No; per the Cornea book, it is an “involutional change”

Arcus juvenilis

It is the congenital version of arcus senilis
Anterior Segment Dysgenesis

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”

- In what pattern does it typically declare itself?
  - It starts at the poles, then spreads circumferentially

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

Arcus senilis
  - aka anterior embryotoxon

Arcus juvenilis
  - Corneal arcus

Peripheral
  - Posterior embryotoxon
  - Axenfeld-Rieger syndrome
What is a posterior embryotoxon?

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What is arcus senilis?
It is the congenital version of arcus senilis
Anterior Segment Dysgenesis

Early arcus senilis
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”

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 80, the prevalence is ~100%

Arcus juvenilis
aka anterior embryotoxon

Arcus senilis

Regarding arcus senilis…

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

Corneal arcus

Axenfeld-Rieger syndrome

Axenfeld-Rieger syndrome

Posterior embryotoxon

Peripheral
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**Anterior Segment Dysgenesis**

**Regarding arcus senilis…**

- **What is its main chemical component?**
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- **Is it usually unilateral, or bilateral?**
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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

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

**Posterior embryotoxon**

**Axenfeld-Rieger syndrome**

**Corneal arcus**

**Arcus juvenilis** aka **anterior embryotoxon**

**Peripheral**
Anterior Segment Dysgenesis

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Arcus juvenilis
aka anterior embryotoxon

Arcus senilis

Arcus juvenilis
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Axenfeld-Rieger syndrome

Posterior embryotoxon

Peripheral

Corneal arcus

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

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

Arcus senilis
Arcus juvenilis
aka
anterior embryotoxon

Corneal arcus

Axenfeld-Rieger syndrome

Peripheral

Posterior embryotoxon
Anterior Segment Dysgenesis

Arcus senilis in older AAM
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?**
- Yes--it is another name for arcus juvenilis

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

**Arcus in an adult <40 places him/her at increased risk of what potentially lethal condition?**
- Coronary artery dz

**Regarding arcus senilis...**

**What is its main chemical component?**
- Cholesterol

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

**Does its prevalence increase with age?**
- Yes; after age 80, the prevalence is ~100%

**Does its prevalence increase before age 40?**
- Yes, before age 40

**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

**What is the underlying mechanism for both the arcus and the CAD in these pts?**
- Familial hyperlipoproteinemia

Another ophthalmic exam finding of hyperlipoproteinemia is often present in such pts. What is it?
- Xanthelasma
What is a posterior embryotoxon?

An anteriorly displaced and thickened Schwalbe’s line/ring

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Arcus juvenilis aka anterior embryotoxon

Arcus senilis

Arcus in an adult <40 places him/her at increased risk of what potentially lethal condition?

Coronary artery dz

Does its prevalence increase with age?

Yes, after age 80, the prevalence is ~100%

What is arcus juvenilis?

It is the congenital version of arcus senilis

Regarding arcus senilis...

What is its main chemical component?

Cholesterol

Is it usually unilateral, or bilateral?

Bilateral

Arcus in an adult <40 places him/her at increased risk of what potentially lethal condition?

Coronary artery dz

Does its prevalence increase with age?

Yes, after age 80, the prevalence is ~100%

What is arcus senilis?

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Regarding arcus senilis...
What is its main chemical component?
- Cholesterol
Is it usually unilateral, or bilateral?
- Bilateral

Arcus in an adult <40 places him/her at increased risk of what potentially lethal condition?
- Coronary artery dz

What is the underlying mechanism for both the arcus and the CAD in these pts?

Does its prevalence increase with age?
- Yes, after age 80, the prevalence is ~100%

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

*Is it usually unilateral, or bilateral?*
Bilateral

*Arcus in an adult <40 places him/her at increased risk of what potentially lethal condition?*
Coronary artery dz

*What is the underlying mechanism for both the arcus and the CAD in these pts?*
Familial hyperlipoproteinemia coupled with high cholesterol

*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
Anterior Segment Dysgenesis

**Anterior embryotoxon**

What is a posterior embryotoxon?

An anteriorly displaced and thickened Schwalbe’s line/ring

Is it always a harbinger of significant pathology?

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In what three situations is it a significant finding?

1) When it is part of the Axenfeld-Rieger syndrome
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Is there such a thing as an anterior embryotoxon?

Yes—It is another name for **arcus juvenilis**

**Arcus juvenilis** aka anterior embryotoxon

**Arcus senilis**

**Corneal arcus**

**Arcus in an adult <40 places him/her at increased risk of what potentially lethal condition?**

Coronary artery dz

**What is its main chemical component?**

Cholesterol

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

Bilateral

**What is the underlying mechanism for both the arcus and the CAD in these pts?**

Familial hyperlipoproteinemia coupled with high cholesterol

Another ophthalmic exam finding of hyperlipoproteinemia is 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**
Anterior Segment Dysgenesis

Regarding arcus senilis...

**What is its main chemical component?**
Cholesterol

**Is it usually unilateral, or bilateral?**
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Regarding arcus senilis…

What is its main chemical component?
Cholesterol

Is it usually unilateral, or bilateral?
Bilateral

Arcus in an adult <40 places him/her at increased risk of what potentially lethal condition?
Coronary artery dz

Well, obviously arcus juvenilis is occurring in someone under 40, so does this mean it is a sign of lipid derangement as well?

No, it is a benign finding

Does its prevalence increase with age?
Yes, after age 80, the prevalence is ~100%

Another ophthalmic exam finding of hyperlipoproteinemia is often present in such pts. What is it?
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What is arcus juvenilis?
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Corneal arcus

Arcus juvenilis
aka
anterior embryotoxon

Arcus senilis

Peripheral

Posterior

Axenfeld-Rieger syndrome

What is arcus senilis?

What is its main chemical component?
Cholesterol

Is it usually unilateral, or bilateral?
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Corneal arcus

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Does its prevalence increase with age?
Yes, after age 80, the prevalence is ~100%
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
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**Is there such a thing as an anterior embryotoxon?**
Yes—this is another name for arcus juvenilis.

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

**Regarding arcus senilis...**
- **What is its main chemical component?**
  Cholesterol
- **Is it usually unilateral, or bilateral?**
  Bilateral
- **Arcus in an adult <40 places him/her at increased risk of what potentially lethal condition?**
  Coronary artery dz

**Does its prevalence increase with age?**
Yes; after age 80, the prevalence is ~100%.

**Does arcus juvenilis occur before age 40?**
Yes, obviously arcus juvenilis is occurring in someone under 40, so does this mean it is a sign of lipid derangement as well? No, it is a benign finding.

**Corneal arcus**

**Arcus juvenilis**
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**What is arcus juvenilis?**
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Is there such a thing as an anterior embryotoxon?
Yes--it is another name for arcus juvenilis.

**Regarding arcus senilis...**

**What are xanthelasmas, that is, what is there 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?
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.

**Arcus juvenilis**
aka anterior embryotoxon

**Arcus senilis**

**Arcus in an adult <40 places him/her at increased risk of what potentially lethal condition?**
Coronary artery dz

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Another ophthalmic exam finding of hyperlipoproteinemia is often present in such pts. What is it?
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It is the congenital version of arcus senilis.
**Anterior Segment Dysgenesis**

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An anteriorly displaced and thickened Schwalbe’s line/ring.

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

**In what three situations is it a significant finding?**

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

Yes--it is another name for arcus juvenilis.

**What is arcus juvenilis?**

It is the congenital version of arcus senilis.

**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.”
- 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 80, the prevalence is ~100%.
- Arcus in an adult <40 places him/her at increased risk of what potentially lethal condition?
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Anterior Segment Dysgenesis

Xanthelasma
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?
Yes—another name for arcus juvenilis.

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

Regarding arcus senilis…

What is arcus senilis, that is, what is there clinical appearance?
They are yellowish plaques located in the medial canthal region, usually on the upper lids.

Do they present unilaterally, or bilaterally?
Bilaterally.

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".

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 80, the prevalence is ~100%.

Arcus in an adult <40 places him/her at increased risk of what potentially lethal condition?
Coronary artery dz.

What is the underlying mechanism for both the arcus and the CAD in these pts?
Familial hyperlipoproteinemia.

Another ophthalmic exam finding of hyperlipoproteinemia is often present in such pts. What is it?
Xanthelasma.

What are xanthelasma, that is, what is there 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?
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.
What are xanthelasmata, that is, what is there clinical appearance?
They are yellowish plaques located in the medial canthal region, usually on the upper lids.

Do they present unilaterally, or bilaterally?
Bilaterally

Regarding arcus senilis...

Does its prevalence increase with age?
Yes, after age 80, the prevalence is ~100%

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

What is arcus senilis?
It is the congenital version of arcus line/ring

Familial hyperlipoproteinemia

Another ophthalmic exam finding of hyperlipoproteinemia is often present in such pts. What is it?
Xanthelasma

Anterior Segment Dysgenesis

Central Axenfeld-Rieger syndrome

Embryotoxon

Posterior keratoconus

Peters anomaly

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?
Yes--it is another name for arcus juvenilis

Peripheral

Arcus juvenilis

aka anterior embryotoxon

Arcus senilis

"Cornea" book, it is an "involutional change"

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"

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?
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Bilaterally

Is it usually unilateral, or bilateral?
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Is it a dystrophy?
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Coronary artery dz

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Familial hyperlipoproteinemia

Another ophthalmic exam finding of hyperlipoproteinemia is often present in such pts. What is it?
Xanthelasma
Anterior Segment Dysgenesis

What is an anterior 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?
Yes--it is another name for arcus juvenilis.

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

Regarding arcus senilis...
What are xanthelasmias, that is, what is there 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.

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.

Xanthelasma

What is arcus senilis?
It is an “involutional change” in the cornea.

Regarding arcus senilis...
What is its main chemical component?
Cholesterol.

Is it usually unilateral, or bilateral?
Bilateral.

Is it a dystrophy?
No.

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.

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Familial hyperlipoproteinemia.

Another ophthalmic exam finding of hyperlipoproteinemia is often present in such pts. What is it?
Xanthelasma.

What are xanthelasmias, 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?
Yes, and when they are, they usually are a sign of lipid derangement.
Anterior Segment Dysgenesis

What is an 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?
Yes--it is another name for arcus juvenilis.

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

Arcus juvenilis
aka anterior embryotoxon

Arcus senilis

Regarding arcus senilis...

What are xanthelasmas, that is, what is there 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.

Do they present unilaterally, or bilaterally?
Bilaterally.

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

What is its main chemical component?
Cholesterol.

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Does it exhibit a racial predilection?
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Does its prevalence increase with age?
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Familial hyperlipoproteinemia.

Another ophthalmic exam finding of hyperlipoproteinemia is often present in such pts. What is it?
Xanthelasma.

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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.

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.
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?
Yes--it is another name for arcus juvenilis

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

Regarding arcus senilis…

What are xanthelasma, that is, what is there 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?

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

What is arcus senilis?
It is an involutional change

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”

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 80, the prevalence is ~100%

Arcus in an adult <40 places him/her at increased risk of what potentially lethal condition?
Coronary artery dz

What is the underlying mechanism for both the arcus and the CAD in these pts?
Familial hyperlipoproteinemia

Another ophthalmic exam finding of hyperlipoproteinemia is often present in such pts. What is it?
Xanthelasma

What are xanthelasmases, that is, what is there 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?
What are xanthelasma(s), that is, what is there 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? No, they can (and often do) appear in individuals with normal lipid panels.

What is arcus juvenilis? It is the congenital version of arcus senilis.
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?
Yes--it is another name for arcus juvenilis

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

What is arcus senilis?
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?
No, they can (and often do) appear in individuals with normal lipid panels

Can they be congenital?

Regarding arcus senilis...

What are xanthelasmata, that is, what is there 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?
No, they can (and often do) appear in individuals with normal lipid panels

Can they be congenital?

Does its prevalence increase with age?
Yes; after age 80, the prevalence is ~100%

What is xanthelasma?
It is a yellowish plaque located in the medial canthal region, usually on the upper lids

Does its prevalence increase with age?
Yes, after age 80, the prevalence is ~100%

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

What is arcus senilis?
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?

Yes--it is another name for arcus juvenilis.

Regarding arcus senilis...

What are xanthelasmas, that is, what is there 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?

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.

What is arcus juvenilis?

It is the congenital version of arcus senilis.

Does its prevalence increase with age?

Yes, after age 80, the prevalence is ~100%.

What is arcus juvenilis? It is the congenital version of arcus senilis.
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?
Yes--it is another name for arcus juvenilis.

Peripheral

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

Regarding arcus senilis...

What are xanthelasma, that is, what is there clinical appearance?
They are yellowish plaques located in the medial canthal region, usually on the upper lids.

This histology--'lipid-filled macrophages'--is often described with other, equivalent terms. What are they?
'Lipid filled' = ?
'Macrophages' = ?

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.

What is xanthelasma?
It is the congenital version of arcus senilis.

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.
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?
Yes—It is another name for arcus juvenilis.

What is arcus senilis?
It is the congenital version of arcus senilis.

Regarding arcus senilis...

What are xanthelasma, that is, what is there clinical appearance?
They are yellowish plaques located in the medial canthal region, usually on the upper lids.

This histology—'lipid-filled macrophages'—is often described with other equivalent terms. What are they?
‘Lipid filled’ = ‘foamy’
‘Macrophages’ = ‘histiocytes’

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.

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

Does its prevalence increase with age?
Yes; after age 80, the prevalence is ~100%

Before age 40

Xanthelasma

A corneal arcus in an adult <40 places him/her at increased risk of what potentially lethal condition?
Coronary artery dz

What is the underlying mechanism for both the arcus and the CAD in these pts?
Familial hyperlipoproteinemia

Another ophthalmic exam finding of hyperlipoproteinemia is often present in such pts. What is it?
Xanthelasma

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.

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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.

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

Does its prevalence increase with age?
Yes; after age 80, the prevalence is ~100% before age 40.
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?

Yes--it is another name for arcus juvenilis.

What is arcus juvenilis?

It is the congenital version of arcus senilis.

Regarding arcus senilis...

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

They are yellowish plaques.

The point being that the terms ‘foamy macrophages,’ ‘lipid-filled (or laden) macrophages,’ ‘foamy histiocytes,’ etc, all mean the same thing, so don’t be misled if you see one term when you’re expecting another.

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.

Can they present unilaterally, or bilaterally?

Bilaterally.

Are they composed of lipid?

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

What is the underlying mechanism for both the arcus and the CAD in these pts?

Familial hyperlipoproteinemia.

Another ophthalmic exam finding of hyperlipoproteinemia is often present in such pts. What is it?

Xanthelasma.

What is xanthelasma?

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?

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

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This histology--‘lipid-filled macrophages,’ ‘lipid-filled (or laden) macrophages,’ ‘foamy histiocytes,’ etc, all mean the same thing, so don’t be misled if you see one term when you’re expecting another.

The point being that the terms ‘foamy macrophages,’ ‘lipid-filled (or laden) macrophages,’ ‘foamy histiocytes,’ etc, all mean the same thing, so don’t be misled if you see one term when you’re expecting another.

‘Lipid filled’ = ‘foamy’

‘Macrophages’ = ‘histiocytes’

The point being that the terms ‘foamy macrophages,’ ‘lipid-filled (or laden) macrophages,’ ‘foamy histiocytes,’ etc, all mean the same thing, so don’t be misled if you see one term when you’re expecting another.

What is arcus juvenilis?

It is the congenital version of arcus senilis.

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

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

**What is arcus senilis?**
Its main chemical component is cholesterol.

Is it usually unilateral, or bilateral?
Bilateral.

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 AAs.

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.

**Xanthelasma**

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.

What is its main chemical component?
Cholesterol.

Is it usually unilateral, or bilateral?
Bilaterally.

Are they composed of lipid?
Yes, and when they are, they usually are a sign of lipid derangement.

What is xanthelasma?
They are yellowish plaques located in the medial canthal region, usually on the upper lids.

Are they composed of lipid?
Yes, and when they are, they usually are a sign of lipid derangement.

Can they be congenital?
Yes, and when they are, they usually are a sign of lipid derangement.

What is the underlying mechanism for both the arcus and the CAD in these pts?
Familial hyperlipoproteinemia.

Another ophthalmic exam finding of hyperlipoproteinemia is often present in such pts. What is it?
Xanthelasma.

Speaking of ‘foamy macrophages’…
What dz comes to mind if, instead of an adult with xanthelasmas, the pt in question was a middle-aged white guy with bilateral panuveitis? First clue--more forthcoming.

First clue--more forthcoming.

Xanthelasma

First clue--more forthcoming.
Anterior Segment Dysgenesis

Speaking of ‘foamy macrophages’…
What dz comes to mind if, instead of an adult with xanthelasmas, the pt in question was a middle-aged white guy with bilateral panuveitis? And a hx of chronic migratory arthritis? Clue #2

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.

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.
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?
Yes--it is another name for arcus juvenilis.

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

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.”

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 AA's.

Does its prevalence increase with age?
Yes; after age 80, the prevalence is ~100%.

Arcus in an adult <40 places him/her at increased risk of what potentially lethal condition?
Coronary artery dz.

What is the underlying mechanism for both the arcus and the CAD in these pts?
Familial hyperlipoproteinemia.

Another ophthalmic exam finding of hyperlipoproteinemia is often present in such pts. What is it?
Xanthelasma.

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.

Are they always present in such pts. What is it?
Xanthelasma.

What is arcus juvenilis?
It is the congenital version of arcus senilis.
Anterior Segment Dysgenesis

Speaking of ‘foamy macrophages’…
What dz comes to mind if, instead of an adult with xanthelasmas, the pt in question was a middle-aged white guy with bilateral panuveitis? And a hx of chronic migratory arthritis? Associated with chronic diarrhea? And CNS symptoms—nystagmus, dementia, coma?

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
Anterior Segment Dysgenesis

Speaking of ‘foamy macrophages’…
What dz comes to mind if, instead of an adult with xanthelasmas, the pt in question was a middle-aged white guy with bilateral panuveitis? And a hx of chronic migratory arthritis? Associated with chronic diarrhea? And CNS symptoms--nystagmus, dementia, coma?

Whipple’s disease

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?

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Whipple’s disease: **Duodenal biopsy** demonstrating **foamy macrophages** in the lamina propria. *This is the pic you’re looking for if you think the answer to an OKAP/Boards question is ‘Whipple’s.’*
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Speaking of ‘foamy macrophages’ part deaux…
What dz comes to mind if, instead of an adult with xanthelasmas, the pt in question was a very young child with unilateral pigmented iris nodules?

Juvenile xanthogranuloma (JXG)

First clue…

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

Corneal arcus

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"
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 AAAs
Does its prevalence increase with age?
Yes; after age 80, the prevalence is ~100%

Arcus in an adult <40 places him/her at increased risk of what potentially lethal condition?
Coronary artery dz
What is the underlying mechanism for both the arcus and the CAD in these pts?
Familial hyperlipoproteinemia
Another ophthalmic exam finding of hyperlipoproteinemia is often present in such pts. What is it?
Xanthelasma

Speaking of ‘foamy macrophages’…
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What is arcus juvenilis?
It is the congenital version of arcus senilis

Arcus juvenilis
aka anterior embryotoxon

Post embryotoxon

Central
dysgenesis

Axenfeld-Rieger syndrome

Posterior keratoconus

Peters anomaly

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?
Yes--it is another name for arcus juvenilis

Arcus senilis

Corneal arcus

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Xanthelasma

Speaking of ‘foamy macrophages’ part deaux…
What dz comes 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 2ndry to those nodules?

Juvenile xanthogranuloma (JXG)

Second clue

What is arcus juvenilis?
It is the congenital version of arcus senilis
Anterior Segment Dysgenesis

Speaking of ‘foamy macrophages’…
What dz comes to mind if, instead of an adult with xanthelasmas, the pt in question was a middle-aged white guy with bilateral panuveitis?
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And heterochromia iridis 2ndry to those nodules?
Along with a nontraumatic hyphema in the affected eye?

Clue #3

Arcus juvenilis
aka anterior embryotoxon

What is arcus juvenilis?
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Another ophthalmic exam finding of hyperlipoproteinemia is often present in such pts. What is it?
Xanthelasma
Before age 40

What are xanthelasmas, that is, what is there 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?
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

Speaking of ‘foamy macrophages’ part deaux…
What dz comes to mind if, instead of an adult with xanthelasmas, the pt in question was a middle-aged white guy with bilateral panuveitis?
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Anterior Segment Dysgenesis

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And heterochromia iridis 2ndry to those nodules?
Along with a nontraumatic hyphema in the affected eye?
Associated with orangish skin papules?

Last clue--the answer is next

Whipple’s disease

xanthelasma

lipid-filled macrophages
Anterior Segment Dysgenesis

Speaking of ‘foamy macrophages’… What dz comes to mind if, instead of an adult with xanthelasmas, the pt in question was a middle-aged white guy with bilateral panuveitis? And a hx of chronic migratory arthritis? Associated with chronic diarrhea? And CNS symptoms--nystagmus, dementia, coma? Whipple’s disease

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? Yes; these typically occur in pts with familial hyperlipoproteinemia. Speaking of ‘foamy macrophages’ part deaux… What dz comes 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 2ndry to those nodules? 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.
Anterior Segment Dysgenesis

Skin papules. The orange color is classic.

Spontaneous hyphema.

Touton giant cells.

Foamy macrophages.

JXG.
Anterior Segment Dysgenesis

Skin papules. The orange color is classic.

Spontaneous hyphema

Foamy macrophages

For more on JXG, see slide-set FELT8
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

Finally: Anterior embryotoxon is another name for arcus juvenilis. Is there an equivalent ‘another name’ for arcus senilis?
Anterior segment dysgenesis

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Yes--anterior **gerontoxon**
**Anterior Segment Dysgenesis**

**Anterior segment dysgenesis**

- **Posterior embryotoxon**
  - 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
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    3) When it is associated with Alagille syndrome

- **Arcus juvenilis**
  - Is there an equivalent ‘another name’ for arcus juvenilis? Yes--anterior gerontoxon

- **Arcus senilis**
  - Is there an equivalent ‘another name’ for arcus senilis? Yes--anterior gerontoxon

- **Peripheral**
  - Posterior embryotoxon
  - Axenfeld syndrome

- **Corneal arcus**
  - Arcus juvenilis aka anterior embryotoxon
  - Arcus senilis aka anterior gerontoxon

Finally: Anterior embryotoxon is another name for arcus juvenilis. Is there an equivalent ‘another name’ for arcus senilis? Yes--anterior gerontoxon.
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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**

<table>
<thead>
<tr>
<th></th>
<th>Embryotoxon</th>
<th>Gerontoxon</th>
</tr>
</thead>
<tbody>
<tr>
<td>Anterior</td>
<td>Arcus juvenilis</td>
<td>Arcus senilis</td>
</tr>
<tr>
<td>Posterior</td>
<td>Posterior embryotoxon</td>
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*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?*
**Anterior Segment Dysgenesis**

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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)

- **Embryotoxon**: An anteriorly displaced and thickened Schwalbe’s line/ring.
- **Gerontoxon**: Another name for arcus juvenilis.

---

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

Yes--it is another name for arcus juvenilis.

**Arcus juvenilis**

It is the congenital version of arcus senilis.

**Arcus senilis**

aka anterior gerontoxon

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

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Yeah, ‘geron-’ as in gerontology, as in old folks.
What features define Axenfeld-Rieger syndrome?

Axenfeld-Rieger syndrome

Peripheral

Posterior embryotoxon

Anterior segment dysgenesis

Megalocornea

Microcornea

Abnormal dentition

Characteristic facies

Periumbilical skin folds

Cardiac valve problems
Anterior segment dysgenesis

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

Peripheral

Posterior embryotoxon

Axenfeld-Rieger syndrome

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
3) Periumbilical skin folds
4) Cardiac valve problems
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

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

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

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‘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

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Anterior Segment Dysgenesis

**Peripheral**

- Posterior embryotoxon

**Axenfeld-Rieger syndrome**

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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.
Anterior Segment Dysgenesis

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**Anterior Segment Dysgenesis**

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What does the term ectropion uveae refer to?

The presence of posterior pigmented iris epithelium on the anterior surface of the iris
Technically speaking, the term a misnomer. Why?
Because the posterior pigmented epithelium derives from neuroectoderm, not uvea.
Anterior Segment Dysgenesis

Anterior segment dysgenesis

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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?
Because the posterior pigmented epithelium derives from neuroectoderm, not uvea
Anterior Segment Dysgenesis

**Peripheral**
- Posterior embryotoxon
- Axenfeld-Rieger syndrome

**Axenfeld-Rieger syndrome**
- 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
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**What corneal abnormalities may be present?**
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Not simultaneously, obviously
Anterior Segment Dysgenesis

Peripheral

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Not simultaneously, obviously
Megalocornea in a 2 y.o. with Axenfeld-Reiger
**Anterior Segment Dysgenesis**

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**Peripheral**

Anterior segment dysgenesis

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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

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Anterior Segment Dysgenesis

Axenfeld-Reiger with microcornea (8.5 mm)
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That pic should have immediately reminded you of another condition (pictured above)—what is it?
Anterior Segment Dysgenesis

Axenfeld-Reiger with microcornea (8.5 mm)

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Essential iris atrophy

Essential iris atrophy is a variant/form of what condition?
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**Anterior Segment Dysgenesis**

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Is it unilateral, or bilateral?
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Does it tend to affect males, or females?
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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)
Axenfeld-Reiger with microcornea (8.5 mm)

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For more on ICE, see slide-set K26
Anterior segment dysgenesis

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Anterior Segment Dysgenesis

(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
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Name two other congenital conditions that include both ocular involvement and abnormal dentition:
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In one word, what sort of condition is IP?
--Phakomatosis

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

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

What nonocular abnormalities may be present?
1) Abnormal dentition
2) Characteristic facies
3) Periumbilical skin folds
4) Cardiac valve problems

Name two other congenital conditions that include both ocular involvement and abnormal dentition:
- Incontinentia pigmenti
- Congenital syphilis

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

What description is commonly applied to the appearance of Hutchinson teeth?
‘Peg shaped’
Anterior Segment Dysgenesis

Congenital syphilis: Hutchinson teeth
Congenital syphilis: Hutchinson teeth

For more on congenital syphilis, see slide-set U16
Anterior segment dysgenesis

Peripheral
- Posterior embryotoxon
- Axenfeld-Rieger syndrome

Central
- Two classic central dysgeneses
- ?
- ?
Anterior Segment Dysgenesis

Anterior segment dysgenesis

Peripheral
- Posterior embryotoxon
- Axenfeld-Rieger syndrome

Central
- Posterior keratoconus
- Peters anomaly

Two classic central dysgeneses
What is posterior keratoconus?

Posterior keratoconus is a focal and discrete indentation of the posterior corneal surface. It is not secondary to a defect in the endothelium and/or Descemet's, as both are usually present and intact. It is rare and affects vision, causing irregular astigmatism, which can be severe enough to result in amblyopia. Most cases are unilateral and sporadic.
**What is posterior keratoconus?**
A focal and discrete indentation of the posterior corneal surface

**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
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?

- Posterior keratoconus
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Is the indentation secondary to a defect in the endothelium and/or Descemet’s?
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Is it common, like regular keratoconus?
No, it is rare

Does it affect vision?
Yes, it causes irregular astigmatism, which can be severe enough to result in amblyopia

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**Anterior Segment Dysgenesis**

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Anterior Segment Dysgenesis
**Anterior Segment Dysgenesis**

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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?
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

Does it require a workup?
No if it's unilateral (usually sporadic)
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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
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**Anterior Segment Dysgenesis**

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Anterior Segment Dysgenesis

Peters anomaly: Hazy cornea
What is Peters anomaly?
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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 mass.

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
P
E
D

Note: Peters anomaly
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?
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 mass.

What is the STUMPED mnemonic for a cloudy cornea in an infant?
- Sclerocornea
- Stromal dystrophy (CHSD)
- Trauma (eg, forcep injury)
- Ulcer
- Mucopolysaccharidosis
- Peters anomaly
- Endothelial dystrophy (CHED)
- Elevated IOP (congenital glaucoma)
- Dermoid of the cornea

Note: There are two S’s and two E’s
Anterior segment dysgenesis

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How might the lens be involved?
--It is often...
--It may be...
--Occasionally it is...

three word location (four words)
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Peters anomaly: Cataractous lens
Anterior segment dysgenesis

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Does it require a workup?
No if it's...unilateral (usually sporadic)Yes if it's...bilateral (do a complete genetic and systemic workup)

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

Is microspherophakia associated with Peters anomaly?
Yes, although only "occasionally" per the BCSC

In a few words, how would you describe the shape of a microspherophakic lens?
The name says it all: the lens is small ('micro') and round ('sphero')

If zonular laxity allows the lens to drift forward, what clinical condition may result?
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Anterior Segment Dysgenesis

Anterior segment dysgenesis

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What common slit-lamp observation owes to the lens’ small size?
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Anterior Segment Dysgenesis
**Anterior Segment Dysgenesis**

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**What common slit-lamp observation owes to the lens’ small size?**
Typically, the entirety of the lens equator can be seen in the pupillary aperture when the pt is widely dilated

**--It**
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How does refractive status manifest the lens' spherical shape?
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Anterior Segment Dysgenesis

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

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- Pupillary block angle-closure glaucoma
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When you hear microspherophakia, don’t think ‘Peters anomaly.’ Instead, what condition should come immediately to mind?

Weill-Marchesani syndrome

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

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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
What is notable about their joints?
They are stiff
**Anterior Segment Dysgenesis**

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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?**
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

**Does it require a workup?**
No if it’s unilateral (usually sporadic)
Yes if it’s bilateral (do a complete genetic and systemic workup)

Hmm… The notion of a ‘small, misshapen’ lens in this context should bring to mind particular condition. What is it?
**Microspherophakia**

Is microspherophakia associated with Peters anomaly?
Yes, although *only “occasionally”* per the BCSC Lens book

**When you hear microspherophakia, what should you think about the lens?**
- It is often cataractous
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When zonular laxity allows the lens to drift forward, what clinical condition may result?
The lens may ‘clog’ the pupillary opening, resulting in pupillary block angle-closure glaucoma

When you hear microspherophakia, don’t think ‘Peters anomaly.’ Instead, what condition should come immediately to mind?
**Weill-Marchesani syndrome**

**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
Anterior Segment Dysgenesis

Weill-Marchesani syndrome: Short stature
Anterior Segment Dysgenesis

Anterior segment dysgenesis

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In a few words, how would you describe the shape of a microspherophakic lens?
The name says it all: the lens is small (‘micro’) and round (‘sphero’)

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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

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

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