Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

Basic distinction used by the BCSC Peds book

Abnormalities of Corneal Transparency
Developmental Abnormalities of the Cornea

Abnormalities of Corneal *Size* or *Shape*  
Abnormalities of Corneal *Transparency*

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Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

- Microcornea
- Cornea plana

Abnormalities of Corneal Transparency

- Megalocornea
What is the definition of megalocornea, ie, how ‘megal’ does it have to be?

Megalocornea?

At age >2 years
At birth

Corneal diameter (mm)

7 8 9 10 11 12 13 14 15
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

- Microcornea
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Abnormalities of Corneal Transparency

What is the definition of megalocornea, ie, how ‘megalo’ does it have to be?
Corneal diameter > 12 mm at birth, or > 13 mm at age 2 years or older
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

- Megalocornea
- Microcornea
- Cornea plana

Abnormalities of Corneal Transparency

- ?
- ?
Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

Megalocornea

Primary

Secondary

Cornea plana

Microcornea

Abnormalities of Corneal Transparency

Developmental Abnormalities of the Cornea
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

Is primary megalocornea a congenital, or acquired condition?

- Congenital—always, and by definition
- Bilaterally—always, and by definition
- Yes; it is X-linked recessive (so is more common in males)
- With what other ocular abnormalities is it associated?
  - Lens abnormalities: Cataract, ectopia lentis
  - Iris abnormalities: Miosis, translucency
- It can be associated with systemic conditions. Which ones?
  - Down syndrome
  - Marfan syndrome
  - Alport syndrome
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Abnormalities of Corneal Size or Shape

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Abnormalities of Corneal

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### Abnormalities of Corneal Size or Shape

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<thead>
<tr>
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- Developmental Abnormalities
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Speaking of cataracts: Is primary megalocornea associated with an increased risk of intraoperative complications during cataract surgery?

Indeed it is

What is it about megalocornea eyes that predisposes them to complications?

(Hint: It's not a corneal issue)

These eyes can have "poor zonular integrity," with all the intra-op problems that entails. (BTW, the quote is from the BCSC Cornea book, which considers this factoid to be a "highlight." The point being, consider it memorization-worthy.)
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(Hint: It’s not a corneal issue)
- These eyes can have “poor zonular integrity,” with all the intra-op problems that entails. (Note: The BCSC Cornea book has made this fact about megalocornea a chapter “highlight.” The point being, it is probably worthy of memorization.)

Megalocornea

Primary

Secondary
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

Is primary megalocornea a congenital, or acquired condition?
Congenital—always, and by definition.

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With what other ocular abnormalities is it associated?
There are many; they include:
- Lens abnormalities: Cataract, ectopia lentis
- Iris abnormalities: Miosis, translucency

What is ectopia lentis?
Displacement of the lens from its normal anatomic position.

These abnormalities can also be associated with systemic conditions. Which ones?
There are many; they include:
- Down syndrome
- Marfan syndrome
- Alport syndrome

Primary Megalocornea

Secondary Megalocornea
Developmental Abnormalities of the Cornea

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Developmental Abnormalities of the Cornea

Ectopia lentis
Abnormalities of Corneal Transparency

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- Fibrillin

What three structures/systems manifest abnormalities in Marfan’s?
- The eye
- The cardiovascular
- The musculoskeletal

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What proportion of Marfan pts manifest ocular abnormalities?
At least 80%

Is megalocornea a common ocular manifestation?
No

OK then, what ocular abnormalities are common?
- High myopia
- Ectopia lentis
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- Does it usually present unilaterally, or bilaterally?
  - Bilaterally—always, and by definition

- Is it an inherited condition? If so, in what manner is it transmitted?
  - Yes; it is X-linked recessive (so is more common in males)

- With what other ocular abnormalities is it associated?
  - Lens abnormalities: Cataract, ectopia lentis
  - Iris abnormalities: Miosis, translucency

- It can be associated with systemic conditions. Which ones?
  - Down syndrome
  - Marfan syndrome
  - Alport syndrome

- What is the name of the protein that is abnormal in Marfan’s?
  - Fibrillin

- What three structures/systems manifest abnormalities in Marfan’s?
  - The eye (duh)
  - The cardiovascular
  - The musculoskeletal

- What proportion of Marfan pts manifest ocular abnormalities?
  - At least 80%

- Is megalocornea a common ocular manifestation?
  - No

- OK then, what ocular abnormalities are common?
  - High myopia
  - Ectopia lentis
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

- Megalocornea
- Microcornea
- Cornea plana

Developmental Abnormalities of the Cornea

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Congenital—always, and by definition

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- Marfan syndrome
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Is megalocornea a congenital, or acquired condition?
Congenital—always, and by definition

Primary Megalocornea

Secondary Megalocornea

Microcornea

Cornea plana
Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea
- Cornea plana

Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

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In which direction do the lenses tend to displace in Marfan’s?
Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea
- Cornea plana

Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

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In which direction do the lenses tend to displace in Marfan’s? Superotemporal
Abnormalities of Corneal Transparency

- Microcornea
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- Cornea plana

Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

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OK then, what ocular abnormalities are common?
- High myopia
- Ectopia lentis

What condition is associated with displacement superonasally?
- Ectopia lentis et pupillae

What condition is associated with displacement inferonasally?
- Homocystinuria

What condition is associated with displacement inferotemporally?
- Nothing I know of

In which direction do the lenses tend to displace in Marfan’s?
- Superotemporal

What three things are primary?
- The eye
- The cardiovascular
- The musculoskeletal
Is megalocornea a common ocular manifestation? No

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In which direction do the lenses tend to displace in Marfan's?
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Marfan Marfan Ectopia lentis et pupillae

Developmental Abnormalities of the Cornea

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Abnormalities of Corneal Size or Shape

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Microcornea
Megalocornea
Cornea plana

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Developmental Abnormalities of the Cornea

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With what other ocular abnormalities is it associated? They include:
- Lens abnormalities: Cataract, ectopia lentis
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It can be associated with systemic conditions. Which ones? They include:
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In which direction do the lenses tend to displace in Marfan’s? Supero-temporal

What condition is associated with displacement superonasally? Ectopia lentis et pupillae

What condition is associated with displacement inferonasally? Nothing I know of

Marfan Ectopia lentis et pupillae
Developmental Abnormalities of the Cornea

Is megalocornea a common ocular manifestation? No

What condition is associated with displacement superonasally? Ectopia lentis et pupillae

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With what other ocular abnormalities is it associated? There are many; they include:

- Lens abnormalities: Cataract, ectopia lentis
- Iris abnormalities: Miosis, translucency

It can be associated with systemic conditions. Which ones? There are many; they include:

- Down syndrome
- Marfan syndrome
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What is the name of the protein that is abnormal in Marfan's? Fibrillin

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What condition is associated with displacement superonasally? Ectopia lentis et pupillae

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In which direction do the lenses tend to displace in Marfan's? Superotemporal
Developmental Abnormalities of the Cornea

**Is primary megalocornea a congenital, or acquired condition?**
- Congenital—always, and by definition
- Bilaterally—always, and by definition

**Is it an inherited condition? If so, in what manner is it transmitted?**
- Yes; it is X-linked recessive (so is more common in males)

**With what other ocular abnormalities is it associated?**
- Many; includes:
  - Lens abnormalities: Cataract, ectopia lentis
  - Iris abnormalities: Miosis, translucency
  - Also associated with systemic conditions: Down syndrome, Marfan syndrome, Alport syndrome

**What is the name of the protein that is abnormal in Marfan’s?**
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Developmental Abnormalities of the Cornea

Abnormalities of Corneal Transparency

Microcornea
Megalocornea
Cornea plana

Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

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There are many; they include:
- Down syndrome
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- Alport syndrome

What sort of condition is Alport syndrome?
A familial oculorenal syndrome

Another familial oculorenal syndrome is often mentioned along with Alport syndrome. What is its eponymous name?
Lowe syndrome

What is the classic presenting sign of the familial oculorenal syndromes (hint: It's nonocular)?
Hematuria

What is the classic lens finding in the familial oculorenal syndromes?
Lenticonus
Abnormalities of Corneal Transparency
- Microcornea
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Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

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Abnormalities of Corneal Size or Shape

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Alport syndrome
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

- Megaocornea
- Microcornea
- Cornea plana

Is secondary megalocornea a congenital, or acquired condition?
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

- Microcornea
- Cornea plana
- Megalocornea

Abnormalities of Corneal Transparency

Is secondary megalocornea a congenital, or acquired condition?
Acquired—always, and by definition
Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

Cornea plana

Microcornea

Megalocornea

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Abnormalities of Corneal Transparency

- Megalocornea
- Microcornea

Abnormalities of Corneal Size or Shape

- Primary
- Secondary

Developmental Abnormalities of the Cornea

- Cornea plana

Is secondary megalocornea a congenital, or acquired condition?
Acquired—always, and by definition

Does it usually present unilaterally, or bilaterally?
It can be either
Developmental Abnormalities of the Cornea

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What is the cause?
Abnormalities of Corneal Transparency

- Microcornea
- Me-}

Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

- Cornea plana
- Microcornea
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Elevated IOP
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What is the cause?
Elevated IOP

Wait a minute—I thought an enlarged globe secondary to elevated IOP is buphthalmos. What’s the difference between secondary megalocornea and buphthalmos?

In buphthalmos, the entire globe (including the cornea) is enlarged, whereas in secondary megalocornea, only the cornea is—the rest of the eye is normally sized

OK, so I see a baby with elevated IOP and big corneas. How do I know whether its buphthalmos vs secondary megalocornea?

By measuring the globes (with ultrasound). In buphthalmos, the eye will be proportionately enlarged; i.e., the deep AC will be accompanied by an enlarged vitreous cavity. In contrast, in secondary megalocornea AC depth will comprise a disproportionately large portion of overall eye length.

With regard to AC depth in secondary megalocornea, what proportion of total eye length are we talking about?
If AC depth is greater than about 20% of total eye length, it’s secondary megalocornea

Think of secondary megalocornea as representing ‘arrested buphthalmos,’ i.e., it affected the anterior segment, but was prevented from affecting the rest of the globe
Abnormalities of Corneal Transparency

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Microcornea

Megalocornea

Cornea plana

Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

Is secondary megalocornea a congenital, or acquired condition?
Acquired—always, and by definition

Does it usually present unilaterally, or bilaterally?
It can be either

What is the cause?
Elevated IOP

Wait a minute—I thought an enlarged globe secondary to elevated IOP is buphthalmos. What’s the difference between secondary megalocornea and buphthalmos?
In buphthalmos, the entire globe (including the cornea) is enlarged, whereas in secondary megalocornea, only the cornea is—the rest of the eye is normally sized.

OK, so I see a baby with elevated IOP and big corneas. How do I know whether its buphthalmos vs secondary megalocornea?
By measuring the globes (with ultrasound). In buphthalmos, the eye will be proportionately enlarged; ie, the deep AC will be accompanied by an enlarged vitreous cavity. In contrast, in secondary megalocornea AC depth will comprise a disproportionately large portion of overall eye length.

With regard to AC depth in secondary megalocornea, what proportion of total eye length are we talking about?

If AC depth is greater than about 20% of total eye length, it’s secondary megalocornea

Think of secondary megalocornea as representing ‘arrested buphthalmos,’ ie, it affected the anterior segment, but was prevented from affecting the rest of the globe.
Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea
- Cornea plana

Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

Is secondary megalocornea a congenital, or acquired condition?
Acquired—always, and by definition

Does it usually present unilaterally, or bilaterally?
It can be either

What is the cause?
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Megalocornea

Primary

Secondary

Is secondary megalocornea a congenital, or acquired condition?
Acquired—always, and by definition

Does it usually present unilaterally, or bilaterally?
It can be either

What is the cause?
Elevated IOP
Abnormalities of Corneal Transparency

Microcornea

Megalocornea

Cornea plana

Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

Is secondary megalocornea a congenital, or acquired condition?
Acquired—always, and by definition

Does it usually present unilaterally, or bilaterally?
It can be either

What is the cause?
Elevated IOP

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Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea
- Cornea plana

Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

Primary

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If AC depth is greater than about 20% of total eye length, it’s secondary megalocornea.

Think of secondary megalocornea as ‘arrested buphthalmos,’ ie, the IOP affected the anterior segment, but not the rest of the globe.
Abnormalities of Corneal Transparency

Microcornea

Megalocornea

Cornea plana

Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

What is the definition of microcornea, ie, how ‘micro’ does it have to be?

Microcornea?

Megalocornea

At age >2 years

At birth

Corneal diameter (mm)

7 8 9 10 11 12 13 14 15
Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea

Abnormalities of Corneal Size or Shape

- Cornea plana

What is the definition of microcornea, ie, how ‘micro’ does it have to be?
Corneal diameter < 9 mm at birth, or < 10 mm at age 2 years or older
Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea

Abnormalities of Corneal Size or Shape

- Does microcornea present unilaterally, or bilaterally?
  - Can cornea also be:
    - Too thick?
    - Hazy?
    - Too flat?

With what ocular conditions is microcornea associated?
- Persistent fetal vasculature (PFV, aka PHPV)
- Congenital cataracts

With what systemic conditions is microcornea associated?
- Ehlers-Danlos syndrome
- Myotonic dystrophy
Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea

Abnormalities of Corneal Size or Shape

- Does microcornea present unilaterally, or bilaterally? It can be either
- Does the cornea also:
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  - Be hazy?
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Developmental Abnormalities of the Cornea

Microcornea

Unilateral

Bilateral
Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

Microcornea

Megalocornea

Developmental Abnormalities of the Cornea

Does microcornea present unilaterally, or bilaterally? It can be either

In addition to being small, can the cornea also be:
-- Too thick?

-- Persistent fetal vasculature (PFV, aka PHPV)
-- Congenital cataracts

-- Ehlers-Danlos syndrome
-- Myotonic dystrophy
Abnormalities of Corneal Translucency

Microcornea

Megalocornea

Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

Abnormalities of Corneal Transparency

Does microcornea present unilaterally, or bilaterally? It can be either.

In addition to being small, can the cornea also be:

--Too thick? No—by definition, the thickness is normal.

--Hazy?
No—by definition, the cornea is clear

--Too flat?
Yes, the cornea is usually flatter than normal

With what ocular conditions is microcornea associated?

--Persistent fetal vasculature (PFV, aka PHPV)
--Congenital cataracts

With what systemic conditions is microcornea associated?

--Ehlers-Danlos syndrome
--Myotonic dystrophy
Abnormalities of Corneal Transparency

Developmental Abnormalities of the Cornea

Abnormalities of
Corneal Size or Shape

Microcornea

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In addition to being small, can the cornea also be:
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Abnormalities of
Corneal Transparency

Megalocornea

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--Ehlers-Danlos syndrome--Myotonic dystrophy
Abnormalities of Corneal Transparency

Microcornea

Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

Megalocornea

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Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

Microcornea

Megalocornea

Abnormalities of Corneal Transparency

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Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

Microcornea

Megalocornea

Abnormalities of Corneal Transparency

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Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

- Microcornea
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- Megalocornea

Abnormalities of Corneal Transparency

- Developmental Abnormalities of the Cornea

A flat cornea implies hyperopia. Is microcornea in fact associated with hyperopia?
Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

Does microcornea present unilaterally, or bilaterally? It can be either.

In addition to being small, can the cornea also be:
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Abnormalities of Corneal Size or Shape

Microcornea

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A flat cornea implies hyperopia. Is microcornea in fact associated with hyperopia? Yes

A flat cornea implies a shallow AC. Is microcornea in fact associated with a shallow AC?
Abnormalities of Corneal Transparency

Megalocornea

Abnormalities of Corneal Size or Shape

Microcornea

Developmental Abnormalities of the Cornea

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--Ehlers-Danlos syndrome--Myotonic dystrophy
Abnormalities of Corneal Transparency

Microcornea

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A flat cornea implies hyperopia. Is microcornea in fact associated with hyperopia? Yes

A flat cornea implies a shallow AC. Is microcornea in fact associated with a shallow AC? Yes

A shallow AC implies increased risk of angle-closure glaucoma. Is microcornea in fact associated with an increased risk of angle-closure glaucoma?
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

Microcornea

Megalocornea

Abnormalities of Corneal Transparency

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Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

Microcornea

Megalocornea

Abnormalities of Corneal Transparency

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What about open-angle glaucoma--does cornea plana convey an increased risk of it?

A shallow AC implies increased risk of angle-closure glaucoma. Is microcornea in fact associated with an increased risk of angle-closure glaucoma? Yes
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

- Microcornea
- Megalocornea

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It can be either

In addition to being small, can the cornea also be:
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A flat cornea implies hyperopia. Is microcornea in fact associated with hyperopia?
Yes

What about open-angle glaucoma—does cornea plana convey an increased risk of it?
Yes. Of the cornea-planan pts who manage to avoid developing angle-closure glaucoma, 20% will go on to develop the open-angle version.

A shallow AC implies increased risk of angle-closure glaucoma. Is microcornea in fact associated with an increased risk of angle-closure glaucoma?
Yes
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

Microcornea

Megalocornea

Abnormalities of Corneal Transparency

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Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

Microcornea

Megalocornea

Developmental Abnormalities of the Cornea

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With what ocular conditions is microcornea associated?
--
--

With what systemic conditions is microcornea associated?
--Ehlers-Danlos syndrome--Myotonic dystrophy
Abnormalities of Corneal Transparency

Microcornea

Does microcornea present unilaterally, or bilaterally?
It can be either

In addition to being small, can the cornea also be:
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--Hazy? No—by definition, the cornea is clear
--Too flat? Yes, the cornea is usually flatter than normal

With what ocular conditions is microcornea associated?
--Persistent fetal vasculature (PFV, aka PHPV)
--Congenital
Does microcornea present unilaterally, or bilaterally? It can be either

*In addition to being small, can the cornea also be:*

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--Persistent fetal vasculature (PFV, aka PHPV)
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Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

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Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

Microcornea

Megalocornea

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-- Persistent fetal vasculature (PFV, aka PHPV)
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With what systemic conditions is microcornea associated?
-- Ehlers-Danlos syndrome
-- Myotonic dystrophy
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape
- Micropia
- Megalocornea

Hol up—you're skipping cornea plana! What's up with that?

Abnormalities of Corneal Transparency

Basic distinction used by the BCSC Peds book

Hol up—you're skipping cornea plana! What's up with that?
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

- Microcornea
- Megalocornea
- Cornea plana

Abnormalities of Corneal Transparency

- Hol up—you’re skipping cornea plana! What’s up with that?
- Patience, Grasshopper—all will be made clear soon
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape
- Microcornea
- Cornea plana
- Megalocornea

Abnormalities of Corneal Transparency

Basic distinction used by the BCSC Peds book

(OK, now answer this)
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape
- Microcornea
- Megalocornea
- Cornea plana

Abnormalities of Corneal Transparency
- Peripheral
- Central

Basic distinction used by the BCSC Peds book

(OK, now answer this)
Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea
- Cornea plana

Abnormalities of Corneal Size or Shape

- Peripheral
  - ?
- Central
  - ?

Developmental Abnormalities of the Cornea
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape
- Microcornea
- Megalocornea
- Cornea plana

Abnormalities of Corneal Transparency
- Peripheral
  - Cornea plana
  - Posterior embryotoxon
- Central
  - Epibulbar dermoid
Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

Cornea plana

Peripheral

Central

Microcornea

Cornea plana

Posterior embryotoxon

Cornea plana belongs in both groups, is why it was saved until now…

No question—carry on
Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

Central

Peripheral

Cornea plana

Microcornea

Cornea plana belongs in both groups, is why it was saved until now…

*Cornea plana has five characteristics, one of which is flat Ks. What are the other four?*

--Flat Ks

--

(The reason there are only three more slots will become apparent on the next slide)
Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea
- Cornea plana

Abnormalities of Corneal Size or Shape

- Peripheral Developmental Abnormalities of the Cornea
  - Central
    - Posterior embryotoxon
  - Peripheral
    - Cornea plana

Cornea plana belongs in both groups, is why it was saved until now...

Cornea plana has five characteristics, one of which is flat Ks. What are the other four?

-- Flat Ks
-- Poorly developed ("indistinct") limbus
-- Shallow AC
-- High refractive status with associated two words

#4
#5
Cornea plana belongs in both groups, is why it was saved until now…

*Cornea plana has five characteristics, one of which is flat Ks. What are the other four?*
--Flat Ks
--Poorly developed (“indistinct”) limbus
--Shallow AC
--High hyperopia with associated accommodative esotropia
Yikes—that is pretty flat, innit?

Cornea plana
Developmental Abnormalities of the Cornea

Yikes—that is pretty flat, innit?

Note the ‘indistinct’ limbus

Cornea plana
Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

Microcornea

Cornea plana

Megalocornea

Cornea plana

Cornea plana belongs in both groups, is why it was saved until now…

Peripheral Developmental Abnormalities of the Cornea

Central

Cornea plana

Cornea plana

What is the average central corneal power of the normal adult cornea?

About 43D

How flat does the central cornea have to be to qualify as cornea plana?

Technically, less than 43D, but don’t get it twisted—Most corneas within shouting distance of 43D are not ‘plana’

OK, what is the typical power of a plana cornea?

Values in the 30-35D range are common

There a pathognomonic corneal curvature—what is it? (Hint: It’s not a specific numeric value.)

If the cornea is the same curvature as the adjacent sclera, the eye is plana.
Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea

Abnormalities of Corneal Size or Shape

- Cornea plana
- Peripheral development abnormalities
- Central development abnormalities

Cornea plana

- Flat Ks
- Poorly developed limbus
- Shallow AC
- High hyperopia with associated accommodative esotropia

Developmental Abnormalities of the Cornea

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Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea

Abnormalities of Corneal Size or Shape

- Cornea plana
- Peripheral Developmental Abnormalities
  - Posterior embryotoxon
  - Epibulbar dermoid
  - Dermolipomas

Central Developmental Abnormalities of the Cornea

Cornea plana belongs in both groups, is why it was saved until now...

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- Flat Ks
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Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea

Abnormalities of Corneal Size or Shape

- Cornea plana
- Peripheral developmental abnormalities
- Central developmental abnormalities

Microcornea

Cornea plana

Peripheral developmental abnormalities

Central developmental abnormalities

**Developmental Abnormalities of the Cornea**

- Abnormalities of Corneal Transparency
- Abnormalities of Corneal Size or Shape

**What is the average central corneal power of the normal adult cornea?**
About 43D

**How flat does the central cornea have to be to qualify as cornea plana?**
Technically, less than 43D, but don’t get it twisted—most corneas within shouting distance of 43D are not ‘plana.’ Plana cornea are much flatter.
Abnormalities of Corneal Transparency

- Microcornea
- Me
cola

Abnormalities of Corneal Size or Shape

- Peripheral Developmental Abnormalities of the Cornea
- Central Developmental Abnormalities of the Cornea

Central Posterior embryotoxon

Cornea plana

Peripheral Cornea plana

What is the average central corneal power of the normal adult cornea?
About 43D

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Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea

Abnormalities of Corneal Size or Shape

- Peripheral Developmental Abnormalities of the Cornea
- Central Developmental Abnormalities of the Cornea
  - Posterior embryotoxon
  - Cornea plana
  - Epibulbar dermoid
  - Dermolipomas

Cornea plana belongs in both groups, why it was saved until now…

Cornea plana has five characteristics, one of which is flat Ks. What are the other four?

- Flat Ks
- Poorly developed (“indistinct”) limbus
- Shallow AC
- High hyperopia with associated accommodative esotropia

What is the average central corneal power of the normal adult cornea?

About 43D

How flat does the central cornea have to be to qualify as cornea plana?

Technically, less than 43D, but don’t get it twisted--most corneas within shouting distance of 43D are not ‘plana.’ Plana cornea are much flatter.

OK, what is the typical power of a plana cornea?

Values in the 30-35D range are common
Developmental Abnormalities of the Cornea

Cornea plana: Keratometry (I know, it’s really blurry)
Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea

Abnormalities of Corneal Size or Shape

- Peripheral Developmental Abnormalities of the Cornea
- Central Developmental Abnormalities of the Cornea

- Posterior embryotoxon
- Epibulbar dermoid
- Dermolipomas

Cornea plana belongs in both groups, is why it was saved until now…

Cornea plana has five characteristics, one of which is flat Ks. What are the other four?

- Flat Ks
- Poorly developed (“indistinct”) limbus
- Shallow AC
- High hyperopia with associated accommodative esotropia

What is the average central corneal power of the normal adult cornea? About 43D

How flat does the central cornea have to be to qualify as cornea plana? Technically, less than 43D, but don’t get it twisted--most corneas within shouting distance of 43D are not ‘plana.’ Plana cornea are much flatter.

OK, what is the typical power of a plana cornea? Values in the 30-35D range are common

There a pathognomonic corneal curvature--what is it? (Hint: It’s not a specific numeric value.)
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- Microcornea
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The key difference is one of corneal transparency. In sclerocornea, the cornea is opaque. In cornea plana, the cornea is clear (relatively speaking).

the same curvature as the adjacent sclera
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a word
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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 anomaly
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No--it is usually too thin and posterior to be seen
Normal angle anatomy: Identify the structures
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Hints forthcoming…

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

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

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Axenfeld-Reiger: Note the posterior embryotoxon (1) with attached iris strands (2)
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‘Angle abnormalities’ suggests an increased risk of glaucoma. Does ARS in fact convey such a risk?

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

Why central-ish?
Deviation from centrality of 1/2 mm is common, and up to 1 mm is considered normal
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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

Corectopia

Posterior embryotoxon
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Transparency

Microcornea
Megalocornea
Cornea plana

Abnormalities of Corneal Size or Shape

Peripheral Developmental Abnormalities of the Cornea

Central Posterior embryotoxon
Cornea plana
Epibulbar dermoid
Dermolipomas

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 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
Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea
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Abnormalities of Corneal Size or Shape

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Axenfeld-Rieger syndrome

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Ectropion uveae
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3) When it is associated with Alagille syndrome

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Not simultaneously, obviously
Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea
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Abnormalities of Corneal Size or Shape

Peripheral Developmental Abnormalities of the Cornea

Central Posterior embryotoxon

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*Not simultaneously, obviously*
Developmental Abnormalities of the Cornea

3 y.o. girl who presented at three months of age with hazy megalocornea, posterior embryotoxon, iris hypoplasia, corectopia, and early-onset severe glaucoma. The horizontal/vertical corneal diameters were 13.0/12.5 mm.
Axenfeld-Reiger with microcornea (8.5 mm)
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea

Abnormalities of Corneal Size or Shape

- Peripheral Developmental Abnormalities of the Cornea
  - Central Posterior embryotoxon
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Axenfeld-Rieger syndrome

Alagille syndrome
Developmental Abnormalities of the Cornea

(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
Developmental Abnormalities of the Cornea

(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
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For more on A-R, see slide-set FELT7

Axenfeld-Reiger syndrome
Developmental Abnormalities of the Cornea

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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
Developmental Abnormalities of the Cornea

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Aniridia. Note the presence of an iris stub/root
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- Microcornea
- Megalocornea
- Cornea plana

Abnormalities of Corneal Size or Shape
- Peripheral Developmental Abnormalities of the Cornea
- Central Posterior embryotoxon
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aniridia

Axenfeld-Rieger syndrome

aniridia

Alagille syndrome

Posterior embryotoxon

Epibulbar dermoid
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Aniridia

Axenfeld-Rieger syndrome

aniridia

Alagille syndrome

Posterior embryotoxon

Epibulbar dermoid
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Peripheral Developmental Abnormalities of the Cornea

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Abnormalities of Corneal Size or Shape

Peripheral Developmental Abnormalities of the Cornea

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- Aniridia
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Aniridia

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- Aniridia
- Alagille syndrome
- Posterior embryotoxon
- Epibulbar dermoid
Developmental Abnormalities of the Cornea

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Yes

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

WAGR complex consists of:
- W
- Aniridia
- A
- R

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

With what developmental complex is aniridia associated?
The WAGR complex

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

What are the three situations in which a posterior embryotoxon is a significant finding?
1) When it is part of the Axenfeld-Rieger syndrome
2) When it is associated with aniridia
3) When it is associated with Alagille syndrome

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
Developmental Abnormalities of the Cornea

WAGR complex: Wilm’s tumor
Developmental Abnormalities of the Cornea

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With what developmental ‘complex’ is aniridia associated?
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Are all aniridia cases at risk for WAGR complex?
No, only those in which the genetic mutation is sporadic


Developmental Abnormalities of the Cornea

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No, only those in which the genetic mutation is sporadic vs familial

Aniridia

Axenfeld-Rieger syndrome

Alagille syndrome

Posterior embryotoxon

Epibulbar dermoid

Central posterior embryotoxon

Abnormalities of corneal transparency

Microcornea
Megalocornea
Cornea plana

Abnormalities of corneal size or shape

Peripheral developmental abnormalities of the cornea

Central posterior embryotoxon

Axenfeld-Rieger syndrome

Aniridia

WAGR complex

Epibulbar dermoid

Cornea plana
Developmental Abnormalities of the Cornea

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

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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.
Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea
- Cornea plana

Abnormalities of Corneal Size or Shape

- Peripheral Developmental Abnormalities of the Cornea
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**Is it always a harbinger of significant pathology?**
No; it is found in about 15% of otherwise normal eyes

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For more on aniridia, see slide-set P17
Developmental Abnormalities of the Cornea

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

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 also common)
Developmental Abnormalities of the Cornea

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

What is the noneponymous name of Alagille syndrome? Arterohepatic dysplasia

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Is it a significant finding? It is significant in the following situations:
1. When it is part of the Axenfeld-Rieger syndrome
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Developmental Abnormalities of the Cornea

Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea
- Cornea plana

Abnormalities of Corneal Size or Shape

Peripheral Developmental Abnormalities of the Cornea

- Central Posterior embryotoxon
- Cornea plana
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Posterior embryotoxon

Epibulbar dermoid
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Transparency

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

What is the non-eponymous 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

To be clear—what is being asked of the eye service, ie, what are you supposed to check for in a ‘rule out Alagille’ consult?

Posterior embryotoxon

What 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

What is the non-eponymous 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 syndrome

Alagille syndrome

Posterior embryotoxon

Epibulbar dermoid
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Transparency

Microcornea

Megalocornea

Cornea plana

Abnormalities of Corneal Size or Shape

Peripheral Developmental Abnormalities of the Cornea

Central Posterior embryotoxon

Cornea plana

Epibulbar dermoid

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

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

To be clear—what is being asked of the eye service, ie, what are you supposed to check for in a ‘rule out Alagille’ consult?
You are being asked to determine whether the infant has a posterior embryotoxon

What is it a significant finding?
Yes; 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

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 syndrome

Posterior embryotoxon
Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea
- Cornea plana

Abnormalities of Corneal Size or Shape

Peripheral Developmental Abnormalities of the Cornea

- Central Posterior embryotoxon
- Epibulbar dermoid

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 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

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? 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 also common)
Abnormalities of Corneal Transparency

Microcornea
Megalocornea
Cornea plana

Abnormalities of Corneal Size or Shape

Peripheral Developmental Abnormalities of the Cornea

Central Posterior embryotoxon
Cornea plana
Epibulbar dermoid

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

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.

What is it a significant finding?
Axenfeld-Rieger syndrome
aniridia
Alagille syndrome

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

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

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.
Developmental Abnormalities of the Cornea

Alagille syndrome: Facies
Abnormalities of Corneal Transparency

Microcornea
Megalocornea
Cornea plana

Abnormalities of Corneal Size or Shape

Peripheral Developmental Abnormalities of the Cornea

Central Posterior embryotoxon
Cornea plana
Epibulbar dermoid

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

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?

What is it a significant finding?
Yes; 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

What is the noneponymous name of Alagille syndrome?
Arteriohepatic dysplasia

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

Alagille syndrome

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

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
--The skeleton
Abnormalities of Corneal Transparency

Microcornea
Megalocornea
Cornea plana

Abnormalities of Corneal Size or Shape

Peripheral Developmental Abnormalities of the Cornea

Central Posterior embryotoxon
Cornea plana
Epibulbar dermoid

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

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:

--The skeleton:
Developmental Abnormalities of the Cornea

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

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 also common)
Developmental Abnormalities of the Cornea

Alagille syndrome: Butterfly vertebrae
Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea
- Cornea plana

Abnormalities of Corneal Size or Shape

Peripheral Developmental Abnormalities of the Cornea

Central Posterior embryotoxon

Epibulbar dermoid

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

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, 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

What is the noneponymous name of Alagille syndrome?
Arteriohepatic dysplasia

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

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

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

In addition to liver, in what three situations is it a significant finding?
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 also common)
Abnormalities of Corneal Transparency:
- Microcornea
- Megalocornea
- Cornea plana

Abnormalities of Corneal Size or Shape:
- Peripheral Developmental Abnormalities of the Cornea
  - Central Posterior embryotoxon
  - Cornea plana
  - Epibulbar dermoid

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

What is the noneponymous name of Alagille syndrome? Arteriohepatic dysplasia

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

What is the classical 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.

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

(Renal, neurologic, and vascular abnormalities are also common.)
Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea
- Cornea plana

Abnormalities of Corneal Size or Shape

Peripheral Developmental Abnormalities of the Cornea

- Central Posterior embryotoxon
- Cornea plana
- Epibulbar dermoid

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 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

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

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

(Renal, neurologic and vascular abnormalities are also common)
Developmental Abnormalities of the Cornea

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

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.

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

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

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

Goldenhar syndrome

In addition to liver, 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 also common)
Developmental Abnormalities of the Cornea

- Abnormalities of Corneal Transparency
  - Microcornea
  - Megalocornea
  - Cornea plana

- Abnormalities of Corneal Size or Shape
  - Peripheral Developmental Abnormalities of the Cornea
    - Posterior embryotoxon
  - Central
    - Posterior embryotoxon
    - Axenfeld-Rieger syndrome
    - Aniridia
    - Alagille syndrome
      - Arterohepatic dysplasia
      - Autosomal dominant, but the expressivity varies widely

- 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 it a significant finding?
  - 1) Axenfeld-Rieger syndrome
  - 2) Aniridia
  - 3) Alagille syndrome

- 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 also common)

- 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

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

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

- Goldenhar syndrome is a craniofacial malformation with butterfly vertebrae as a finding.
Abnormalities of Corneal Transparency

Microcornea
Megalocornea
Cornea plana

Abnormalities of Corneal Size or Shape

Peripheral Developmental Abnormalities of the Cornea

Central Posterior embryotoxon
Cornea plana
Epibulbar dermoid

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

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

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 also common)

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?
Oculoauriculo-vertebral (OAV) syndrome
Goldenhar syndrome: Limbal (epibulbar) dermoids; lid coloboma

Goldenhar syndrome: Hemifacial microsomia

Goldenhar: Ear abnormalities
Goldenhar: Limbal (epibulbar) dermoids; lid coloboma

Goldenhar syndrome: Hemifacial microsomia

Developmental Abnormalities of the Cornea

For more on Goldenhar, see slide-set P22
Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

- Microcornea
- Cornea plana
- Megalocornea

Abnormalities of Corneal Transparency

Peripheral
- Cornea plana
- Posterior embryotoxon
- Epibulbar dermoid

Central

Developmental Abnormalities of the Cornea

Are epibulbar dermoids hamartomas, or choristomas?
Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape
- Cornea plana
  - Microcornea
  - Megalocornea

Abnormalities of Central Posterior embryotoxon
- Cornea plana
- Posterior embryotoxon
- Epibulbar dermoid

Peripheral Developmental Abnormalities of the Cornea

Are epibulbar dermoids hamartomas, or choristomas?
Choristomas
Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

Microcornea

Cornea plana

Megalocornea

Developmental Abnormalities of the Cornea

Peripheral

Central

- Cornea plana
- Posterior embryotoxon
- Epibulbar dermoid

Are epibulbar dermoids hamartomas, or choristomas?

Choristomas

What is the difference between a hamartoma and choristoma?
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape
- Microcornea
- Megalocornea
- Cornea plana

Abnormalities of Corneal Transparency
- Peripheral
  - Cornea plana
  - Posterior embryotoxon
  - Epibulbar dermoid
- Central

Are epibulbar dermoids hamartomas, or choristomas?

Choristomas

What is the difference between a hamartoma and choristoma?
A hamartoma is a nest of abnormal cells in their normal location, whereas a choristoma is the opposite—normal cells in an abnormal location
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape
- Microcornea
- Cornea plana
- Megalocornea

Abnormalities of Corneal Transparency
- Peripheral
  - Cornea plana
  - Posterior embryotoxon
  - Epibulbar dermoid
- Central

Are epibulbar dermoids hamartomas, or choristomas?

Choristomas

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Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea

Abnormalities of Corneal Size or Shape

- Cornea plana

Peripheral Developmental Abnormalities of the Cornea

- Cornea plana
- Posterior embryotoxon
- Epibulbar dermoid

Choristomas

Are epibulbar dermoids hamartomas, or choristomas?

What is the difference between a hamartoma and choristoma?

A hamartoma is a nest of abnormal cells in their normal location, whereas a choristoma is the opposite—normal cells in an abnormal location.

So then, from what structure do the 'normal' choristoma cells of an epibulbar dermoid derive?
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Transparency

Microcornea

Megalocornea

Cornea plana

Peripheral

Central

- Cornea plana
- Posterior embryotoxon
- Epibulbar dermoid

Are epibulbar dermoids hamartomas, or choristomas?

Choristomas

What is the difference between a hamartoma and choristoma?

A hamartoma is a nest of abnormal cells in their normal location, whereas a choristoma is the opposite—normal cells in an abnormal location.

So then, from what structure do the ‘normal’ choristoma cells of an epibulbar dermoid derive?

The eyelid (the embryologic eyelid, that is)
Abnormalities of Corneal Transparency

- Cornea plana
- Posterior embryotoxon
  - Epibulbar dermoid

Abnormalities of Corneal Size or Shape

- Megalocornea
- Microcornea
- Cornea plana

Peripheral

Central

Are epibulbar dermoids hamartomas, or choristomas?
Choristomas

With what syndrome are epibulbar dermoids associated?
Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

Peripheral Developmental Abnormalities of the Cornea

Central Developmental Abnormalities of the Cornea

- Cornea plana
- Posterior embryotoxon
- Epibulbar dermoid

Are epibulbar dermoids hamartomas, or choristomas?
Choristomas

With what syndrome are epibulbar dermoids associated?
Goldenhar
Developmental Abnormalities of the Cornea

Goldenhar syndrome: Limbal (epibulbar) dermoids, lid coloboma

Goldenhar syndrome: Hemifacial microsomia

Goldenhar: Ear abnormalities
Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

Microcornea

Megalocornea

Cornea plana

Peripheral

Central

- Cornea plana
- Posterior embryotoxon
- Epibulbar dermoid

Are epibulbar dermoids hamartomas, or choristomas?
Choristomas

With what syndrome are epibulbar dermoids associated?
Goldenhar

Where on the ocular surface are epibulbar dermoids typically located?
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape
- Microcornea
- Megalocornea
- Cornea plana

Abnormalities of Corneal Transparency
- Peripheral
  - Cornea plana
  - Posterior embryotoxon
  - Epibulbar dermoid
- Central

Are epibulbar dermoids hamartomas, or choristomas? Choristomas

With what syndrome are epibulbar dermoids associated? Goldenhar

Where on the ocular surface are epibulbar dermoids typically located? At the limbus
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape
- Microcornea
- Cornea plana
- Megalocornea

Abnormalities of Corneal Transparency
- Peripheral
  - Cornea plana
  - Posterior embryotoxon
  - Epibulbar dermoid
- Central

Are epibulbar dermoids hamartomas, or choristomas?
Choristomas

With what syndrome are epibulbar dermoids associated?
Goldenhar

Where on the ocular surface are epibulbar dermoids typically located?
At the limbus

By what other name are epibulbar dermoids commonly known?
Limbal dermoids
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

- Microcornea
- Cornea plana
- Megalocornea

Abnormalities of Corneal Transparency

Peripheral
- Cornea plana
- Posterior embryotoxon
  - Epibulbar dermoid

Central

Are epibulbar dermoids hamartomas, or choristomas? Choristomas

With what syndrome are epibulbar dermoids associated? Goldenhar

Where on the ocular surface are epibulbar dermoids typically located? At the limbus

By what other name are epibulbar dermoids commonly known? Limbal dermoids
Developmental Abnormalities of the Cornea

Goldenhar syndrome: Epibulbar (aka ‘limbal’) dermoid
Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape
- Microcornea
- Cornea plana
- Megalocornea
- Peripheral
  - Cornea plana
  - Posterior embryotoxon
  - Epibulbar dermoid
- Central
  - ?
  - ?
  - ?
  - ?
  - ?
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

- Microcornea
- Megalocornea
- Cornea plana

Abnormalities of Corneal Transparency

Peripheral
- Cornea plana
- Posterior embryotoxon
- Epibulbar dermoid

Central
- S
- T
- U
- M
- P
- E
- D
Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

Central

Peripheral
- Cornea plana
- Posterior embryotoxon
- Epibulbar dermoid

STUMPED? What that even mean?
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

- Microcornea
- Cornea plana
- Megalocornea

Abnormalities of Corneal Transparency

Peripheral
- Posterior embryotoxon
- Epibulbar dermoid

Central
- Cornea plana

STUMPED? What that even mean?
It’s a reference to the infamous STUMPED mnemonic for remembering the DDx for a cloudy cornea in an infant.
Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

Megalocornea

Microcornea

Cornea plana

Peripheral

- Cornea plana
- Posterior embryotoxon
- Epibulbar dermoid

Central

STUMPED

STUMPED? What that even mean?
It’s a reference to the infamous STUMPED mnemonic for remembering the DDx for a cloudy cornea in an infant.
Developmental Abnormalities of the Cornea

- S
- T
- U
- M
- P
- E
- D

Start here
Developmental Abnormalities of the Cornea

- Sclerocornea
- T
- U
- M
- P
- E
- D

Next
Developmental Abnormalities of the Cornea

- **S**clerocornea
- **T**rauma (endothelial; ie, from forceps)
  
  *(Tears in Descemet’s membrane works too)*

- **U**ndefined
- **M**apped
- **P**refixed
- **E**lected
- **D**ocumented
Developmental Abnormalities of the Cornea

- **S**clerocornea
- **T**rauma (endothelial; ie, from forceps)
- **U**
Developmental Abnormalities of the Cornea

- Sclerocornea
- Trauma (endothelial; ie, from forceps)
- Ulcer
- M
- P
- E
- D

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Developmental Abnormalities of the Cornea

- Sclerocornea
- Trauma (endothelial; ie, from forceps)
- Ulcer
- Metabolic disorders
- P
- E
- D
Developmental Abnormalities of the Cornea

- Sclerocornea
- Trauma (endothelial; ie, from forceps)
- Ulcer
- Metabolic disorders
- Peters anomaly
- E
- D
Developmental Abnormalities of the Cornea

- Sclerocornea
- Trauma (endothelial; ie, from forceps)
- Ulcer
- Metabolic disorders
- Peters anomaly
- Endothelial dystrophy (CHED)

(CHED = congenital hereditary endothelial dystrophy)

/Edit works too
Developmental Abnormalities of the Cornea

- Sclerocornea
- Trauma (endothelial; ie, from forceps)
- Ulcer
- Metabolic disorders
- Peters anomaly
- Endothelial dystrophy (CHED)
Developmental Abnormalities of the Cornea

- Sclerocornea
- Trauma (endothelial; ie, from forceps)
- Ulcer
- Metabolic disorders
- Peters anomaly
- Endothelial dystrophy (CHED)
- Dermoid of the cornea
Developmental Abnormalities of the Cornea

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For more on the STUMPED mnemonic, see slide-set K9