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 neurocristopathy
Anterior Segment Dysgenesis

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

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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
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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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Which wave involves which future structure?
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Second wave ➔ Iris stroma
Third wave ➔ Corneal stroma (keratocytes)
Anterior Segment Dysgenesis

Anterior segment dysgenesis

A very basic anatomic distinction

? ?
Anterior Segment Dysgenesis

Anterior segment
dysgenesis

A very basic
anatomic distinction

Peripheral  Central
Anterior Segment Dysgenesis

Anterior segment dysgenesis

Peripheral

Central

Two classic peripheral dysgeneses
Anterior Segment Dysgenesis

**Anterior segment dysgenesis**

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

- **Central**

**Two classic peripheral dysgeneses**
Anterior Segment Dysgenesis

**Anterior segment dysgenesis**

Peripheral

- Posterior embryotoxon
- Axenfeld-Rieger syndrome

*What is a posterior embryotoxon?*

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 Alagille syndrome
3. ‘Rule out Alagille syndrome’

Is there such a thing as an anterior embryotoxon?

Yes; arcus senilis is the anterior embryotoxon.
Anterior Segment Dysgenesis

**What is a posterior embryotoxon?**
An anteriorly displaced and thickened Schwalbe’s line/ring

Peripheral

- **Posterior embryotoxon**
- **Axenfeld syndrome**
Anterior Segment Dysgenesis

Anterior segment dysgenesis

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
- Normally not apparent during slit-lamp examination
- Often 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

Anterior segment dysgenesis

Peripheral

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Normal angle anatomy: Identify the structures
Normal angle anatomy: Identify the structures
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Interestingly, all three of these begin with the letter ‘A’
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Yes; arcus senilis is the anterior embryotoxon

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

Are all aniridia cases at risk for WAGR complex?

- No, only those in which the genetic mutation is sporadic
Why is the term ‘aniridia’ technically a misnomer? Because a rudimentary iris root is always present.
Aniridia. Note the presence of an iris stub/root
Anterior Segment Dysgenesis

Anterior segment dysgenesis

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**Anterior segment dysgenesis**

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Is aniridia usually unilateral, or bilateral? It is almost always bilateral.

Is nystagmus commonly associated with aniridia? Yes.

Is this a sensory or a motor nystagmus? Sensory.

Is it a jerk, or a pendular nystagmus? Pendular.

What is a posterior embryotoxon? An anteriorly displaced and thickened Schwalbe's line/ring.

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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.

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Anterior Segment Dysgenesis

Anterior segment dysgenesis

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   Yes.

4. **Is this a sensory or a motor nystagmus?**
   Sensory.

5. **What anatomic abnormalities are responsible for the poor vision in aniridia?**
   Foveal and optic nerve hypoplasia.

6. **Is there such a thing as an anterior embryotoxon?**
   Yes; arcus senilis is the anterior embryotoxon.

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   Yes.

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11. **What anatomic abnormalities are responsible for the poor vision in aniridia?**
    Foveal and optic nerve hypoplasia.

12. **What are the three situations in which an anterior embryotoxon is 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.

13. **Is the term ‘aniridia’ technically a misnomer?**
    Because a rudimentary iris root is always present.

14. **Is aniridia usually unilateral, or bilateral?**
    It is almost always bilateral.

15. **Is nystagmus commonly associated with aniridia?**
    Yes.

16. **With what developmental ‘complex’ is aniridia associated?**
    The WAGR complex.

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

18. **Is this a sensory or a motor nystagmus?**
    Sensory.

19. **Is it a jerk, or a pendular nystagmus?**
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**Anterior Segment Dysgenesis**

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Aniridia
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*With what developmental ‘complex’ is aniridia associated?*
The WAGR complex.

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

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WAGR complex consists of:
- Wilms tumor
- Aniridia
- Genitourinary abnormalities
- Retardation

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

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

 retardation
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.
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- Are all aniridia cases at risk for WAGR complex?
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- Is there such a thing as an anterior embryotoxon?
  - Yes; arcus senilis is the anterior 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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The PAX6 gene

What other ocular abnormalities are associated with defects of the PAX6 gene?
I’m glad you asked…

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The PAX6 gene.

**What other ocular abnormalities are associated with defects of the PAX6 gene?**
Axenfeld-Rieger syndrome, aniridia, Alagille syndrome.
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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?
What four ocular abnormalities are attributed to the PAX6 gene?

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

- Peters anomaly
- Anirida (duh)
- Congenital cataract
- (say it out loud)
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
- Anirida (duh)
- Congenital cataract
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We know that corneal opacities and foveal hypoplasia are associated with aniridia…
What four ocular abnormalities are attributed to the PAX6 gene?

- Peters anomaly
- Aniridia (duh)
- Congenital cataracts
- Foveal hypoplasia

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

We know that corneal opacities and foveal hypoplasia are associated with aniridia…
What four ocular abnormalities are attributed to the PAX6 gene?

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

We know that corneal opacities and foveal hypoplasia are associated with aniridia…
What sort of gene is PAX6 anyway?

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
- Anirida (duh)
- Congenital cataract
- Hyphoplasia

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

What sort of gene is PAX6 anyway? A homeobox gene.
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 sort of gene is PAX6 anyway?
A homeobox gene

What is a homeobox gene?
One that regulates morphogenesis

What four ocular abnormalities are attributed to the PAX6 gene?

Peter's anomaly

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Congenital cataract

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

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?

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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

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?

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.

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

--The heart: Septal defects, PDA, and tetralogy of Fallot are common
--The skeleton: The classic finding is 'butterfly vertebrae'
Renal, neurological and vascular abnormalities are common as well.
Anterior Segment Dysgenesis

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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) 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; 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.

What is the classic presentation?
An infant with jaundice who presents to the eye service as a ‘rule out Alagille syndrome’ consult.
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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.

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

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

The skeleton: The classic finding is ‘butterfly vertebrae’.

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Alagille syndrome
Alagille syndrome: Facies
Anterior Segment Dysgenesis

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Alagille syndrome: Butterfly vertebrae
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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
**Anterior Segment Dysgenesis**

*Anterior segment dysgenesis*

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Rieger syndrome
Anterior Segment Dysgenesis

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Significant finding?
Rieger syndrome
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Anterior Segment Dysgenesis

**Anterior segment dysgenesis**

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

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Anterior Segment Dysgenesis

Anterior segment dysgenesis

Peripheral

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**Anterior Segment Dysgenesis**

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**Arcus juvenilis**

aka

**anterior embryotoxon**

**What is arcus juvenilis?**

- It is the congenital version of **arcus senilis**
Anterior Segment Dysgenesis

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Corneal arcus

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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%
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Bilateral.

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

What is arcus senilis?
It is an involutional change, affecting the peripheral cornea.

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

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Yes, men are more likely to develop it.

Does it exhibit a racial predilection?
Yes, it is more common in African Americans.

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**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

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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? Cholesterol

Is it usually unilateral, or bilateral? Bilateral

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

Arcus juvenilis aka anterior embryotoxon

Corneal arcus

Peripheral

Axenfeld-Rieger syndrome

Posterior embryotoxon

Central

Posterior keratoconus

Peters anomaly

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

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  - Yes, men are more likely to develop it

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Arcus juvenilis aka anterior embryotoxon

Arcus senilis
Anterior Segment Dysgenesis

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Arcus juvenilis

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Arcus senilis

Corneal arcus

Peripheral

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Does it exhibit a racial predilection?

Yes, it is more common in African Americans

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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

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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?

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

Arcus juvenilis
aka anterior embryotoxon

Arcus senilis
Corneal arcus

Axenfeld-Rieger syndrome

Posterior embryotoxon

Peripheral

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

What is arcus juvenilis?
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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

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

Corneal arcus

Arcus juvenilis aka anterior embryotoxon

Arcus senilis

Axenfeld-Rieger syndrome

Peripheral

Posterior embryotoxon

Before age 40
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
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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

Does it usually present before age 40?
Yes—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

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

Anterior segment dysgenesis

Central

Axenfeld-Rieger syndrome

Posterior keratoconus

Peters anomaly

What is a posterior embryotoxon?

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

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

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?

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

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3. When it is associated with Alagille syndrome.

Is there such a thing as an anterior embryotoxon?
Yes—another name for arcus juvenilis.

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% before age 40.

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?
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3) When it is associated with Alagille syndrome

Is there such a thing as an anterior embryotoxon?
Yes—the same ring/ring 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?
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Is there such a thing as an anterior embryotoxon?
Yes--it is another name for arcus juvenilis

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

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

Is it a harbinger of significant pathology?
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%

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

Does arcus juvenilis occur in young adults?
Yes, before age 40

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

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.

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

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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

**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?**
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**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.

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.
Anterior Segment Dysgenesis

Regarding arcus senilis...

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

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

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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 are xanthelasmas, that is, what is there clinical appearance?
They are yellowish plaques 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.

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 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; it is a congenital 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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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 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.
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.

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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.

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.
Anterior Segment Dysgenesis

What is an anterior embryotoxon?

It is another name for arcus juvenilis, also known as anterior embryotoxon.

What is arcus juvenilis?

It is the congenital version of arcus senilis.

What is arcus senilis?

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.

Does its prevalence increase with age?

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

What is xanthelasma?

It is often present in such pts. What is it?

Xanthelasma.

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

Xanthelasma.

What is familial hyperlipoproteinemia?

The underlying mechanism for both the arcus and the CAD in these pts is familial hyperlipoproteinemia.

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Bilaterally.

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Can they be congenital?

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

What is coronary artery disease?

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 disease.

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Anterior Segment Dysgenesis

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? Yes, 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.

Arcus senilis

Arcus juvenilis
aka anterior embryotoxon
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?
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Does its prevalence increase with age?
Yes; after age 80, the prevalence is ~100%.

Does arcus juvenilis place an individual at increased risk of what potentially lethal condition?
Coronary artery dz.

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What is arcus juvenilis?
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Anterior Segment Dysgenesis

Regarding arcus senilis...

**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?**
No; they can (and often do) appear in individuals with normal lipid panels

**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

Arcus juvenilis
aka anterior embryotoxon

Arcus senilis
Anterior Segment Dysgenesis

Regarding arcus senilis...

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What is arcus juvenilis?
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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 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 senilis?**

It 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

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Does its prevalence increase with age?
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before age 40
Anterior Segment Dysgenesis

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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 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.
Anterior Segment Dysgenesis

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.

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

‘Lipid filled’ = ‘foamy’
‘Macrophages’ = ‘histiocytes’

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 senilis
Arcus juvenilis aka anterior embryotoxon

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.
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 xanthelasmas, that is, what is their clinical appearance?
They are yellowish plaques up front.

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 senilis?
It is an involutional change.

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

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

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? 

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.
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

Arcus juvenilis aka anterior embryotoxon

Arcus senilis

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

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...
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 AAs.

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

Does its prevalence increase before age 40?
No.

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

Speaking of ‘foamy macrophages’...
What disease 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? Need 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; xanthelasma 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

Speaking of ‘foamy macrophages’…
What disease comes to mind if, instead of an adult with xanthelasmas, the patient in question was a middle-aged white guy with bilateral panuveitis?
And a history 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.

What is xanthelasma?
It is often present in such patients. What is it?

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

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

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What is arcus juvenilis?

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Arcus juvenilis

aka anterior embryotoxon

Arcus senilis
Anterior Segment Dysgenesis

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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...
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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?
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What are xanthelasmas, that is, what is there clinical appearance?
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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 very young child with unilateral pigmented iris nodules?
Juvenile xanthogranuloma (JXG).

First clue...

Whipple’s disease
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?
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Whipple’s disease

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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?
And heterochromia iridis 2ndry to those nodules?
Second clue

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

Posterior embryotoxon

Axenfeld-Rieger syndrome
Peters anomaly

Central

Peripheral

Embryotoxon

Arcus juvenilis
aka anterior embryotoxon

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

Arcus senilis
Corneal arcus

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What is arcus juvenilis?
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Anterior Segment Dysgenesis

What is a posterior embryotoxon?
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And heterochromia iridis 2ndry to those nodules?
Along with a nontraumatic hyphema in the affected eye?"
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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?
Whipple's disease.

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?
And a hx of chronic migratory arthritis?
Associated with chronic diarrhea?
And CNS symptoms--nystagmus, dementia, coma?
Whipple's disease.

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'...
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 As.

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 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'... What dz comes to mind if, instead of an adult with xanthelasm, 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.

Speaking of 'foamy macrophages' part deaux... What dz comes to mind if, instead of an adult with xanthelasm, 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).
Anterior Segment Dysgenesis

**Anterior segment dysgenesis**

- **Posterior embryotoxon**
- Axenfeld-Rieger syndrome
- **Corneal arcus**
  - Arcus juvenilis (aka anterior embryotoxon)
  - Arcus senilis (aka anterior gerontoxon)

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**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-Rieger syndrome

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Yes--anterior **gerontoxon**

**Gerontoxon?**
Anterior Segment Dysgenesis

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Gerontoxon?
Yeah, ‘geron-’ as in gerontology, as in old folks
Anterior Segment Dysgenesis

Peripheral

Posterior embryotoxon

Axenfeld-Rieger syndrome

What features define Axenfeld-Rieger syndrome?

1. Posterior embryotoxon with attached iris strands
2. Iris hypoplasia
3. 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
Anterior Segment Dysgenesis

What features define Axenfeld-Rieger syndrome?
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Posterior embryotoxon

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Normal iris strands attached to SS
Abnormal iris strands attached to posterior embryotoxon in A-R
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?

Axenfeld-Rieger syndrome

Peripheral

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

Peripheral

Anterior segment dysgenesis

Axenfeld-Rieger syndrome

Posterior embryotoxon

What features define Axenfeld-Rieger syndrome?
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‘Angle abnormalities’ suggests an increased risk of glaucoma. Does ARS in fact convey such a risk?

Megalocornea
Microcornea

What corneal abnormalities may be present?

Abnormal dentition
Characteristic facies
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What is the lifetime risk of developing glaucoma?
50%
Anterior Segment Dysgenesis

Anterior segment dysgenesis

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The lifetime risk of developing glaucoma is 50%.
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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
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Corectopia
Anterior Segment Dysgenesis

**Peripheral**

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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.
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2) Not simultaneously, obviously

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Peripheral

Axenfeld-Rieger syndrome

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**Anterior Segment Dysgenesis**

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**Axenfeld-Rieger syndrome**

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*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.
A-R with microcornea (8.5 mm)
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(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**

**Peripheral**

- Posterior embryotoxon

**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
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
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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?
A 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

**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
Anterior Segment Dysgenesis

Anterior segment dysgenesis

Peripheral
- Posterior embryotoxon
- Axenfeld-Rieger syndrome

Central
- Posterior keratoconus
  - Also known as:
    - Circumscribed posterior keratoconus
    - Centralized posterior keratoconus
    - Posterior corneal depression
    - Von Hippel internal ulcer
- Peters anomaly

Note: Posterior keratoconus is also known as:
- Circumscribed posterior keratoconus
- Centralized posterior keratoconus
- Posterior corneal depression
and, most colorfully, as
- Von Hippel internal ulcer
Anterior Segment Dysgenesis

Anterior segment dysgenesis

What is posterior keratoconus?

Central

Posterior keratoconus

Peters anomaly
Anterior Segment Dysgenesis

What is posterior keratoconus?
A focal and discrete indentation of the posterior corneal surface

Central

Posterior keratoconus

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Anterior Segment Dysgenesis

**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?*

Central

- Posterior keratoconus
- Peters anomaly
Anterior Segment Dysgenesis

**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?**
No, both are usually present and intact

Central

Posterior keratoconus

Peters anomaly
Anterior Segment Dysgenesis

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Does it affect vision?
Anterior Segment Dysgenesis

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No, it is very rare

Does it affect vision?
Yes, it causes irregular astigmatism, which can be severe enough to result in amblyopia
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 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)
**Anterior Segment Dysgenesis**

*Anterior segment dysgenesis*

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

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Peters anomaly
Peters anomaly
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Peters anomaly: Hazy cornea
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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
T
M
P
E
D
Peters anomaly

Central

Peters anomaly

Peripheral
Anterior Segment Dysgenesis

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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

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Anterior segment dysgenesis

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How might the lens be involved?
--It is often...
--It may be... to the...
--Occasionally it is... size, shape, and location (four words)
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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
Peters anomaly: Cataractous lens
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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

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?

The lens may ‘clog’ the pupillary opening, resulting in pupillary block angle-closure glaucoma.
Anterior Segment Dysgenesis

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Typically, the entirety of the lens equator can be seen in the pupillary aperture when the pt is widely dilated

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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 Lens book

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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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 mess.

How might the lens be involved?
-- It is often cataractous
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Anterior segment dysgenesis

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