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

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

What is a neurocristopathy?
A congenital/developmental abnormality owing to flawed neural-crest cell migration or differentiation
**Anterior Segment Dysgenesis**

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

A neurocristopathy

What is a neurocristopathy?
A congenital/developmental abnormality owing to flawed neural-crest cell migration or differentiation

What is/are neural crest cells?
In terms of the fundamental embryological disorder involved, anterior segment dysgenesis is what sort of condition?
A neurocristopathy

What is a neurocristopathy?
A congenital/developmental abnormality owing to flawed neural-crest cell migration or differentiation

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

What is a neurocristopathy?
A congenital/developmental abnormality owing to flawed neural-crest cell migration or differentiation

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

Neural-crest-cell migration concerning the anterior segment occurs in three ‘waves.’
Which wave involves which future structure?
First wave ➔
Second wave ➔
Third wave ➔
In terms of the fundamental embryological disorder involved, anterior segment dysgenesis is what sort of condition?
A neurocristopathy

What is a neurocristopathy?
A congenital/developmental abnormality owing to flawed neural-crest cell migration or differentiation

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

Neural-crest-cell migration concerning the anterior segment occurs in three ‘waves.’
Which wave involves which future structure?
First wave → Corneal endothelium
Second wave →
Third wave →
In terms of the fundamental embryological disorder involved, anterior segment dysgenesis is what sort of condition?
A **neurocristopathy**

**What is a neurocristopathy?**
A congenital/developmental abnormality owing to flawed neural-crest cell migration or differentiation

**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

**Neural-crest-cell migration concerning the anterior segment occurs in three ‘waves.’**
Which wave involves which future structure?
- **First wave** → **Corneal endothelium**
- **Second wave** → **Iris stroma**
- **Third wave** →
In terms of the fundamental embryological disorder involved, anterior segment dysgenesis is what sort of condition?
A neurocristopathy

What is a neurocristopathy?
A congenital/developmental abnormality owing to flawed neural-crest cell migration or differentiation

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

Neural-crest-cell migration concerning the anterior segment occurs in three ‘waves.’
Which wave involves which future structure?
First wave ➔ Corneal endothelium
Second wave ➔ Iris stroma
Third wave ➔ Corneal stroma (keratocytes)
Anterior Segment Dysgenesis

Anterior segment dysgenesis

A very basic anatomic distinction

?  ?
Anterior segment dysgenesis

A very basic anatomic distinction

Peripheral Central
Anterior segment dysgenesis

Peripheral

Central

Two classic peripheral dysgeneses
Anterior segment dysgenesis

Peripheral

- Posterior embryotoxon
- Two classic peripheral dysgeneses

Central

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.

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 (rule out Alagille syndrome)

Is there such a thing as an anterior embryotoxon?

Yes; arcus senilis is the anterior embryotoxon.
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

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 familial aniridia
3) When it is associated with Alagille syndrome

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

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

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
Normal angle anatomy: Identify the structures
Normal angle anatomy: Identify the structures
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

Is it normally apparent during slit-lamp examination?
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
Posterior embryotoxon
**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?*
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

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

Anterior segment dysgenesis

Peripheral

Posterior embryotoxon

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?
Anterior segment dysgenesis

Peripheral

Posterior embryotoxon

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

Is it always a harbinger of significant pathology?
No; it is found in about 15% of otherwise normal eyes

In what three situations is it a significant finding?
1) 
2) 
3)
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

Interestingly, all three of these begin with the letter ‘A’
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

Interestingly, all three of these begin with the letter ‘A’
Anterior segment dysgenesis

**Anterior segment dysgenesis**

*Why is the term ‘aniridia’ technically a misnomer?*

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

*Is it always a harbinger of significant pathology?*

*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**

*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.
Anterior Segment Dysgenesis

Anterior segment dysgenesis

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

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

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

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; 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.
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; 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

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
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?

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; 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.

**Anterior segment dysgenesis**

**Anterior Segment Dysgenesis**

- **What is a posterior embryotoxon?**
  - An anteriorly displaced and thickened Schwalbe's line/ring
  - Is it always a harbinger of significant pathology?
    - No; it is found in about 15% of otherwise normal eyes
  - In what three situations is it a significant finding?
    1. When it is part of the Axenfeld-Rieger syndrome
    2. When it is associated with aniridia
    3. When it is associated with Alagille syndrome

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

**Mental note of aniridia’s ocular associations:**
--Nystagmus
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.

Is this a sensory or a motor nystagmus? Sensory.

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

Is this a sensory or a motor nystagmus? Sensory.

Anterior segment dysgenesis

Peripheral Central

Posterior

embryotoxon

Axenfeld-Rieger syndrome

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

Is there such a thing as an anterior embryotoxon? 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.

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

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.

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, and 3) When it is associated with Alagille syndrome.

Is there such a thing as an anterior embryotoxon? 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.

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.

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; 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.

Is this a sensory or a motor nystagmus?
Sensory.

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

- Is this a sensory or a motor nystagmus?
  - Sensory

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

- 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; 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

- Is this a sensory or a motor nystagmus?
  - Sensory

- What anatomic abnormalities are responsible for the poor vision in aniridia?
Anterior Segment Dysgenesis

**Anterior segment dysgenesis**

- What is a posterior embryotoxon?
  - An anteriorly displaced and thickened Schwalbe's line/ring
- Is it always a harbinger of significant pathology?
  - No; it is found in about 15% of otherwise normal eyes
- In what three situations is it a significant finding?
  1. When it is part of the Axenfeld-Rieger syndrome
  2. When it is associated with aniridia
  3. When it is associated with Alagille syndrome
- Is there such a thing as an anterior embryotoxon?
  - 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
- Is this a sensory or a motor nystagmus?
  - Sensory
- 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
- What anatomic abnormalities are responsible for the poor vision in aniridia?
  - Foveal and optic nerve hypoplasia
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; 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

Is this a sensory or a motor nystagmus?
Sensory

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

Mental note of aniridia’s ocular associations:
--Nystagmus
--Foveal hypoplasia
--ON hypoplasia
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?**
Aniridia is associated with the Axenfeld-Rieger syndrome and Alagille syndrome.
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.

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, 2) When it is associated with aniridia, 3) When it is associated with Alagille syndrome.
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; 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
WAGR complex consists of:
W: Wilms tumor
A: Aniridia
G: Genitourinary abnormalities
R: Retardatio
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; 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.

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

Axenfeld-Rieger syndrome
Alagille syndrome
Anterior segment dysgenesis

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

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

Is nystagmus commonly associated with aniridia? Yes.

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

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

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

WAGR complex consists of:
Wilms tumor
Retardation

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

WAGR complex: Wilm’s tumor
**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

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

**Is there such a thing as an anterior embryotoxon?**
Yes; arcus senilis

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

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

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

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

Is there such a thing as an anterior embryotoxon?
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.

Defects involving what gene are the cause of aniridia?
Defects involving the PAX6 gene are the cause of aniridia.

What other ocular abnormalities are associated with defects of the PAX6 gene?
Anterior segment dysgenesis.
**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

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

- **Defects involving what gene are the cause of aniridia?**
  - The PAX6 gene

- **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; arcus senilis is the anterior embryotoxon

- **Why is the term ‘aniridia’ technically a misnomer?**
  - Because a rudimentary iris root is always present

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

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

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

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

- **Defects involving what gene are the cause of aniridia?**
  - The PAX6 gene
**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.

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

**Defects involving what gene are the cause of aniridia?**
The PAX6 gene.

**What other ocular abnormalities are associated with defects of the PAX6 gene?**
- 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; 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
Defects involving what gene are the cause of aniridia?
The PAX6 gene
What other ocular abnormalities are associated with defects of the PAX6 gene?
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)
Anterior Segment Dysgenesis

What four ocular abnormalities are attributed to the PAX6 gene?

- Peters anomaly
- Anirida (duh)
- Congenital cataract (say it out loud)
- 6
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
- Foveal hypoplasia

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

Peters anomaly
Anirida (duh)
Congenital cataracts
Foveal hypoplasia

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

…but are cataracts associated with it as well?
What four ocular abnormalities are attributed to the PAX6 gene?

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

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

…but are cataracts associated with it as well? Indeed they are
What four ocular abnormalities are attributed to the PAX6 gene?

- Foveal hypoplasia
- Peters anomaly
- Congenital cataracts

Mental note of aniridia’s ocular associations:
- Nystagmus
- Foveal hypoplasia
- ON hypoplasia
- Peters anomaly
- Congenital cataracts

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

…but are cataracts associated with it as well? Indeed they are
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
- 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
- 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?

Peters anomaly
Hirida (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
- Foveal hypoplasia

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

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

One that regulates morphogenesis

What sort of gene is PAX6 anyway?

A homeobox gene
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.

*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.

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.

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.

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? 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?*
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.

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

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?

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
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
- Peters anomaly
- Foveal and optic nerve hypoplasia
- Glaucoma
- Cataracts
Nystagmus

limbal stem cell deficiency

Peters anomaly

foveal and optic nerve hypoplasia

glaucoma

cataracts

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

One final point regarding aniridia:
Nystagmus

Limbal stem cell deficiency

Peters anomaly

Foveal and optic nerve hypoplasia

Glaucoma

Cataracts

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

One final point regarding aniridia:
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; 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.

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.
What is the noneponymous name of Alagille syndrome?
Arterohepatic dysplasia

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; 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

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.
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; 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
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.
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; 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

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**

**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; 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 vis a vis us eye dentists?**
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

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; 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 vis a vis us eye dentists?
An infant with jaundice who presents to the eye service as a ‘rule out Alagille syndrome’ consult.
**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; *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 vis a vis us eye dentists?
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**
What is the noneponymous name of Alagille syndrome?
Arterohepatic dysplasia

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

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

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.
Alagille syndrome: Facies
Anterior segment dysgenesis

What is the noneponymous name of Alagille syndrome?
Arterohepatic dysplasia

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

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

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?
--
--
What is the noneponymous name of Alagille syndrome?
Arterohepatic dysplasia

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

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

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
--The skeleton

What is the 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; 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 vis a vis us eye dentists?
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
--The skeleton
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; arcus senilis is the anterior embryotoxon
What is the noneponymous name of Alagille syndrome?
Arteriohepatic dysplasia
How is it inherited?
Autosomal dominant, but the expressivity varies widely
What is the classic presentation vis a vis us eye dentists?
An infant with jaundice who presents to the eye service as a ‘rule out Alagille syndrome’ consult
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? How are they affected?
--The heart:
--The skeleton:

Anterior Segment Dysgenesis

Anterior segment dysgenesis
Anterior Segment Dysgenesis

Anterior segment dysgenesis

What is the noneponymous name of Alagille syndrome?
Arterohepatic dysplasia

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

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

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? How are they affected?
--The heart: Septal defects, PDA, and tetralogy of Fallot are common
--The skeleton: The classic finding is ‘butterfly vertebrae’
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; 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 vis a vis us eye dentists?
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? How are they affected?
--The heart: Septal defects, PDA, and tetralogy of Fallot are common
--The skeleton: The classic finding is ‘butterfly vertebrae’
(Renal, neurologic and vascular abnormalities are common as well.)
Alagille syndrome: Butterfly vertebrae
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; 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

Alagille pts have a characteristic facial appearance—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? How are they affected?
--The heart: Septal defects, PDA, and tetralogy of Fallot are common
--The skeleton: The classic finding is 'butterfly vertebrae'

(Renal, neurologic and vascular abnormalities are common as well.)

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

Rieger syndrome

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—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? How are they affected?
--The heart: Septal defects, PDA, and tetralogy of Fallot are common
--The skeleton: The classic finding is 'butterfly vertebrae'

(Renal, neurologic and vascular abnormalities are common as well.)

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—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? How are they affected?
--The heart: Septal defects, PDA, and tetralogy of Fallot are common
--The skeleton: The classic finding is 'butterfly vertebrae'

(Renal, neurologic 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
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; 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

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? How are they affected?
--The heart: Septal defects, PDA, and tetralogy of Fallot are common
--The skeleton: The classic finding is 'butterfly vertebrae'
(Renal, neurologic and vascular abnormalities are common as well.)

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

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

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; 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

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? How are they affected?

--The heart: Septal defects, PDA, and tetralogy of Fallot are common
--The skeleton: The classic finding is 'butterfly vertebrae'

(Renal, neurologic and vascular abnormalities are common as well.)

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

two words
Anterior Segment Dysgenesis

Anterior segment dysgenesis

What is the noneponymous name of Alagille syndrome?
Arterohepatic dysplasia

How is it inherited?
Autosomal dominant

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—'Triangular.' They have a broad forehead and a pointy chin.

In addition to liver, eye and face findings, what other organs are commonly affected? How are they affected?
--The heart: Septal defects, PDA, and tetralogy of Fallot are common
--The skeleton: The classic finding is 'butterfly vertebrae'

(Renal, neurologic and vascular abnormalities are common as well.)
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; 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

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? How are they affected?
--The heart: Septal defects, PDA, and tetralogy of Fallot are common
--The skeleton: The classic finding is ‘butterfly vertebrae’
(Renal, neurologic and vascular abnormalities are common as well.)

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

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; 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

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? How are they affected?
--The heart: Septal defects, PDA, and tetralogy of Fallot are common
--The skeleton: The classic finding is 'butterfly vertebrae'

(Renal, neurologic and vascular abnormalities are common as well.)
Goldenhar: Limbal (epibulbar) dermoids; lid coloboma (OCULO-auriculo-vertebral syndrome)

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

Goldenhar syndrome: Hemifacial microsomia
Anterior Segment Dysgenesis

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

Goldenhar syndrome: Hemifacial microsomia

For more on Goldenhar, see slide-set P22

Goldenhar: Ear abnormalities (Oculo-AURICULO-vertebral syndrome)
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 normal eyes.

In what three situations is it a significant finding?
1) When it is part of the Axenfeld-Rieger syndrome
2) When it is associated with aniridia
3) When it is associated with Alagille syndrome

Is there such a thing as an anterior embryotoxon?
Anterior Segment Dysgenesis

Anterior segment dysgenesis

Peripheral

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

**What is 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**
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**
Anterior Segment Dysgenesis

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 AAs.

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.
It is not always a harbinger of significant pathology.
About 15% of otherwise normal eyes have it.
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 its main chemical component?
Cholesterol.

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

Cholesterol

Bilateral
No; per the Cornea book, it is an "involutional change"

It starts at the poles, then spreads circumferentially.

Yes, men are more likely to develop it.

Yes, it is more common in AAs.

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
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 African Americans.

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

**Regarding arcus senilis...**

What is its main chemical component?
- Cholesterol

Is it usually unilateral, or bilateral?
- Bilateral

What is arcus senilis?
- 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 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

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)

Arcus juvenilis aka anterior embryotoxon
Arcus senilis
Corneal arcus
Posterior embryotoxon
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.

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 patient has unilateral arcus?

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

What is arcus senilis?

It is an involutional change with cholesterol as its main chemical component.

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%.

What condition should be suspected if a patient has unilateral arcus?
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—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

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.
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

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

Arcus juvenilis aka anterior embryotoxon

Corneal arcus
Anterior Segment Dysgenesis

Regarding arcus senilis...

What is its main chemical component?
Cholesterol

Is it usually unilateral, or bilateral?
Bilateral

Is it a dystrophy?

Arcus juvenilis aka anterior embryotoxon

Arcus senilis

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

Concerning 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%
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”

What is arcus senilis?
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”
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

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?

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

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

Early 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 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

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

Arcus juvenilis aka anterior embryotoxon

Arcus senilis

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

Arcus senilis

Corneal arcus

Peripheral

Axenfeld-Rieger syndrome

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

Arcus senilis

Arcus juvenilis aka anterior embryotoxon

Arcus juvenilis

Corneal arcus

Periperal

Axenfeld-Rieger syndrome

Posterior embryotoxon

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

- **Anterior embryotoxon**
- **Posterior embryotoxon**
- **Axenfeld-Rieger syndrome**
- **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

### 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%
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.

What is arcus senilis?
It is an involutional change in the cornea, typically more common in elderly men and AA populations.
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

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

Corneal arcus
Arcus juvenilis aka anterior embryotoxon
Arcus senilis
Posterior embryotoxon
Axenfeld-Rieger syndrome
Peripheral
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—this is another name for arcus juvenilis.

What is arcus senilis?

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

**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 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

- 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%

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

**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

**Corneal arcus**

Arcus juvenilis aka anterior embryotoxon

**Axenfeld-Rieger syndrome**

**Peripheral**

Posterior embryotoxon

**Corneal arcus**

Arcus juvenilis aka anterior embryotoxon

**Arcus senilis**

Before age 40

**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

**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

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%

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

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

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

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

**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

**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

**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 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 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.

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 coupled with high cholesterol.

What is arcus senilis?

It is the congenital version of arcus juvenilis.
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

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

Arcus juvenilis
aka anterior embryotoxon

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 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?

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
What is 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

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

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.
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—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.

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.

What is arcus juvenilis? (before age 40)

It is the congenital version of arcus senilis.

What is arcus senilis? (after age 40)

The congenital version of arcus senilis.

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%.

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

Xanthelasma.
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.

**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

**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.
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%

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
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.

What is arcus senilis? 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 AA.

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

Other 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.

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 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.

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.
Anterior Segment Dysgenesis

Xanthelasma
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 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.

What is arcus senilis?

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.

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

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--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 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% before age 40.

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 senilis?

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.

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.

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 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 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%.

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

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

What is arcus senilis?
It is a dystrophy.

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.

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 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.

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?

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

What is arcus senilis?
It is a dystrophy.

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.

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

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 are xanthelasmans, 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 the involutional change of arcus juvenilis

Xanthelasma

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?

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

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
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

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 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 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?

Regarding arcus senilis...

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%

Xanthelasma

Before age 40

What is arcus senilis?
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? Yes--it is another name for arcus juvenilis

What is arcus juvenilis? It is the congenital version of 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

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

What is arcus senilis? It is the 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 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

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
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%

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

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

Before age 40

Speaking of ‘xanthelasmas’…What dz comes to mind if an adult presents with what appear to be xanthelasmas, but:
The xanthelasmas are ulcerated?
Necrobiotic xanthogranuloma

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

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

The above are 3 of 4 conditions that fall under what heading?
The 'adult xanthogranulomatous diseases'

In three words, how would you characterize these conditions?
They are non-Langerhans cell histocytoses
Anterior Segment Dysgenesis

Speaking of ‘xanthelasmas’…What dz comes to mind if an adult presents with what appear to be xanthelasmas, but: The xanthelasmas are ulcerated? Necrobiotic xanthogranuloma

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? 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
Necrobiotic xanthogranuloma
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%

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

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

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

Speaking of ‘xanthelasmas’…What dz comes to mind if an adult presents with what appear to be xanthelasmas, but:

The xanthelasmas are ulcerated? Necrobiotic xanthogranuloma

The pt c/o adult-onset asthma?

Necrobiotic xanthogranuloma and the adult-onset asthma with periocular xanthogranuloma

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

The above are 3 of 4 conditions that fall under what heading?

The ‘adult xanthogranulomatous diseases’

In three words, how would you characterize these conditions?

They are non-Langerhans cell histocytoses
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%.

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 ‘xanthelasmas’…What dz comes to mind if an adult presents with what appear to be xanthelasmas, but:
The xanthelasmas are ulcerated? Necrobiotic xanthogranuloma
The pt c/o adult-onset asthma? Adult-onset asthma with periocular xanthogranuloma

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?
Often present in such pts. 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.
Necrobiotic xanthogranuloma

Adult-onset asthma with periocular xanthogranuloma
Anterior Segment Dysgenesis

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

The xanthelasmas are ulcerated? Necrobiotic xanthogranuloma
The pt c/o adult-onset asthma? Adult-onset asthma with periocular xanthogranuloma
The pt is severely systemically ill (by the same dz process)?

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

Arcus juvenilis
aka anterior embryotoxon

Arcus senilis

What is arcus senilis?
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

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
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?
It is the involutional change of the cornea
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 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
Speaking of ‘xanthelasmas’…What dz comes to mind if an adult presents with what appear to be xanthelasmas, but:
The xanthelasmas are ulcerated? Necrobiotic xanthogranuloma
The pt c/o adult-onset asthma? Adult-onset asthma with periocular xanthogranuloma
The pt is severely systemically ill (by the same dz process)? Erdheim-Chester dz
Anterior Segment Dysgenesis

Necrobiotic xanthogranuloma

Erdheim-Chester disease

Adult-onset asthma with periocular xanthogranuloma
Anterior Segment Dysgenesis

Speaking of ‘xanthelasmas’…What dz comes to mind if an adult presents with what appear to be xanthelasmas, but:

The xanthelasmas are ulcerated? Necrobiotic xanthogranuloma
The pt c/o adult-onset asthma? Adult-onset asthma with periocular xanthogranuloma
The pt is severely systemically ill (by the same dz process)? Erdheim-Chester dz

The above are 3 of 4 conditions that fall under what heading?

Arcus juvenilis aka anterior embryotoxon
Arcus senilis

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

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?

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 arcus juvenilis?
It is the congenital version of arcus senilis.
Speaking of ‘xanthelasmas’… What dz comes to mind if an adult presents with what appear to be xanthelasmas, but:
The xanthelasmas are ulcerated? Necrobiotic xanthogranuloma
The pt c/o adult-onset asthma? Adult-onset asthma with periocular xanthogranuloma
The pt is severely systemically ill (by the same dz process)? Erdheim-Chester dz

The above are 3 of 4 conditions that fall under what heading?
The ‘adult xanthogranulomatous diseases’

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
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.

Archus juvenilis

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

Peripheral

What is arcus senilis?
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?
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 often present in such pts. What is it?
Xanthelasma.

What is the fourth adult xanthogranulomatous disease?
It is called simply adult-onset xanthogranuloma, and presents like the "asthma" type (but without the asthma, duh).

The above are 3 of 4 conditions that fall under what heading?
The ‘adult xanthogranulomatous diseases’

Speaking of ‘xanthelasmas’…What dz comes to mind if an adult presents with what appear to be xanthelasmas, but:
The xanthelasmas are ulcerated? Necrobiotic xanthogranuloma
The pt c/o adult-onset asthma? Adult-onset asthma with periocular xanthogranuloma
The pt is severely systemically ill (by the same dz process)? Erdheim-Chester dz

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 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?
It is the adult version of arcus juvenilis

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

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

What is the fourth adult xanthogranulomatous disease?

The above are 3 of 4 conditions that fall under what heading?
The ‘adult xanthogranulomatous diseases’
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%.

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.

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.

In three words, how would you characterize these conditions?

They are non-Langerhans cell histocytoses.

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

The xanthelasmas are ulcerated? Necrobiotic xanthogranuloma
The pt c/o adult-onset asthma? Adult-onset asthma with periocular xanthogranuloma
The pt is severely systemically ill (by the same dz process)? Erdheim-Chester dz

The above are 3 of 4 conditions that fall under what heading?
The ‘adult xanthogranulomatous diseases’

In three words, how would you characterize these conditions?
**Anterior Segment Dysgenesis**

Speaking of ‘xanthelasmas’…What dz comes to mind if an adult presents with what appear to be xanthelasmas, but:

The xanthelasmas are ulcerated? Necrobiotic xanthogranuloma
The pt c/o adult-onset asthma? Adult-onset asthma with periocular xanthogranuloma
The pt is severely systemically ill (by the same dz process)? Erdheim-Chester dz

The above are 3 of 4 conditions that fall under what heading?
The ‘adult xanthogranulomatous diseases’

In three words, how would you characterize these conditions?
They are non-Langerhans cell...
Anterior Segment Dysgenesis

Speaking of ‘xanthelasmas’…What dz comes to mind if an adult presents with what appear to be xanthelasmas, but:
- The xanthelasmas are ulcerated? Necrobiotic xanthogranuloma
- The pt c/o adult-onset asthma? Adult-onset asthma with periocular xanthogranuloma
- The pt is severely systemically ill (by the same dz process)? Erdheim-Chester dz

The above are 3 of 4 conditions that fall under what heading?
The ‘adult xanthogranulomatous diseases’

In three words, how would you characterize these conditions?
They are non-Langerhans cell histiocytoses

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

Are they always a harbinger of elevated serum lipids?
No, they can (and often do) appear in individuals with normal lipid panels

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

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

What is arcus senilis?
Arcus senilis is a yellowish deposit around the cornea, usually bilaterally

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?
Yes, but below age 40, the prevalence decreases

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

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

Speaking of ‘xanthelasmas’…What dz comes to mind if an adult presents with what appear to be xanthelasmas, but:
The xanthelasmas are ulcerated? Necrobiotic xanthogranuloma
The pt c/o adult-onset asthma? Adult-onset asthma with periocular xanthogranuloma
The pt is severely systemically ill (by the same dz process)? Erdheim-Chester dz

In three words, how would you characterize these conditions?
They are non-Langerhans cell histiocytoses

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

Are they always a harbinger of elevated serum lipids?
No, they can (and often do) appear in individuals with normal lipid panels

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

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

Arcus juvenilis
aka
anterior embryotoxon

Arcus senilis

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 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

Arcus juvenilis aka anterior embryotoxon

Arcus senilis

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%

What is arcus juvenilis?

It is the congenital version of arcus senilis

Xanthelasma

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?

Familial hyperlipoproteinemia

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

Xanthelasma

Speaking of ‘xanthelasmas’…What dz comes to mind if an adult presents with what appear to be xanthelasmas, but:

The xanthelasmas are ulcerated? Necrobiotic xanthogranuloma

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

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

The above are 3 of 4 conditions that fall under what heading?

The ‘adult xanthogranulomatous diseases’

In three words, how would you characterize these conditions?

They are non-Langerhans cell histiocytoses

Do the lesions of adult xanthogranulomatous dz contain lipid-filled macrophages a la xanthelasma?

Indeed they do

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 be congenital?

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

Can they be congenital?

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

Can they be congenital?

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

Can they be congenital?

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

Can they be congenital?

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

Can they be congenital?

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

Can they be congenital?

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

Can they be congenital?

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

Can they be congenital?

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

Can they be congenital?

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

Can they be congenital?

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

Can they be congenital?

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

Can they be congenital?

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

Can they be congenital?

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

Can they be congenital?

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

Can they be congenital?

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

Can they be congenital?

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

Can they be congenital?

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

Can they be congenital?

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

Can they be congenital?

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

Can they be congenital?

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

Can they be congenital?

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

Can they be congenital?
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%

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 ‘xanthelasmas’…What dz comes to mind if an adult presents with what appear to be xanthelasmas, but:
The xanthelasmas are ulcerated? Necrobiotic xanthogranuloma
The pt c/o adult-onset asthma? Adult-onset asthma with periocular xanthogranuloma
The pt is severely systemically ill (by the same dz process)? Erdheim-Chester dz

The above are 3 of 4 conditions that fall under what heading?
The ‘adult xanthogranulomatous diseases’

Do the lesions of adult xanthogranulomatous dz contain lipid-filled macrophages a la xanthelasma?
Indeed they do

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 the lesions of adult xanthogranulomatous dz contain lipid-filled macrophages a la xanthelasma?
Indeed they do

xanthelasma

Does its prevalence increase with age?
Yes, after age 80, the prevalence is ~100% 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 lid. 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. Xanthelasma is often present in such pts. 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

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.

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' = '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 Xanthelasma?

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

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 are xanthelasmas, that is, what is their clinical appearance?
They are yellowish plaques located in the medial canthal region, usually on the upper eyelids.

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%.

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

Arcus juvenilis
aka
ante造成了 embryotoxon

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.

Arcus juvenilis
aka
ante造成了 embryotoxon

Arcus senilis

Peripheral

Regarding arcus senilis...

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

First clue--more forthcoming

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%

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

Arcus senilis

Post embryotoxon

Arcus juvenilis
aka
anterior embryotoxon
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 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

Speaking of ‘foamy macrophages’…
What dz comes to mind if, instead of an adult with xanthelasmamas, 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 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%

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

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.
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.’*
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.’*
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.

First clue…

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)
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?

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)

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

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

Are they always a harbinger of elevated serum lipids?

Speaking of ‘foamy macrophages’ part 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?

Clue #3

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

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

Are they always a harbinger of elevated serum lipids?

Speaking of ‘foamy macrophages’ part 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

Speaking of ‘foamy macrophages’…
What dz comes to mind if, instead of an adult with xanthelasmatas, 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?

Speaking of ‘foamy macrophages’ part deaux…
What dz comes to mind if, instead of an adult with xanthelasmatas, 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.

Foamy macrophages

Spontaneous hyphema

JXG

For more on JXG, see slide-set FELT8

cells
Anterior segment dysgenesis

- Posterior embryotoxon
- 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 normal eyes.

In what three situations is it a significant finding?
1) When it is part of the Axenfeld-Rieger syndrome
2) When it is associated with aniridia
3) When it is associated with Alagille syndrome

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

**Anterior segment dysgenesis**

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

**Anterior segment dysgenesis**

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

**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?**
Yes--anterior *gerontoxon*
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 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?
Yes--anterior gerontoxon

Gerontoxon?
Yeah, ‘geron-’ as in gerontology, as in old folks
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.

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

Gerontoxon?
Yeah, ‘geron-‘ as in gerontology, as in old folks.

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

<table>
<thead>
<tr>
<th>Anterior</th>
<th>Embryotoxon</th>
<th>Gerontoxon</th>
</tr>
</thead>
<tbody>
<tr>
<td>Arcus juvenilis</td>
<td>Arcus senilis</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Posterior</th>
<th>Embryotoxon</th>
<th>Gerontoxon</th>
</tr>
</thead>
<tbody>
<tr>
<td>Posterior embryotoxon</td>
<td>Nope</td>
<td></td>
</tr>
</tbody>
</table>

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)
Anterior Segment Dysgenesis

What features define Axenfeld-Rieger syndrome?

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

Peripheral

Posterior embryotoxon

Axenfeld-Rieger syndrome

Nonocular abnormalities:
1) Abnormal dentition
2) Characteristic facies
3) Periumbilical skin folds
4) Cardiac valve problems
Anterior Segment Dysgenesis

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

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

Where does ARS rank as a cause of iris hypoplasia?
Anterior Segment Dysgenesis

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

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

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

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

What is the lifetime risk of developing glaucoma?
50%
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?
- Corectopia
- Ectropion uveae
- Cryptless, glassy surface

What corneal abnormalities may be present?
- Megalocornea
- Microcornea

What nonocular abnormalities may be present?
- Abnormal dentition
- Characteristic facies
- Periumbilical skin folds
- Cardiac valve problems

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

50%
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**

‘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

Peripheral

Axenfeld-Rieger syndrome

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

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

What is the lifetime risk of developing glaucoma? 50%
What features define Axenfeld-Rieger syndrome?
Posterior embryotoxon with attached iris strands + iris hypoplasia + angle abnormalities

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

Peripheral

Axenfeld-Rieger syndrome

Posterior embryotoxon
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?
- Corectopia
- Ectropion uveae
- Cryptless, glassy surface

What corneal abnormalities may be present?
- Megalocornea
- Microcornea

What nonocular abnormalities may be present?
- Abnormal dentition
- Characteristic facies
- Periumbilical skin folds
- Cardiac valve problems

What is corectopia?
The displacement of the pupil from its normal central-ish location. Deviation from centrality of 1/2 mm is common, and up to 1 mm is considered normal.
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

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

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

Corectopia
Anterior Segment Dysgenesis

Peripheral

Posterior embryotoxon

Axenfeld-Rieger syndrome

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

Why central-ish?

What other iris abnormalities may be present?
1) Corectopia
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
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

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

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

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.
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 does the term ectropion uveae refer to?
The presence of posterior pigmented iris epithelium on the anterior surface of the iris
Anterior Segment Dysgenesis

Ectropion uveae
Anterior Segment Dysgenesis

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

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?
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 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

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) Microcornea
2) Not simultaneously, obviously

Peripheral

Axenfeld-Rieger syndrome
**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

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

<table>
<thead>
<tr>
<th></th>
<th>Horizontal diameter at birth (mm)</th>
<th>Horizontal diameter at ≥2 years (mm)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Megalocornea</td>
<td>&gt;?</td>
<td></td>
</tr>
<tr>
<td>Microcornea</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
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

<table>
<thead>
<tr>
<th></th>
<th>Horizontal diameter at birth (mm)</th>
<th>Horizontal diameter at ≥2 years (mm)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Megalocornea</td>
<td>&gt;12</td>
<td></td>
</tr>
<tr>
<td>Microcornea</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
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

<table>
<thead>
<tr>
<th></th>
<th>Horizontal diameter at birth (mm)</th>
<th>Horizontal diameter at ≥2 years (mm)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Megalocornea</td>
<td>&gt;12</td>
<td>&gt;?</td>
</tr>
<tr>
<td>Microcornea</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
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

<table>
<thead>
<tr>
<th>Condition</th>
<th>Horizontal diameter at birth (mm)</th>
<th>Horizontal diameter at ≥2 years (mm)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Megalocornea</td>
<td>&gt;12</td>
<td>&gt;13</td>
</tr>
<tr>
<td>Microcornea</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
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

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

<table>
<thead>
<tr>
<th>Condition</th>
<th>Horizontal diameter at birth (mm)</th>
<th>Horizontal diameter at ≥2 years (mm)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Megalocornea</td>
<td>&gt;12</td>
<td>&gt;13</td>
</tr>
<tr>
<td>Microcornea</td>
<td>&lt;?</td>
<td></td>
</tr>
</tbody>
</table>
Anterior Segment Dysgenesis

**Peripheral**
- Posterior embryotoxon

**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

<table>
<thead>
<tr>
<th></th>
<th>Horizontal diameter at birth (mm)</th>
<th>Horizontal diameter at ≥2 years (mm)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Megalocornea</td>
<td>&gt;12</td>
<td>&gt;13</td>
</tr>
<tr>
<td>Microcornea</td>
<td>&lt;9</td>
<td></td>
</tr>
</tbody>
</table>
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.

<table>
<thead>
<tr>
<th></th>
<th>Horizontal diameter at birth (mm)</th>
<th>Horizontal diameter at ≥2 years (mm)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Megalocornea</td>
<td>&gt;12</td>
<td>&gt;13</td>
</tr>
<tr>
<td>Microcornea</td>
<td>&lt;9</td>
<td>&lt;?</td>
</tr>
</tbody>
</table>
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**

<table>
<thead>
<tr>
<th></th>
<th>Horizontal diameter at birth (mm)</th>
<th>Horizontal diameter at ≥2 years (mm)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Megalocornea</td>
<td>&gt;12</td>
<td>&gt;13</td>
</tr>
<tr>
<td>Microcornea</td>
<td>&lt;9</td>
<td>&lt;10</td>
</tr>
</tbody>
</table>
Axenfeld-Reiger with microcornea (8.5 mm)
That pic should have immediately reminded you of another condition (pictured above)—what is it?

Axenfeld-Reiger with microcornea (8.5 mm)
Axenfeld-Reiger with microcornea (8.5 mm)

Anterior Segment Dysgenesis

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

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

Essential iris atrophy

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

Axenfeld-Reiger with microcornea (8.5 mm)
Anterior Segment Dysgenesis

Axenfeld-Reiger with microcornea (8.5 mm)

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

Essential iris atrophy is a variant/form of what condition?
Iridocorneal endothelial (ICE) syndrome
Anterior Segment Dysgenesis

Axenfeld-Reiger with microcornea (8.5 mm)

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

Essential iris atrophy is a variant/form of what condition?
Iridocorneal endothelial (ICE) syndrome

How do we know that the ARS pic isn’t actually ICE?
Anterior Segment Dysgenesis

Axenfeld-Reiger with microcornea (8.5 mm)

----

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

Essential iris atrophy is a variant/form of what condition?
Iridocorneal endothelial (ICE) syndrome

How do we know that the ARS pic isn’t actually ICE?
Because ICE is not associated with microcornea
Anterior Segment Dysgenesis

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

Essential iris atrophy is a variant/form of what condition?
Iridocorneal endothelial (ICE) syndrome

How do we know that the ARS pic isn’t actually ICE?
Because ICE is not associated with microcornea

Is ICE sporadic, or inherited?
Sporadic

Is it unilateral, or bilateral?
Unilateral

Does it tend to affect males, or females?
Females
Anterior Segment Dysgenesis

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

Essential iris atrophy is a variant/form of what condition?
Iridocorneal endothelial (ICE) syndrome

How do we know that the ARS pic isn't actually ICE?
Because ICE is not associated with microcornea

Is ICE sporadic, or inherited?
Sporadic
Anterior Segment Dysgenesis

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

Essential iris atrophy

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

Iridocorneal endothelial (ICE) syndrome

Is ICE sporadic, or inherited?
Sporadic

Is it unilateral, or bilateral?

Is it unilateral, or bilateral?

How do we know that the ARS pic isn’t actually ICE?
Because ICE is not associated with microcornea
That pic should have immediately reminded you of another condition (pictured above)—what is it?
Essential iris atrophy

Essential iris atrophy is a variant/form of what condition?
Iridocorneal endothelial (ICE) syndrome

How do we know that the ARS pic isn’t actually ICE?
Because ICE is not associated with microcornea
Anterior Segment Dysgenesis

Is ICE sporadic, or inherited?
Sporadic

Is it unilateral, or bilateral?
Unilateral

Does it tend to affect males, or females?
Females

Essential iris atrophy is a variant/form of what condition?
Iridocorneal endothelial (ICE) syndrome

How do we know that the ARS pic isn’t actually ICE?
Because ICE is not associated with microcornea.
Axenfeld-Reiger with microcornea (8.5 mm)

Anterior Segment Dysgenesis

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

Essential iris atrophy is a variant/form of what condition?
Iridocorneal endothelial (ICE) syndrome

Is ICE sporadic, or inherited?  
Sporadic

Is it unilateral, or bilateral?  
Unilateral

Does it tend to affect males, or females?  
Females

How do we know that the ARS pic isn’t actually ICE?  
Because ICE is not associated with microcornea
Axenfeld-Reiger with microcornea (8.5 mm)

Anterior Segment Dysgenesis

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

Essential iris atrophy

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

Iridocorneal endothelial (ICE) syndrome

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

Because ICE is not associated with microcornea

Is ICE sporadic, or inherited?

Sporadic

Is it unilateral, or bilateral?

Unilateral

Does it tend to affect males, or females?

Females

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)
Anterior Segment Dysgenesis

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

Essential iris atrophy is a variant/form of what condition? Iridocorneal endothelial (ICE) syndrome

How do we know that the ARS pic isn’t actually ICE? Because ICE is not associated with microcornea

Is ICE sporadic, or inherited? Sporadic

Is it unilateral, or bilateral? Unilateral

Does it tend to affect males, or females? Females

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)

For more on ICE, see slide-set K26
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)
2)
3)
4)
Anterior Segment Dysgenesis

Peripheral

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
3) Periumbilical skin folds
4) Cardiac valve problems
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
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
3) Periumbilical skin folds

Name two other congenital conditions that include both ocular involvement and abnormal dentition:
--
--
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

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

Peripheral

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
3) Periumbilical skin folds

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

Bloch-Sulzberger syndrome

Incontinentia pigmenti

Congenital syphilis

Name two other congenital conditions that include both ocular involvement and abnormal dentition:
Incontinentia pigmenti
Congenital syphilis
**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?**
- Corectopia
- Ectropion uveae
- Cryptless, glassy surface

**What corneal abnormalities may be present?**
- Megalocornea
- Microcornea

**What nonocular abnormalities may be present?**
- Abnormal dentition
- Characteristic facies
- Periumbilical skin folds
- Cardiac valve problems

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

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

**Bloch-Sulzberger 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
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

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

What is the eponymous name for IP?
Bloch-Sulzberger 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
3) Periumbilical skin folds

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

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

What is the eponymous name for IP?
Bloch-Sulzberger 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
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

Very broadly, what is a phakomatosis?

In one word, what sort of condition is IP?
- Phakomatosis

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

Very broadly, what is a phakomatosis? A congenital condition characterized by abnormalities of the eyes and skin

In one word, what sort of condition is IP? Phakomatosis

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

Very broadly, what is a phakomatosis?
A congenital condition characterized by abnormalities of the eyes and skin

In one word, what sort of condition is IP?
- Phakomatosis

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

Very broadly, what is a phakomatosis?
A congenital condition characterized by abnormalities of the eyes and skin

What is the classic skin finding associated with incontinentia pigmenti?
'Splashed paint' appearance

In one word, what sort of condition is IP?
Phakomatosis

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

What is the classic skin finding associated with incontinentia pigmenti?
'Splashed paint' appearance

Very broadly, what is a phakomatosis?
A congenital condition characterized by abnormalities of the eyes and skin

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

What is the classic skin finding associated with incontinentia pigmenti?
'Splashed paint' appearance

In one word, what sort of condition is IP?
Phakomatosis

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

What is the classic skin finding associated with incontinentia pigmenti?
'Splashed paint' appearance

Very broadly, what is a phakomatosis?
A congenital condition characterized by abnormalities of the eyes and skin
### 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?**
- Corectopia
- Ectropion uveae
- Cryptless, glassy surface

**What corneal abnormalities may be present?**
- Megalocornea
- Microcornea

**What nonocular abnormalities may be present?**
- Abnormal dentition
- Characteristic facies
- Periumbilical skin folds
- Cardiac valve problems

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

**In one word, what sort of condition is IP?**
- Phakomatosis

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

**Very broadly, what is a phakomatosis?**
A congenital condition characterized by abnormalities of the eyes and skin

**What is the classic skin finding associated with incontinentia pigmenti?**
‘Splashed paint’ appearance
Incontinentia pigmenti: Splashed-paint appearance
Incontinentia pigmenti: Splashed-paint appearance
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

Abnormal dentition

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

What are two other congenital conditions that include both ocular involvement and abnormal dentition:
1) Incontinentia pigmenti
2) Congenital syphilis
Anterior Segment Dysgenesis

- Axenfeld-Rieger syndrome
  - Posterior embryotoxon with attached iris strands + iris hypoplasia + angle abnormalities

What other iris abnormalities may be present?
- Corectopia
- Ectropion uveae
- Cryptless, glassy surface

What corneal abnormalities may be present?
- Megalocornea
- Microcornea

What nonocular abnormalities may be present?
- Abnormal dentition
- Characteristic facies
- Periumbilical skin folds
- Cardiac valve problems

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

Abnormal dentition

What features define Axenfeld-Rieger syndrome?

- Peripheral
- Posterior embryotoxon

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

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

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

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
3) Periumbilical skin folds

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’

Abnormal dentition

Two other congenital conditions that include both ocular involvement and abnormal dentition:
Incontinentia pigmenti

Congenital syphilis
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?

Anterior Segment Dysgenesis

Central

Posterior keratoconus

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

Posterior keratoconus

Anterior Segment Dysgenesis

Central

Peters anomaly
Anterior Segment Dysgenesis

Posterior keratoconus
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?
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.
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

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

Are most cases unilateral, or bilateral?
Unilateral

Are most cases familial, or sporadic?
Sporadic
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

Is it common, like regular keratoconus?
No, it is rare
**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

*Is it common, like regular keratoconus?*
No, it is rare

*Does it affect vision?*
**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

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
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

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

Are most cases unilateral, or bilateral?

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

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

Are most cases unilateral, or bilateral?
Unilateral

Anterior Segment Dysgenesis

Central

Posterior keratoconus

Peters anomaly
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

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

Are most cases unilateral, or bilateral?
Unilateral

Are most cases familial, or sporadic?
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

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

Are most cases unilateral, or bilateral?
Unilateral

Are most cases familial, or sporadic?
Sporadic
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
-- 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)
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.
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
Peters anomaly
Anterior segment dysgenesis

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

How does it present?
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

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.

Does it require a workup?
No if it's unilateral (usually sporadic). Yes if it's bilateral (do a complete genetic and systemic workup).
Peters anomaly: Hazy cornea
Anterior segment dysgenesis

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

How does it present?
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)

What is the STUMPED mnemonic for a cloudy cornea in an infant?
- S = Sclerocornea
- T = Stromal dystrophy (CHSD)
- U = Trauma (eg, forcep injury)
- M = Ulcer
- P = Peters anomaly
- E = Endothelial dystrophy (CHED); Elevated IOP (congenital glaucoma)

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

What is the STUMPED mnemonic for a cloudy cornea in an infant?
Sclerocornea; Stromal dystrophy (CHSD)
Trauma (e.g., 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

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?
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... 
--It may be... to the ... 
--Occasionally it is... size, shape, and location (four words)
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
--It may be...adherent to the posterior corneal defect
--Occasionally it is...small, misshapen, and displaced into the AC
Peters anomaly: Cataractous lens
Anterior segment dysgenesis

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

How does it present?
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

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

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?

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

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

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
--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

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

**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
--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 a 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

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
--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

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

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
-- 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 a 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’)

What common slit-lamp observation owes to the lens’ small size?
-- Occasionally it is...small, misshapen, and displaced into the AC
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
--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

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’)

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

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

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’)

How does refractive status manifest the lens’ spherical shape?
--It is usually highly myopic
--Occasionally it is...small, misshapen, and displaced into the AC
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
--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

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")

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

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

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?
--It may ‘clog’ the pupillary opening, resulting in pupillary block angle-closure glaucoma
--Occasionally it is small, misshapen, and displaced into the AC
Anterior segment dysgenesis

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
-- 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

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

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

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

How does it present?
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, 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?
The lens may 'clog' the pupillary opening, resulting in pupillary block angle-closure glaucoma.
Anterior segment dysgenesis

What is Peters anomaly?

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

How does it present?

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 a 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, 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?

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

Occasionally it is...small, misshapen, and displaced into the AC
Anterior segment dysgenesis

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
--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

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.

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
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
--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

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.

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
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
-- 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 a 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 you are thinking of what?
A small, round lens

What is microspherophakia associated with Peters anomaly?
Yes, although only "occasionally" per the BCSC Lens book

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
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
--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

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

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
Weill-Marchesani syndrome: Short fingers

Anterior Segment Dysgenesis
What is Peters anomaly?
A defect of the posterior central cornea, including the absence of Descemet's and subjacent endothelium. Adhesions extending from the iris to the posterior corneal defect are often present.

How does it present?
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 a 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

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?
Occasionally it is... small, misshapen
Anterior segment dysgenesis

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

How does it present?
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 a 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

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
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
--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 a 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

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

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

Weill-Marchesani syndrome
Quite short
What is notable about their digits?
They are short as well
What is notable about their joints?
They are stiff

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

Does Peters anomaly require a workup?
No if it’s…unilateral (usually sporadic)
Yes if it’s…bilateral (do a complete genetic and systemic workup)
**Anterior Segment Dysgenesis**

**Anterior segment dysgenesis**

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

*How does it present?*
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 Peters anomaly require a workup?*
**No** if it’s...
**Yes** if it’s...
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
--It may be...adherent to the posterior corneal defect
--Occasionally it is...small, misshapen, and displaced into the AC

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