Developmental Abnormalities of the Cornea

Basic distinction used by the BCSC Peds book

Abnormalities of Corneal Size or Shape

Abnormalities of Corneal Transparency
Developmental Abnormalities of the Cornea

Abnormalities of Corneal *Size* or *Shape*

Abnormalities of Corneal *Transparency*

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Abnormalities of Corneal *Size* or *Shape*

Abnormalities of Corneal *Transparency*
Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

- Microcornea
- Cornea plana

Abnormalities of Corneal Transparency

Developmental Abnormalities of the Cornea
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

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

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

Megalocornea?

At age >2 years
At birth

<table>
<thead>
<tr>
<th>Corneal diameter (mm)</th>
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<tbody>
<tr>
<td>7 8 9 10 11 12 13 14 15</td>
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</table>
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

- Microcornea
- Cornea plana
- Megalocornea

What is the definition of megalocornea, ie, how ‘megalο’ 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
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Abnormalities of Corneal Transparency

- ?
- ?
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

- Microcornea
- Megalocornea
  - Primary
  - Secondary
  - Cornea plana

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Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

Is primary megalocornea a congenital, or acquired condition?

Primary

Megalocornea

Secondary

Is it 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?

- 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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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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Primary Developmental Abnormalities of the Cornea

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Megalocornea

Primary

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Abnormalities of Corneal Size or Shape

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

With what other ocular abnormalities is it associated?  
There are many; they include:
--Lens abnormalities: Cataract, ectopia lentis
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What is megalocornea?
Large cornea

Primary megalocornea usually presents in both eyes.

Secondary megalocornea may occur after trauma or surgery.

Megalocornea can be associated with:
--Cataracts
--Iris abnormalities

With what other systemic conditions is it associated?  
There are many; they include:
--Down syndrome
--Marfan syndrome
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Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

Primary Megalocornea

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What is ectopia lentis?

Displacement of the lens from its normal anatomic position

Secondary Megalocornea

Primary

Secondary
Abnormalities of Corneal Transparency

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Megalocornea

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 system
The musculoskeletal system

Other Abnormalities

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- Microcornea
- Meilocornea
- Cornea plana

Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

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What three structures/systems manifest abnormalities in Marfan's?
--The eye
--The cardiovascular
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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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What are some other ocular abnormalities associated with megalocornea?
--Lens abnormalities: Cataract, ectopia lentis
--Iris abnormalities: Miosis, translucency

What systemic conditions may be associated with megalocornea?
--Down syndrome
--Marfan syndrome
--Alport syndrome

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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
- 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

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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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What condition is associated with displacement inferonasally?
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Marfan

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Marfan

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**Primary Developmental Abnormalities of the Cornea**

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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

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Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

- Megalocornea
  - Primary
  - Secondary

- Microcornea

- Cornea plana

Abnormalities of Corneal Transparency

Is secondary megalocornea a congenital, or acquired condition?
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Abnormalities of Corneal Size or Shape

- Megalocornea
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Developmental Abnormalities of the Cornea

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Developmental Abnormalities of the Cornea

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Abnormalities of Corneal Transparency

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What is the cause?
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Elevated IOP

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

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

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Megalocornea
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 it’s *buphthalmos* vs secondary megalocornea?

**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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*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 it’s *buphthalmos* vs secondary megalocornea?*

**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

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 it’s 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.

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

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What is the cause?
Elevated IOP

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

- **Microcornea**
- **Megalocornea**
- **Cornea plana**

Developmental Abnormalities of the Cornea

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**What is the cause?**

Elevated IOP

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**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.
Abnormalities of Corneal Transparency

- **Microcornea**
- **Megalocornea**
- **Cornea plana**

Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

**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

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Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea
- Cornea plana

Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

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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

Abnormalities of Corneal Size or Shape

Cornea plana

Microcornea

Megalocornea

**Developmental Abnormalities of the Cornea**

**Abnormalities of Corneal Size or Shape**

**Cornea plana**

**Microcornea**

**Megalocornea**

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

- **Microcornea**: At age >2 years or At birth, Corneal diameter < 9 mm at birth, or < 10 mm at age 2 years or older

**Microcornea?**

- Megalocornea

- Corneal diameter (mm): 7, 8, 9, 10, 11, 12, 13, 14, 15
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.
Developmental Abnormalities of the Cornea

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

- **Microcornea**

  Does microcornea present unilaterally, or bilaterally?

- **Megalocornea**

Abnormalities of Corneal **Transparency**

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

With what **systemic** conditions is microcornea associated?

- Ehlers-Danlos syndrome
- Myotonic dystrophy
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

Microcornea

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

Megalocornea

Abnormalities of Corneal Transparency

Persistent fetal vasculature (PFV, aka PHPV)

Congenital cataracts

Ehlers-Danlos syndrome

Myotonic dystrophy
Abnormalities of Corneal Size or Shape

Microcornea

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

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

Abnormalities of Corneal Transparency

Developmental Abnormalities of the Cornea

Megalocornea
Abnormalities of Corneal Size or Shape

- Microcornea
  - 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
Abnormalities of Corneal Transparency

Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

Microcornea

Megalocornea

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?
Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

Megalocornea

Microcornea

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? No—by definition, the thickness is normal
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--Persistent fetal vasculature (PFV, aka PHPV)
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With what systemic conditions is microcornea associated?
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Abnormalities of Corneal Transparency

Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

Microcornea

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?

Megalocornea

With what ocular conditions is microcornea associated?
--Persistent fetal vasculature (PFV, aka PHPV)
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Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

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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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A flat cornea implies hyperopia. Is microcornea in fact associated with hyperopia?
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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Does microcornea present unilaterally, or bilaterally? It can be either

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

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? Yes

Abnormalities of Corneal Transparency
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

Microcornea

Megalocornea

Abnormalities of Corneal Transparency

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Abnormalities of Corneal Size or Shape

Microcornea

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Megalocornea

Abnormalities of Corneal Transparency

Developmental Abnormalities of the Cornea
Abnormalities of Corneal Size or Shape

Microcornea

Megalocornea

Developmental Abnormalities of the Cornea

Abnormalities of Corneal Transparency

**Does microcornea present unilaterally, or bilaterally?**
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-- **Too thick?** No—by definition, the thickness is normal
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Abnormalities of Corneal Transparency

Developmental Abnormalities of the Cornea

Abnormalities of 
Corneal Size or Shape

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Microcornea

Megalocornea

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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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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

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
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- **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

- **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-plana 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

Abnormalities of Corneal Transparency

Megalocornea

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Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

Microcornea

Megalocornea

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- Congenital cataracts
- **With what systemic conditions is microcornea associated?**
  - Ehlers-Danlos syndrome
  - Myotonic dystrophy
Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

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

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*With what ocular conditions is microcornea associated?*

-- Persistent fetal vasculature (PFV, aka PHPV)

-- Congenital cataracts

*With what systemic conditions is microcornea associated?*

--

--
Developmental Abnormalities of the Cornea

Abnormalities of Corneal \textit{Size} or \textit{Shape}

Microcornea

Megalocornea

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

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With what ocular conditions is microcornea associated?
--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

- Microcornea
- Cornea plana
- Megalocornea

Abnormalities of Corneal Transparency

Hold on--you skipped cornea plana! What’s up with that?
Hold on--you skipped cornea plana! What’s up with that? Patience, Grasshopper--all will be made clear soon. Now, where were we?
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

? ?
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
Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

- Cornea plana
- Microcornea
- Megalocornea

Abnormalities of Corneal Transparency

- Peripheral
  - ?
  - ?
  - ?

- Central
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

Abnormalities of Corneal Size or Shape

- Microcornea
- Cornea plana

Abnormalities of Corneal Transparency

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

Microcornea

Megalocornea

Cornea plana

Morphologic Abnormalities

Peripheral Developmental Abnormalities of the Cornea

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What is the average central corneal power in adults?

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 is 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

Cornea plana

Abnormalities of Corneal Size or Shape

Peripheral Developmental Abnormalities of the Cornea

Central 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 of the Cornea

- Central Posterior embryotoxon
- Cornea plana
- Epibulbar dermoid
- Dermolipomas

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Abnormalities of Corneal Transparency

Microcornea
Megalocornea
Cornea plana

Abnormalities of Corneal Size or Shape

Peripheral Developmental Abnormalities of the Cornea

Central Developmental Abnormalities of the Cornea

Cornea plana

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About 43D

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Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

- Microcornea
- Megacornea
- Cornea plana

Abnormalities of Corneal Transparency

- Peripheral Developmental Abnormalities
- Central

Corneal plana

- Flat Ks
- Poorly developed (“indistinct”) limbus
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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

**Cornea plana**

- Flat Ks
- Poorly developed (“indistinct”) limbus
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**What is the average central corneal power in adults?**
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

Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

Cornea plana

Peripheral Developmental Abnormalities of the Cornea

Central Developmental Abnormalities of the Cornea

Microcornea

Megalocornea

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 in adults?
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But if the cornea is the same curvature as the adjacent sclera, how is that not simply sclerocornea? How do you differentiate between these conditions?

The difference is one of corneal transparency:

In sclerocornea, the cornea is opaque
In cornea plana, the cornea is clear
Abnormalities of Corneal Transparency

Microcornea
- Megalocornea

Abnormalities of Corneal Size or Shape
- Cornea plana

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- a word
- its antonym

**the same curvature as the adjacent sclera**
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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
2) When it is associated with aniridia
3) When it is associated with Alagille syndrome
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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.'
Developmental Abnormalities of the Cornea

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Hints forthcoming…

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Peripheral Developmental Abnormalities of the Cornea

Cornea plana

Epibulbar dermoid

Posterior embryotoxon
Developmental Abnormalities of the Cornea

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Posterior embryotoxon

 Syndromes: Axenfeld-Rieger, Alagille, Aniridia

Epibulbar dermoid

Cornea plana
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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?**
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**What nonocular abnormalities may be present?**
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2) Characteristic facies
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Where does ARS rank as a cause of iris hypoplasia?
It is the most common
Developmental Abnormalities of the Cornea

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Axenfeld-Rieger syndrome

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What is the lifetime risk of developing glaucoma?
50%
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Axenfeld-Rieger syndrome

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

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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Posterior embryotoxon with attached iris strands + iris hypoplasia + angle abnormalities.

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

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

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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
2) When it is associated with aniridia
3) When it is associated with Alagille syndrome

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

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

Is nystagmus commonly associated with aniridia?
Yes

With what developmental complex is aniridia associated?
The WAGR complex

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

Posterior embryotoxon
Epibulbar dermoid
Developmental Abnormalities of the Cornea

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

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- Wilms tumor
- Aniridia
- Genitourinary abnormalities
- Retardation

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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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Abnormalities of corneal transparency

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Peripheral Developmental Abnormalities of the Cornea

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Arterohepatic dysplasia

How is it inherited?

Autosomal dominant, but the expressivity varies widely

What is the classic presentation?

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'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
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Abnormalities of Corneal Transparency

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Posteriors embryotoxon

Axenfeld-Rieger syndrome

Epibulbar dermoid
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Another syndrome of ophthalmic concern includes butterfly vertebrae as a finding. What is it?
Goldenhar syndrome
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In two words, what sort of condition is Goldenhar?
A craniofacial malformation

What is the noneponymous name for Goldenhar syndrome?
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  - Cornea plana
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- Central

Are epibulbar dermoids hamartomas, or choristomas?
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- Choristomas

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A hamartoma is a nest of abnormal cells in their normal location, whereas a choristoma is the opposite--normal cells in an abnormal location.
Are epibulbar dermoids hamartomas, or choristomas?

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**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

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**Are epibulbar dermoids hamartomas, or choristomas?**

Choristomas

*What is the difference between a hamartoma and choristoma?*

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*So then, from what structure do the ‘normal’ choristoma cells of an epibulbar dermoid derive?*
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The eyelid (the embryologic eyelid, that is)
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With what syndrome are epibulbar dermoids associated?
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By what other name are epibulbar dermoids commonly known?
- Limbal dermoids
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STUMPED? What that even mean?
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STUMPED? What that even mean?

It’s a reference to the infamous STUMPED mnemonic intended to aid in remembering the DDx for a cloudy cornea in an infant.

(For details, see the slide-set of the same name.)