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

With regard to lens ‘displacement’—what do the following terms mean?
--Sublux(at)ed:
--Lux(at)ed:
What is ectopia lentis?
Displacement of the lens from its normal anatomic position

With regard to lens ‘displacement’—what do the following terms mean?
--Sublux(ate)ed: The lens is partially displaced, but remains in the ‘general area’
--Lux(ate)ed:
Ectopia Lentis

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

With regard to lens 'displacement'—what do the following terms mean?

--**Sublux(at)ed**: The lens is partially displaced, but remains in the 'general area'
--**Lux(at)ed**: The lens is completely displaced from its normal position

What iris finding is classic for a subluxed lens?

Iridodonesis
A 'tremulous' iris
What is ectopia lentis?
Displacement of the lens from its normal anatomic position

With regard to lens 'displacement'—what do the following terms mean?
--Sublux(at)ed: The lens is partially displaced, but remains in the 'general area'
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What iris finding is classic for a subluxed lens?
Iridodonesis
What is ectopia lentis?
Displacement of the lens from its normal anatomic position

With regard to lens 'displacement'—what do the following terms mean?

--**Sublux(at)ed**: The lens is partially displaced, but remains in the 'general area'

--**Lux(at)ed**: The lens is displaced beyond the confines of the 'general area'

*What iris finding is classic for a subluxed lens?*

Iridodonesis

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

With regard to lens ‘displacement’—what do the following terms mean?

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With regard to lens ‘displacement’—what do the following terms mean?
--Sublux(at)ed: The lens is partially displaced, but remains in the ‘general area’
--Lux(at)ed: The lens is dislocated--completely removed from the pupillary aperture. All zonular attachments have been disrupted.
Ectopia Lentis

Aphakic

Lens resting on the retina

b-scan: lens on ONH
Ectopia Lentis

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

With regard to lens ‘displacement’—what do the following terms mean?
--Sublux(at)ed: The lens is partially displaced, but remains in the ‘general area’
--Lux(at)ed: The lens is dislocated--completely removed from the pupillary aperture. All zonular attachments have been disrupted.

The *Peds* book also uses the term *ectopic* here.
The BCSC series divides ectopia lentis into three categories:
The BCSC series divides ectopia lentis into three categories:
What are the four **congenital** causes covered in the BCSC?
What are the four congenital causes covered in the BCSC?
What are the five developmental causes covered in the BCSC?
Ectopia Lentis

**Congenital**
- Marfan
- Aniridia
- Ectopia lentis et pupillae
- Simple ectopia lentis (congenital type)

**Developmental**
- Marfan
- Homocystinuria
- Microspherophakia
- Buphthalmos 2° to congenital glaucoma
- Simple ectopia lentis (late-onset type)

**Acquired**

---

What are the five developmental causes covered in the BCSC?
What are the two **acquired** causes covered in the BCSC?
**What are the two acquired causes covered in the BCSC?**

**Acquired**
- Trauma
- (Pseudo)exfoliation syndrome

**Developmental**
- Marfan
- Homocystinuria
- Microspherophakia
- Buphthalmos 2° to congenital glaucoma
- Simple ectopia lentis (late-onset type)

**Congenital**
- Marfan
- Aniridia
- Ectopia lentis et pupillae
- Simple ectopia lentis (congenital type)
Which is the most common heritable cause of ectopia lentis?
Which is the most common heritable cause of ectopia lentis? Marfan
Ectopia Lentis

Congenital

- Marfan

Developmental

- Marfan

Acquired

- Trauma

Simple ectopia lentis (congenital type)

Buphthalmos 2º to congenital glaucoma

Simple ectopia lentis (late-onset type)

By the way—why does Marfan appear under both Congenital and Developmental headings?
By the way—why does Marfan appear under both Congenital and Developmental headings? Because although ectopia lentis in Marfan is clearly a developmental issue, it is believed to be present at birth—ie, congenital—in the majority of cases.
Ectopia Lentis

- Congenital
  - Marfan
- Developmental
  - Marfan
- Acquired
  - Trauma
  - (Pseudo)exfoliation syndrome

What protein is abnormal in Marfan’s?

(late-onset type)
What protein is abnormal in Marfan’s? Fibrillin

- Congenital
  - Marfan

- Developmental
  - Marfan

- Acquired
  - Trauma
  - (Pseudo)exfoliation syndrome

Ectopia Lentis

Simple ectopia lentis (congenital type)

Ectopia lentis et pupillae

Trauma

Aniridia

Buphthalmos 2o to congenital glaucoma

Microspherophakia (Pseudo)exfoliation syndrome

What protein is abnormal in Marfan’s? Fibrillin

(late-onset type)
Ectopia Lentis

Congenital
- Marfan

Developmental
- Marfan

Acquired
- Trauma
- (Pseudo)exfoliation syndrome

What protein is abnormal in Marfan's?
Fibrillin

What is the inheritance pattern?
AD, although family history is negative in 15% of cases
Q/A

Ectopia Lentis

Congenital

Marfan

Developmental

Marfan

Acquired

Trauma

(Pseudo)exfoliation syndrome

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Fibrillin

What is the inheritance pattern?
AD, although family history is negative in % of cases

(late-onset type)
Ectopia Lentis

- Congenital
  - Marfan
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  - Marfan
- Acquired
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(late-onset type)
What protein is abnormal in Marfan’s?
Fibrillin

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

(late-onset type)
Ectopia Lentis

What protein is abnormal in Marfan’s?
Fibrillin

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

(late-onset type)
What protein is abnormal in Marfan’s? 
Fibrillin

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

What cardiovascular abnormalities are common? 
-- 
-- 
--
Ectopia Lentis

**Congenital**
- Marfan

**Developmental**
- Marfan

**Acquired**
- Trauma
- (Pseudo)exfoliation syndrome

What protein is abnormal in Marfan’s?
Fibrillin

What three structures/systems manifest abnormalities in Marfans?
- The eye (duh)
- **The cardiovascular**
- The musculoskeletal

What cardiovascular abnormalities are common?
- Dilatation of the aortic root and descending aorta
- Aortic aneurysms
- Mitral valve prolapse

Indeed they are—they are responsible for the significantly shortened lifespan of Marfan pts
What protein is abnormal in Marfan’s?  
Fibrillin

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

What cardiovascular abnormalities are common?  
--Dilatation of the aortic root and descending aorta  
--Aortic aneurysms  
--Mitral valve prolapse

Are these abnormalities clinically significant?
**Ectopia Lentis**

- **Congenital**
  - Marfan

- **Developmental**
  - Marfan

- **Acquired**
  - Trauma
  - (Pseudo)exfoliation syndrome

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**What protein is abnormal in Marfan’s?**
- Fibrillin

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Indeed they are—they are responsible for the significantly shortened lifespan of Marfan pts

How significant is the lifespan shortening?

Marfan

Congenital

Developmental

Acquired

Trauma

(Pseudo)exfoliation syndrome
**Q/A**

**Ectopia Lentis**

**Congenital**
- Marfan

**Developmental**
- Marfan

**Acquired**
- Trauma
- (Pseudo)exfoliation syndrome

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**What protein is abnormal in Marfan’s?**
Fibrillin

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**Are these abnormalities clinically significant?**
Indeed they are—they are responsible for the **significantly shortened lifespan of Marfan pts**

---

**How significant is the lifespan shortening?**
Quite. The life-expectancy of Marfan pts is about **half** that of the so-called normal population.
Ectopia Lentis

- **Congenital**
  - Marfan

- **Developmental**
  - Marfan

- **Acquired**
  - Trauma
  - (Pseudo)exfoliation syndrome

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**What protein is abnormal in Marfan’s?**
- Fibrillin

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- The eye (duh)
- **The cardiovascular**
- The musculoskeletal

**What cardiovascular abnormalities are common?**
- Dilatation of the aortic root and descending aorta
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**Are these abnormalities clinically significant?**
- Indeed they are—they are responsible for the **significantly shortened lifespan of Marfan pts**

**How significant is the lifespan shortening?**
- Quite. The life-expectancy of Marfan pts is about half that of the so-called normal population.
Marfan syndrome: Aortic dissection

Ectopia Lentis
Ectopia Lentis

Congenital
- Marfan

Developmental
- Marfan

Acquired
- Trauma
- (Pseudo)exfoliation syndrome

What protein is abnormal in Marfan’s?
- Fibrillin

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

What musculoskeletal abnormalities are common?
--?
--?
--?
Ectopia Lentis

Congenital
- Marfan

Developmental
- Marfan

Acquired
- Trauma
- (Pseudo)exfoliation syndrome

What protein is abnormal in Marfan’s?
Fibrillin

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

What musculoskeletal abnormalities are common?
-- Arachnodactyly
-- Hypermobile joints
-- Sternum deformities (eg, two-us word-ums)
Ectopia Lentis

**Congenital**
- Marfan

**Developmental**
- Marfan

**Acquired**
- Trauma
- (Pseudo)exfoliation syndrome

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**What protein is abnormal in Marfan’s?**
- Fibrillin

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

---

**What musculoskeletal abnormalities are common?**
- Arachnodactyly
- Hypermobile joints
- Sternum deformities (eg, pectus excavatum)
Arachnodactyly

Ectopia Lentis

Hypermobile joints

Pectus excavatum

Marfan syndrome
Ectopia Lentis

**Congenital**
- Marfan

**Developmental**
- Marfan

**Acquired**
- Trauma
- (Pseudo)exfoliation syndrome

What protein is abnormal in Marfan’s?
Fibrillin

What three systems manifest abnormalities in Marfans?
- The eye (duh)
- The cardiovascular
- The musculoskeletal

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

Congenital
- Marfan

Developmental
- Marfan

Acquired
- Trauma
- (Pseudo)exfoliation syndrome

What protein is abnormal in Marfan’s?
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What three systems manifest abnormalities in Marfans?
- The eye (duh)
- The cardiovascular
- The musculoskeletal

What proportion of Marfan pts manifest ocular abnormalities?
- At least 80%
What protein is abnormal in Marfan’s? Fibrillin

What three systems/membranes manifest abnormalities in Marfans?
-- The eye (duh)
-- The cardiovascular
-- The musculoskeletal

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

Other than ectopia lentis, what ocular structural abnormalities are often present?
--
--
Ectopia Lentic

**Congenital**
- Marfan

**Developmental**
- Marfan

**Acquired**
- Trauma
- (Pseudo)exfoliation syndrome

**What protein is abnormal in Marfan’s?**
Fibrillin

**What three systems manifest abnormalities in Marfans?**
--The eye (duh)
--The cardiovascular
--The musculoskeletal

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

**Other than ectopia lentis, what ocular structural abnormalities are often present?**
--Microspherophakia
--Increased axial length
What protein 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%

Other than ectopia lentis, what ocular structural abnormalities are often present?
--- Microspherophakia
--- Increased axial length

Of the three (ectopia lentis, m’phakia, increased axial length), which is most likely to be present?
Ectopia Lentis

**Congenital**
- Marfan

**Developmental**
- Marfan

**Acquired**
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- (Pseudo)exfoliation syndrome

What protein is abnormal in Marfan’s?
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What three systems manifest abnormalities in Marfans?
- The eye (duh)
- The cardiovascular
- The musculoskeletal

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

Other than **ectopia lentis**, what ocular structural abnormalities are often present?
- Microspherophakia
- Increased axial length

Of the three (ectopia lentis, m’phakia, increased axial length), which is most likely to be present?
- Ectopia lentis
Ectopia Lentis

Congenital
  - Marfan

Developmental
  - Marfan

Acquired
  - Trauma
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What protein is abnormal in Marfan’s? Fibrillin

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

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

Other than ectopia lentis, what ocular structural abnormalities are often present?
  -- Microspherophakia
  -- Increased axial length

What percentage of Marfan patients develop ectopia lentis? 50 - 80%
Ectopia Lentis

**Congenital**
- Marfan

**Developmental**
- Marfan

**Acquired**
- Trauma
  - (Pseudo)exfoliation syndrome

**What protein is abnormal in Marfan’s?**
- Fibrillin

**What three structures/systems manifest abnormalities in Marfans?**
- The eye (duh)
- The cardiovascular
- The musculoskeletal

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

**Other than ectopia lentis, what other ocular abnormalities are often present?**
- Microspherophakia
- Increased axial length

**What percentage of Marfan patients develop ectopia lentis?**
- 50 - 80
Marfan syndrome: Ectopia lentis
Ectopia Lentis

Marfan pts tend to be high myopes. Give three reasons why.

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What protein is abnormal in Marfan?
Fibrillin

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

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

Other than ectopia lentis, what ocular structural abnormalities are often present?
--Microspherophakia
--Increased axial length

Marfan pts tend to be high myopes. Give three reasons why.

--

--

--

A spectacle-corrected Marfan patient c/o difficulty reading—why?
The zonules in Marfan are stretched, and stretched zonules do not transmit accommodative forces well.

Re managing Marfan:
Poor accommodation implies what?
Bifocals may be needed at an early age.

Why not just bust up in there and do a clear lens extraction with IOL?
Marfan patients are at very high risk for vitreous loss as well as retinal detachment.
**Ectopia Lentis**

*Marfan pts tend to be high myopes. Give three reasons why.*

--Increased axial length → **axial** myopia
--Microspherophakia → increased lens curvature → **lenticular** myopia
--Ectopia lentis → Lens subluxation → looking through highly curved peripheral lens → **lens periphery** myopia

**Congenital**

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*What protein is abnormal in Marfans?*

Fibrillin

*What three structures/systems manifest abnormalities in Marfans?*

--The eye (duh)
--The cardiovascular
--The musculoskeletal

*What proportion of Marfan pts manifest ocular abnormalities?*

At least 80%

*Other than ectopia lentis, what ocular structural abnormalities are often present?*

--Microspherophakia
--Increased axial length

---

Marfan patients tend to be high myopes. Give three reasons why.

--Increased axial length → **axial** myopia
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A spectacle-corrected Marfan patient reports difficulty reading—why?

The zonules in Marfan are **stretched**, and stretched zonules do **not** transmit accommodative forces well.

Re managing Marfans: Poor accommodation implies what?

Bifocals may be needed at an early age.

Why not just bust up in there and do a clear lens extraction with IOL?

Marfan patients are at very high risk for vitreous loss as well as retinal detachment.
Marfan pts tend to be high myopes. Give three reasons why.
--Increased axial length → axial myopia
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I made this term up; a better name is spherical-aberration myopia
Marfan pts tend to be high myopes. Give three reasons why.
- Increased axial length → axial myopia
- Microspherophakia → increased lens curvature → lenticular myopia
- Ectopia lentis → Lens subluxation → looking through highly curved peripheral lens → lens periphery myopia
- Increased corneal curvature?

What protein is abnormal in Marfan?
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What proportion of Marfan pts manifest ocular abnormalities?
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Other than ectopia lentis, what ocular structural abnormalities are often present?
- Microspherophakia
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Why not just bust up in there and do a clear lens extraction with IOL?

Marfan patients are at very high risk for vitreous loss as well as retinal detachment.

What about the cornea in Marfan pts--does it contribute to their high myopia?

No—in fact, the cornea in Marfan tends to be flatter than normal.
What protein is abnormal in Marfan?
- Fibrillin

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

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

Other than ectopia lentis, what ocular structural abnormalities are often present?
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-- Increased corneal curvature? No!

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What about the cornea in Marfan pts--does it contribute to their high myopia?

No—in fact, the cornea in Marfan tends to be flatter than normal.

Is the cornea flatter because it has a smaller-than-normal diameter (ie, microcornea)?

What protein is abnormal in Marfans?

Fibrillin

What three structures/systems manifest abnormalities in Marfans?

--The eye (duh)
--The cardiovascular
--The musculoskeletal

What proportion of Marfan pts manifest ocular abnormalities?

At least 80%

Other than ectopia lentis, what ocular structural abnormalities are often present?

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--Increased corneal curvature? No!

What about the cornea in Marfan pts--does it contribute to their high myopia?

No—in fact, the cornea in Marfan tends to be flatter than normal.

Is the cornea flatter because it has a smaller-than-normal diameter (ie, microcornea)?
**Ectopia Lentis**

Marfan pts tend to be high myopes. Give three reasons why.
--Increased axial length $\rightarrow$ axial myopia
--Microspherophakia $\rightarrow$ increased lens curvature $\rightarrow$ lenticular myopia
--Ectopia lentis $\rightarrow$ Lens subluxation $\rightarrow$ looking through highly curved peripheral lens $\rightarrow$ lens periphery myopia
--Increased corneal curvature? **No!**

**What about the cornea in Marfan pts--does it contribute to their high myopia?**
No—in fact, the cornea in Marfan tends to be **flatter** than normal.

**Is the cornea flatter because it has a smaller-than-normal diameter (ie, microcornea)?**
No—in fact, if anything, corneal diameter in Marfan’s tends to be **larger** than normal (ie, megalocornea).

**What protein is abnormal in Marfans?**
Fibrillin

**What three structures/systems manifest abnormalities in Marfans?**
--The eye *(duh)*
--The cardiovascular
--The musculoskeletal

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

Other than **ectopia lentis**, what ocular structural abnormalities are often present?
--Microspherophakia
--Increased axial length

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--Increased corneal curvature? **No!**
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--Increased axial length → **axial** myopia
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A spectacle-corrected Marfan patient c/o difficulty reading—why?

**Marfan**

**Congenital**

Marfan

What protein is abnormal in Marfan?
Fibrillin

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

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

Other than ectopia lentis, what ocular structural abnormalities are often present?
--Microspherophakia
--Increased axial length

A spectacle-corrected Marfan patient c/o difficulty reading—why?

The zonules in Marfan are stretched, and stretched zonules do not transmit accommodative forces well.

Re managing Marfans: Poor accommodation implies what?
Bifocals may be needed at an early age.

Why not just bust up in there and do a clear lens extraction with IOL?
Marfan patients are at very high risk for vitreous loss as well as retinal detachment.
Marfan pts tend to be high myopes. Give three reasons why.
- Increased axial length → axial myopia
- Microspherophakia → increased lens curvature → lenticular myopia
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A spectacle-corrected Marfan patient c/o difficulty reading—why?
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A spectacle-corrected Marfan patient c/o difficulty reading—why?
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**Ectopia Lentis**

**Marfan pts tend to be high myopes. Give three reasons why.**
--Increased axial length \(\rightarrow\) **axial** myopia
--Microspherophakia \(\rightarrow\) increased lens curvature \(\rightarrow\) **lenticular** myopia
--Ectopia lentis \(\rightarrow\) Lens subluxation \(\rightarrow\) looking through highly curved peripheral lens \(\rightarrow\) **lens periphery** myopia

**A spectacle-corrected Marfan patient c/o difficulty reading—why?**
The zonules in Marfan are **s t r e t c h e d**, and stretched zonules do not transmit accommodative forces well

**Re managing Marfan’s: Poor accommodation implies what?**

---

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

**Other than ectopia lentis, what ocular structural abnormalities are often present?**
--Microspherophakia
--Increased axial length

---

**What protein is abnormal in Marfan?**
Fibrillin

---

**What three structures/systems manifest abnormalities in Marfans?**
--The eye (duh)
--The cardiovascular
--The musculoskeletal

---

**Congenital**

- Marfan

---

**Q**
**Ectopia Lentis**

Marfan pts tend to be high myopes. Give three reasons why.

- Increased axial length → **axial** myopia
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A spectacle-corrected Marfan patient c/o difficulty reading—why?
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Re managing Marfan’s: Poor accommodation implies what?
Bifocals may be needed at an early age

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**Congenital**

- Marfan

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**Ectopia Lentis**

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*A spectacle-corrected Marfan patient c/o difficulty reading—why?*
The zonules in Marfan are **stretched**, and stretched zonules do not transmit accommodative forces well

**Re managing Marfan’s: Poor accommodation implies what?**
Bifocals may be needed at an early age

**Why not just bust up in there and do a clear lens extraction with IOL?**

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A spectacle-corrected Marfan patient c/o difficulty reading—why? The zonules in Marfan are stretched, and stretched zonules do not transmit accommodative forces well.

Re managing Marfan's: Poor accommodation implies what? Bifocals may be needed at an early age.

Why not just bust up in there and do a clear lens extraction with IOL?
Ectopia Lentis

Marfan pts tend to be high myopes. Give three reasons why.
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A spectacle-corrected Marfan patient c/o difficulty reading—why? The zonules in Marfan are **stretched**, and stretched zonules do not transmit accommodative forces well

**Re managing Marfan's:** Poor accommodation implies what? Bifocals may be needed at an early age

**Why not just bust up in there and do a clear lens extraction with IOL?** Marfan patients are at very high risk for vitreous loss as well as retinal detachment

What protein is abnormal in Marfan?
Fibrillin

Other than ectopia lentis, what ocular structural abnormalities are often present?
--Microspherophakia
--Increased axial length

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

What three structures/systems manifest abnormalities in Marfan?
--The eye (duh)
--The cardiovascular
--The musculoskeletal
Which is the most common heritable cause of ectopia lentis?
Which is the most common heritable cause of ectopia lentis?

Homocystinuria
Ectopia Lentis

**Congenital**
- Marfan
- Aniridia

**Developmental**
- Marfan
- Homocystinuria

**Acquired**
- Trauma
- (Pseudo)exfoliation syndrome

---

*Homocystinuria is due to faulty metabolism for what amino acid? (hint: it’s not cysteine)*
Homocystinuria is due to faulty metabolism for what amino acid? (hint: it's not cysteine)
Methionine
Homocystinuria is due to faulty metabolism for what amino acid? (hint: it's not cysteine) Methionine

Then why isn't it called ‘methioninuria’?
Homocystinuria is due to faulty metabolism for what amino acid? (hint: it’s not cysteine) Methionine

Then why isn’t it called ‘methioninuria’? Because faulty methionine metabolism manifests as excessive serum homocysteine, which spills into the urine—hence, homocystinuria
Homocystinuria is due to faulty metabolism for what amino acid? (hint: it’s not cysteine) Methionine

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Because faulty methionine metabolism manifests as excessive serum homocysteine, which spills into the urine—hence, homocystinuria

What four structures/systems manifest abnormalities in homocystinuria?
-- The eye (duh)
--
Homocystinuria is due to faulty metabolism for what amino acid? (hint: it’s not cysteine) Methionine

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Because faulty methionine metabolism manifests as excessive serum homocysteine, which spills into the urine—hence, homocystinuria

What four structures/systems manifest abnormalities in homocystinuria?
--The eye (duh)
--The cardiovascular
--The musculoskeletal
--The CNS
Ectopia Lentis

Congenital
- Marfan
- Aniridia

Developmental
- Marfan
- Homocystinuria

Acquired
- Trauma
- (Pseudo)exfoliation syndrome

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What four structures/systems manifest abnormalities in homocystinuria?
- The eye (duh)
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- The musculoskeletal
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Note that these were the three structures/systems affected in Marfan
Ectopia Lentis

- Congenital
  - Marfan
  - Aniridia
- Developmental
  - Homocystinuria
- Acquired
  - Trauma
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**What four structures/systems manifest abnormalities in homocystinuria?**
- The eye *(duh)*
- The cardiovascular
- The musculoskeletal
- The CNS

*Note that these were the three structures/systems affected in Marfan*
Ectopia Lentis

- Congenital
  - Marfan
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What four structures/systems manifest abnormalities in homocystinuria?
- The eye (duh)
- The cardiovascular
- The musculoskeletal
- The CNS

What is the classic habitus in homocystinuria?
Tall and thin, with arachnodactyly and sternum abnormalities

Note that these were the three structures/systems affected in Marfan
Homocystinuria is due to faulty metabolism for what amino acid? (hint: it’s not cysteine) Methionine

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What is the classic habitus in homocystinuria? Tall and thin, with arachnodactyly and sternum abnormalities.

How about their coloration? They tend to be fair-skinned and fair-haired.

What four structures/systems manifest abnormalities in homocystinuria?

--The eye (duh)
--The cardiovascular
--The musculoskeletal
--The CNS

Note that these were the three structures/systems affected in Marfan.
Ectopia Lentis

**Congenital**
- Marfan
- Aniridia

**Developmental**
- Marfan
- Homocystinuria

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*How about their coloration?*
They tend to be fair-skinned and fair-haired

*What four structures/systems manifest abnormalities in homocystinuria?*
- The eye (duh)
- The cardiovascular
- The musculoskeletal
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Ectopia Lentis

Homocystinuria
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What proportion of homocystinuria pts manifest ocular abnormalities?

- The eye (duh)
- The cardiovascular
- The musculoskeletal
- The CNS
Homocystinuria is due to faulty metabolism for what amino acid? (hint: it’s not cysteine)

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Because faulty methionine metabolism manifests as excessive serum homocysteine, which spills into the urine—hence, homocystinuria

What four structures/systems manifest abnormalities in homocystinuria?
--The eye (duh)
--The cardiovascular
--The musculoskeletal
--The CNS

What proportion of homocystinuria pts manifest ocular abnormalities?
About 80%
Ectopia Lentis

Congenital
- Marfan
- Aniridia

Developmental
- Marfan
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What proportion of homocystinuria pts manifest ocular abnormalities? About 80%

Other than ectopia lentis, are other ocular structural abnormalities often present?
Homocystinuria is due to faulty metabolism for what amino acid? *(hint: it's not cysteine)*

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Other than ectopia lentis, are other ocular structural abnormalities often present? Nah—ectopia lentis is pretty much it
Homocystinuria: Ectopia lentis
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What proportion of homocystinuria pts manifest ocular abnormalities?
About 80%

What four structures/systems manifest abnormalities in homocystinuria?
The eye (duh)
The cardiovascular
The musculoskeletal
The CNS

Other than ectopia lentis, what ocular structural abnormalities are often present?
Ectopia lentis

During which stage of life does subluxation typically occur?
Childhood (in most cases prior to age 10 years)
Ectopia Lentis

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--The musculoskeletal
--The CNS

Other than ectopia lentis, what ocular structural abnormalities are often present?
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About 80%

*Other than ectopia lentis, what ocular structural abnormalities are often present?*
Nah—ectopia lentis is pretty much it

*What sight-threatening complication can result from ectopia lentis?*
Secondary angle-closure glaucoma

*During which stage of life does subluxation typically occur?*
Childhood (in most cases prior to age 10 years)
Ectopia Lentis

**Homocystinuria**

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Ectopia Lentis

Buphthalmos in a homocystinuria pt with ectopia lentis
Homocystinuria is due to faulty metabolism for what amino acid? (hint: it’s not cysteine)

Methionine

What sight-threatening complication can result from ectopia lentis?

Secondary angle-closure glaucoma

For more on secondary angle-closure glaucoma stemming from ectopia lentis, see slide-set G16

Other than ectopia lentis, what ocular structural abnormalities are often present?

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Ectopia lentis

During which stage of life does subluxation typically occur?

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-- The eye (duh)
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-- The musculoskeletal
-- The CNS

*What cardiovascular abnormalities are common?*
**Ectopia Lentis**

**Congenital**
- Marfan
- Aniridia

**Developmental**
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What four structures/systems manifest abnormalities in homocystinuria?
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What cardiovascular abnormalities are common?
- HTN
- Cardiomegaly
- Thromboembolism
- CAD
- CVA
Homocystinuria is due to faulty metabolism for what amino acid? (hint: it's not cysteine)
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--HTN
--Cardiomegaly
--Thromboembolism
--CAD
--CVA

What proportion of untreated homocystinurics will experience a significant vascular event prior to age 30?
An astonishing 50%! (And the majority will experience it prior to age 20)
Ectopia Lentes

Homocystinuria is due to faulty metabolism for what amino acid? (hint: it’s not cysteine) Methionine

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- Aniridia

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--The cardiovascular
--The musculoskeletal
--The CNS

What cardiovascular abnormalities are common?
--HTN
--Cardiomegaly
--Thromboembolism
--CAD
--CVA

What relatively common medical event greatly increases the risk of a thromboembolic event?
- General anesthesia
Homocystinuria is due to faulty metabolism for what amino acid? (hint: it's not cysteine) Methionine

Then why isn’t it called ‘methioninuria’?

Because faulty methionine metabolism manifests as excessive serum homocysteine, which spills into the urine—hence, homocystinuria

What four structures/systems manifest abnormalities in homocystinuria?

--The eye (duh)
--The cardiovascular
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--The CNS

What cardiovascular abnormalities are common?

--HTN
--Cardiomegaly
--Thromboembolism
--CAD
--CVA

What relatively common medical event greatly increases the risk of a thromboembolic event?

General anesthesia. It is vital to identify homocystinurics prior to surgery!
Homocystinuria is due to faulty metabolism for what amino acid? (hint: it's not cysteine) Methionine

Then why isn’t it called ‘methioninuria’? Because faulty methionine metabolism manifests as excessive serum homocysteine, which spills into the urine—hence, homocystinuria.

What four structures/systems manifest abnormalities in homocystinuria?
--The eye (duh)
--The cardiovascular
--The musculoskeletal
--The CNS

In addition to arachnodactyly and sternum deformities, there is a musculoskeletal abnormality classic for homocystinuria. What is it?
Ectopia Lentis

**Developmental**
- Marfan
- Homocystinuria

**Acquired**
- Trauma
- (Pseudo)exfoliation syndrome

**Congenital**
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**Homocystinuria**

*Homocystinuria is due to faulty metabolism for what amino acid? (hint: it’s not cysteine)*  
Methionine

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*What four structures/systems manifest abnormalities in homocystinuria?*
---
- The eye (duh)
- The cardiovascular
- **The musculoskeletal**
- The CNS

*In addition to arachnodactyly and sternum deformities, there is a musculoskeletal abnormality classic for homocystinuria. What is it?*
Early osteoporosis
Homocystinuria is due to faulty metabolism for what amino acid? (hint: it’s not cysteine) Methionine

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Because faulty methionine metabolism manifests as excessive serum homocysteine, which spills into the urine—hence, homocystinuria

What four structures/systems manifest abnormalities in homocystinuria?
-- The eye (duh)
-- The cardiovascular
-- The musculoskeletal
-- The CNS

How early are we talking about?

Early osteoporosis
Homocystinuria is due to faulty metabolism for what amino acid? (hint: it's not cysteine)
Methionine

Then why isn’t it called ‘methioninuria’?
Because faulty methionine metabolism manifests as excessive serum homocysteine, which spills into the urine—hence, homocystinuria

What four structures/systems manifest abnormalities in homocystinuria?
--- The eye (duh)
--- The cardiovascular
--- The musculoskeletal
--- The CNS

How early are we talking about?
Crazy early. Osteoporotic changes are common in childhood (and have been reported in toddlers)
Early osteoporosis

In addition to arachnodactyly and sternum deformities, there is a musculoskeletal abnormality classic for homocystinuria.
What is it?
Homocystinuria is due to faulty metabolism for what amino acid? (hint: it’s not cysteine)
Methionine

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What four structures/systems manifest abnormalities in homocystinuria?
--The eye (duh)
--The cardiovascular
--The musculoskeletal
--The CNS

What CNS abnormalities are common?
--
Ectopia Lentis

Congenital
- Marfan
- Aniridia

Developmental
- Marfan
- Homocystinuria

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- Trauma
- (Pseudo)exfoliation syndrome

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What four structures/systems manifest abnormalities in homocystinuria?
--The eye (duh)
--The cardiovascular
--The musculoskeletal
--The CNS

What CNS abnormalities are common?
--Seizures
--Mental retardation
Ectopia Lentis

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Methionine

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Because faulty methionine metabolism manifests as excessive homocysteine, which spills into the urine—hence, homocystinuria

What four structures/systems manifest abnormalities in homocystinuria?
--The eye (duh)
--The cardiovascular
--The musculoskeletal
--The CNS

So, MOST homocystinurics have these nonocular problems:
--M
--O
--S
--T
Ectopia Lentis

**Congenital**
- Marfan
- Aniridia

**Developmental**
- Marfan
- Homocystinuria

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What four structures/systems manifest abnormalities in homocystinuria?
- The eye (duh)
- The cardiovascular
- The musculoskeletal
- The CNS

So, **MOST** homocystinurics have these nonocular problems:
- Mental retardation
- Osteoporosis
- Seizures
- Thrombosis of the systemic vasculature
Ectopia Lentis

**Congenital**
- Marfan
- Aniridia

**Developmental**
- Marfan
- Homocystinuria

**Acquired**
- Trauma
- (Pseudo)exfoliation syndrome

**Homocystinuria** is due to faulty metabolism for what amino acid? *(hint: it’s not cysteine)*
Methionine

*Then why isn’t it called ‘methioninuria’?*
Because faulty methionine metabolism manifests as excessive serum homocysteine, which spills into the urine—hence, homocystinuria

*Are there steps that can be taken to mitigate the effects of the faulty methionine metabolism?*
- The cardiovascular
- The musculoskeletal
- The CNS
Homocystinuria is due to faulty metabolism for what amino acid? (hint: it’s not cysteine) Methionine

Then why isn’t it called ‘methioninuria’?

Because faulty methionine metabolism manifests as excessive serum homocysteine, which spills into the urine—hence, homocystinuria

Are there steps that can be taken to mitigate the effects of the faulty methionine metabolism?

Indeed there are. A diet low in amino acid (and high in amino acid), along with supplemental vitamin , can significantly reduce the rate of ectopia (and systemic probs as well)

-- The cardiovascular
-- The musculoskeletal
-- The CNS
Homocystinuria is due to faulty metabolism for what amino acid? (hint: it's not cysteine)
Methionine

Then why isn’t it called ‘methioninuria’?
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Are there steps that can be taken to mitigate the effects of the faulty methionine metabolism?
Indeed there are. A diet low in methionine (and high in cysteine), along with supplemental vitamin B₆, can significantly reduce the rate of ectopia (and systemic probs as well)

-- The cardiovascular
-- The musculoskeletal
-- The CNS
As is likely apparent by now, Marfan and homocystinuria share a number of features—but not all. Let’s run through them and see which are which.
Marfan syndrome

- Associated with cataracts
- Lens subluxes up
- Lens subluxes down
- Tall
- Thin habitus
- Fair-haired
- Zonules stretched
- Zonules broken
- Associated with microspherophakia
- Ectopia usually bilateral and symmetric
- Chest-wall deformities

Ectopia Lentis

Homocystinuria

↑ risk thromboembolism

Divvy up the characteristics.
Some go both ways.

(Start here, work down the list)
Ectopia Lentis

Marfan syndrome

Homocystinuria

Do these simultaneously

Lens subluxes up
Lens subluxes down

Tall
Thin habitus
Fair-haired

Zonules stretched
Zonules broken
Associated with microspherophakia

Ectopia usually bilateral and symmetric
Chest-wall deformities

↑ risk thromboembolism
Q/A

**Ectopia Lentis**

- Associated with cataracts
- Lens subluxes **up**
- Lens subluxes **down**

**Marfan syndrome**
- Tall
- Thin habitus
- Fair-haired
- Zonules *stretched*
- Zonules *broken*
- Associated with microspherophakia
- Ectopia usually bilateral and symmetric
- Chest-wall deformities

**Homocystinuria**
- ↑ risk *thromboembolism*
- Lens subluxes **down**
Q/A

**Ectopia Lentis**

**Marfan syndrome**

**Homocystinurua**

↑ risk thromboembolism

Lens subluxes *up*

- Tall
- Thin habitus
- Fair-haired
- Zonules *stretched*
- Zonules *broken*
- Associated with microspherophakia
- Ectopia usually bilateral and symmetric
- Chest-wall deformities

Lens subluxes *down*

- Tall
- Both
Q/A

Marfan syndrome

Homocystinuria

↑ risk thromboembolism

Ectopia Lentis

Associated with cataracts

Lens subluxes up

Lens subluxes down

Tall

Thin habitus

Fair-haired

Zonules stretched

Zonules broken

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Ectopia usually bilateral and symmetric

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Homocystinuria
Q/A

Marfan syndrome

Homocystinuria

↑ risk thromboembolism

Ectopia Lentis

Associated with cataracts

Associated with cataracts

Lens subluxes up

Lens subluxes down

Tall

Tall

Thin habitus

Thin habitus

Fair-haired

Do these simultaneously

Zonules stretched

Zonules broken

Associated with microspherophakia

Ectopia usually bilateral and symmetric

Chest-wall deformities

↑ risk thromboembolism
Q/A

**Marfan syndrome**

- Lens subluxes *up*
- Tall
- Thin habitus
- Zonules *stretched*

**Homocystinuria**

- Lens subluxes *down*
- Tall
- Thin habitus
- Fair-haired
- Zonules *broken*

**Ectopia Lentsis**

- ↑ risk *thromboembolism*

Associated with:

- microspherophakia
- Ectopia usually bilateral and symmetric
- Chest-wall deformities
Q/A

**Ectopia Lentis**

- Associated with cataracts
- Lens subluxes **up**
- Tall
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**Homocystinuria**

- **↑ risk thromboembolism**
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Ectopia Lentis

Marfan syndrome

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**Ectopia Lentis**

- **Marfan syndrome**
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  - Ectopia usually bilateral and symmetric
  - Chest-wall deformities

- **Homocystinuria**
  - Associated with cataracts
  - Lens subluxes **down**
  - Tall
  - Thin habitus
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↑ risk **thromboembolism**
Marfan syndrome

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Homocystinuria

- Lens subluxes *down*
- ↑ risk *thromboembolism*
- Tall
- Thin habitus
- Fair-haired
- Zonules *broken*
- Associated with microspherophakia
- Ectopia usually bilateral and symmetric
- Chest-wall deformities

Note the Key Differences!
In a few words, how would you describe the shape of a microspherophakic lens?
In a few words, how would you describe the shape of a microspherophakic lens? The name says it all: the lens is small (‘micro’) and round (‘sphero’).
Microspherophakia. Note the small size, extreme curvature of the lens
In a few words, how would you describe the shape of a microspherophakic lens? The name says it all: the lens is small (‘micro’) and round (‘sphero’).

What common slit-lamp observation owes to the lens’ small size?
In a few words, how would you describe the shape of a microspherophakic lens? The name says it all: the lens is small (‘micro’) and round (‘sphero’).

What common slit-lamp observation owes to the lens’ small size? Typically, the entirety of the lens equator can be seen in the pupillary aperture when the pt is widely dilated.
Microspherophakia. With mydriasis, the lens is able to fit through the pupillary aperture
In a few words, how would you describe the shape of a microspherophakic lens? The name says it all: the lens is small (‘micro’) and round (‘sphero’).

Pts with microspherophakia are almost always high myopes. Why?
In a few words, how would you describe the shape of a microspherophakic lens? The name says it all: the lens is small (‘micro’) and round (‘sphero’).

Pts with microspherophakia are almost always high myopes. Why? Because the lens is small, it has a short radius of curvature.
In a few words, how would you describe the shape of a microspherophakic lens? The name says it all: the lens is **small** (‘micro’) and **round** (‘sphero’).

*Pts with microspherophakia are almost always high myopes. Why?* Because the lens is small, it has a short radius of curvature. Further, because it is spherical, it is more curved than is a normal lens.
Ectopia Lentis

- Congenital
  - Marfan
  - Aniridia
  - Ectopia lentis et pupillae
  - Simple ectopia lentis

- Developmental
  - Marfan
  - Homocystinuria
  - Microspherophakia

- Acquired
  - Trauma
  - (Pseudo)exfoliation syndrome

---

In a few words, how would you describe the shape of a microspherophakic lens? The name says it all: the lens is small (‘micro’) and round (‘sphero’).

*Pts with microspherophakia are almost always high myopes. Why? Because the lens is small, it has a short radius of curvature. Further, because it is spherical, it is more curved than is a normal lens. These two factors give the m'spheric lens vastly more converging power than a normal lens possesses.*
In a few words, how would you describe the shape of a microspherophakic lens? The name says it all: the lens is small (‘micro’) and round (‘sphero’).

How does this differ from ‘run of the mill’ high myopia?

is spherical, it is more curved than is a normal lens. These two factors give the m’spheric lens vastly more converging power than a normal lens possesses.
In a few words, how would you describe the shape of a microspherophakic lens? The name says it all: the lens is small (‘micro’) and round (‘sphero’).

How does this differ from ‘run of the mill’ high myopia? Most cases of high myopia are due to excessive length of the optical axis (so-called ‘axial myopia’) is spherical, it is more curved than is a normal lens. These two factors give the m'spheric lens vastly more converging power than a normal lens possesses.
With what condition is microspherophakia most frequently associated?
With what condition is microspherophakia most frequently associated?

Weill-Marchesani syndrome
Ectopia Lentis

Congenital
- Marfan
- Aniridia
- Ectopia lentis et pupillae

Developmental
- Marfan
- Homocystinuria
- Microspherophakia

Acquired
- Trauma
- (Pseudo)exfoliation syndrome

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

**What are the findings in Weill-Marchesani?**
With what condition is microspherophakia most frequently associated? Weill-Marchesani syndrome

What are the findings in Weill-Marchesani?
Patients with Weill-Marchesani have:
- short stature
- short fingers
- stiff joints

(Think of it as the opposite of Marfan syndrome)
Ectopia Lentis

- Congenital
  - Marfan
  - Aniridia
  - Ectopia lentis et pupillae
  - Simple ectopia lentis (congenital type)
- Developmental
  - Marfan
  - Homocystinuria
  - Microspherophakia
  - Ectopia lentis et pupillae (late-onset type)
- Acquired
  - Trauma
  - (Pseudo)exfoliation syndrome

With what condition is microspherophakia most frequently associated?

- Weill-Marchesani syndrome

**What are the findings in Weill-Marchesani?**
Patients with Weill-Marchesani have:

...short stature
Weill-Marchesani syndrome: Short stature
Q/A

Ectopia Lentis

- Congenital
  - Marfan
  - Aniridia
  - Ectopia lentis et pupillae
  - Simple ectopia lentis (congenital type)

- Developmental
  - Marfan
  - Homocystinuria
  - Microspherophakia
  - Ectopia lentis et pupillae
  - Simple ectopia lentis (late-onset type)

- Acquired
  - Trauma
  - (Pseudo)exfoliation syndrome

With what condition is microspherophakia most frequently associated?

Weill-Marchesani syndrome

What are the findings in Weill-Marchesani?

Patients with Weill-Marchesani have:

- short stature
- short fingers
- stiff joints

(Take it as the opposite of Marfan syndrome)
With what condition is microspherophakia most frequently associated?

Weill-Marchesani syndrome

What are the findings in Weill-Marchesani?
Patients with Weill-Marchesani have:

...short stature
...short fingers
Ectopia Lentis

Weill-Marchesani syndrome: Short fingers
Ectopia Lentis

Congenital
- Marfan
- Aniridia
- Ectopia lentis et pupillae

Developmental
- Marfan
- Homocystinuria
- Microspherophakia

Acquired
- Trauma
- (Pseudo)exfoliation syndrome

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**With what condition is microspherophakia most frequently associated?**

Weill-Marchesani syndrome

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**What are the findings in Weill-Marchesani?**

Patients with Weill-Marchesani have:
- short stature
- short fingers
- stiff joints
With what condition is microspherophakia most frequently associated?

Weill-Marchesani syndrome

What are the findings in Weill-Marchesani?

Patients with Weill-Marchesani have:

...short stature
...short fingers
...stiff joints
**Ectopia Lentsis**

**Congenital**
- Marfan
- Aniridia
- Ectopia lentis et pupillae
- Simple ectopia lentis (congenital type)

**Developmental**
- Marfan
- Homocystinuria
- Microspherophakia

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

---

**What are the findings in Weill-Marchesani?**
Patients with Weill-Marchesani have:
- ***short*** stature
- ***short*** fingers
- ***stiff*** joints

(Think of it as the opposite of Marfan syndrome)
Ectopia Lentis

Congenital
- Marfan
  - Aniridia
  - Ectopia lentis et pupillae
- Simple ectopia lentis

Developmental
- Marfan
- Microspherophakia
- Homocystinuria

Acquired
- Trauma
- (Pseudo)exfoliation syndrome

**With what condition is microspherophakia most frequently associated?**

**Weill-Marchesani syndrome**

What are the findings in Weill-Marchesani?
Patients with Weill-Marchesani have:

...short stature (Tall stature)
...short fingers (Long fingers)
...stiff joints (Lax joints)
(Think of it as the opposite of Marfan syndrome)
Weill-Marchesani syndrome

Marfan syndrome

Ectopia Lentis
Ectopia Lentis

Congenital
- Marfan
- Aniridia
- Ectopia lentis et pupillae
- Simple ectopia lentis

Developmental
- Marfan
- Homocystinuria
- Microspherophakia

Acquired
- Trauma
- (Pseudo)exfoliation syndrome

**Weill-Marchesani syndrome**

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

*Weill-Marchesani is strongly associated with microspherophakia. With what conditions is microspherophakia *occasionally* associated?*
Ectopia Lentis

**Congenital**
- Marfan
- Aniridia
- Ectopia lentis et pupillae

**Developmental**
- Marfan
- Homocystinuria
- Microspherophakia

**Acquired**
- Trauma
- (Pseudo)exfoliation syndrome

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

Weill-Marchesani is strongly associated with microspherophakia. With what conditions is *microspherophakia occasionally* associated?
- Congenital rubella, Lowe syndrome, Alport syndrome, Marfan syndrome and Peters anomaly
Ectopia Lentis

Ruby LAMP is a mnemonic for the other conditions associated with microspherophakia:

Ruby = Rubella

Lowe syndrome
Alport syndrome
Marfan syndrome
Peters anomaly

With what condition is microspherophakia most frequently associated?

Weill-Marchesani syndrome

Weill-Marchesani is strongly associated with microspherophakia. With what conditions is microspherophakia occasionally associated?

Congenital rubella, Lowe syndrome, Alport syndrome, Marfan syndrome and Peters anomaly

In three words (including syndrome), what are Lowe and Alport syndromes? Familial oculorenal syndromes.

What is their classic (nonocular) presenting sign? Hematuria.

Microspherophakia is not the classic lens finding in the oculorenal syndromes (and should not be the first one out of your mouth if pimped about them). What is? Lenticonus.
Weill-Marchesani is strongly associated with microspherophakia. With what conditions is microspherophakia occasionally associated?

Congenital rubella, Lowe syndrome, Alport syndrome, Marfan syndrome and Peters anomaly.

What is their classic (nonocular) presenting sign?

Hematuria.

Microspherophakia is not the classic lens finding in the oculorenal syndromes (and should not be the first one out of your mouth if pimped about them). What is?

Lenticonus.

In three words (including syndrome), what are Lowe and Alport syndromes?

Familial oculorenal syndromes.
Simple ectopia lentis (late-onset type) is strongly associated with microspherophakia. With what conditions is microspherophakia occasionally associated?

- Congenital rubella
- Lowe syndrome
- Alport syndrome
- Marfan syndrome
- Peters anomaly

In three words (including syndrome), what are Lowe and Alport syndromes?

Familial oculorenal syndromes

What is their classic (nonocular) presenting sign?

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Lenticonus
Weill-Marchesani is strongly associated with microspherophakia. With what conditions is microspherophakia occasionally associated?

Congenital rubella, Lowe syndrome, Alport syndrome, Marfan syndrome and Peters anomaly.

In three words (including syndrome), what are Lowe and Alport syndromes?

Familial oculorenal syndromes

What is their classic (nonocular) presenting sign?

Hematuria

Microspherophakia occasionally associated.

Congenital rubella, Lowe syndrome, Alport syndrome, Marfan syndrome and Peters anomaly.
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Simple ectopia lentis (late-onset type)

Weill-Marchesani is strongly associated with microspherophakia. With what conditions is microspherophakia occasionally associated?

- Congenital rubella
- Lowe syndrome
- Alport syndrome
- Marfan syndrome
- Peters anomaly

Ectopia Lentis

In three words (including syndrome), what are Lowe and Alport syndromes?

Familial oculorenal syndromes

What is their classic (nonocular) presenting sign?

Hematuria

Microspherophakia is not the classic lens finding in the oculorenal syndromes (and should not be the first one out of your mouth if pimped about them). What is?

Lenticonus

With what condition is microspherophakia occasionally associated?

Congenital rubella, Lowe syndrome, Alport syndrome, Marfan syndrome and Peters anomaly.
Anterior lenticous in Alport syndrome
Simple ectopia lentis (late-onset type) is strongly associated with microspherophakia. With what condition is microspherophakia occasionally associated? Congenital rubella, Lowe syndrome, Alport syndrome, Marfan syndrome and Peters anomaly.

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

In three words (not including a form of), what is Peters anomaly? A form of anterior segment dystgenesis.
Ectopia Lentis

**Congenital**
- Marfan
- Aniridia
- Ectopia lentis et pupillae

**Developmental**
- Marfan
- Homocystinuria
- **Microspherophakia**

**Acquired**
- Trauma
- (Pseudo)exfoliation syndrome

---

Weill-Marchesani is strongly associated with microspherophakia. With what conditions is microspherophakia occasionally associated?

**Weill-Marchesani syndrome**

Simple ectopia lentis

**Peters anomaly**

In three words (not including a form of), what is Peters anomaly? A form of anterior segment dysgenesis.
Simple ectopia lentis (late-onset type) is strongly associated with microspherophakia. With what conditions is microspherophakia occasionally associated?

- Congenital rubella
- Lowe syndrome
- Alport syndrome
- Marfan syndrome
- Peters anomaly

Weill-Marchesani syndrome is associated with microspherophakia.

In three words (not including a form of), what is Peters anomaly?

A form of anterior segment dysgenesis

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

A neurocristopathy is a congenital/developmental abnormality owing to flawed neural-crest cell migration or differentiation.
Ectopia Lentis

**Congenital**
- Marfan
- Aniridia
- Ectopia lentis et pupillae

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

*In three words (not including a form of), what is Peters anomaly?*
A form of anterior segment dysgenesis
Simple ectopia lentis (late-onset type) is strongly associated with microspherophakia. With what conditions is microspherophakia occasionally associated?

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- Lowe syndrome
- Alport syndrome
- Marfan syndrome
- Peters anomaly

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

A neurocristopathy

What is a neurocristopathy?

A congenital/developmental abnormality owing to flawed neural-crest cell migration or differentiation

In three words (not including a form of), what is Peters anomaly?

A form of anterior segment dysgenesis
Simple ectopia lentis (late-onset type)

Weill-Marchesani is strongly associated with microspherophakia. With what conditions is microspherophakia occasionally associated?

- Congenital rubella
- Lowe syndrome
- Alport syndrome
- Marfan syndrome
- Peters anomaly

With what condition is microspherophakia most frequently associated?

Weill-Marchesani syndrome

In three words (not including a form of), what is Peters anomaly?

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

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What is a neurocristopathy?

A congenital/developmental abnormality owing to flawed neural-crest cell migration or differentiation
Weill-Marchesani syndrome is strongly associated with microspherophakia. With what conditions is microspherophakia occasionally associated?

- Congenital rubella
- Lowe syndrome
- Alport syndrome
- Marfan syndrome
- Peters anomaly

How does Peters anomaly present?

As a corneal opacity at birth (it’s in the STUMPED mnemonic). The opacity ranges in severity from a faint haze to an opaque, elevated and vascularized mess.
How does Peters anomaly present?

With a defect of the posterior central cornea, including the absence of Descemet’s and subjacent endothelium. Adhesions extending from the iris to the posterior corneal defect are often present.
1. Defect of the posterior central cornea, including the absence of Descemet’s and subjacent endothelium

2. Adhesions extending from the iris to the posterior corneal defect

Peters anomaly

Ectopia Lentis
Simple ectopia lentis (late-onset type) is strongly associated with microspherophakia. With what conditions is microspherophakia occasionally associated?

Congenital rubella, Lowe syndrome, Alport syndrome, Marfan syndrome and Peters anomaly.

With what condition is microspherophakia most frequently associated?

Weill-Marchesani syndrome.

How does Peters anomaly present?

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

What is the classic clinical scenario for Peters?

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

### Congenital
- Marfan
- Aniridia
- Ectopia lentis et pupillae

### Developmental
- Marfan
- Homocystinuria
- Microspherophakia

### Acquired
- Trauma
- (Pseudo)exfoliation syndrome

---

**How does Peters anomaly present?**
With a defect of the posterior central cornea, including the absence of Descemet’s and subjacent endothelium. Adhesions extending from the iris to the posterior corneal defect are often present.

**What is the classic clinical scenario for Peters?**
A corneal opacity noted at birth (it’s in the STUMPED mnemonic). The opacity ranges in severity from a faint haze to an opaque, elevated and vascularized mess.
Peters anomaly: Hazy cornea
Simple ectopia lentis (late-onset type) is strongly associated with microspherophakia. With what conditions is microspherophakia occasionally associated?

- Congenital rubella
- Lowe syndrome
- Alport syndrome
- Marfan syndrome
- Peters anomaly

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

- Sclerocornea
- Stromal dystrophy (CHSD)
- Turcot
- Usher syndrome
- Peters anomaly
- Elevated IOP (congenital glaucoma)

Note: There are two S’s and two E’s. What is the classic clinical scenario for Peters?

A corneal opacity noted at birth (it’s in the STUMPED mnemonic). The opacity ranges in severity from a faint haze to an opaque, elevated and vascularized mess.
Simple ectopia lentis (late-onset type) is strongly associated with microspherophakia. With what conditions is microspherophakia occasionally associated?

- Congenital rubella
- Lowe syndrome
- Alport syndrome
- Marfan syndrome
- Peters anomaly

What is the classic clinical scenario for Peters?

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

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

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

Note: There are two S’s and two E’s.
Why is the term ‘aniridia’ technically a misnomer?

Because a rudimentary iris root is always present

Is aniridia usually unilateral, or bilateral?

It is almost always bilateral

Is nystagmus commonly associated with aniridia?

Yes

With what developmental ‘complex’ is aniridia associated?

The WAGR complex

Are all aniridia cases at risk for WAGR complex?

No, only those in which the genetic mutation is sporadic
Ectopia Lentis

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

Congenital

- Marfan
- Aniridia
- Ectopia lentis
- Simple ectopia lentis (congenital type)
- Simple ectopia lentis (late-onset type)
- Ectopia lentis et pupillae
- Trauma
- Aniridia
- Buphthalmos 2o to congenital glaucoma
- Microspherophakia
- (Pseudo)exfoliation syndrome
- Marfan
- Simple ectopia lentis (congenital type)

Acquired
Ectopia Lentis

Aniridia. Note the presence of an iris stub/root
**Ectopia Lentis**

- **Congenital**
  - Marfan
  - Aniridia
  - Ectopia lentis
  - Simple ectopia lentis (congenital type)
  - Ectopia lentis et pupillae
  - Trauma
  - Homocystinuria
  - (Pseudo)exfoliation syndrome
  - Congenital glaucoma

- **Acquired**
  - Buphthalmos
  - Valsalva

---

**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.

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**Is nystagmus commonly associated with aniridia?**

Yes.

---

**With what developmental ‘complex’ is aniridia associated?**

The WAGR complex.

---

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

No, only those in which the genetic mutation is sporadic.
**Ectopia Lentis**

- **Congenital**
  - Marfan
  - Aniridia
    - Ectopia lentis
      - Simple ectopia lentis (congenital type)
    - (Pseudo)exfoliation syndrome
  - Trauma
    - Buphthalmos 2o to congenital glaucoma
- **Acquired**
  - Homocystinuria
  - Marfan
  - Simple ectopia lentis (late-onset type)

---

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

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

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

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

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

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?

Congenital
- Marfan
- Aniridia
- Ectopia lentis
  - Simple ectopia lentis (congenital type)

Acquired
- Trauma
- (Pseudo)exfoliation syndrome

Marfan

Aniridia

Ectopia lentis
Ectopia Lentis

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

Congenital

Marfan

Aniridia

Ectopia lentis

Simple ectopia lentis (congenital type)

Acquired

Trauma

(pseudo)exfoliation syndrome

Marfan

Homocystinuria

Simple ectopia lentis (late-onset type)
Ectopia Lentis

- Congenital
  - Marfan
  - Aniridia
  - Ectopia lentis
    - Simple (congenital type)
  - Ectopia lentis et pupillae
  - Trauma
  - Aniridia
  - Buphthalmos
  - 2o to congenital glaucoma
  - Microspherophakia
  - (Pseudo)exfoliation syndrome

- Acquired

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 common with aniridia?
Yes

Is this a sensory or a motor nystagmus?
Sensory
Ectopia Lentis

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

Is aniridia usually unilateral, or bilateral?
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Is nystagmus commonly associated with aniridia?
Yes

Is this a sensory or a motor nystagmus?
Sensory
Ectopia Lentis

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 a sensory or a motor nystagmus?
Sensory

What anatomic abnormalities are responsible for the poor vision in aniridia?
(hint: It's not the lack of an iris)

Fovealand optic nerve hypoplasia
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 this a sensory or a motor nystagmus? Yes. Sensory.

What anatomic abnormalities are responsible for the poor vision in aniridia? (hint: It's not the lack of an iris) Foveal and optic nerve hypoplasia.
Ectopia Lentis

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

Congenital
- Marfan
- Aniridia
- Ectopia lentis
- Simple ectopia lentis (congenital type)
- Ectopia lentis et pupillae
- Trauma
- Aniridia
- Buphthalmos 2o to congenital glaucoma
- Microspherophakia
- (Pseudo)exfoliation syndrome

Acquired
- Homocystinuria

Marfan
**Ectopia Lentis**

### Congenital

- **Marfan**
- **Aniridia**
  - Ectopia lentis
  - Simple ectopia lentis (congenital type)

### Acquired

- Trauma
- (pseudo)exfoliation syndrome

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

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Yes.

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The WAGR complex.
**Ectopia Lentis**

- **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** consists of:
  - W: **W** (Werner syndrome)
  - A: **A** (Aniridia)
  - G: **G** (Genitourinary abnormalities)
  - R: **R** (Retroperitoneal fibrosis)

**WAGR complex**

- **Congenital**
  - Marfan
  - Aniridia
  - Ectopia lentis
  - Simple ectopia lentis (congenital type)

- **Acquired**
  - Trauma
  - (Pseudo)exfoliation syndrome

- **Marfan**
Ectopia Lentis

**Congenital**
- Marfan
- Aniridia
- Ectopia lentis
- Simple ectopia lentis (congenital type)

**Acquired**
- Trauma
- Aniridia
- Buphthalmos
to congenital glaucoma
- Microspherophakia
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**Is nystagmus commonly associated with aniridia?**
Yes.

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

(WAGR complex)
Ectopia Lentis

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

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

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

Congenital
- Marfan
- Aniridia
- Ectopia lentis
- Simple ectopia lentis (congenital type)

Acquired
- Trauma
- (pseudo)exfoliation syndrome

Q
Ectopia Lentis

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

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

Congenital
- Marfan
- Aniridia
- Ectopia lentis
- Simple ectopia lentis (congenital type)

Acquired
- Trauma
- (pseudo)exfoliation syndrome

Marfan
- Homocystinuria
- Simple ectopia lentis (late-onset type)
**WAGR complex: Wilm’s tumor**

**Ectopia Lentis**
**Ectopia Lentis**

- **Congenital**
  - Marfan
  - Aniridia
  - Ectopia lentis
    - Simple ectopia lentis (congenital type)

- **Acquired**
  - Trauma
  - Homocystinuria
  - Simple ectopia lentis (late-onset type)
  - (pseudo)exfoliation syndrome

---

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Yes.

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

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

**Ectopia Lentis**

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

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

*Is nystagmus commonly associated with aniridia?*
Yes

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

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

---

**Congenital**

- Marfan
- Aniridia
  - Ectopia lentis
  - Simple ectopia lentis (congenital type)

**Acquired**

- Trauma
- (pseudo)exfoliation syndrome

**Marfan**

**Homocystinuria**

**Simple ectopia lentis** (congenital type)
Ectopia Lentis

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

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

Is nystagmus commonly associated with aniridia?
Yes

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

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

**Congenital**

- Marfan
- **Aniridia**
- Ectopia lentis
- Simple ectopia lentis (congenital type)

**Acquired**

- Trauma
- Homocystinuria
- Ectopia lentis et pupillae
- Microspherophakia
- (Pseudo)exfoliation syndrome
- Marfan
- Simple ectopia lentis (late-onset type)

For more info on aniridia and/or the WAGR complex, see slide-set P17
Nystagmus

Ectopia Lentis

We mentioned that aniridia is associated with nystagmus...

One final point regarding aniridia:
Nystagmus

...but you need to know the other eye findings associated with it, namely:

One final point regarding aniridia:

Ectopia Lentis

(Hints forthcoming)
Nystagmus

**Ectopia Lentis**

A corneal issue

Hypoplasia of two structures

Angle-related condition

Anterior seg issue

**Q/A**

...but you need to know the other eye findings associated with it, namely:

One final point regarding aniridia:
Nystagmus

- True

Aniridia is associated with limbal stem cell deficiency
- True

- Presents unilaterally and bilaterally in roughly equal rates  False; it is almost always bilateral

- The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris root is present  False; it’s a misnomer because a rudimentary iris root is always present

- Aniridia is strongly associated with foveal and optic nerve hypoplasia  True

- Patients complain of (and infants suffer from) photophobia  True

- Familial cases are at risk for Wilms tumor  False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex

- Aniridia is associated with glaucoma  True

- Aniridia is associated with early-onset cataracts  True

---

The takeaway point: Don’t think of aniridia as an iris condition! The BCSC characterizes it as a panophthalmic disorder.

One final point regarding aniridia:

...but you need to know the other eye findings associated with it, namely:

- Ectopia Lentis
- Foveal and optic nerve hypoplasia
- Glaucoma
- Cataracts

---
Nystagmus

limbal stem cell deficiency

Ectopia Lentis

The takeaway point: Don’t think of aniridia as an iris condition! The BCSC characterizes it as a panophthalmic disorder

One final point regarding aniridia:

- glaucoma
- cataracts
- foveal and optic nerve hypoplasia

Aniridia is strongly associated with foveal and optic nerve hypoplasia

True

Patients complain of (and infants suffer from) photophobia

True

Familial cases are at risk for Wilms tumor

False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex

Aniridia is associated with glaucoma

True

Aniridia is associated with early-onset cataracts

True
What is simple ectopia lentis?

- Simple ectopia lentis (congenital type)
- Simple ectopia lentis (late-onset type)
**What is simple ectopia lentis?**
A genetic condition the hallmark of which is subluxation of the lens.
Ectopia Lentis

**What is simple ectopia lentis?**
A genetic condition the hallmark of which is subluxation of the lens.

**How common is it?**

---

**Congenital**
- Marfan
- Aniridia
- Ectopia lentis et pupillae
  - Simple ectopia lentis (congenital type)

---

- Simple ectopia lentis (late-onset type)

---

congenital glaucoma
What is simple ectopia lentis?
A genetic condition the hallmark of which is subluxation of the lens

How common is it?
It is uncommon

**Congenital**
- Marfan
- Aniridia
- Ectopia lentis et pupillae
  - Simple ectopia lentis (congenital type)
- Simple ectopia lentis (late-onset type)
  - Congenital glaucoma

**Ectopia Lentis**
What is simple ectopia lentis?
A genetic condition the hallmark of which is subluxation of the lens

How common is it?
It is uncommon

Is it unilateral, or bilateral?

Congenital
- Marfan
- Aniridia
- Ectopia lentis et pupillae
- Simple ectopia lentis (congenital type)

Late-onset type
- Simple ectopia lentis (late-onset type)
What is simple ectopia lentis?
A genetic condition the hallmark of which is subluxation of the lens

How common is it?
It is uncommon

Is it unilateral, or bilateral?
Bilateral

Congenital
- Marfan
- Aniridia
- Ectopia lentis et pupillae
- Simple ectopia lentis (congenital type)

Late-onset
- Simple ectopia lentis (late-onset type)
What is simple ectopia lentis?
A genetic condition the hallmark of which is subluxation of the lens

How common is it?
It is uncommon

Is it unilateral, or bilateral?
Bilateral

In what direction are the lenses displaced?
Superotemporal

What is Ectopia Lentis?
What is simple ectopia lentis?
A genetic condition the hallmark of which is subluxation of the lens.

How common is it?
It is uncommon.

Is it unilateral, or bilateral?
Bilateral.

In what direction are the lenses displaced?
Superotemporal.

Simple ectopia lentis (congenital type)
What is simple ectopia lentis?
A genetic condition the hallmark of which is subluxation of the lens

How common is it?
It is uncommon

Is it unilateral, or bilateral?
Bilateral

In what direction are the lenses displaced?
Superotemporal

Assuming the age-of-onset for the congenital type is birth [ed.note: it is], when does lens displacement occur in the late-onset type?

Simple ectopia lentis (late-onset type)
**Ectopia Lentis**

- **What is simple ectopia lentis?**
  A genetic condition the hallmark of which is subluxation of the lens.

- **How common is it?**
  It is uncommon.

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

- **In what direction are the lenses displaced?**
  Superotemporal.

**Congenital**

- Marfan
- Aniridia
- Ectopia lentis et pupillae
- Simple ectopia lentis (congenital type)

**Late-Onset**

- Simple ectopia lentis (late-onset type)

Assuming the age-of-onset for the congenital type is birth [ed.note: it is], when does lens displacement occur in the late-onset type? After age 20 years.
What is ectopia lentis et pupillae?

Ectopia lentis et pupillae is a genetic condition whose hallmark is the displacement of the pupil and (microspherophakic) lens. It is very rare. The pupils are bilateral and typically have two further abnormalities:

- They are very miotic, and dilate poorly.
- They are slit-like in shape.

In what direction are the pupils and lenses displaced? The pupils are displaced inferotemporally, and the lenses are displaced superonasally.
What is ectopia lentis et pupillae?
A genetic condition the hallmark of which is the displacement of the pupil and (microspherophakic) lens.

**Congenital**
- Marfan
- Aniridia
- Simple ectopia lentis (congenital type)

**Ectopia lentis et pupillae**
- Microspherophakia
- Buphthalmos 2° to congenital glaucoma
- Simple ectopia lentis (late-onset type)
What is ectopia lentis et pupillae?
A genetic condition the hallmark of which is the displacement of the pupil and (microspherophakic) lens

How common is it?

Congenital
- Marfan
- Aniridia
- Ectopia lentis et pupillae
- Simple ectopia lentis (congenital type)

Bilateral

Ectopia lentis et pupillae

Microspherophakia
- Buphthalmos 2° to congenital glaucoma
- Simple ectopia lentis (late-onset type)
What is ectopia lentis et pupillae?
A genetic condition the hallmark of which is the displacement of the pupil and (microspherophakic) lens

How common is it?
It is very rare

Congenital
- Marfan
- Aniridia
- Ectopia lentis et pupillae
  - Simple ectopia lentis (congenital type)

Ectopia lentis et pupillae

Microspherophakia
- Buphthalmos 2o to congenital glaucoma
- Simple ectopia lentis (late-onset type)
What is ectopia lentis et pupillae?
A genetic condition the hallmark of which is the displacement of the pupil and (microspherophakic) lens

How common is it?
It is very rare

Is it unilateral, or bilateral?

Bilateral
What is ectopia lentis et pupillae?
A genetic condition the hallmark of which is the displacement of the pupil and (microspherophakic) lens

How common is it?
It is very rare

Is it unilateral, or bilateral?
Bilateral

**Congenital**
- Marfan
- Aniridia
  - Ectopia lentis et pupillae
  - Simple ectopia lentis (congenital type)

**Acquired**
- Marfan
- Aniridia
  - Simple ectopia lentis (late-onset type)
  - Microspherophakia
  - Buphthalmos 2° to congenital glaucoma
  - Ectopia lentis et pupillae
  - Homocystinuria

- Simple ectopia lentis
- Trauma
- Aniridia
- Buphthalmos 2° to congenital glaucoma
- Ectopia lentis et pupillae
- Homocystinuria

The pupils typically have two further abnormalities—what are they?
- They are very miotic, and dilate poorly
- They are slit-like in shape

In what direction are the pupils and lenses displaced?
In opposite directions—pupils inferotemporal, lenses superonasal
**What is ectopia lentis et pupillae?**
A genetic condition the hallmark of which is the displacement of the pupil and (microspherophakic) lens.

**How common is it?**
It is very rare.

**Is it unilateral, or bilateral?**
Bilateral.

**The pupils typically have two further abnormalities—what are they?**
--?
--?

**Congenital**

- Marfan
- Aniridia
- **Ectopia lentis et pupillae**
- Simple ectopia lentis (congenital type)
- Microspherophakia
- Buphthalmos 2° to congenital glaucoma
- Simple ectopia lentis (late-onset type)
**What is ectopia lentis et pupillae?**
A genetic condition the hallmark of which is the displacement of the pupil and (microspherophakic) lens.

**How common is it?**
It is very rare.

**Is it unilateral, or bilateral?**
Bilateral.

**The pupils typically have two further abnormalities—what are they?**
- They are very miotic and dilate poorly.

**In what direction are the pupils and lenses displaced?**
In opposite directions—pupils inferotemporal, lenses superonasal.
What is ectopia lentis et pupillae?
A genetic condition the hallmark of which is the displacement of the pupil and (microspherophakic) lens.

How common is it?
It is very rare.

Is it unilateral, or bilateral?
Bilateral.

The pupils typically have two further abnormalities—what are they?
--They are very miotic, and dilate poorly.
--?
Ectopia lentis et pupillae. Note the tee-tiny pupils (ignore the LPIs).
**Q**

What is ectopia lentis et pupillae?
A genetic condition the hallmark of which is the displacement of the pupil and (microspherophakic) lens.

How common is it?
It is very rare.

Is it unilateral, or bilateral?
Bilateral.

The pupils typically have two further abnormalities—what are they?
--They are very miotic, and dilate poorly.
--They are slit-like in shape.

**Congenital**

- Marfan
- Aniridia
- **Ectopia lentis et pupillae**
  - Simple ectopia lentis (congenital type)
- Microspherophakia
- Buphthalmos 2° to congenital glaucoma
- Simple ectopia lentis (late-onset type)
What is ectopia lentis et pupillae?
A genetic condition the hallmark of which is the displacement of the pupil and (microspherophakic) lens

How common is it?
It is very rare

Is it unilateral, or bilateral?
Bilateral

The pupils typically have two further abnormalities—what are they?
-- They are very miotic, and dilate poorly
-- They are slit-like in shape

Congenital
- Marfan
- Aniridia
- Simple ectopia lentis (congenital type)

Ectopia lentis et pupillae
- Microspherophakia
- Buphthalmos 2o to congenital glaucoma
- Simple ectopia lentis (late-onset type)
Ectopia lentis et pupillae: Bilateral inferonasal displacement of slit(ish) pupils
What is ectopia lentis et pupillae?
A genetic condition the hallmark of which is the displacement of the pupil and (microspherophakic) lens

How common is it?
It is very rare

Is it unilateral, or bilateral?
Bilateral

The pupils typically have two further abnormalities—what are they?
--They are very miotic, and dilate poorly
--They are slit-like in shape

In what direction are the pupils and lenses displaced?

Congenital
- Marfan
- Aniridia
- Simple ectopia lentis (congenital type)

Ectopia lentis et pupillae

Microspherophakia
- Buphthalmos 2° to congenital glaucoma
- Simple ectopia lentis (late-onset type)
**Q/A**

**What is ectopia lentis et pupillae?**
A genetic condition the hallmark of which is the displacement of the pupil and (microspherophakic) lens

**How common is it?**
It is very rare

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

**The pupils typically have two further abnormalities—what are they?**
--They are very miotic, and dilate poorly
--They are slit-like in shape

**In what direction are the pupils and lenses displaced?**
In opposite directions—pupils superonasal, lenses superonasal
**What is ectopia lentis et pupillae?**
A genetic condition the hallmark of which is the displacement of the pupil and (microspherophakic) lens.

**How common is it?**
It is very rare.

**Is it unilateral, or bilateral?**
Bilateral.

**The pupils typically have two further abnormalities—what are they?**
--They are very miotic, and dilate poorly
--They are slit-like in shape

**In what direction are the pupils and lenses displaced?**
In opposite directions—pupils inferotemporal, lenses superonasal.

---

**Congenital**
- Marfan
- Aniridia
- **Ectopia lentis et pupillae**
- Simple ectopia lentis (congenital type)
- Microspherophakia
- Buphthalmos 2° to congenital glaucoma
- Simple ectopia lentis (late-onset type)
Ectopia lentis et pupillae: Pupil displaced inferonasal; lens, superotemporal
**What is ectopia lentis et pupillae?**
A genetic condition the hallmark of which is the displacement of the pupil and (microspherophakic) lens

**How common is it?**
It is very rare

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

The pupils typically have two further abnormalities—what are they?
--They are very miotic, and dilate poorly
--They are slit-like in shape

In what direction are the pupils and lenses displaced?
In opposite directions—pupils inferotemporal, lenses superonasal

Can the pupils and lenses be so displaced that the lens isn’t located within the pupillary aperture?

In opposite directions—pupils inferotemporal, lenses superonasal
What is ectopia lentis et pupillae? 
A genetic condition the hallmark of which is the displacement of the pupil and (microspherophakic) lens

How common is it? 
It is very rare

Is it unilateral, or bilateral? 
Bilateral

The pupils typically have two further abnormalities—what are they? 
--They are very miotic, and dilate poorly 
--They are slit-like in shape

In what direction are the pupils and lenses displaced? 
In opposite directions—pupils inferotemporal, lenses superonasal

Can the pupils and lenses be so displaced that the lens isn’t located within the pupillary aperture? 
They can indeed

In opposite directions—pupils inferotemporal, lenses superonasal

Congenital
- Marfan
- Aniridia
- Ectopia lentis et pupillae
  - Simple ectopia lentis (congenital type)
- Microspherophakia
  - Buphthalmos 2° to congenital glaucoma
  - Simple ectopia lentis (late-onset type)
**What is ectopia lentis et pupillae?**
A genetic condition the hallmark of which is the displacement of the pupil and (microspherophakic) lens

**How common is it?**
It is very rare

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

Next, let’s review the direction of displacement for several causes of ectopia lentis

In what direction are the pupils and lenses displaced?
In opposite directions—pupils inferotemporal, lenses superonasal

- **Microspherophakia**
- **Buphthalmos 2° to congenital glaucoma**
- **Simple ectopia lentis (late-onset type)**
Ectopia Lentis

Lens subluxation: Which conditions are associated with displacement in which directions?
Lens subluxation: Which conditions are associated with displacement in which directions?

Marfan; Simple ectopia lentis

Ectopia Lentis

Marfan; Simple ectopia lentis
Lens subluxation: Which conditions are associated with displacement in which directions?
Lens subluxation: Which conditions are associated with displacement in which directions?

*Marfan; Simple ectopia lentis*

*Ectopia lentis et pupillae*

*Marfan; Simple ectopia lentis*
Lens subluxation: Which conditions are associated with displacement in which directions?

Marfan; Simple ectopia lentis

Ectopia lentis et pupillae

Marfan; Simple ectopia lentis

(one)
Lens subluxation: Which conditions are associated with displacement in which directions?

- Marfan; Simple ectopia lentis
- Ectopia lentis et pupillae
- Homocystinuria
- Marfan; Simple ectopia lentis
Lens subluxation: Which conditions are associated with displacement in which directions?

- Marfan; Simple ectopia lentis
- Ectopia lentis et pupillae
- Homocystinuria
- (?)

- (?)
- Marfan; Simple ectopia lentis
Lens subluxation: Which conditions are associated with displacement in which directions?

- **Marfan; Simple ectopia lentis**
- **Ectopia lentis et pupillae**
- **Homocystinuria**
- **Marfan; Simple ectopia lentis**

(noting I know of)