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*
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

Abnormalities of Corneal Transparency
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

- Microcornea
- Cornea plana
- Megalocornea

Abnormalities of Corneal Transparency
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

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

Corneal diameter (mm)

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

Abnormalities of Corneal Size or Shape

- Microcornea
- Cornea plana
- Megalocornea

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
Developmental Abnormalities of the Cornea

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

- Megalocornea
  - Primary
  - Secondary
- Microcornea
- Cornea plana

Abnormalities of Corneal **Transparency**
Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea
- Cornea plana

Abnormalities of Corneal Size or Shape

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?

- 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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Primary Developmental Abnormalities of the Cornea

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How does 'iris translucency' manifest during slit-lamp exam?

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  - As iris transillumination during retroillumination
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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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cataract
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Primary

Secondary

Megalocornea

Megalocornea
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

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Primary Me
galocornea

Secondary Cataract

There are many; they include:

Primary Abnormalities of Corneal Transparency

Microcornea

Megalocornea

Cornea plana
Abnormalities of Corneal Transparency

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Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

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

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Another, potentially blinding, ocular abnormality is a late-onset association with primary megalocornea—what is it?

Glaucoma
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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
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Transparency

- Microcornea
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Abnormalities of Corneal Size or Shape

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

Is megalocornea a common ocular manifestation?
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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)

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

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 is Marfan syndrome associated with?
- Down syndrome
- Marfan syndrome
- Alport syndrome
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

- Microcornea
- Megalocornea
- Cornea plana

Abnormalities of Corneal Abnormalities

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With what other ocular abnormalities is it associated?
- Lens abnormalities: Cataract, ectopia lentis
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It can be associated with systemic conditions. Which ones?
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Is megalocornea a common ocular manifestation?
No

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Developmental Abnormalities of the Cornea

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### Developmental Abnormalities of the Cornea

#### Abnormalities of Corneal Size or Shape

**Developmental Abnormalities of the Cornea**

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**Primary Megalocornea**

- **What is the name of the protein that is abnormal in Marfan's?** Fibrillin
- **What three structures/systems manifest abnormalities in Marfan's?**
  1. The eye
  2. The cardiovascular
  3. The musculoskeletal

**Secondary Megalocornea**

- **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 proportion of Marfan pts manifest ocular abnormalities?** At least 80%
- **Is megalocornea a common ocular manifestation?** No
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Abnormalities of Corneal Transparency

- Microcornea
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Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

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- No

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

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

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- Microcornea
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Developmental Abnormalities of the Cornea

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Developmental Abnormalities of the Cornea

- Primary
- Secondary

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What three entities are affected in Marfan’s?
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Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

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

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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
Developmental Abnormalities of the Cornea

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--Wheatann syndrome
Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea
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Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

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Developmental Abnormalities of the Cornea

Megalocornea

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

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Acquired—always, and by definition
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

- Megalocornea
  - Primary
  - Secondary

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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 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.

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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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.
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.

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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Abnormalities of Corneal Transparency

Microcornea

Megalocornea

Cornea plana

Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

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Acquired—always, and by definition

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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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Elevated IOP
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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

Primary vs 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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- Microcornea
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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
- Cornea plana
- Megalocornea

Abnormalities of Corneal Transparency

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
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 Size or Shape

Does microcornea present unilaterally, or bilaterally?

Developmental Abnormalities of the Cornea

Abnormalities of Corneal Transparency

Microcornea

Megalocornea

Does microcornea present unilaterally, or bilaterally?

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

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

Abnormalities of Corneal Size or Shape

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

Microcornea

Megalocornea

Abnormalities of Corneal Transparency

Does microcornea present unilaterally, or bilaterally?

It can be either

--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
Developmental Abnormalities of the Cornea

Microcornea

Unilateral

Bilateral
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?

Abnormalities of Corneal *Transparency*

Megalocornea

--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

Megalocornea

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Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

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Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

Megalocornea

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Abnormalities of Corneal Transparency

Developmental Abnormalities of the Cornea
Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

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Microcornea

Megalocornea

Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

Ehlers-Danlos syndrome
Myotonic dystrophy
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?
Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

Microcornea

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

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

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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

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Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

Microcornea

Megalocornea

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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

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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.

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Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

Microcornea

Megalocornea

Abnormalities of Corneal Transparency

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Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

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With what ocular conditions is microcornea associated?

--
--
Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

Megalocornea

Microcornea

Abnormalities of Corneal Transparency

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

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

Abnormalities of Corneal Size or Shape

Microcornea

Megalocornea

Abnormalities of Corneal Transparency

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With what systemic conditions is microcornea associated?
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Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

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Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape

- Microcornea
- Cornea plana
- Megalocornea

Abnormalities of Corneal Transparency

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

Basic distinction used by the BCSC Peds book
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape
- Microcornea
- Megalocornea

Abnormalities of Corneal Transparency

Cornea plana

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
- Megalocornea
- Cornea plana

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
- Cornea plana
- Megalocornea

Abnormalities of Corneal Transparency
- Peripheral
- Central

Basic distinction used by the BCSC Peds book

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

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

Abnormalities of Corneal Transparency
  - Peripheral
    - ?
    - ?
    - ?
  - Central
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
Cornea plana belongs in both groups, is why it was saved until now…
Abnormalities of Corneal Transparency

Microcornea

Megalocornea

Cornea plana

Peripheral Developmental Abnormalities of the Cornea

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**

**Abnormalities of Corneal Size or Shape**

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

**Central Developmental Abnormalities of the Cornea**

- **Cornea plana**
- Posterior embryotoxon

---

*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

#4 #5
Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

Microcornea

Cornea plana

Peripheral

Posterior embryotoxon

Central

Abnormalities of Corneal Transparency

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 hyperopia with associated accommodative esotropia
Developmental Abnormalities of the Cornea

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

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--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'

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

Abnormalities of Corneal Size or Shape

Microcornea

Megalocornea

Cornea plana

Peripheral Developmental Abnormalities of the Cornea

Posterior embryotoxon

Cornea plana

Dermolipomas

Epibulbar dermoid

Central Developmental Abnormalities of the Cornea

Central

Cornea plana

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

Flat Ks

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

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’

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
- Meig's cornea plana
- Abnormalities of corneal size or shape

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Cornea plana: Keratometry (I know, it’s really blurry)
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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 key difference is one of corneal transparency
In sclerocornea, the cornea is opaque
In cornea plana, the cornea is clear (relatively speaking)
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a word
its antonym

the same curvature as the adjacent sclera
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- Cornea plana
- 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

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
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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?

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Developmental Abnormalities

Normal angle anatomy: Identify the structures
Developmental Abnormalities

Normal angle anatomy: Identify the structures

Schwalbe’s Line
TM (non pigmented)
TM (pigmented)
SS
CB
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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.

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
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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?

Axenfeld-Rieger syndrome

- posterior embryotoxon
- Axenfeld-Rieger syndrome

- angle abnormalities

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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
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

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2) When it is associated with aniridia
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What is corectopia?
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What other iris abnormalities may be present?
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- Microcornea
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Abnormalities of Corneal Size or Shape

Peripheral Developmental Abnormalities of the Cornea
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The presence of posterior pigmented iris epithelium on the anterior surface of the iris.

Axenfeld-Rieger syndrome
Posterior embryotoxon

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Dermolipomas

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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.
Developmental Abnormalities of the Cornea

Axenfeld-Reiger with microcornea (8.5 mm)
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(A) Facial photograph showing maxillary hypoplasia, thin upper lip, and broad nasal bridge
(B) Left eye with corectopia
(C) Right eye with posterior embryotoxon
(D) Dental anomalies, including maxillary hypodontia
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Axenfeld-Reiger syndrome
Developmental Abnormalities of the Cornea

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

Axenfeld-Reiger syndrome
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No, only those in which the genetic mutation is sporadic

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Aniridia. Note the presence of an iris stub/root
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**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
Developmental Abnormalities of the Cornea

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

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With what developmental complex is aniridia associated?
The WAGR complex consists of:

- W
- Aniridia
- A
- G
- R

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 posterior embryotoxon?
An anteriorly displaced and thickened Schwalbe's line/ring

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The WAGR complex

WAGR complex consists of:

- W
- Aniridia
- A
- G
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Posterior embryotoxon

Epibulbar dermoid
Developmental Abnormalities of the Cornea

What is a posterior embryotoxon?
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It is almost always bilateral

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With what developmental complex is aniridia associated?
The WAGR complex consists of:
- Wilms tumor
- Aniridia
- Genitourinary abnormalities
- Retardation

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Because a rudimentary iris root is always present

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The WAGR complex

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

Posterior embryotoxon

Epibulbar dermoid
Developmental Abnormalities of the Cornea

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

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An anteriorly displaced and thickened Schwalbe’s line/ring

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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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The WAGR complex

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No, only those in which the genetic mutation is sporadic
Developmental Abnormalities of the Cornea

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The WAGR complex

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

Aniridia

Axenfeld-Rieger syndrome

Alagille syndrome

Epibulbar dermoid

Posterior embryotoxon

Peripheral Developmental Abnormalities of the Cornea

Cornea plana

Central Posterior embryotoxon

Microcornea

Megalocornea
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**

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

Abnormalities of Corneal Transparency

Microcornea

Megalocornea

Cornea plana

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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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--The heart: Septal defects, PDA, and tetralogy of Fallot are common
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Developmental Abnormalities of the Cornea

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Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea

Abnormalities of Corneal Size or Shape

Peripheral Developmental Abnormalities of the Cornea

- Central Posterior embryotoxon
- Cornea plana
- Epibulbar dermoid

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Abnormalities of Corneal Size or Shape

Peripheral Developmental Abnormalities of the Cornea

Central Posterior embryotoxon

Epibulbar dermoid

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Alagille syndrome

Posterior embryotoxon

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Abnormalities of Corneal Size or Shape

Peripheral Developmental Abnormalities of the Cornea

Central Posterior embryotoxon

Cornea plana

Epibulbar dermoid

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Posterior embryotoxon
Developmental Abnormalities of the Cornea

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

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What is it a significant finding? In three situations:
1) When it is part of the Axenfeld-Rieger syndrome
2) When it is associated with aniridia
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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.

What is the characteristic face shape of Alagille syndrome? ‘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?
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- 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.)

Posterior embryotoxon
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

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?

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**
- **Cornea plana**
- **Epibulbar dermoid**

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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
Developmental Abnormalities of the Cornea

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

Abnormalities of Corneal Size or Shape

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

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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?

Posteriors embryotoxon

Axenfeld-Rieger syndrome

Alagille syndrome

Epibulbar dermoid
Developmental Abnormalities of the Cornea

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An anteriorly displaced and thickened Schwalbe's line/-ring

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--The skeleton: The classic finding is ‘butterfly vertebrae’
(Renal, neurologic and vascular abnormalities are also common)
Developmental Abnormalities of the Cornea

Alagille syndrome: Facies
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

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Developmental Abnormalities of the Cornea

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

What is the non-epponymous name of Alagille syndrome?
Arteriohepatic dysplasia

How is it inherited?
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Abnormalities of Cornea

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--The heart:
--The skeleton:
Abnormalities of Corneal Transparency

- Microcornea
- Megalocornea
- Cornea plana

Abnormalities of Corneal Size or Shape

Developmental Abnormalities of the Cornea

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

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

Alagille syndrome: Butterfly vertebrae
Abnormalities of the Cornea

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

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Another syndrome of opthalmic 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
Developmental Abnormalities of the Cornea

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

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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 Posterior embryotoxon
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Abnormalities of Corneal Transparency

- Microcornea
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Central Posterior embryotoxon

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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: 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)

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

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

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Developmental Abnormalities of the Cornea

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- Megalocornea
- Cornea plana

Abnormalities of Corneal Size or Shape

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

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

What is the noneponymous name for Goldenhar syndrome?
Oculoauriculovertebral (OAV) syndrome
Goldenhar syndrome: Hemifacial microsomia

Goldenhar: Limbal (epibulbar) dermoids; lid coloboma

Goldenhar: Ear abnormalities
Developmental Abnormalities of the Cornea

Goldenhar: Limbal (epibulbar) dermoids; lid coloboma

For more on Goldenhar, see slide-set P22

Goldenhar syndrome: Hemifacial microsomia

Goldenhar: Ear abnormalities
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?
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
Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

- Cornea plana
- Microcornea
- Megalocornea

Peripheral Developmental Abnormalities of the Cornea

- Posterior embryotoxon
- Epibulbar dermoid

Abnormalities of Corneal Transparency

- Cornea plana

Are epibulbar dermoids hamartomas, or choristomas?

Choristomas

What is the difference between a hamartoma and choristoma?
Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape
- Microcornea
- Megalocornea

Cornea plana

Peripheral Developmental Abnormalities of the Cornea
- Posterior embryotoxon
- Epibulbar dermoid

Central Developmental Abnormalities of the Cornea
- Cornea plana

Are epibulbar dermoids hamartomas, or choristomas?

What is the difference between a hamartoma and choristoma?
- One of them is a nest of abnormal cells in their normal location, whereas the other is the opposite--normal cells in an abnormal location.
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

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

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?
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)
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?
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
Developmental Abnormalities of the Cornea

Goldenhar syndrome: Limbal (epibulbar) dermoids; lid coloboma

Goldenhar syndrome: Hemifacial microsomia

Goldenhar: Ear abnormalities
Developmental Abnormalities of the Cornea

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- 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
Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

Microcornea

Megalocornea

Cornea plana

Peripheral Developmental Abnormalities of the Cornea

Central Developmental Abnormalities of the Cornea

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?
At the limbus
Developmental Abnormalities of the Cornea

Abnormalities of Corneal Size or Shape
- Microcornea
- Megalocornea

Abnormalities of Corneal Transparency

Central
- Posterior embryotoxon

Peripheral
- 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?
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
- Megalocornea
- Cornea plana

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

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 Size or Shape

- Microcornea
- Megalocornea
- Cornea plana

Abnormalities of Corneal Transparency

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

Abnormalities of Corneal Size or Shape

- Microcornea
- Cornea plana
- Megalocornea

Peripheral Developmental Abnormalities of the Cornea
- Posterior embryotoxon
- Epibulbar dermoid

Central
- Cornea plana

Abnormalities of Corneal Transparency
Developmental Abnormalities of the Cornea

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

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

STUMPED? What that even mean?
Abnormalities of Corneal Transparency

Abnormalities of Corneal Size or Shape

Megalocornea

Microcornea

Cornea plana

Peripheral

Central

- Cornea plana
- Posterior embryotoxon
- Epibulbar dermoid

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

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

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

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

Start here
Developmental Abnormalities of the Cornea

- Sclerocornea
- T
- U
- M
- P
- E
- D
Developmental Abnormalities of the Cornea

- Sclerocornea
- Trauma (endothelial; ie, from forceps)  
  
  (Tears in Descemet’s membrane works too)
Developmental Abnormalities of the Cornea

- Sclerocornea
- Trauma (endothelial; ie, from forceps)
- U
- M
- P
- E
- D
Developmental Abnormalities of the Cornea

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

PED
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)
- D (Edema works too)
Developmental Abnormalities of the Cornea

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

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

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

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