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(ated):
--Lux(ated):
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(ated): The lens is partially displaced, but remains in the ‘general area’
--Lux(ated):
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(ated)**: The lens is partially displaced, but remains in the ‘general area’
-- **Lux(ated)**: The lens is fully displaced from the ‘general area’

**What iris finding is classic for a subluxed lens?**
What is ectopia lentis?
Displacement of the lens from its normal anatomic position

With regard to lens 'displacement':
-- **Sublux(at)ed**: The lens is partially displaced, but remains in the 'general area'
-- **Lux(at)ed**: The lens is displaced out of the normal anatomic position

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(ated): 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 iridodonesis?
What is ectopia lentis?
Displacement of the lens from its normal anatomic position

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What iris finding is classic for a subluxed lens?
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What is iridodonesis?
A ‘tremulous’ iris
What is ectopia lentis?
Displacement of the lens from its normal anatomic position

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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(ated): The lens is partially displaced, but remains in the ‘general area’
--Lux(ated): 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

Luxated 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’

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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?
Ectopia Lentis

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

What are the four **congenital** causes covered in the BCSC?
What are the five developmental causes covered in the BCSC?
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?
Which is the most common heritable cause of ectopia lentis?
Which is the most common heritable cause of ectopia lentis? Marfan
By the way—why does Marfan appear under both Congenital and Developmental headings?

- Simple ectopia lentis (congenital type)
- Buphthalmos 2° to congenital glaucoma
- Simple ectopia lentis (late-onset type)
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
  - Simple ectopia lentis
- Developmental
  - Marfan
  - Ectopia lentis et pupillae
- Acquired
  - Trauma
  - (Pseudo)exfoliation syndrome

What protein is abnormal in Marfan’s?

(late-onset type)
Ectopia Lentis

Congenital
- Marfan

Developmental
- Marfan

Acquired
- Trauma
- (Pseudo)exfoliation syndrome

What protein is abnormal in Marfan’s? Fibrillin

(late-onset type)
Ectopia Lentis

**Congenital**
- Marfan

**Developmental**
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- Trauma
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**What protein is abnormal in Marfan?**
- Fibrillin

**What is the inheritance pattern?**
- AD, although family history is negative in 15% of cases

(late-onset type)
Q/A

Ectopia Lentis

Congenital
- Marfan

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What protein is abnormal in Marfan's?
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(late-onset type)
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(late-onset type)
What protein is abnormal in Marfan’s?
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What three structures/systems manifest abnormalities in Marfan’s?
-- The eye (duh)
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-- (late-onset type)
Ectopia Lentis

**Congenital**
- Marfan

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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

(late-onset type)
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  - Marfan

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--- The musculoskeletal

What cardiovascular abnormalities are common?
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What three structures/systems manifest abnormalities in Marfans?
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**Ectopia Lentis**

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

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Are these abnormalities clinically significant?

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

**Congenital**
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**How significant is the lifespan shortening?**

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Q/A

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

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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

Marfan syndrome: Aortic dissection
Ectopia Lentis

- Congenital
  - Marfan
- Developmental
  - Marfan
- Acquired
  - Trauma
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What protein is abnormal in Marfan's?
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What three structures/systems manifest abnormalities in Marfan’s?
- The eye (duh)
- The cardiovascular
- The musculoskeletal

What musculoskeletal abnormalities are common?
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- ?
- ?
What protein is abnormal in Marfan's?  
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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)
Ectopia Lentis

- **Congenital**
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Ectopia Lentis

Marfan syndrome: Arachnodactyly
Ectopia Lentis

Marfan syndrome: Hypermobile joints
Ectopia Lentis

Marfan syndrome: Pectus excavatum
**Ectopia Lentis**

**Congenital**
- Marfan

**Developmental**
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- (Pseudo)exfoliation syndrome

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

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

Other than ectopia lentis, what ocular structural abnormalities are often present?
- Microspherophakia
- Increased axial length
What protein is abnormal in Marfan's? Fibrillin

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- The cardiovascular
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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
- Increased axial length

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

**Congenital**
- Marfan

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

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

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

- Congenital
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What protein is abnormal in Marfan's?
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--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%
What protein is abnormal in Marfan's? Fibrillin

What three systems manifest abnormalities in Marfan's?
- The eye (duh)
- The cardiovascular system
- The musculoskeletal system

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

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

What percentage of Marfan patients develop ectopia lentis? 50 - 80%
Marfan syndrome: Ectopia lentis
Marfan pts tend to be high myopes. Give three reasons why.
--
--
--

What protein is abnormal in Marfan?
Fibrillin

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

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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
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

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

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.
**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

I made this term up; a better name is **spherical-aberration** myopia

---

**Congenital**

<table>
<thead>
<tr>
<th>Marfan</th>
</tr>
</thead>
</table>

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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**Ectopia Lentis**

Marfan pts tend to be high myopes. Give three reasons why.
- Increased axial length → axial myopia
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--Increased corneal curvature?

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

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.

---

**Congenital**

Marfan

What protein is abnormal in Marfan?
Fibrillin

What three systems manifest abnormalities in Marfan?
--The eye (due to ectopia lentis)
--The cardiovascular
--The musculoskeletal

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

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Increased corneal curvature?

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.

Marfan patients c/o difficulty reading—why?
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What proportion of Marfan pts manifest ocular abnormalities?
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Ectopia Lentis

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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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Is the cornea flatter because it has a smaller-than-normal diameter (ie, microcornea)?
No—in fact, the cornea in Marfan tends to be flatter than normal.

Marfan pts tend to be high myopes. Give three reasons why.
--Increased axial length → axial myopia
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**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
- Increased corneal curvature? No!

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

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)

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

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

**What three structures/systems manifest abnormalities in Marfan's?**
- The eye (due to increased axial length, microspherophakia, and ectopia lentis)
- The cardiovascular system
- The musculoskeletal system

**What proportion of Marfan patients 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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A spectacle-corrected Marfan patient c/o difficulty reading—why?

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

**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%

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- Increased axial length

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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.
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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?*

---

**Congenital**

- **Marfan**

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

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- The cardiovascular
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**Q**
Ectopia Lentis

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

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

**Congenital**
- Marfan

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 Marfans?
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What three structures/systems manifest abnormalities in Marfans?
--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
Homocystinuria is due to faulty metabolism for what amino acid? (hint: it’s not cysteine)
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
Ectopia Lentis

Homocystinuria

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

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

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

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Ectopia Lentis

Homocystinuria
Ectopia Lentis

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What proportion of homocystinuria pts manifest ocular abnormalities?

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

**Congenital**
- Marfan
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**Homocystinuria**

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**Other than ectopia lentis, are other ocular structural abnormalities often present?**
Ectopia Lentis

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Developmental
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--The CNS

Other than ectopia lentis, are other ocular structural abnormalities often present?
Nah—ectopia lentis is pretty much it
Ectopia Lentis

Homocystinuria: Ectopia lentis
Ectopia Lentis

**Congenital**
- Marfan
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During which stage of life does subluxation typically occur?
Childhood (in most cases prior to age 10 years)
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What proportion of homocystinuria pts manifest ocular abnormalities?
About 80%

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

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

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

End-stage glaucoma in a homocystinuria pt with ectopia lentis
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For more on secondary angle-closure glaucoma stemming from ectopia lentis, see slide-set G16

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

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

What cardiovascular abnormalities are common?
--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 Lentis

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What relatively common medical event greatly increases the risk of a thromboembolic event?
General anesthesia. It is vital to identify homocystinurics prior to surgery!
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--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

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--**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

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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?
Crazy early. Osteoporotic changes are common in childhood (and have been reported in toddlers)

What is it?
Early osteoporosis
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)
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-- The eye (duh)
-- The cardiovascular
-- The musculoskeletal
-- The CNS

What CNS abnormalities are common?
--
--
--
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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
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

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

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 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.
Ectopia Lentis

Marfan syndrome

Homocystinuria

(Start here, work down the list)

Divvy up the characteristics. Some go both ways.

↑ risk thromboembolism

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
Q/A

**Ectopia Lentis**

**Marfan syndrome**

**Homocystinuria**

↑ risk *thromboembolism*

- 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

Do these simultaneously
**Q/A**

**Marfan syndrome**

- Associated with cataracts
- Zonules stretched
- Zonules broken
- Associated with microspherophakia
- Ectopia usually bilateral and symmetric
- Chest-wall deformities

**Homocystinuria**

- Associated with microspherophakia
- ↑ risk thromboembolism

**Ectopia Lentis**

- Lens subluxes up
- Lens subluxes down
- Tall
- Thin habitus
- Fair-haired
- Ectopia usually bilateral and symmetric
- Chest-wall deformities
Ectopia Lentis

Marfan syndrome

Homocystinuria

↑ risk thromboembolism

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Lens subluxes down
**Q/A**

**Ectopia Lentis**

**Marfan syndrome**

**Homocystinuria**

- **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**

**Marfan syndrome**

**Homocystinuria**

↑ risk **thromboembolism**

Lens subluxes *up*

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

Lens subluxes *down*

- Tall
- Thin habitus
- Fair-haired
- Zonules *broken*
Q/A

**Marfan syndrome**

- Associated with cataracts
- Lens subluxes *up*
- Tall
- Thin habitus
- Zonules *stretched*

**Homocystinuria**

- Associated with microspherophakia
- Ectopia usually bilateral and symmetric
- Chest-wall deformities
- Hiring thromboembolism

**Ectopia Lentis**

- Both
- Thromboembolism

- Associated with cataracts
- Lens subluxes *down*
- Tall
- Thin habitus
- Fair-haired
- Zonules *broken*
**Q/A**

**Ectopia Lentis**

**Marfan syndrome**

- Lens subluxes *up*
- Tall
- Thin habitus
- Zonules *stretched*
- Associated with microspherophakia
- Ectopia usually bilateral and symmetric
- Chest-wall deformities

**Homocystinuria**

- Lens subluxes *down*
- Tall
- Thin habitus
- Fair-haired
- Zonules *broken*
- Associated with microspherophakia
- ↑ risk *thromboembolism*

**Associated with cataracts**

- Lens subluxes
- Ectopia usually bilateral and symmetric
- Chest-wall deformities
Ectopia Lentis

Marfan syndrome

Homocystinuria

↑ risk thromboembolism

Lens subluxes up

Lens subluxes down

Tall

Thin habitus

Tall

Thin habitus

Fair-haired

Zonules stretched

Zonules broken

Associated with microspherophakia

Associated with microspherophakia

Ectopia usually bilateral and symmetric

Ectopia usually bilateral and symmetric

Chest-wall deformities
**Marfan syndrome**

- Lens subluxes **up**
- Tall
- Thin habitus
- Zonules **stretched**
- Associated with microspherophakia
- Ectopia usually bilateral and symmetric
- Chest-wall deformities

**Homocystinuria**

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

**Ectopia Lentis**
Ectopia Lentis

Marfan syndrome

- Associated with cataracts
- Lens subluxes \textit{up}
- Tall
- Thin habitus
- Fair-haired
- Zonules \textit{stretched}
- Associated with microspherophakia
- Ectopia usually bilateral and symmetric
- Chest-wall deformities

Homocystinuria

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

\textbf{Note the Key Differences!}
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?
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’).
Ectopia Lentis

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
Ectopia Lentis

Microspherophakia. With mydriasis, the lens is able to fit through the pupillary aperture
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?
Ectopia Lentis

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?
Ectopia Lentis

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

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

Simple ectopia lentis (late-onset type)
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

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?
Patients with Weill-Marchesani have:
…short stature
Ectopia Lentis

Weill-Marchesani syndrome: Short stature
**Q/A**

**Ectopia Lentis**

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

**Developmental**
- Marfan
- Homocystinuria
- Microspherophakia

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

---

**Weill-Marchesani syndrome**

*With what condition is microspherophakia most frequently associated?*

*What are the findings in Weill-Marchesani?*

Patients with Weill-Marchesani have:
- **short** stature
- **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**

*What are the findings in Weill-Marchesani?*

Patients with Weill-Marchesani have:

...short stature
...short fingers
Weill-Marchesani syndrome: Short fingers

Ectopia Lentis
Q/A

Ectopia Lentis

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

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

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

---

**Simple ectopia lentis**

---

**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
...short joints
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**

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

Patients with Weill-Marchesani have:

... **short** stature
... **short** fingers
... **stiff** joints

---
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?*

Patients with Weill-Marchesani have:
- **short** stature
- **short** fingers
- **stiff** joints

(Think of 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  (Tall stature)
- **short** fingers  (Long fingers)
- **stiff**  joints  (Lax joints)

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

Weill-Marchesani syndrome

Marfan 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

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

- Congenital
  - Marfan
  - Aniridia
  - Ectopia lentis et pupillae
  - Simple ectopia lentis
- Developmental
  - Marfan
  - Homocystinuria
- Acquired
  - Trauma
  - (Pseudo)exfoliation syndrome

Weill-Marchesani syndrome

Simple ectopia lentis

With what condition is microspherophakia most frequently associated?

Weill-Marchesani syndrome

Weill-Marchesani is strongly associated with microspherophakia. With what conditions is microspherophakia exceptionally 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**

---

**Microspherophakia**

**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

Congenital

Developmental

Acquired

Marfan

Marfan

Trauma

Weill-Marchesani syndrome

Lowe syndrome

Alport syndrome

Peters anomaly

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 and 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
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.

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

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
- Peters anomaly

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

Familial oculorenal syndromes

What is their classic (nonocular) presenting sign?

Lenticonus
Simple ectopia lentis (late-onset type)

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

With what condition is microspherophakia most frequently associated?

Weill-Marchesani syndrome

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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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?

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Lenticonus
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?

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

What conditions can microspherophakia occasionally be associated with?

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

Anterior lenticonus in Alport syndrome
Weill-Marchesani syndrome 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 (not including a form of), what is Peters anomaly? A form of anterior segment dystgenesis.
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 (not including a form of), what is Peters anomaly? A form of anterior segment dysgenesis

Weill-Marchesani syndrome is strongly associated with microspherophakia. 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
- 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

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

With what condition is microspherophakia most frequently associated?

- Weill-Marchesani syndrome

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

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

A neurocristopathy

What is 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) 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?

A form of anterior segment dysgenesis

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

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.
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

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

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?
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?
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

170 Q

Ectopia Lentis

Congenital
- Marfan
- Aniridia

Developmental
- Marfan

Acquired
- Trauma
- (Pseudo)exfoliation syndrome
- Buphthalmos due to congenital glaucoma
- Microspherophakia

What is the classic clinical scenario for Peters anomaly?

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)
- TUMP
- Peters anomaly
- Elevated IOP (congenital glaucoma)
- Dermoid of the cornea

Note: There are two S’s and two E’s.
Simple ectopia lentis

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

171

Ectopia Lentis

Congenital

- Marfan
- Aniridia

Developmental

- Marfan

Acquired

- Trauma

Ectopia lentis et pupillae

Trauma

Aniridia

Buphthalmos 20 to congenital glaucoma

Microspherophakia

(Pseudo)exfoliation syndrome

Marfan

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 (e.g., forceps 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)
- (Pseudo)exfoliation syndrome
- Trauma

**Acquired**
- Congenital glaucoma
- Aniridia
- Buphthalmos
  2° to congenital glaucoma
- Homocystinuria
- **Marfan**
- Simple ectopia lentis
  (late-onset type)
Aniridia. Note the presence of an iris stub/root
**Ectopia Lentis**

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

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

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

### Trauma
- (Pseudo)exfoliation syndrome
- Trauma
- Buphthalmos 2o to congenital glaucoma

### Acquired
- Homocystinuria
- Marfan

### Developed
- Ectopia Lentis
### Ectopia Lentis

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

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

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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.
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
- Simple ectopia lentis (late-onset type)

Ectopia Lentis
179

Ectopia Lentis

Congenital

- Simple ectopia lentis
- Marfan
- Aniridia

Acquired

- Trauma
- Homocystinuria
- Buphthalmos
- 2o to congenital glaucoma
- Microspherophakia
- Pseudoexfoliation syndrome

Simple ectopia lentis (congenital type)

Marfan

Aniridia

- 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 there nystagmus usually unilateral or bilateral?
  - Yes

- Is this a sensory or a motor nystagmus?
  - Sensory

- With what developmental 'complex' is aniridia usually 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)

**Marfan**

**Aniridia**

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

Is this a sensory or a motor nystagmus?
Sensory

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

**Congenital**
- Marfan
- Aniridia

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

**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.

**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.

**Is aniridia associated with a specific developmental complex?**
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.

Is this a sensory or a motor nystagmus?
Sensory.

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

(hint: It’s not the lack of an iris)
**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?**
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 Lentes**

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

**Acquired**
- Trauma
- Homocystinuria
- Buphthalmos 2o to congenital glaucoma
- Microspherophakia
- (Pseudo)exfoliation syndrome

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

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

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

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

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

WAGR complex

Marfan

Aniridia

Ectopia lentis et pupillae

Trauma

Aniridia

Buphthalmos 2o to congenital glaucoma

Microspherophakia

(Pseudo)exfoliation syndrome

Ectopia Lentis
**Ectopia Lentis**

**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
- 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.

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

**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.
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

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

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

- **Acquired**
  - Trauma
  - Homocystinuria
  - Buphthalmos 2o to congenital glaucoma
  - Microspherophakia
  - (Pseudo)exfoliation syndrome

- **Marfan**

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

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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** 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
Ectopia Lentis

WAGR complex: Wilm’s tumor
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
**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.
A \[ Ectopia \ Lentis \]

**Congenital**
- Marfan
- Aniridia
- Ectopia lentis
- 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

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

We mentioned that aniridia is associated with nystagmus…

One final point regarding aniridia:

Ectopia Lentis
Nystagmus

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

One final point regarding aniridia:

Ectopia Lentis

(Hints forthcoming)
Q/A

Nystgamus

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

One final point regarding aniridia:

Ectopia Lentis

A corneal issue

Hypoplasia of two structures

Angle-related condition

Anterior seg issue

196
Nystagmus

limbal stem cell deficiency

foveal and optic nerve hypoplasia

glaucoma

cataracts

Ectopia Lentis

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

One final point regarding aniridia:
Nystagmus

limbal stem cell deficiency

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

foveal and optic nerve hypoplasia

glaucoma

cataracts

One final point regarding aniridia:

Ectopia Lentis

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
Ectopia Lentis

What is simple ectopia lentis?

Congenital

- Marfan
- Aniridia
- Ectopia lentis et pupillae

Simple ectopia lentis (congenital 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

Congenital

- Marfan
- Aniridia
- Ectopia lentis et pupillae

- Simple ectopia lentis (congenital 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?**
Uncommon.

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

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

**Congenital glaucoma**
A genetic condition the hallmark of which is subluxation of the lens

How common is it?
It is uncommon
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)**

---

- **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

**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

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

**In what direction are the lenses displaced?**

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

**Ectopia Lentis**
- Trauma
- Aniridia
- Buphthalmos 2o to congenital glaucoma
- Microspherophakia
- (Pseudo)exfoliation syndrome
- Marfan
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)

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

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?
**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

*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

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

---

**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?**

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.
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)

What is it called?
Ectopia lentis et pupillae

How common is it?
Very rare

Is it unilateral, or bilateral?
Bilateral

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

**Congenital**

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

**Acquired**

- Simple ectopia lentis (late-onset type)
- Buphthalmos 2º to congenital glaucoma
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
- Simple ectopia lentis (congenital type)

Ectopia lentis et pupillae

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

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Bilateral

The pupils typically have two further abnormalities—what are they?
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--They are slit-like in shape
Ectopia Lentis

Ectopia lentis et pupillae
**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
**Q/A**

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

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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

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

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

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

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

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

Marfan; Simple ectopia lentis

Ectopia Lentis
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Marfan; Simple ectopia lentis

Ectopia lentis et pupillae

Homocystinuria

Marfan; Simple ectopia lentis
Ectopia lentis

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

Marfan; Simple ectopia lentis

Ectopia lentis et pupillae

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 (?)

 (?)
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

(nothing I know of)

(nothing I know of)