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Anterior Segment Dysgenesis

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What is a neurocristopathy?
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Anterior Segment Dysgenesis

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Which wave involves which future structure?
First wave ➔
Second wave ➔
Third wave ➔
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Anterior Segment Dysgenesis

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Normal Neural Crest Migration

**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

Anterior segment dysgenesis

Peripheral

Central

A very basic anatomic distinction
Anterior Segment Dysgenesis

Anterior segment dysgenesis

Peripheral

Central

Two classic peripheral dysgeneses

?
Anterior Segment Dysgenesis

Anterior segment dysgenesis

Peripheral
- Posterior embryotoxon
- Axenfeld-Rieger syndrome

Central

Two classic peripheral dysgeneses
**Anterior Segment Dysgenesis**

**Anterior segment dysgenesis**

Peripheral

- Posterior embryotoxon
- Axenfeld-Rieger syndrome

**What is a posterior embryotoxon?**

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**

- **Posterior embryotoxon**
  - An anteriorly displaced and thickened Schwalbe’s line/ring
- **Peripheral**
- **Axenfeld-Rieger syndrome**

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Anterior Segment Dysgenesis

Anterior segment dysgenesis

Peripheral

- Posterior embryotoxon
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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
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Normal angle anatomy: Identify the structures
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Anterior Segment Dysgenesis

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3) When it is associated with Alagille syndrome.

Interestingly, all three of these begin with the letter ‘A’.
Anterior Segment Dysgenesis

**Anterior segment dysgenesis**

Peripheral

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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
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Aniridia. Note the presence of an iris stub/root
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Anterior segment dysgenesis

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- Is this a sensory or a motor nystagmus?
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- Is it a jerk, or a pendular nystagmus?
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Anterior Segment Dysgenesis

Anterior segment dysgenesis

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

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WAGR complex consists of:
W
A
G
R

Anterior Segment Dysgenesis

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

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

### Anterior segment dysgenesis

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Anterior Segment Dysgenesis

**Anterior segment dysgenesis**

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

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

**What is the noneponymous name for Wilms tumor (ie, what sort of tumor is it)?**
- A nephroblastoma
WAGR complex: Wilm’s tumor
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No, only those in which the genetic mutation is sporadic

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

There are four main abnormalities, and the term PAX6 acts as its own mnemonic. Start with the ‘P’ and make your way down…
What four ocular abnormalities are attributed to the PAX6 gene?
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- Peters anomaly
- Anirida (duh)
- X
- 6
What four ocular abnormalities are attributed to the PAX6 gene?

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

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

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

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 cataract
- Foveal hypoplasia

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

We know that corneal opacities and foveal hypoplasia are associated with anirida...
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- Anirida (duh)
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…*but are cataracts associated with it as well?* Indeed they are.

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*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
- Aniridia (duh)
- Congenital cataract
- Hypoplasia of the fovea

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

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

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

Why is sporadic aniridia associated with Wilms tumor, but not familial aniridia?
The PAX6 gene and the Wilms tumor gene (called WT1) are adjacent to one another on chromosome 11p. Inherited genetic abnormalities leading to familial aniridia are located within the PAX6 gene itself, and thus do not affect the viability of the nearby WT1. In contrast, sporadic cases of aniridia are usually the result of the wholesale deletion of a chunk of genetic material in the PAX6 ‘neighborhood.’ And since WT1 is its next-door neighbor, it is often affected as well by these deletions.

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

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

Are all aniridia cases at risk for WAGR complex? No, only those in which the genetic mutation is sporadic.
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Because of the PAX6/WT1 spatial relationship, all infants presenting with sporadic aniridia must undergo genetic screening for the Wilms tumor defect.

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

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

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

Via periodic renal ultrasound

*How often, and for how long?*

---

*Are all aniridia cases at risk for WAGR complex?*

No, only those in which the genetic mutation is **sporadic**.
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*If a child tests positive for the Wilms tumor defect, how should they be screened for Wilms tumor?*  
Via periodic renal ultrasound

*How often, and for how long?*  
Every 3 months until age 7 years
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.

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?
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.
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Yes, it can be associated with Peters anomaly

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Renal, neurological and vascular abnormalities are common as well.
### 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.

**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.

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

**What organ is also affected?**
Renal, neurological and vascular abnormalities are common as well.
Alagille syndrome: Facies
Anterior Segment Dysgenesis

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Another syndrome of ophthalmic concern includes butterfly vertebrae as a finding. What is it?
Rieger syndrome
In two words, what sort of condition is Goldenhar?
A craniofacial malformation
What is the noneponymous name for Goldenhar syndrome?
Oculo-auricular-vertebral (OAV) syndrome
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Peripheral

Posterior embryotoxon

Axenfeld syndrome

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

Regarding arcus senilis...

What is its main chemical component?

Arcus juvenilis
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Arcus senilis
Corneal arcus
Axenfeld-Rieger syndrome
Posterior embryotoxon

Peripheral

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**Anterior Segment Dysgenesis**

- Axenfeld-Rieger syndrome
- Peters anomaly

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**What is arcus juvenilis?**

It is the congenital version of arcus senilis.

**Regarding arcus senilis...**

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

**Arcus senilis**

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

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

Bilateral

**Is it a dystrophy?**

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

**In what pattern does it typically declare itself?**

It starts at the poles, then spreads circumferentially.

**Does it exhibit a gender predilection?**

Yes, men are more likely to develop it.

**Does it exhibit a racial predilection?**

Yes, it is more common in AAs.

**Does its prevalence increase with age?**

Yes; after age 80, the prevalence is ~100%.
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Axenfeld-Rieger syndrome

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Carotid occlusive dz, or ocular ischemic syndrome (OIS)
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**If arcus is a sign of carotid occlusion or OIS, which side is occluded/ischemic--the side with the arcus, or the side without the arcus?**

- The side without the arcus

**Arcus senilis**

- It is a sign of ocular ischemia

**Arcus juvenilis**

- It is the congenital version of arcus senilis

**Arcus juvenilis aka anterior embryotoxon**

- It is found in about 15% of otherwise normal eyes

**Axenfeld-Rieger syndrome**

- It is associated with aniridia

**Alagille syndrome**

- It is associated with posterior keratoconus

**Central embryotoxon**

- It is anteriorly displaced and thickened Schwalbe's line/ring

**Posterior embryotoxon**

- It is found in about 15% of otherwise normal eyes

**Peripheral embryotoxon**

- It is another name for arcus juvenilis

**Corneal arcus**

- It starts at the poles, then spreads circumferentially

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

**Anterior segment dysgenesis**

Central

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
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What is arcus juvenilis?

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Regarding arcus senilis...

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

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

- **Is it a dystrophy?**
  - No; per the *Cornea* book, it is an “involutional change”

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  - Yes, men are more likely to develop it

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- **Does its prevalence increase with age?**
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What is the underlying mechanism for both the arcus and the CAD in these pts?
Familial hyperlipoproteinemia

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

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**Peripheral**

- Posterior embryotoxon
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Arcus senilis

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**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.

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

Xanthelasma

Do xanthelasma present unilaterally, or bilaterally?
Bilaterally.

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

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

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

Arcus senilis

Arcus juvenilis
aka
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What is arcus senilis?

Xanthelasma

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

What is arcus juvenilis, that is, what is there clinical appearance?
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

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

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It is the congenital version of arcus senilis

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

**Arcus juvenilis** aka anterior embryotoxon

**Arcus senilis**

**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?**

**Xanthelasma**

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

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**Arcus senilis**
Anterior Segment Dysgenesis

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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

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

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

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

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

Arcus senilis
Arcus juvenilis aka anterior embryotoxon

Xanthelasma

Familial hyperlipoproteinemia

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

What is xanthelasma?
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Do they present unilaterally, or bilaterally?
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Yes; after age 80, the prevalence is ~100%

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

What is arcus senilis?
It is the involutional change

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

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Anterior Segment Dysgenesis

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?

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

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

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

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

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**What is arcus juvenilis?**
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**Regarding arcus senilis...**

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

*Do they present unilaterally, or bilaterally?*
Bilaterally.

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

*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% before age 40.

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

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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.

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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

Regarding arcus senilis...

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

Do they present unilaterally, or bilaterally? Bilaterally.

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

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

Can they be congenital? Yes, and when they are, they usually are a sign of lipid derangement often present in such pts. What is it? Xanthelasma.

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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 there clinical appearance?
  - They are yellowish plaques located in the medial canthal region, usually on the upper eyelid.

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Anterior Segment Dysgenesis

What is a posterior embryotoxon?
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No; it is found in about 15% of otherwise normal eyes

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Yes--it is another name for arcus juvenilis

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

Regarding arcus senilis...

What 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

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

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

Regarding arcus senilis...
Anterior Segment Dysgenesis

Regarding arcus senilis...

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

Lipid-filled macrophages

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

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

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

Xanthelasma

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

Lipid-filled macrophages

This histology—lipid-filled macrophages—is often present in such pts. What is it? Xanthelasma

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

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 usually located in the medial canthal region, upper lids.

Lipid-filled macrophages

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

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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%
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 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

Arcus juvenilis aka anterior embryotoxon

Arcus senilis

Post 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
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Is there such a thing as an anterior embryotoxon?
Yes--it is another name for arcus juvenilis

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

Arcus senilis
Anterior Segment Dysgenesis

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

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 and the xanthelasmas are 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? 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, often present in such pts. What is it?

Xanthelasma

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

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

Speaking of ‘foamy macrophages’…
What disease comes to mind if, instead of an adult with xanthelasmas, the 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.
The 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?
Whipple’s disease

Are they composed of lipid?
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Can they be congenital?
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What is it?
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Arcus juvenilis aka anterior embryotoxon

Arcus senilis
Anterior Segment Dysgenesis

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Whipple’s disease

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First clue…

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

Speaking of ‘foamy macrophages’…
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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.

Speaking of ‘foamy macrophages’ part deaux…
What dz comes to mind if, instead of an adult with xanthelasmas, the pt in question was a very young child with unilateral pigmented iris nodules?
And heterochromia iridis 2ndry to those nodules?
Juvenile xanthogranuloma (JXG)

Second clue

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

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

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.

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 '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?
Whipple’s disease.

Are they always a harbinger of elevated serum lipids?
Last clue--the answer is next.

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

Speaking of ‘foamy macrophages’… What disease comes to mind if, instead of an adult with xanthelasmas, the 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.

Speaking of ‘foamy macrophages’ part deaux… What disease 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

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 an equivalent ‘another name’ for arcus senilis?
Yes--anterior gerontoxon

Finally: Anterior embryotoxon is another name for arcus juvenilis.

Arcus juvenilis aka anterior embryotoxon
Arcus senilis aka?
Anterior Segment Dysgenesis

Anterior segment dysgenesis

Peripheral

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 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

 Peripheral

 Posterior embryotoxon

 Axenfeld-Rieger syndrome

 Corneal arcus

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 aka anterior embryotoxon

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 Gerontoxon?
Anterior Segment Dysgenesis

**Anterior segment dysgenesis**

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

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

What features define Axenfeld-Rieger syndrome?

Peripheral

Posterior embryotoxon

Axenfeld-Rieger syndrome

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

Peripheral

Posterior embryotoxon

Axenfeld-Rieger syndrome

What features define Axenfeld-Rieger syndrome?
Posterior embryotoxon with attached iris strands + iris hypoplasia + angle abnormalities
Normal iris strands attached to SS
Abnormal iris strands attached to posterior embryotoxon in A-R
Anterior Segment Dysgenesis

**Anterior segment dysgenesis**

- What features define Axenfeld-Rieger syndrome?
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- Where does ARS rank as a cause of iris hypoplasia?
  - It is the most common
Anterior Segment Dysgenesis

**Posterior embryotoxon**

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**Peripheral**

**Iris hypoplasia**

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

50%
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Lifetime risk of developing glaucoma: 50%
Anterior Segment Dysgenesis

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

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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 is a misnomer. Why?
Because the posterior pigmented epithelium derives from neuroectoderm, not uvea.
Anterior Segment Dysgenesis

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**Peripheral**

- Posterior embryotoxon
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3 y.o. girl who presented at three months of age with hazy megalocornea, posterior embryotoxon, iris hypoplasia, corectopia with early onset severe glaucoma. The horizontal/vertical corneal diameters were 13.0/12.5 mm.
A-R with microcornea (8.5 mm)
Anterior Segment Dysgenesis

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

Axenfeld-Reiger syndrome
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In one word, what sort of condition is IP?

- A phakomatosis

What is the eponymous name for IP?

- Bloch-Sulzberger syndrome
Anterior Segment Dysgenesis

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Anterior Segment Dysgenesis

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

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

What corneal abnormalities may be present?
1) Megalocornea
2) Microcornea

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

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

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

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

**Anterior segment dysgenesis**

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**Anterior segment dysgenesis**

- Peripheral
- Posterior embryotoxon
- Axenfeld-Rieger syndrome

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Anterior Segment Dysgenesis

Two classic central dysgeneses
Anterior Segment Dysgenesis

Anterior segment dysgenesis

Peripheral
- Posterior embryotoxon
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Central
- Posterior keratoconus
- Peters anomaly

Two classic central dysgeneses
Anterior Segment Dysgenesis

Anterior segment dysgenesis

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

What is posterior keratoconus?

Central

Posterior keratoconus

Peters anomaly
Anterior Segment Dysgenesis

Anterior segment dysgenesis

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

Central

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Anterior Segment Dysgenesis

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

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

Central

Posterior keratoconus

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**Anterior Segment Dysgenesis**

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**Posterior keratoconus**

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How does it present?

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

How might the lens be involved?

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

Does it require a workup?

No if it's unilateral (usually sporadic)
Yes if it's bilateral (do a complete genetic and systemic workup)
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2. Adhesions extending from the iris to the posterior corneal defect

Peters anomaly
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Peters anomaly: Hazy cornea
Anterior Segment Dysgenesis

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

STUMPED mnemonic
- Sclerocornea
- Stromal dystrophy (CHSD)
- Trauma (eg, forcep injury)
- Ulcer
- Mucopolysaccharidosis
- Peters anomaly
- Endothelial dystrophy (CHED); Elevated IOP (congenital glaucoma)
- Dermoid of the cornea

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

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Peripheral Central
Embryotoxon
Axenfeld-Rieger syndrome
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Peters anomaly: Cataractous lens
Anterior Segement Dysgenesis

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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

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What common slit-lamp observation owes to the lens’ small size?

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

-- Occasionally it is… **small**, misshapen, and displaced into the AC
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--It is usually highly myopic

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With what condition is microspherophakia most frequently associated?
Weill-Marchesani syndrome

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
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

With what condition is microspherophakia most frequently associated? Weill-Marchesani syndrome

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Type 1  Type 2

Peters anomaly
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