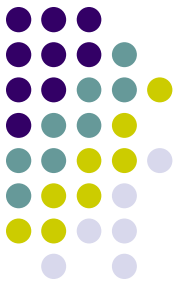


Q

## Ectopia Lentis

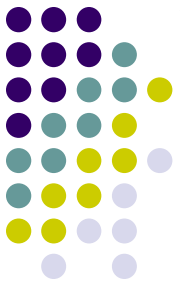
*What is ectopia lentis?*



# A

## Ectopia Lentis

2



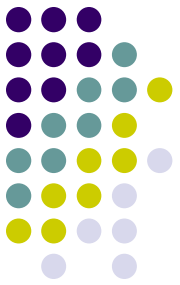
*What is* ectopia lentis?

Displacement of the lens from its normal anatomic position

# Q

## Ectopia Lentis

3



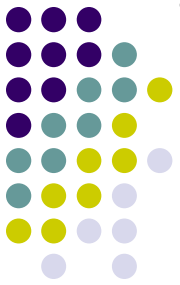
*What is ectopia lentis?*

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?*

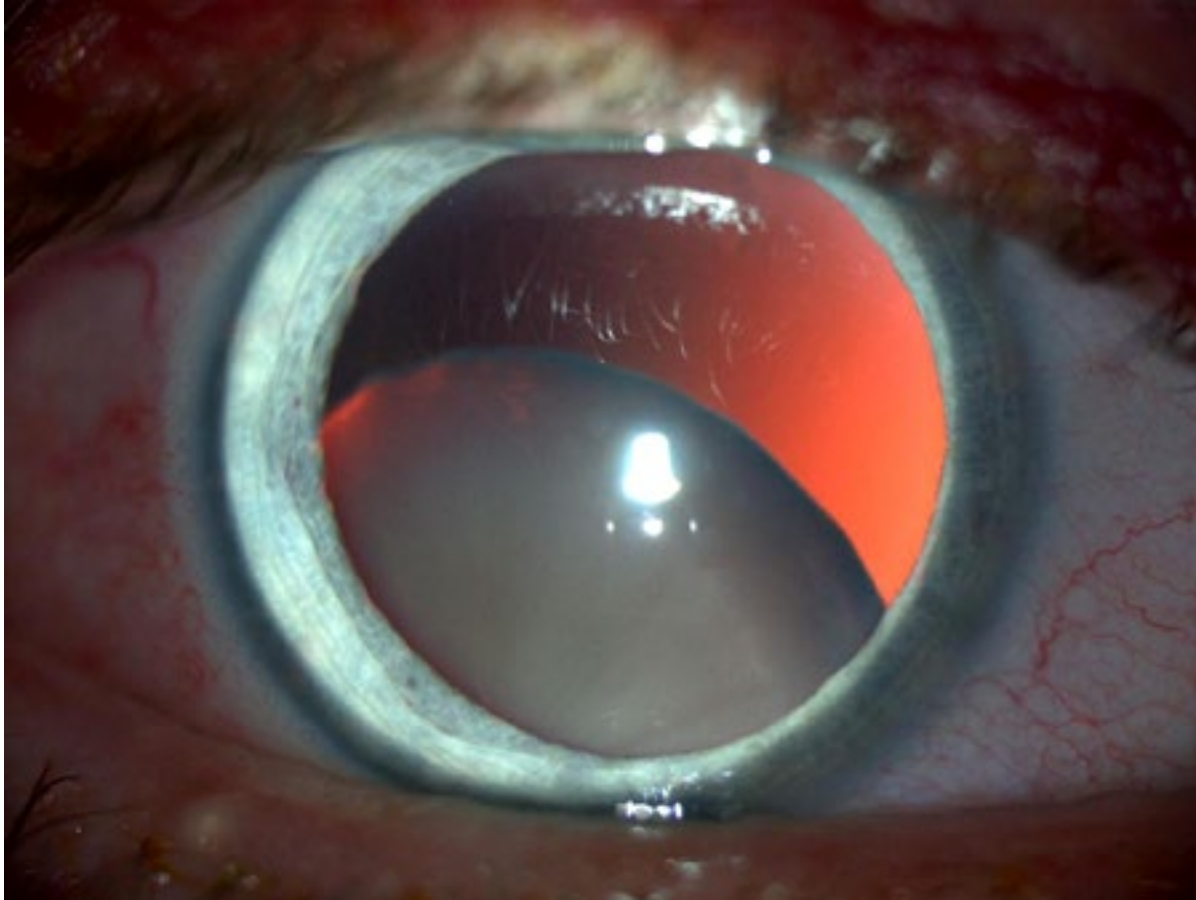
Displacement of the lens from its normal anatomic position

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

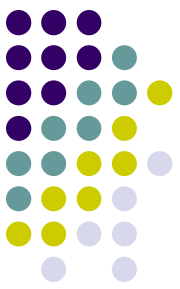
--*Sublux(at)ed*: The lens is partially displaced, but remains in the 'general area'

--*Lux(at)ed*:

# Ectopia Lentis

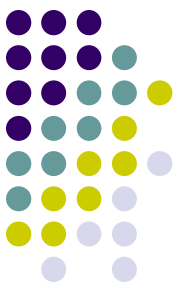


Subluxed lens



# Q

## Ectopia Lentis



*What is ectopia lentis?*

Displacement of the lens from its normal anatomic position

*With regard to lens ‘displacement’*

--**Sublux(at)ed**: The

--Lux(at)ed:

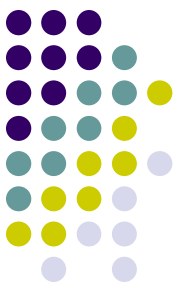
*What iris finding is classic for a subluxed lens?*

*mean?*

the ‘general area’

# A

## Ectopia Lentis



7

*What is ectopia lentis?*

Displacement of the lens from its normal anatomic position

*With regard to lens ‘displacement’*

--**Sublux(at)ed**: The

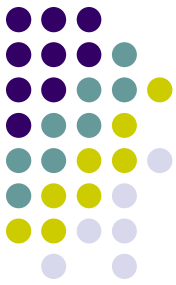
--**Lux(at)ed**:

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

Iridodonesis

*mean?*

the ‘general area’



*What is ectopia lentis?*

Displacement of the lens from its normal anatomic position

*With regard to lens ‘luxation’*

--**Sublux(at)ed**: The

--Lux(at)ed:

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

Iridodonesis

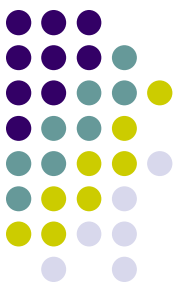
*What is iridodonesis?*

*mean?*

the ‘general area’

# A

## Ectopia Lentis



*What is ectopia lentis?*

Displacement of the lens from its normal anatomic position

*With regard to lens ‘*

--**Sublux(at)ed**: The

--**Lux(at)ed**:

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

Iridodonesis

*What is iridodonesis?*

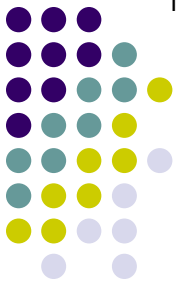
A ‘tremulous’ iris

*mean?*

the ‘general area’

# A

## Ectopia Lentis



*What is ectopia lentis?*

Displacement of the lens from its normal anatomic position

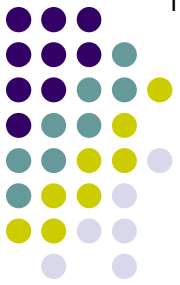
*With regard to lens 'displacement'—what do the following terms mean?*

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

--*Lux(at)ed*:

# A

## Ectopia Lentis



*What is ectopia lentis?*

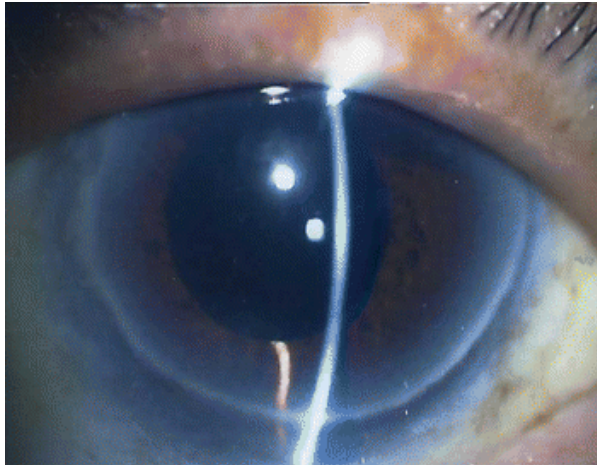
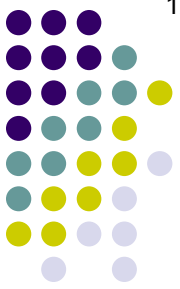
Displacement of the lens from its normal anatomic position

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

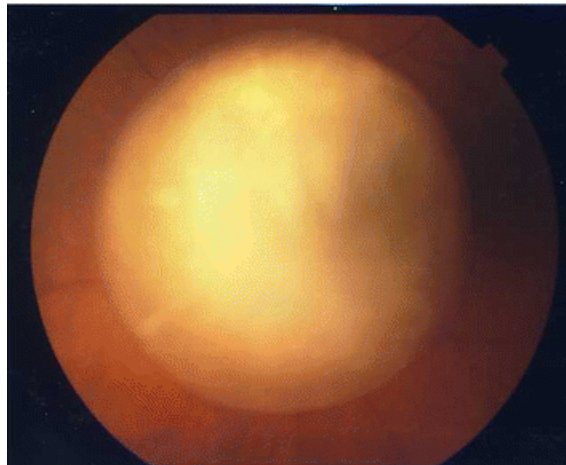
--*Sublux(at)ed*: The lens is partially displaced, but remains in the 'general area'

--*Lux(at)ed*: The lens is dislocated--completely removed from the pupillary aperture. All zonular attachments have been disrupted.

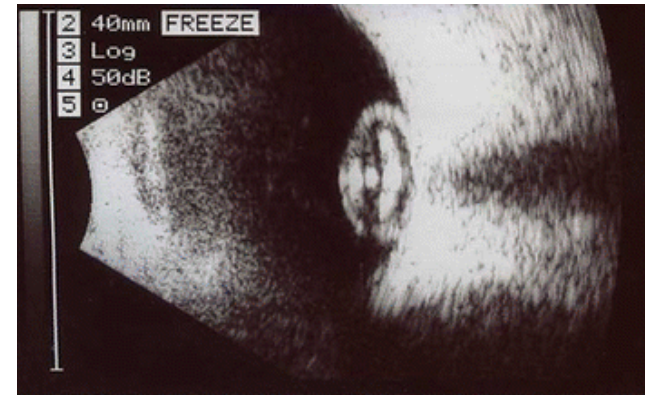
# Ectopia Lentis



Aphakic

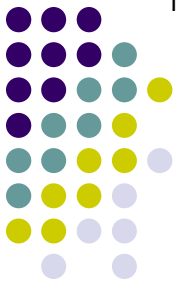


Lens resting on the retina



b-scan: lens on ONH

Luxated lens



## Ectopia Lentis

The *Peds* book also uses

*What is the term ectopic here*

Displacement of the lens from its normal anatomic position

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

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

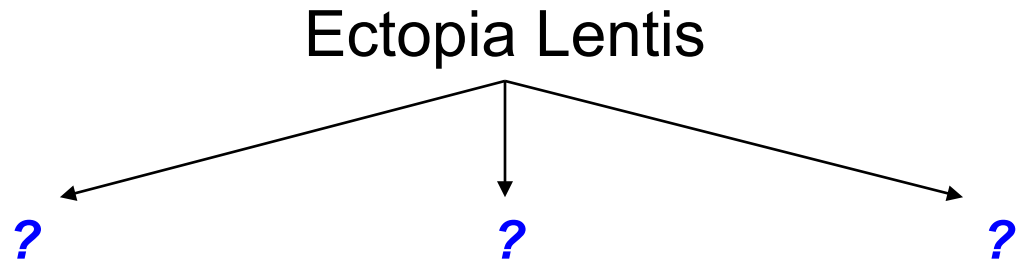
--***Lux(ate)d***: The lens is dislocated--completely removed from the pupillary aperture. All zonular attachments have been disrupted.

Q

## Ectopia Lentis



14

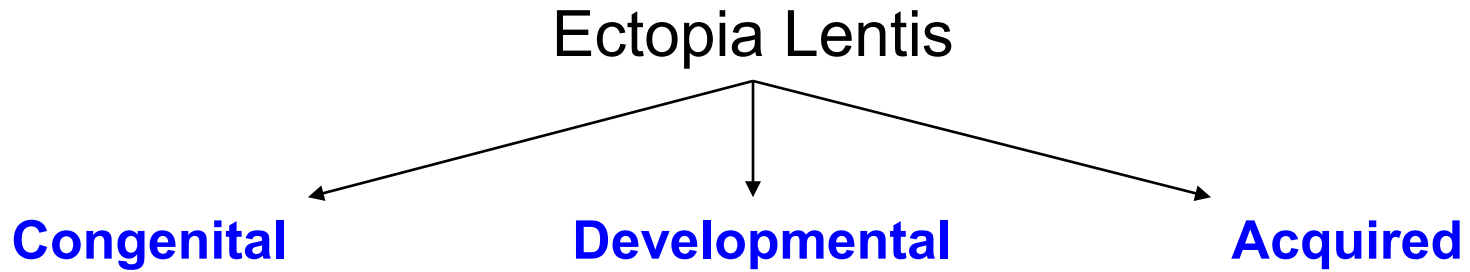
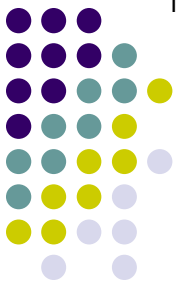


*The BCSC series divides ectopia lentis into three categories:*

# A

## Ectopia Lentis

15

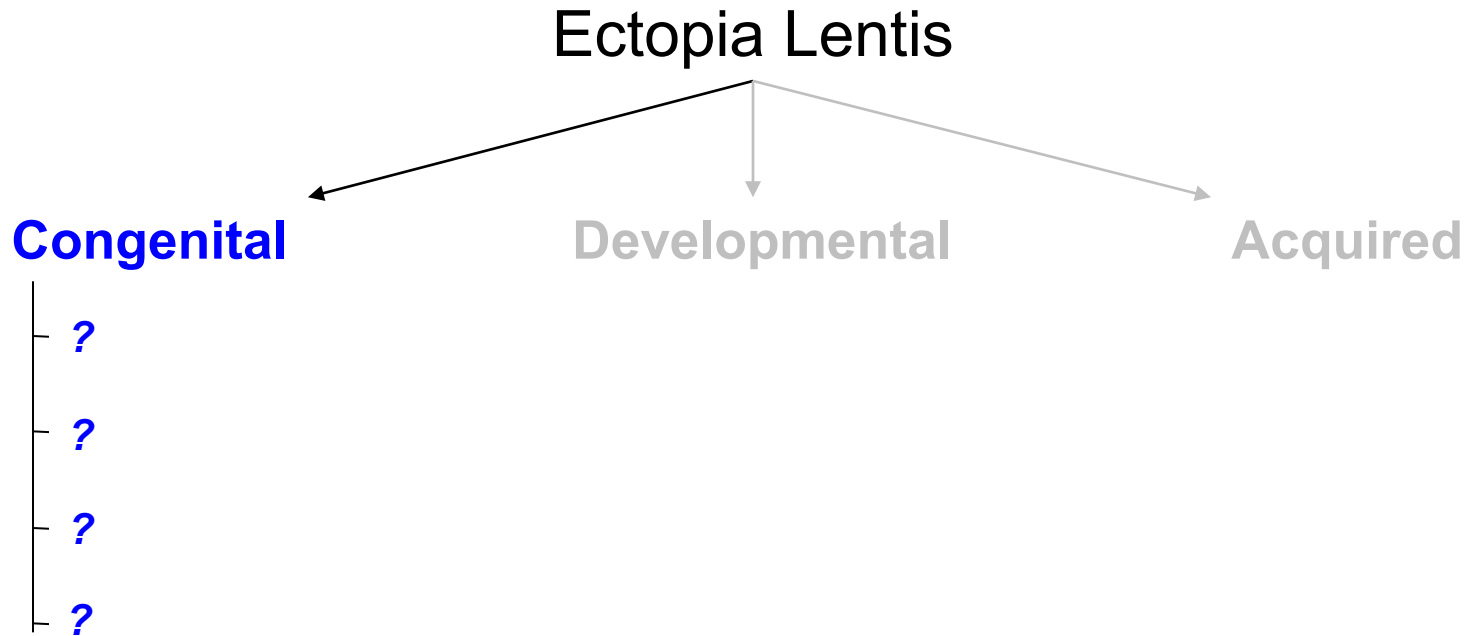
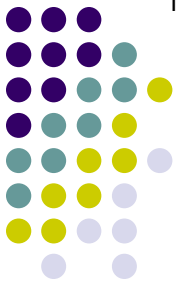


*The BCSC series divides ectopia lentis into three categories:*

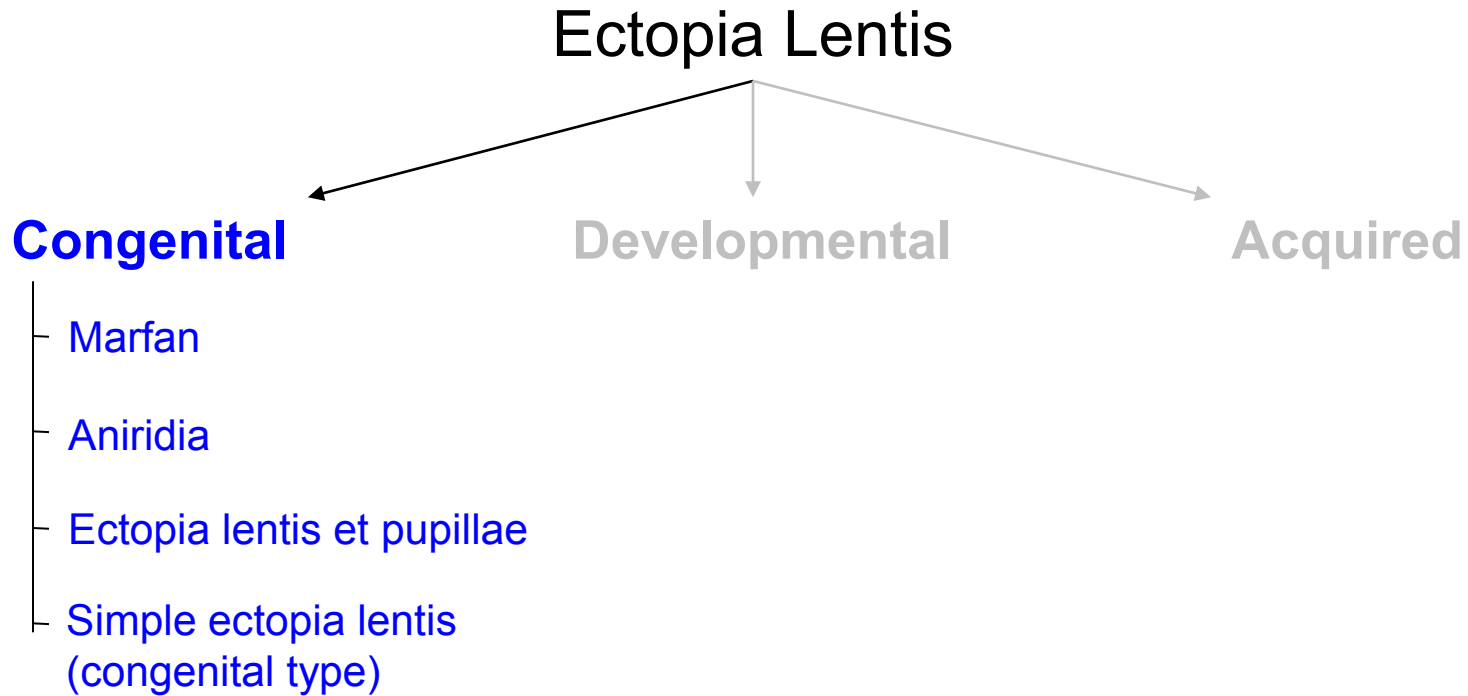
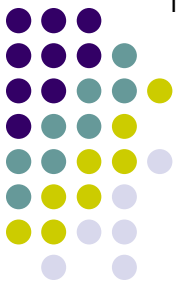
Q

## Ectopia Lentis

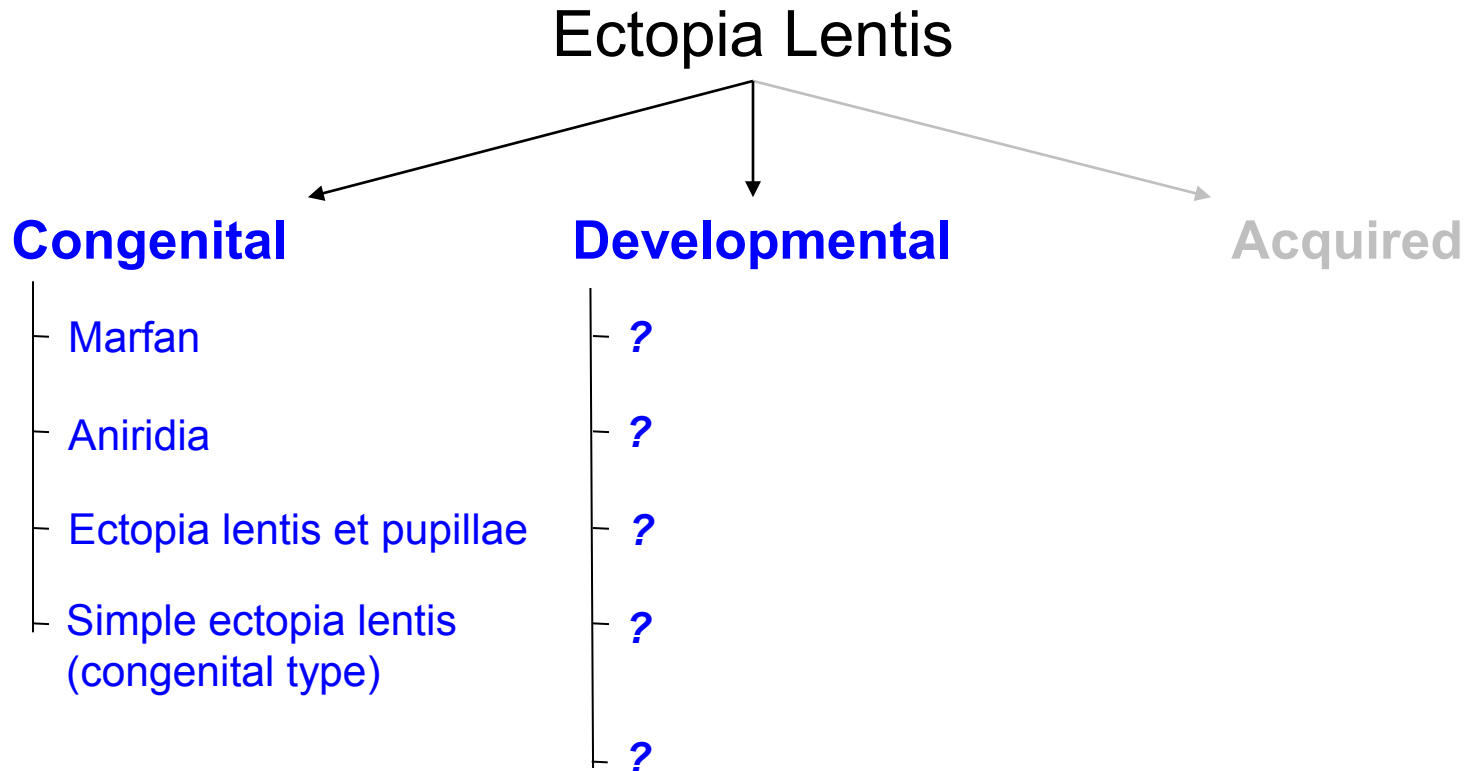
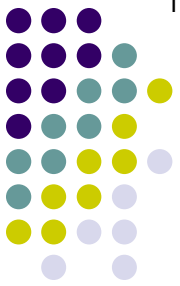
16



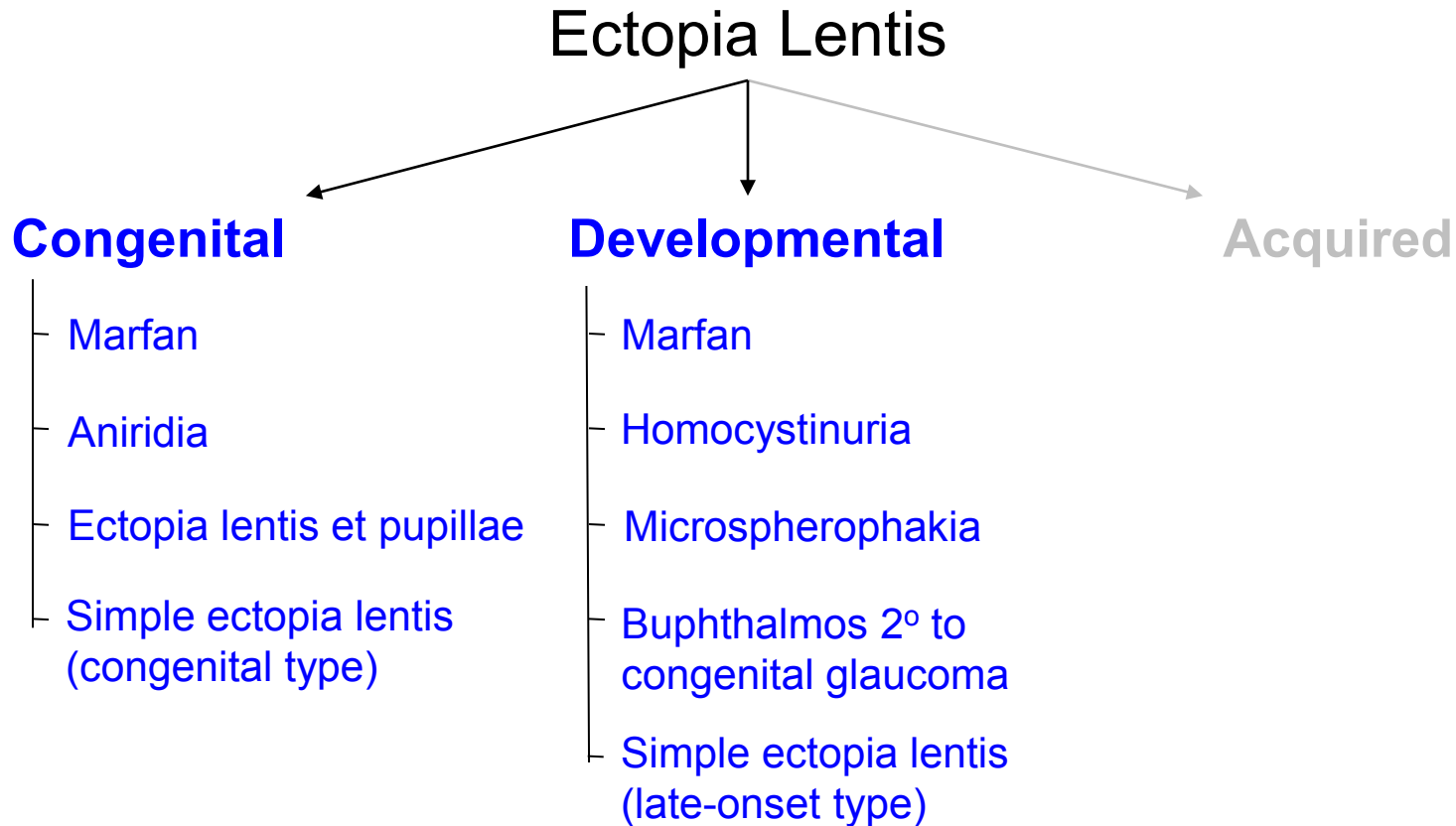
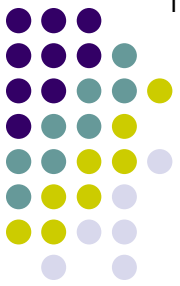
What are the four **congenital** causes covered in the BCSC?



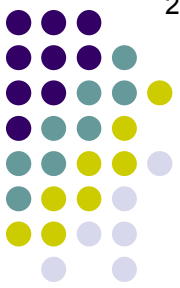
What are the four **congenital** causes covered in the BCSC?



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



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



## Ectopia Lentis

### Congenital

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

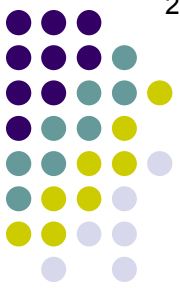
### Developmental

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

### Acquired

- ?
- ?

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



## Ectopia Lentis

### Congenital

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

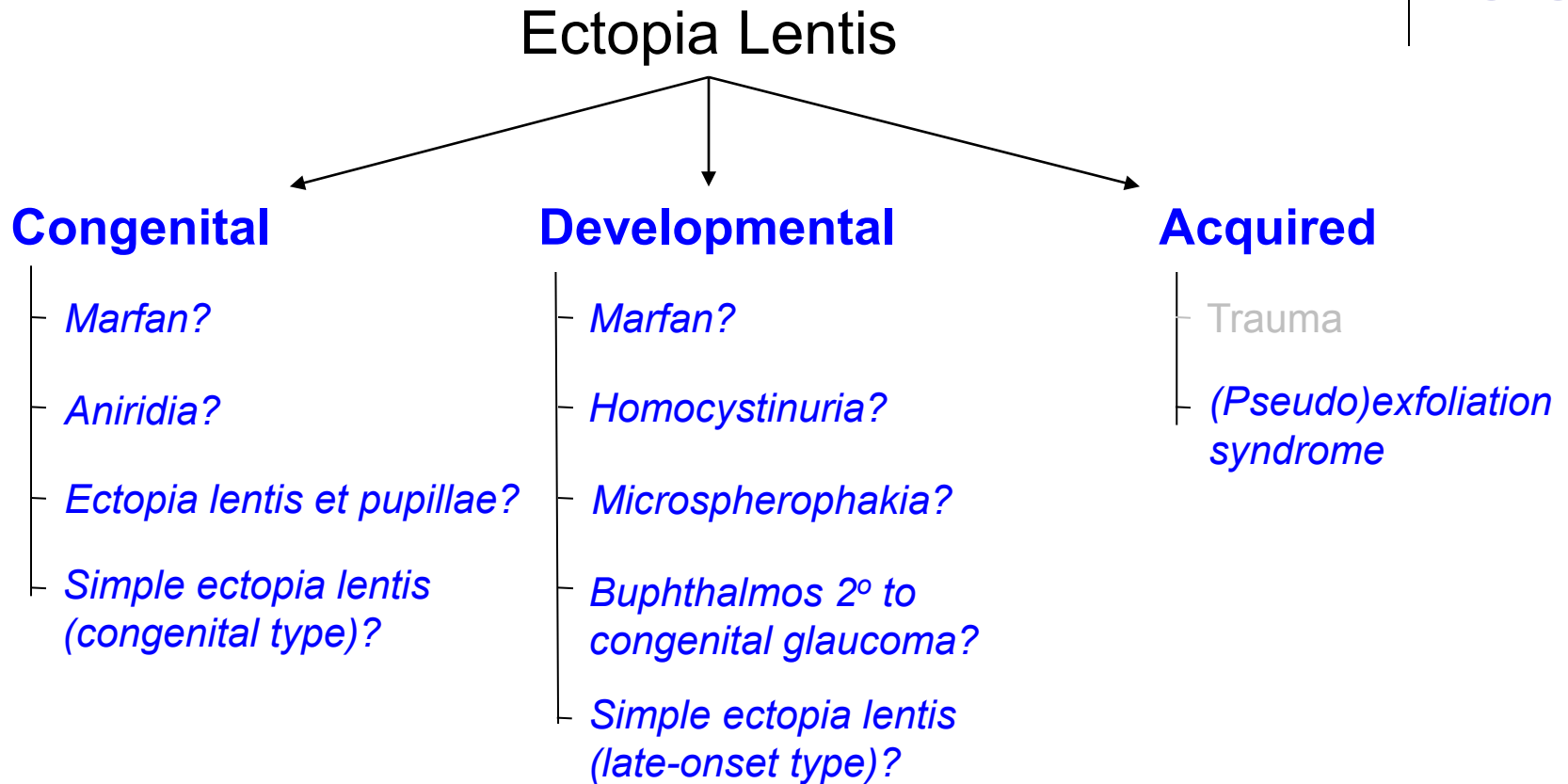
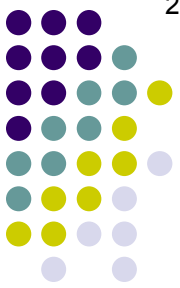
### Developmental

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

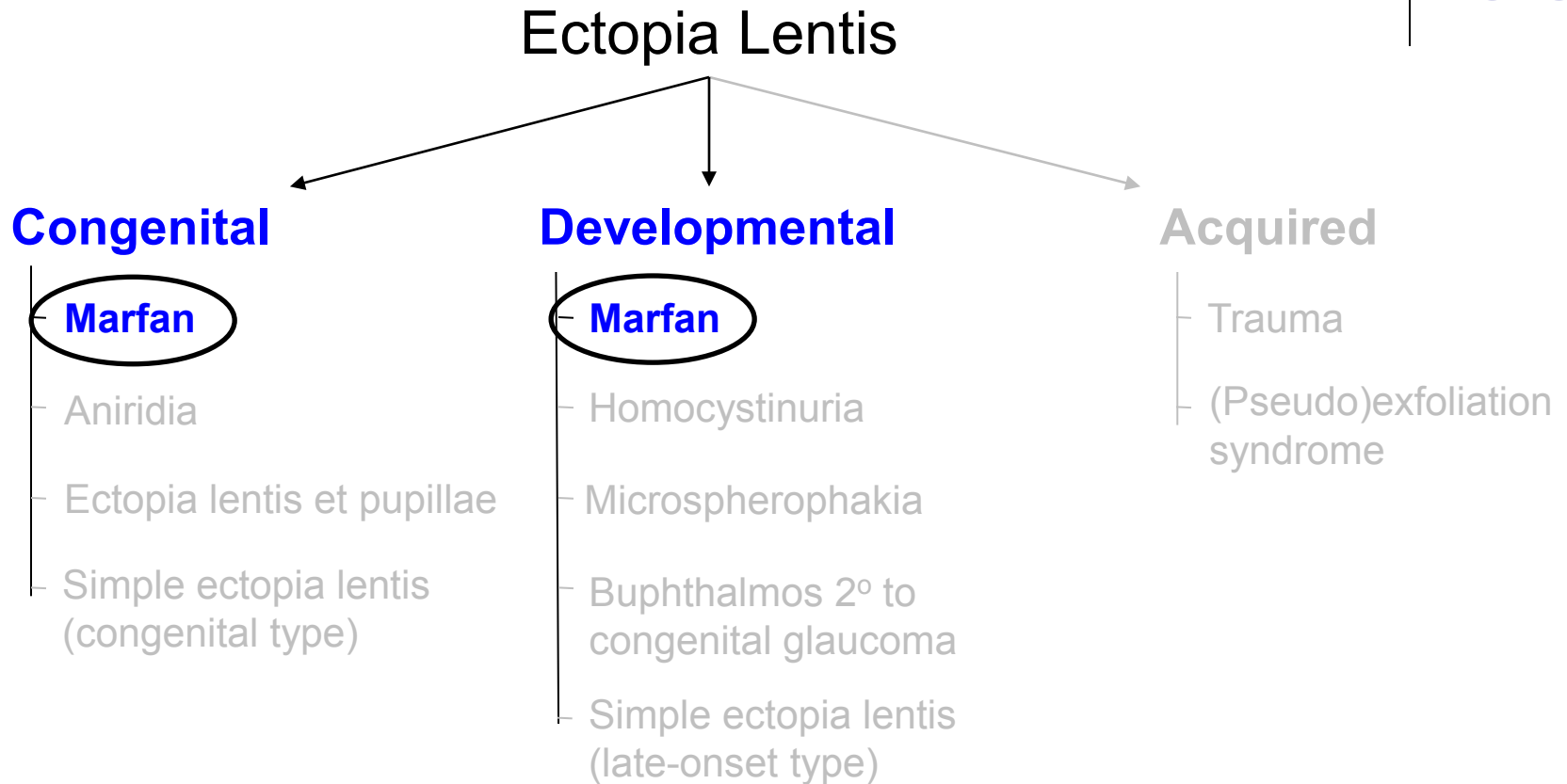
### Acquired

- Trauma
- (Pseudo)exfoliation syndrome

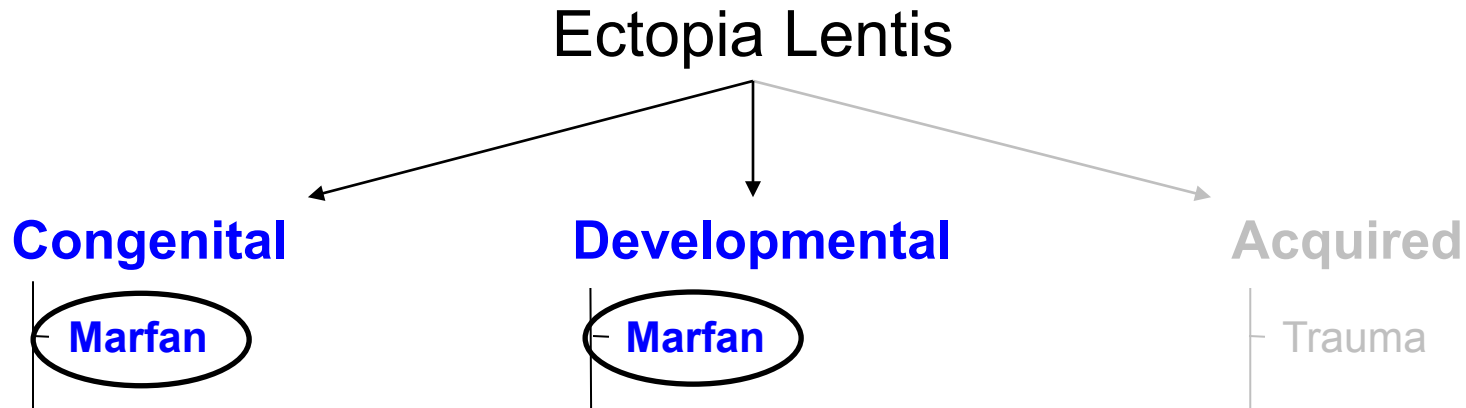
*What are the two **acquired** causes covered in the BCSC?*



*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?*

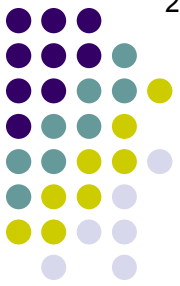
Simple ectopia lentis  
(congenital type)

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

# A

## Ectopia Lentis

25



### Ectopia Lentis

#### Congenital

Marfan

#### Developmental

Marfan

#### Acquired

Trauma

*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*

Simple ectopia lentis  
(congenital type)

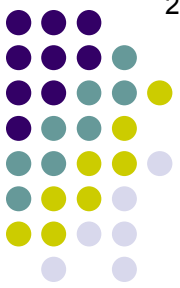
Buphthalmos 2° to  
congenital glaucoma

Simple ectopia lentis  
(late-onset type)

Q

## Ectopia Lentis

26



### Ectopia Lentis

#### Congenital

Marfan

#### Developmental

Marfan

#### Acquired

Trauma

(Pseudo)exfoliation  
syndrome

*What protein is abnormal in Marfan's?*

(late-onset type)

A

## Ectopia Lentis



27

### Ectopia Lentis

#### Congenital

Marfan

#### Developmental

Marfan

#### Acquired

Trauma

(Pseudo)exfoliation  
syndrome

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

(late-onset type)

Q

## Ectopia Lentis



28

### Ectopia Lentis

#### Congenital

Marfan

#### Developmental

Marfan

#### Acquired

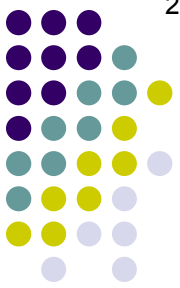
Trauma

(Pseudo)exfoliation syndrome

What is the inheritance pattern?

What protein is involved?  
**Fibrillin**

(late-onset type)



### Ectopia Lentis

#### Congenital

Marfan

#### Developmental

Marfan

#### Acquired

Trauma

(Pseudo)exfoliation syndrome

What is the inheritance pattern?

AD, although family history is negative in % of cases

(late-onset type)

What protein is involved in Marfan's?  
Fibrillin

A

## Ectopia Lentis



30

### Ectopia Lentis

#### Congenital

Marfan

#### Developmental

Marfan

#### Acquired

Trauma

(Pseudo)exfoliation syndrome

*What is the inheritance pattern?*

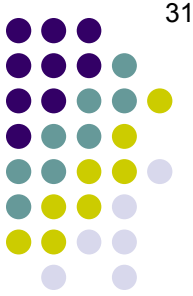
AD, although family history is negative in 15% of cases

*What protein is involved?*  
**Fibrillin**

(late-onset type)

Q

## Ectopia Lentis



31

### Ectopia Lentis

#### Congenital

Marfan

#### Developmental

Marfan

#### Acquired

Trauma

(Pseudo)exfoliation  
syndrome

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

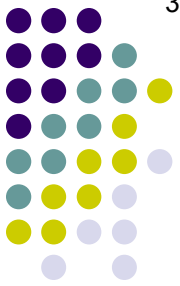
*What three structures/systems manifest abnormalities in Marfan's?*  
--The eye (duh)  
--  
--

(late-onset type)

A

## Ectopia Lentis

32



### Ectopia Lentis

#### Congenital

— Marfan

#### Developmental

— Marfan

#### Acquired

— Trauma

— (Pseudo)exfoliation syndrome

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

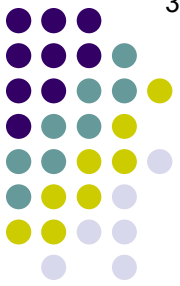
*What three structures/systems manifest abnormalities in Marfan's?*  
--The eye (duh)  
--The cardiovascular  
--The musculoskeletal

(late-onset type)

Q

## Ectopia Lentis

33



### Ectopia Lentis

#### Congenital

Marfan

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

#### Developmental

Marfan

*What three structures/sys*  
--The eye (duh)  
--**The cardiovascular**  
--The musculoskeletal

*What cardiovascular abnormalities are common?*

--  
--  
--

#### Acquired

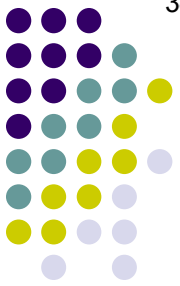
Trauma

(Pseudo)exfoliation  
syndrome

A

## Ectopia Lentis

34



### Ectopia Lentis

#### Congenital

Marfan

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

#### Developmental

Marfan

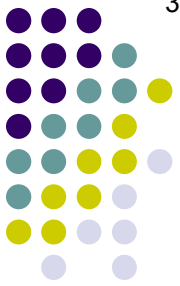
*What three structures/systems are affected?*  
--The eye (duh)  
--**The cardiovascular**  
--The musculoskeletal

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

#### Acquired

Trauma

(Pseudo)exfoliation syndrome



## Ectopia Lentis

### Congenital

— Marfan

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

### Developmental

— Marfan

*What three structures/systems are affected?*  
--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?*

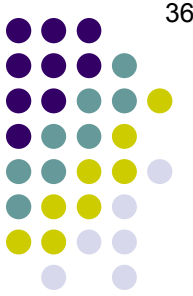
### Acquired

— Trauma

— (Pseudo)exfoliation syndrome

# A

## Ectopia Lentis



36

### Ectopia Lentis

#### Congenital

— Marfan

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

#### Developmental

— Marfan

*What three structures/systems are affected?*  
--The eye (duh)  
--**The cardiovascular**  
--The musculoskeletal

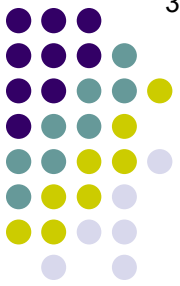
*What cardiovascular abnormalities are common?*  
--Dilatation of the aortic root and descending aorta  
--Aortic aneurysms  
--Mitral valve prolapse

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

#### Acquired

— Trauma

— (Pseudo)exfoliation syndrome



## Ectopia Lentis

### Congenital

— Marfan

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

*What three structures/sys*  
--The eye (duh)  
--**The cardiovascular**  
--The musculoskeletal

### Developmental

— Marfan

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

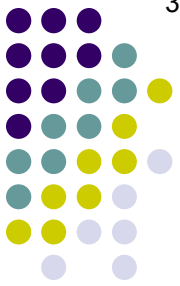
*Are these abnormalities clinically significant?*  
Indeed they are—they are responsible for the  
**significantly shortened lifespan of Marfan pts**

*How significant is the lifespan shortening?*

### Acquired

— Trauma

— (Pseudo)exfoliation syndrome



### Ectopia Lentis

#### Congenital

Marfan

What protein is abnormal in Marfan's?  
Fibrillin

What three structures/sys  
--The eye (duh)  
--**The cardiovascular**  
--The musculoskeletal

#### Developmental

Marfan

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

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  
% that of the so-called normal population.

#### Acquired

Trauma

(Pseudo)exfoliation  
syndrome

# A

## Ectopia Lentis



39

### Ectopia Lentis

#### Congenital

— Marfan

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

*What three structures/sys*  
--The eye (duh)  
--**The cardiovascular**  
--The musculoskeletal

#### Developmental

— Marfan

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

*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.

#### Acquired

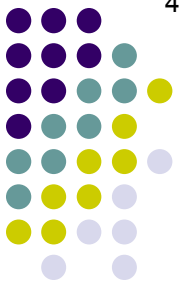
— Trauma

— (Pseudo)exfoliation syndrome

## Ectopia Lentis



Marfan syndrome: Aortic dissection



## Ectopia Lentis

### Congenital

— Marfan

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

*What three structures/systems manifest abnormalities in Marfan's?*

--The eye (duh)

--The cardiovascular

--**The musculoskeletal**

### Developmental

— Marfan

*What musculoskeletal abnormalities are common?*

--?

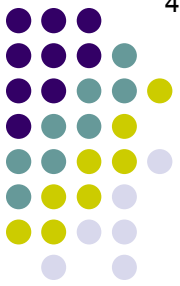
--?

--?

### Acquired

— Trauma

— (Pseudo)exfoliation  
syndrome



### 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?

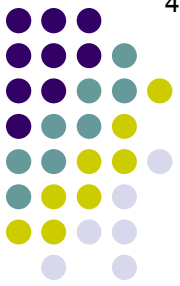
- Arachnodactyly
- Hypermobility joints
- Sternum deformities (eg,

two-us word-ums )

# A

## Ectopia Lentis

43



### 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

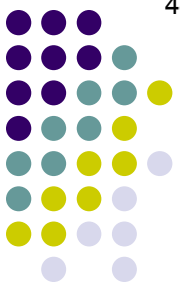
--**The musculoskeletal**

*What musculoskeletal abnormalities are common?*

--Arachnodactyly

--Hypermobility joints

--Sternum deformities (eg, pectus excavatum )



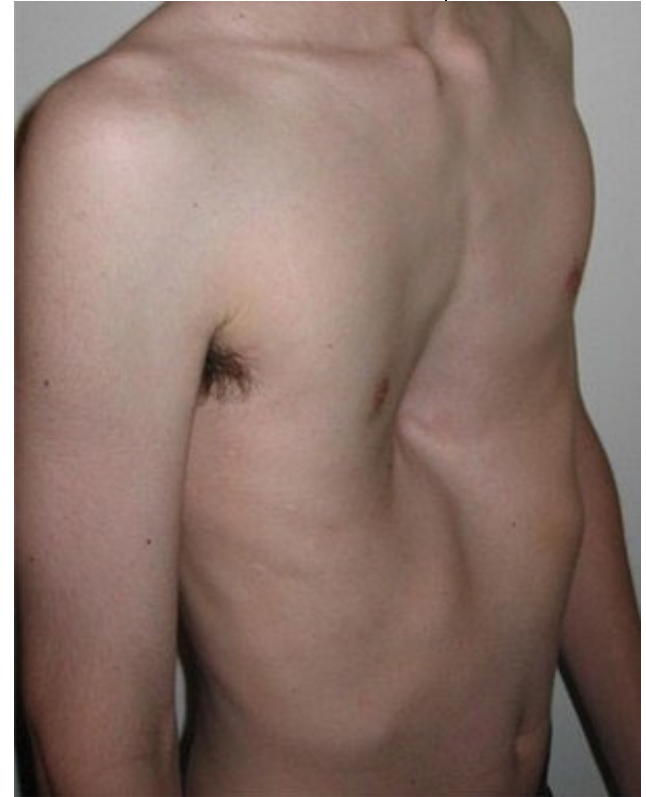
## Ectopia Lentis



Arachnodactyly



Hypermobile joints



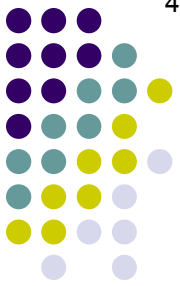
Pectus excavatum

Marfan syndrome

Q

## Ectopia Lentis

45



### Ectopia Lentis

#### Congenital

Marfan

What protein is abnormal in Marfan's?  
Fibrillin

What three systems are affected in Marfan's?  
--The eye (dislocation)  
--The cardiovascular system  
--The musculoskeletal system

#### Developmental

Marfan

What proportion of Marfan pts manifest ocular abnormalities?

#### Acquired

Trauma

(Pseudo)exfoliation syndrome

A

## Ectopia Lentis



46

### Ectopia Lentis

#### Congenital

Marfan

What protein is abnormal in Marfan's?  
Fibrillin

What three systems are affected in Marfan's?  
--The eye (dislocation)  
--The cardiovascular system  
--The musculoskeletal system

#### Developmental

Marfan

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

#### Acquired

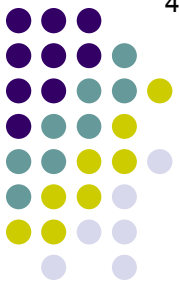
Trauma

(Pseudo)exfoliation syndrome

Q

## Ectopia Lentis

47



### Ectopia Lentis

#### Congenital

Marfan

#### Developmental

Marfan

#### Acquired

Trauma

(Pseudo)exfoliation syndrome

What protein is abnormal in Marfan's?  
Fibrillin

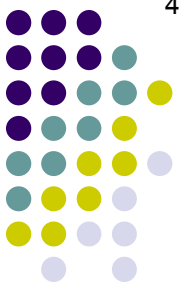
What three st  
--The eye (du  
--The cardio  
--The muscul

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

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

--

--



# A

## Ectopia Lentis

### Ectopia Lentis

#### Congenital

Marfan

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

*What three st*  
--**The eye** (du  
--The cardio  
--The muscul

#### Developmental

Marfan

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At least 80%

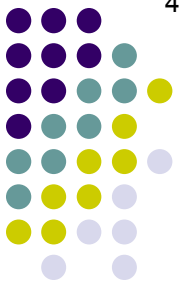
*Other than ectopia lentis, what ocular structural abnormalities are often present?*

--Microspherophakia  
--Increased axial length

#### Acquired

Trauma

(Pseudo)exfoliation syndrome



## Ectopia Lentis

### Congenital

Marfan

What protein is abnormal in Marfan's?  
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What three st

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

Other than **ectopia lentis**, what ocular structural abnormalities  
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--**Microspherophakia**  
--**Increased axial length**

### Developmental

Marfan

### Acquired

Trauma

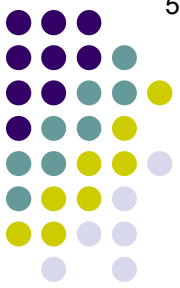
(Pseudo)exfoliation  
syndrome

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

A

# Ectopia Lentis

50



## Ectopia Lentis

### Congenital

Marfan

### Developmental

Marfan

### Acquired

Trauma

(Pseudo)exfoliation syndrome

What protein is abnormal in Marfan's?  
Fibrillin

What three st...  
--The eye (du

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

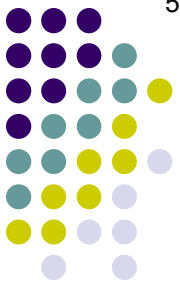
--Microspherophakia  
--Increased axial length

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

Q

## Ectopia Lentis

51



### Ectopia Lentis

#### Congenital

Marfan

What protein is abnormal in Marfan's?  
Fibrillin

What three st

--The eye (du

--The cardio

--The muscul

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

Other than **ectopia lentis**, what are often present?

--Microspherophakia  
--Increased axial length

#### Developmental

Marfan

**Ectopia lentis**

#### Acquired

Trauma

(Pseudo)exfoliation syndrome

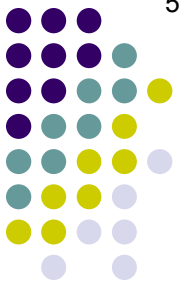
What percentage of Marfan patients develop ectopia lentis?

Of (microspherophakia, increased axial length), which is most likely to be present?

A

## Ectopia Lentis

52



### Ectopia Lentis

#### Congenital

Marfan

What protein is abnormal in Marfan's?  
Fibrillin

What three st

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--The cardio

--The muscul

What proportion of Marfan pts manifest ocular abnormalities?  
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Other than **ectopia lentis**, what  
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--Increased axial length

#### Developmental

Marfan

**Ectopia lentis**

#### Acquired

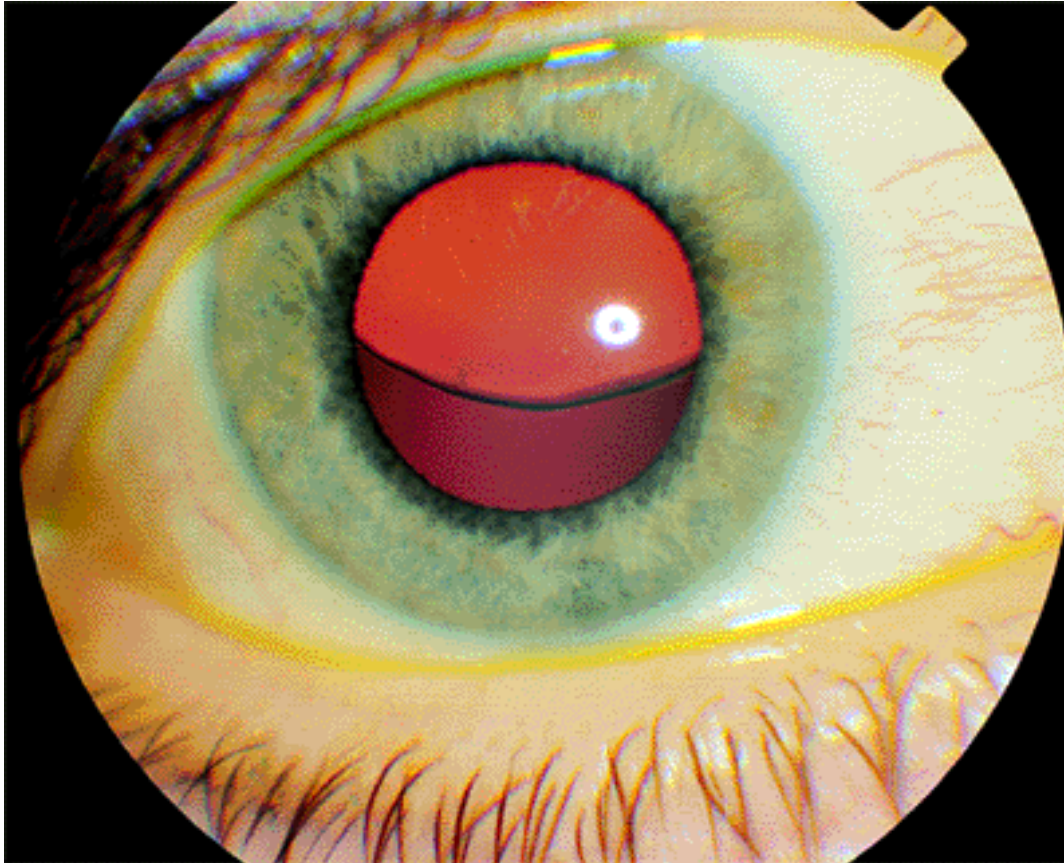
Trauma

(Pseudo)exfoliation  
syndrome

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

Of (microspherophakia, increased axial length), which is most likely to be present?

## Ectopia Lentis



Marfan syndrome: Ectopia lentis



Q

## Ectopia Lentis

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

--  
--  
--

### Congenital

Marfan

*What protein is abnormal in Marfan?*  
Fibrillin

*What three structures are affected in Marfan?*  
--**The eye** (du

--The cardiovascular system  
--The musculoskeletal system

*What proportion of Marfan patients have ectopia lentis?*  
At least 80%

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

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--Increased axial length



# A

## 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

Marfan

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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

I made this term up;  
a better name is

**spherical-aberration myopia**

## Congenital

Marfan

What protein is abnormal in Marfan?  
Fibrillin

What three structural abnormalities are present in Marfan?  
At least 80%

--**The eye** (du)

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--The musculoskeletal

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

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## Ectopia Lentis

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- Ectopia lentis→Lens subluxation→looking through highly curved peripheral lens→lens periphery myopia
- Increased corneal curvature?

## Congenital

### Marfan

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

*What protein is abnormal in Marfan?*  
Fibrillin

*What three structures are affected in Marfan?*  
At least 80%

--The eye (du)

--The cardiovascular

--The musculoskeletal

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

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--Increased axial length



A

## Ectopia Lentis

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- Microspherophakia→increased lens curvature→lenticular myopia
- Ectopia lentis→Lens subluxation→looking through highly curved peripheral lens→lens periphery myopia
- Increased corneal curvature? No!

## Congenital

Marfan

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 are affected in Marfan?  
At least 80%

- The eye (du)
- The cardiovascular
- The musculoskeletal

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

- Microspherophakia
- Increased axial length

## 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!

### Congenital

#### Marfan

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 Marfan?  
Fibrillin

What three structural abnormalities are present in Marfan?  
At least 80%

- The eye (du)
- The cardiovascular system
- The musculoskeletal system

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

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- Increased axial length

A

## Ectopia Lentis

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- Increased axial length**→axial myopia
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- Ectopia lentis**→Lens subluxation→looking through highly curved peripheral lens→**lens periphery** myopia
- Increased corneal curvature? No!*

## Congenital

### Marfan

*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?*  
Fibrillin

*What three structural abnormalities are present?*  
At least 80%

- The eye** (du)
- The cardiovascular
- The muscular

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

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- Increased axial length**

Q

## Ectopia Lentis



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

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Marfan

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## Congenital

Marfan

*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 Marfan?*  
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*What three structures are affected in Marfan?*  
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--The musculoskeletal system

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## Congenital

### Marfan

*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?*

*What protein is abnormal in Marfan?*  
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## Congenital

### Marfan

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

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Bifocals may be needed at an early age

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Q

## Ectopia Lentis



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#### Marfan

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

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## A

## Ectopia Lentis



## Congenital

## Marfan

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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

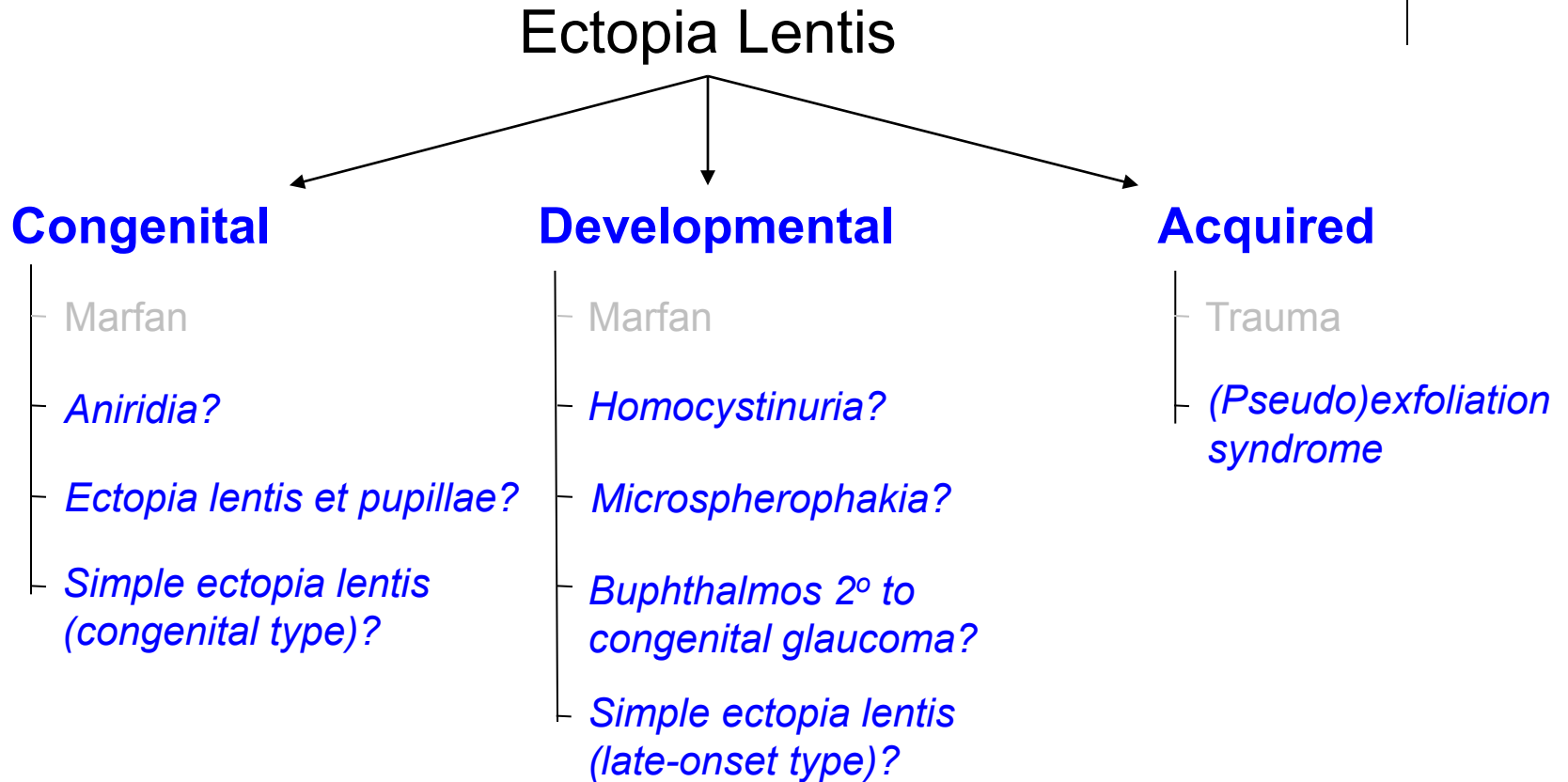
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*What proportion of Marfan patients have ectopia lentis?*  
At least 80%

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

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- Increased axial length

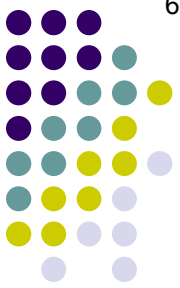


*second*  
*Which is the most common heritable cause of ectopia lentis?*

A

## Ectopia Lentis

68



### 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

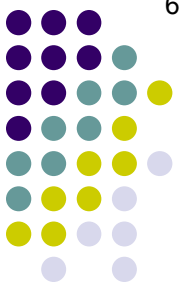
- Trauma
- (Pseudo)exfoliation syndrome

*second*  
**Which is the most common heritable cause of ectopia lentis?**  
**Homocystinuria**

Q

## Ectopia Lentis

69



### Ectopia Lentis

#### Congenital

- Marfan
- Aniridia

#### Developmental

- Marfan
- **Homocystinuria**

#### Acquired

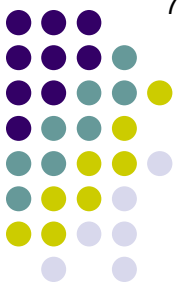
- Trauma
- (Pseudo)exfoliation syndrome

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

# A

## Ectopia Lentis

70



### Ectopia Lentis

#### Congenital

- Marfan
- Aniridia

#### Developmental

- Marfan
- **Homocystinuria**

#### Acquired

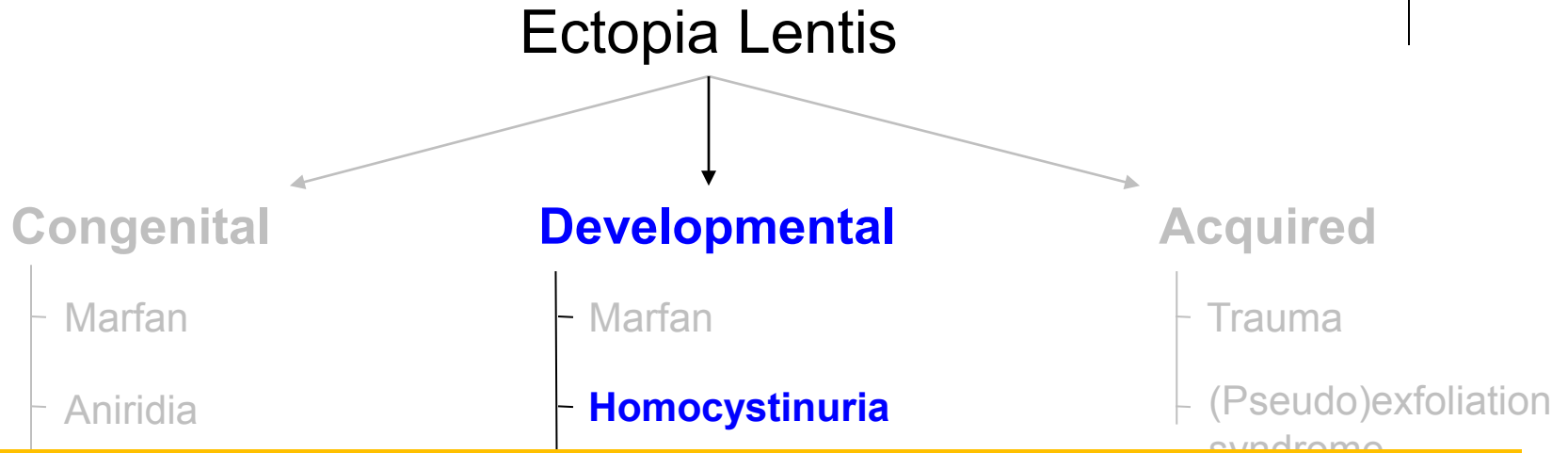
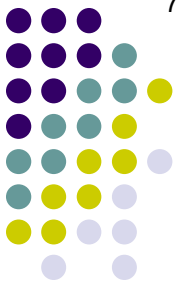
- Trauma
- (Pseudo)exfoliation syndrome

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

Q

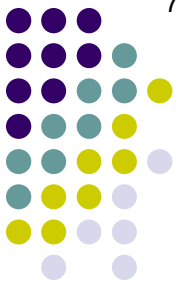
## Ectopia Lentis

71

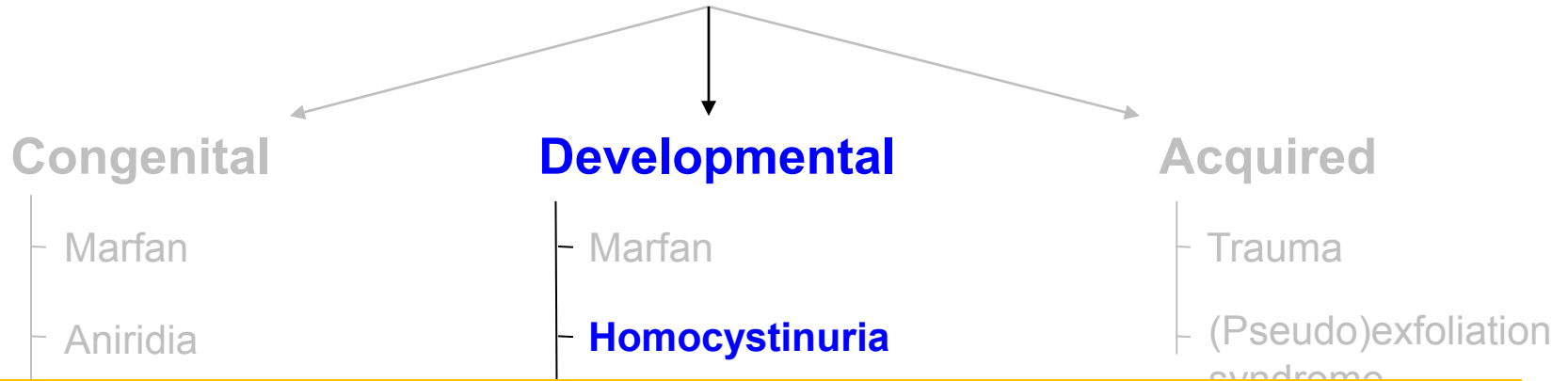


*Homocystinuria is due to faulty metabolism for what amino acid? (hint: it's not cysteine)*  
Methionine

*Then why isn't it called 'methioninuria'?*



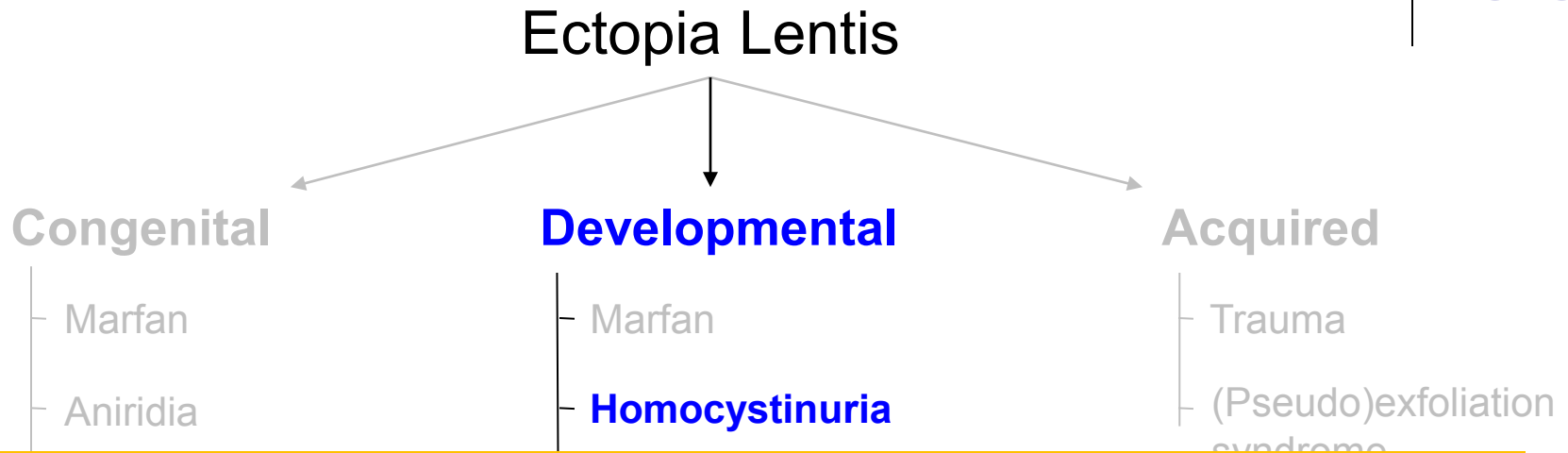
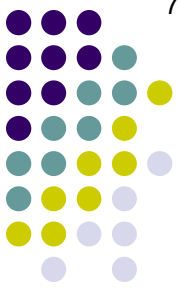
## Ectopia Lentis



*Homocystinuria is due to faulty metabolism for what amino acid? (hint: it's not cysteine)*  
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*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)*  
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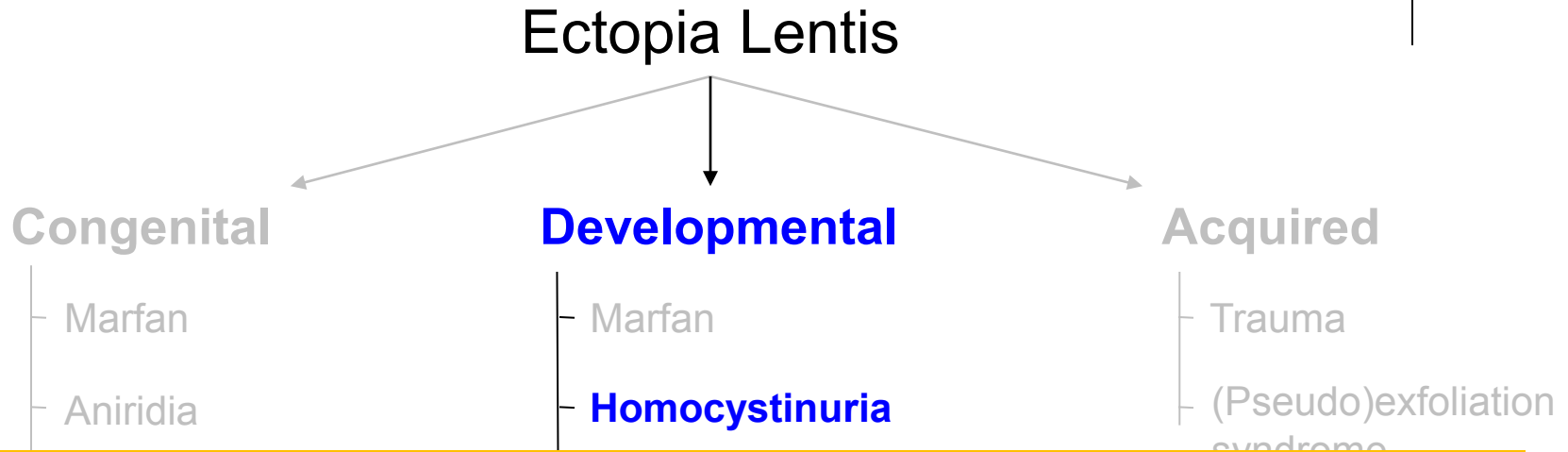
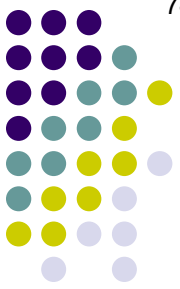
*What four structures/systems manifest abnormalities in homocystinuria?*

--The eye (duh)

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--

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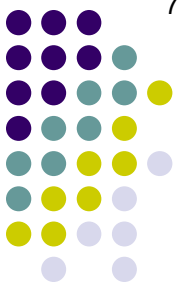
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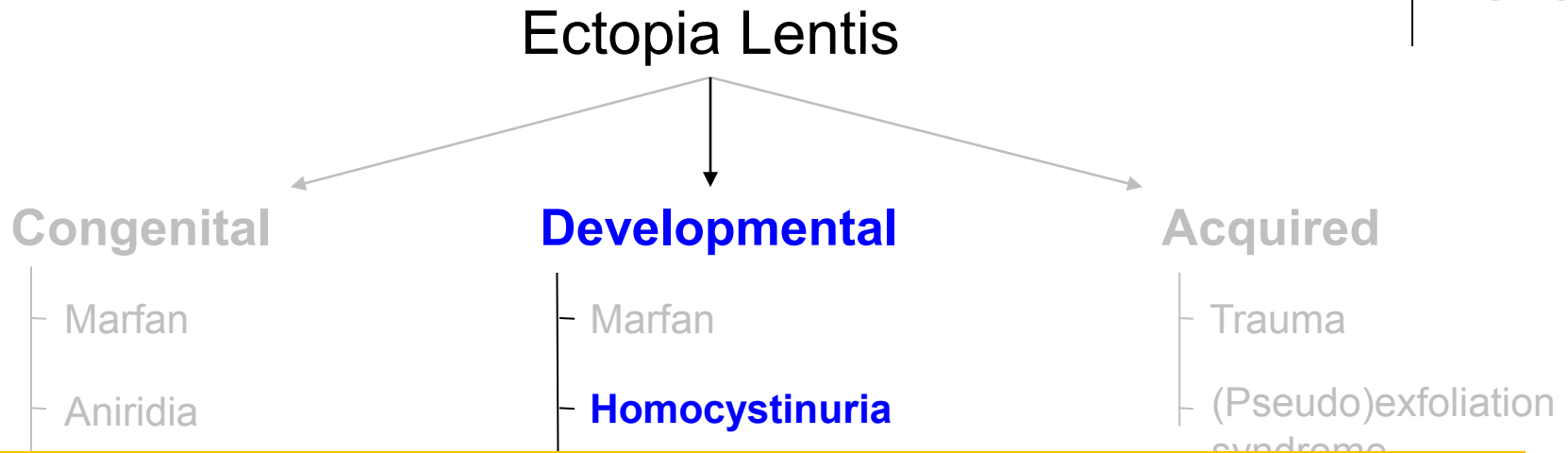
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*What four structures/systems manifest abnormalities in homocystinuria?*

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



# Ectopia Lentis



*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

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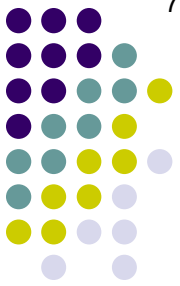
--The eye (duh)

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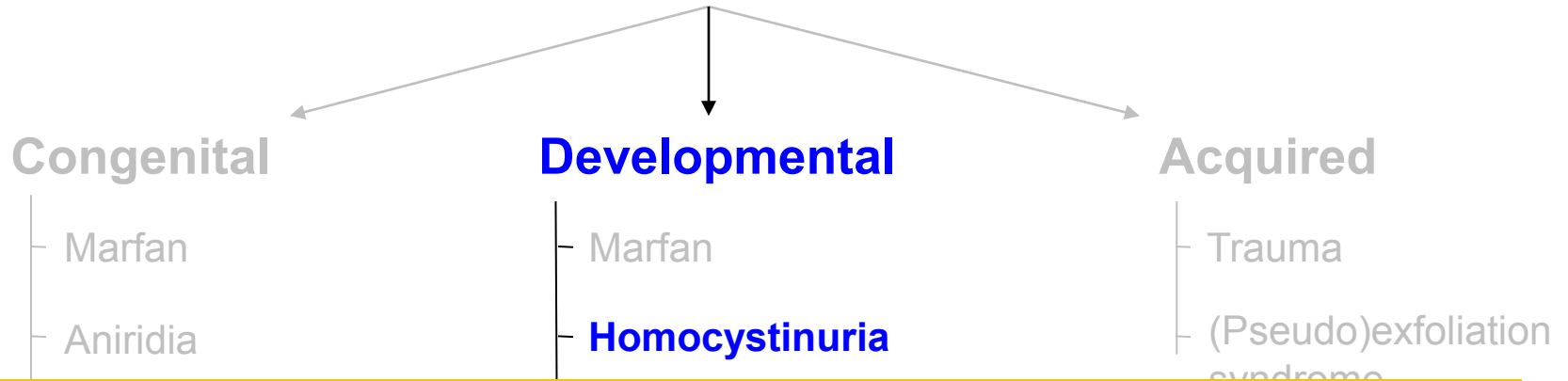
--The musculoskeletal

--The CNS

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



## Ectopia Lentis



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

*What is the classic habitus in homocystinuria?*

*Then why isn't it called 'methioninuria'?*  
Because faulty methionine metabolism leads to homocysteine, which spills into the urine—hence, homocystinuria.

*What four structures/systems manifest abnormalities in homocystinuria?*

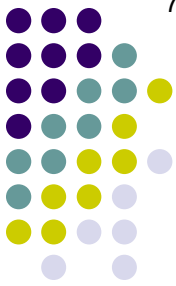
- The eye (duh)
- The cardiovascular
- The musculoskeletal
- The CNS

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

A

# Ectopia Lentis

77



## 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

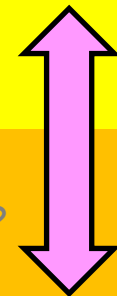
*Then why isn't it called 'methioninuria'?*  
Because faulty methionine metabolism produces homocysteine, which spills into the urine—hence, homocystinuria

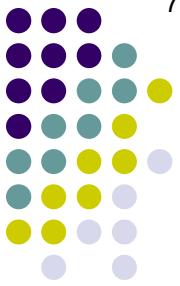
*What is the classic habitus in homocystinuria?*  
Tall and thin, with arachnodactyly and sternum abnormalities

*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**

### Acquired

- Trauma
- (Pseudo)exfoliation syndrome

*Homocystinuria is due to faulty metabolism for what amino acid? (hint: it's not cysteine)*  
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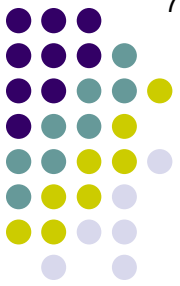
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*How about their coloration?*

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- Marfan
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### Acquired

- Trauma
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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'?*

Because faulty methionine metabolism leads to homocystine, 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?*

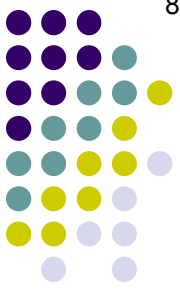
--The eye (duh)

--The cardiovascular

--The musculoskeletal

--The CNS

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



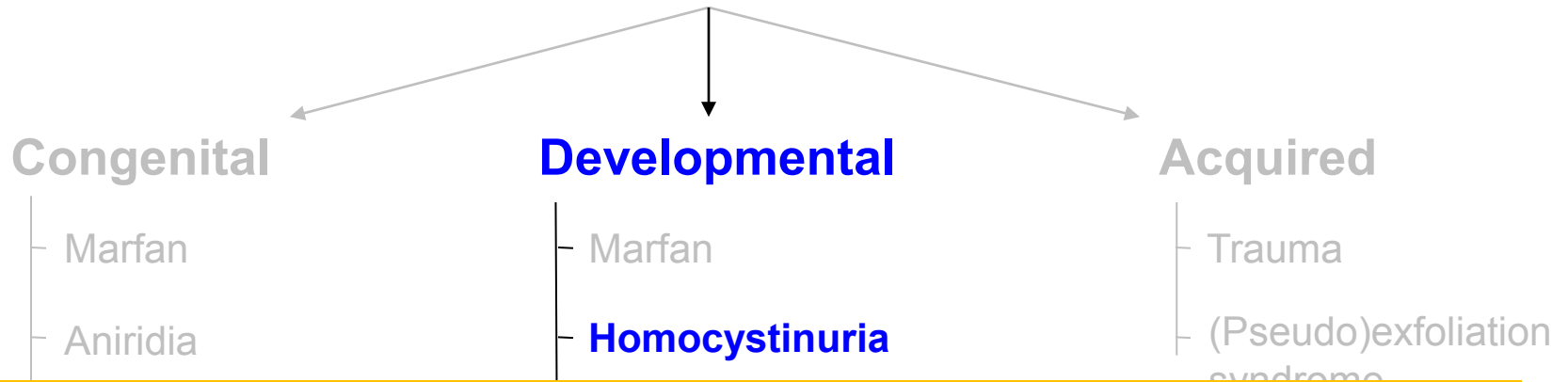
## Ectopia Lentis



Homocystinuria



## Ectopia Lentis



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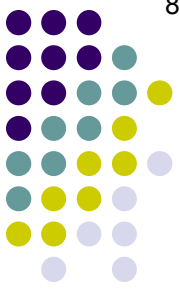
*Then why isn't it called 'methioninuria'?*

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

*What proportion of homocystinuria pts manifest ocular abnormalities?*

*What four structures are affected?*

- The eye (dislocation)
- The cardiovascular system
- The musculoskeletal system
- The CNS



## Ectopia Lentis

### Congenital

- Marfan
- Aniridia

### Developmental

- Marfan
- **Homocystinuria**

### Acquired

- Trauma
- (Pseudo)exfoliation syndrome

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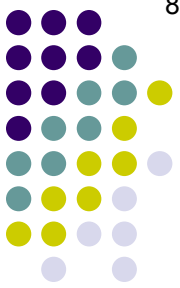
Because faulty methionine metabolism manifests as excessive serum homocysteine, which spills into the urine—hence, homocystinuria

*What proportion of homocystinuria pts manifest ocular abnormalities?*

About 80%

*What four structures are affected?*

- The eye** (dislocation)
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- The musculoskeletal system
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## Ectopia Lentis

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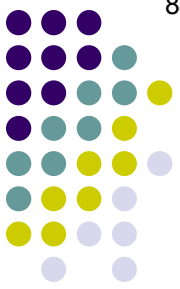
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*Other than ectopia lentis, are other ocular structural abnormalities often present?*



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--The cardiovascular system

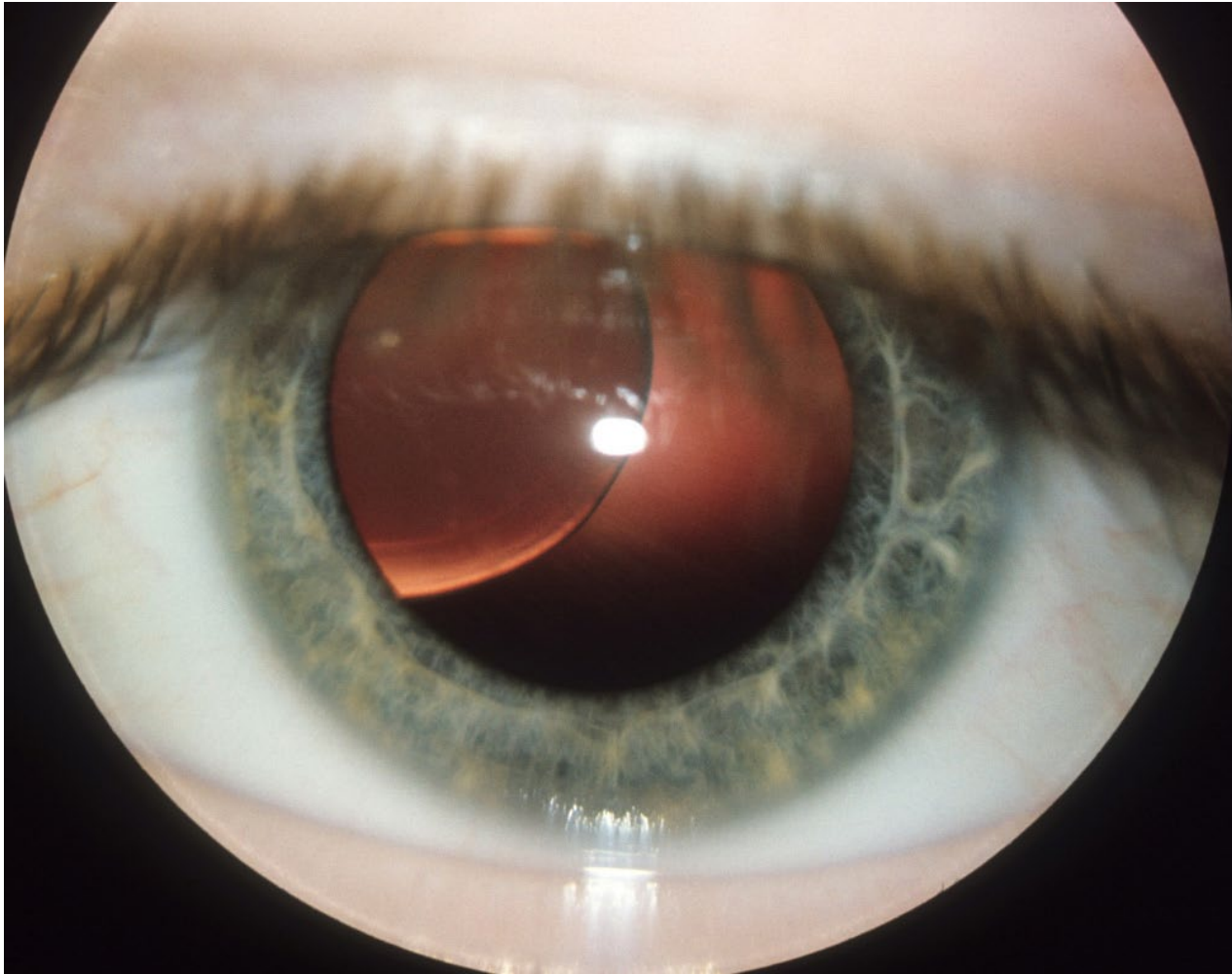
--The musculoskeletal system

--The CNS

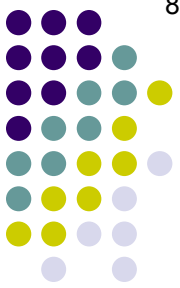
*Other than ectopia lentis, are other ocular structural abnormalities often present?*

Nah—ectopia lentis is pretty much it

## Ectopia Lentis



Homocystinuria: Ectopia lentis



## Ectopia Lentis

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- Marfan
- Aniridia

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- **Homocystinuria**

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- The CNS

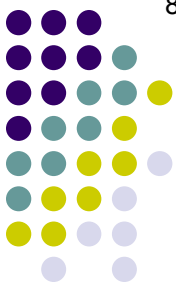
Nah—**ectopia lentis** is present

*During which stage of life does subluxation typically occur?*

A

# Ectopia Lentis

87



## Ectopia Lentis

### Congenital

- Marfan
- Aniridia

### Developmental

- Marfan
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### Acquired

- Trauma
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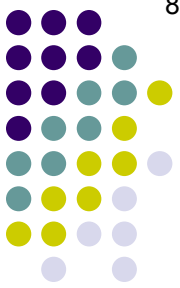
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About 80%

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- The eye** (dislocation)
- The cardiovascular system
- The musculoskeletal system
- The CNS

**ectopia lentis**

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



## 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

*What sight-threatening complication can result from ectopia lentis?*

*Then what?*

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

*What proportion of homocystinuria pts manifest ocular abnormalities?*

About 80%

*What four structures?*

--**The eye** (displacement)

--The cardiovascular system

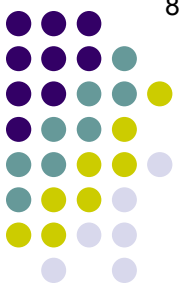
--The musculoskeletal system

--The CNS

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

Nah—**ectopia lentis** is present

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



A

# Ectopia Lentis

## 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

*What sight-threatening complication can result from ectopia lentis?*

Secondary angle-closure glaucoma

*Then what is the metabolic basis for this?*  
Because faulty methionine metabolism manifests as excessive serum homocysteine, which spills into the urine—hence, homocystinuria

*What proportion of homocystinuria pts manifest ocular abnormalities?*

About 80%

*What four structures are affected?*

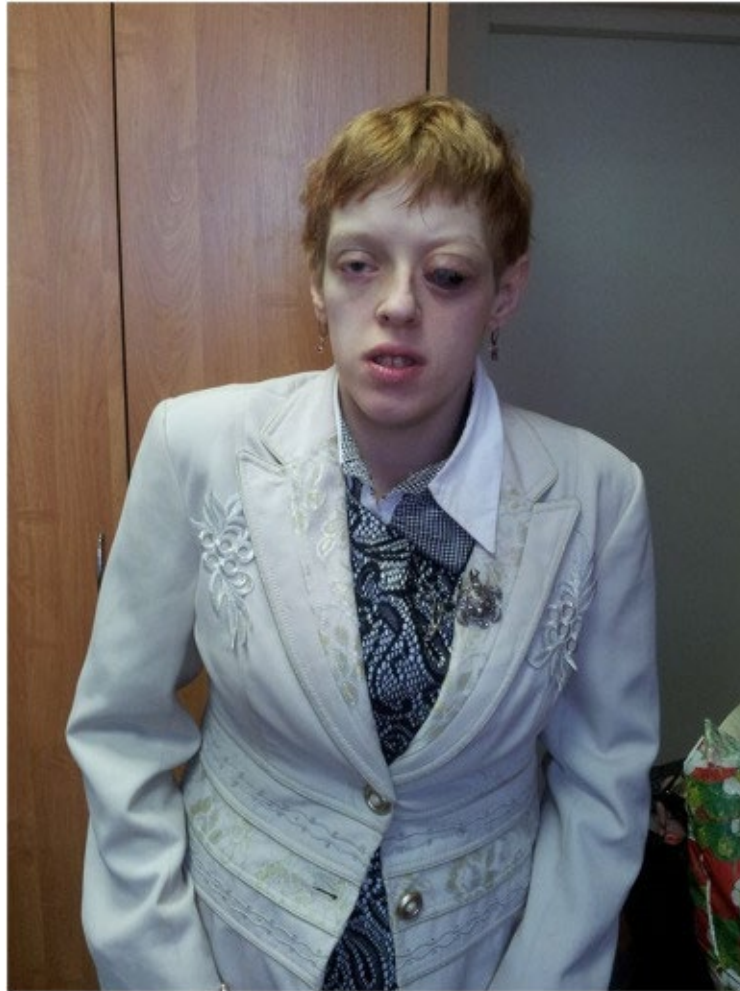
- The eye** (displacement)
- The cardiovascular system
- The musculoskeletal system
- The CNS

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

Nah—**ectopia lentis** is the primary abnormality

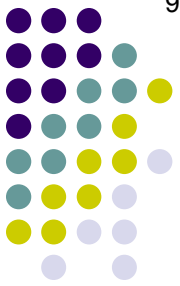
*During which stage of life does subluxation typically occur?*  
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## Ectopia Lentis



Buphthalmos in a homocystinuria pt with ectopia lentis

# Ectopia Lentis



## Ectopia Lentis

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- Aniridia

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- Marfan
- **Homocystinuria**

### Acquired

- Trauma
- (Pseudo)exfoliation syndrome

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**

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

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

- The eye (or
- The cardiovascular
- The muscular
- The CNS

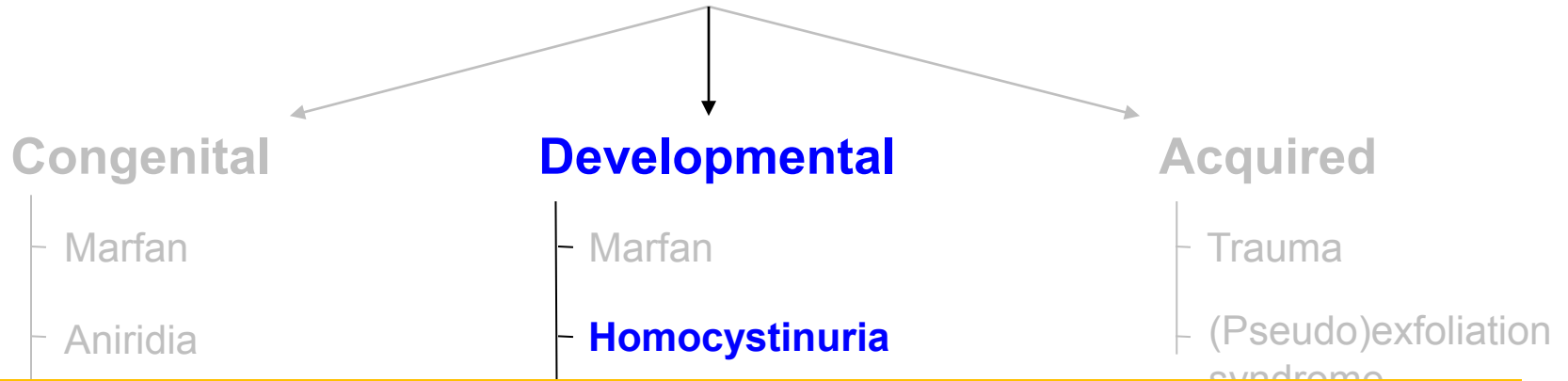
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## Ectopia Lentis



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

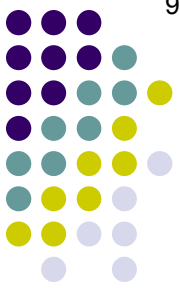
Because faulty methionine metabolism manifests as excessive serum homocysteine, which spills into the urine—hence, homocystinuria

*What four structures/systems are commonly affected by homocystinuria?*

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

*What cardiovascular abnormalities are common?*

- 
- 
- 
- 
-



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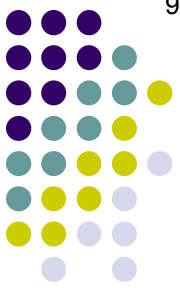
*What four structures/sy*

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

*What cardiovascular abnormalities are common?*

- HTN
- Cardiomegaly
- Thromboembolism
- CAD
- CVA

*ria?*



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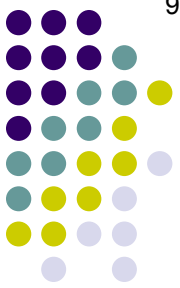
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*What proportion of untreated homocystinurics will experience a significant vascular event prior to age 30?*



A

## Ectopia Lentis

### Ectopia Lentis

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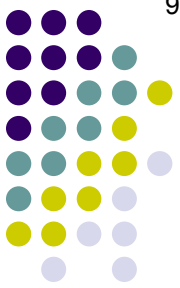
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*What cardiovascular abnormalities are common?*

- HTN
- Cardiomegaly
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*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)*



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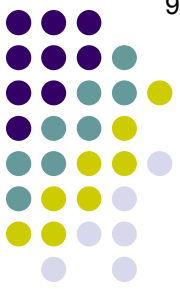
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- Thromboembolism**
- CAD
- CVA

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



## Ectopia Lentis

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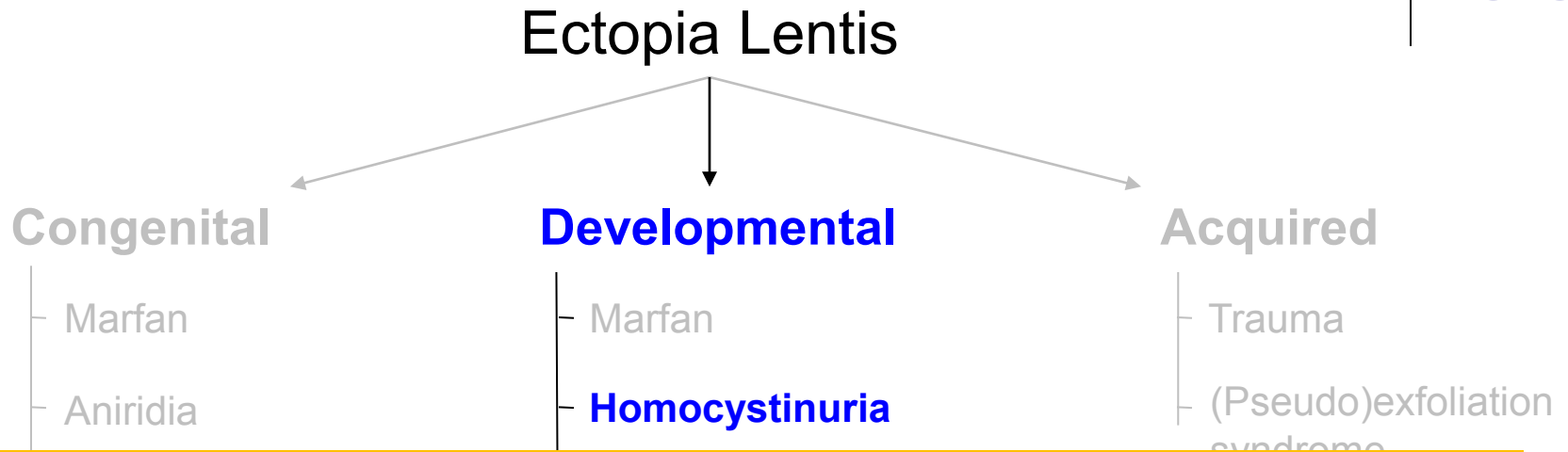
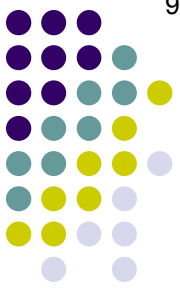
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- The musculoskeletal
- The CNS

*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.** 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

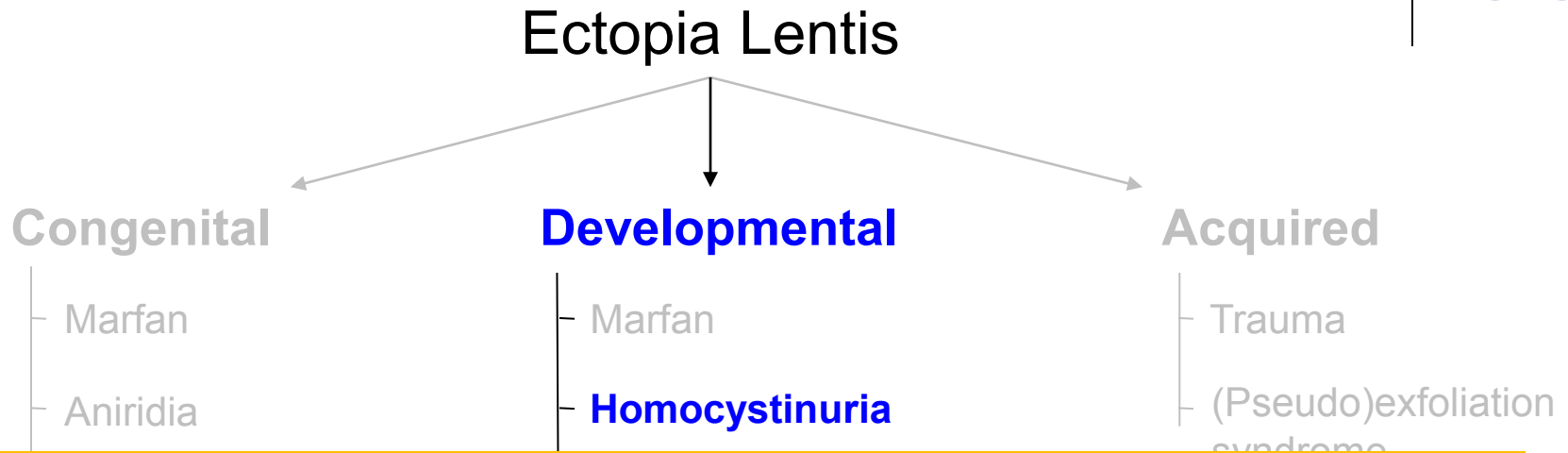
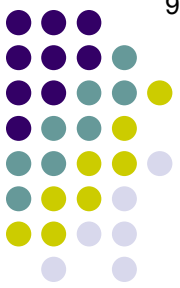
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*What four structures/systems manifest abnormalities in homocystinuria?*

- 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?*



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- The CNS

*In addition to arachnodactyly and sternum deformities, there is a musculoskeletal abnormality classic for homocystinuria.*

*What is it?*

Early osteoporosis

Q

# Ectopia Lentis

100



## Ectopia Lentis

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

*How early are we talking about?*

*What four structures*

--The eye (duh)

--The cardiovascular

--**The musculoskeletal**

--The CNS

*ties, there is a musculoskeletal abnormality classic for homocystinuria.*

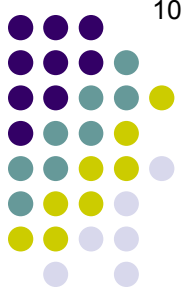
*What is it?*

**Early osteoporosis**

A

# Ectopia Lentis

101



## Ectopia Lentis

### Congenital

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### Developmental

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*How early are we talking about?*

*What four structures are affected?* Crazy early. Osteoporotic changes are common in childhood

--The eye (duh) (and have been reported in toddlers)

--The cardiovascular system

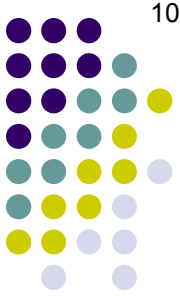
--**The musculoskeletal** system

--The CNS

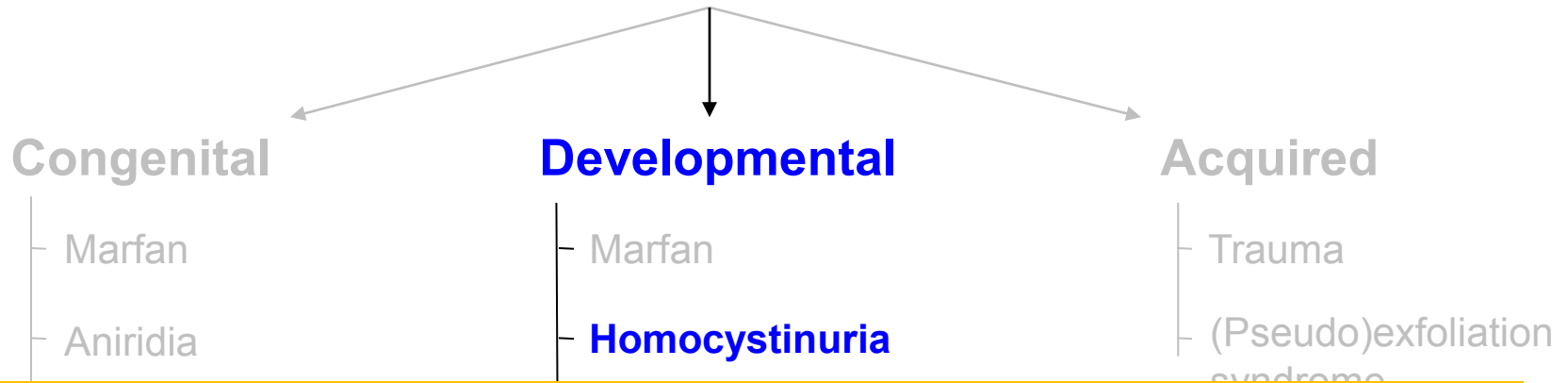
**Early osteoporosis**

*is a musculoskeletal abnormality classic for homocystinuria.*

*What is it?*



## Ectopia Lentis



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*What four structures/systems manifest abnormalities in homocystinuria?*

--The eye (duh)

--The cardiovas

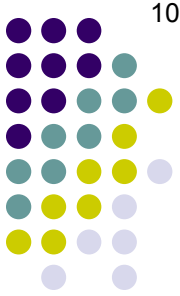
--The musculos

--**The CNS**

*What CNS abnormalities are common?*

--

--



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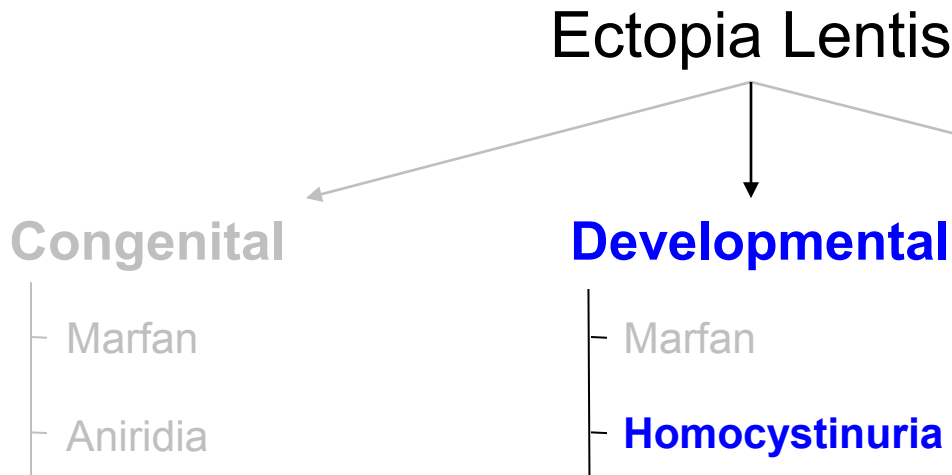
- The eye (duh)
- The cardiovas
- The musculos
- The CNS**

*What CNS abnormalities are common?*

- Seizures
- Mental retardation

Q

# Ectopia Lentis



H  
O  
M  
O  
C  
Y  
S  
T  
I  
N  
U  
R  
I  
A

*Homocystinuria is due to faulty metabolism for what amino acid? Methionine*

*Then why isn't it called 'methioninuria'?*

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

*What four structures/systems manifest abnormalities in homocystinuria?*

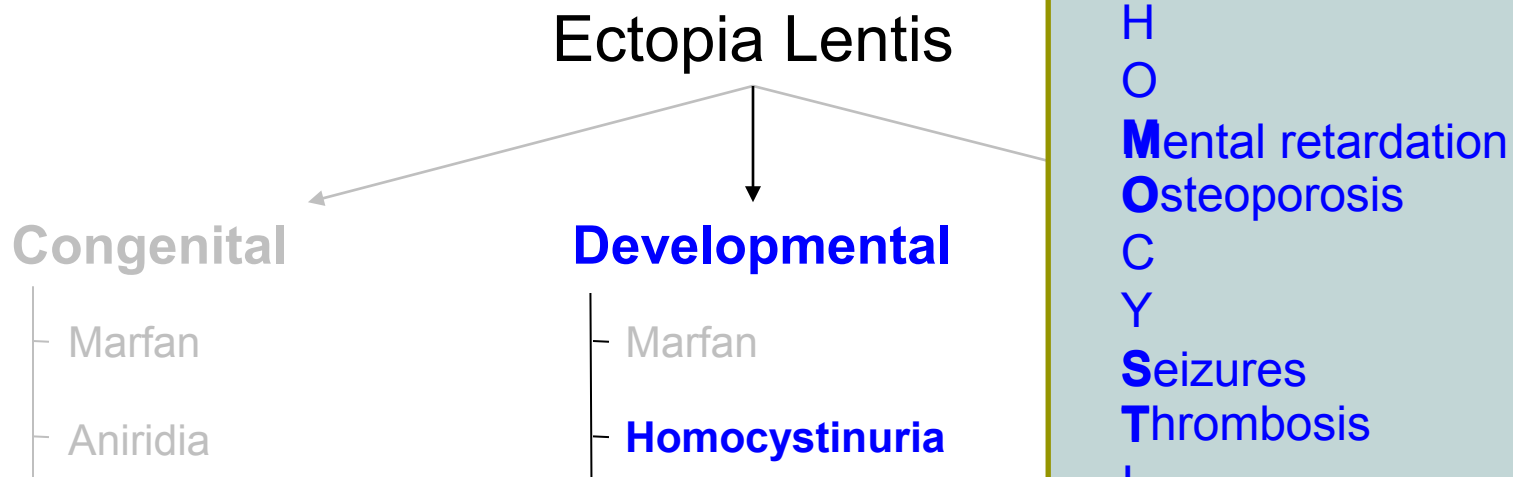
- The eye (duh)
- The cardiovascular**
- The musculoskeletal**
- The CNS**

*So, **MOST** homocystinurics have these nonocular problems:*

- M
- O
- S
- T

A

# Ectopia Lentis



*Homocystinuria is due to faulty metabolism for what amino acid?*  
Methionine

*Then why isn't it called 'methioninuria'?*

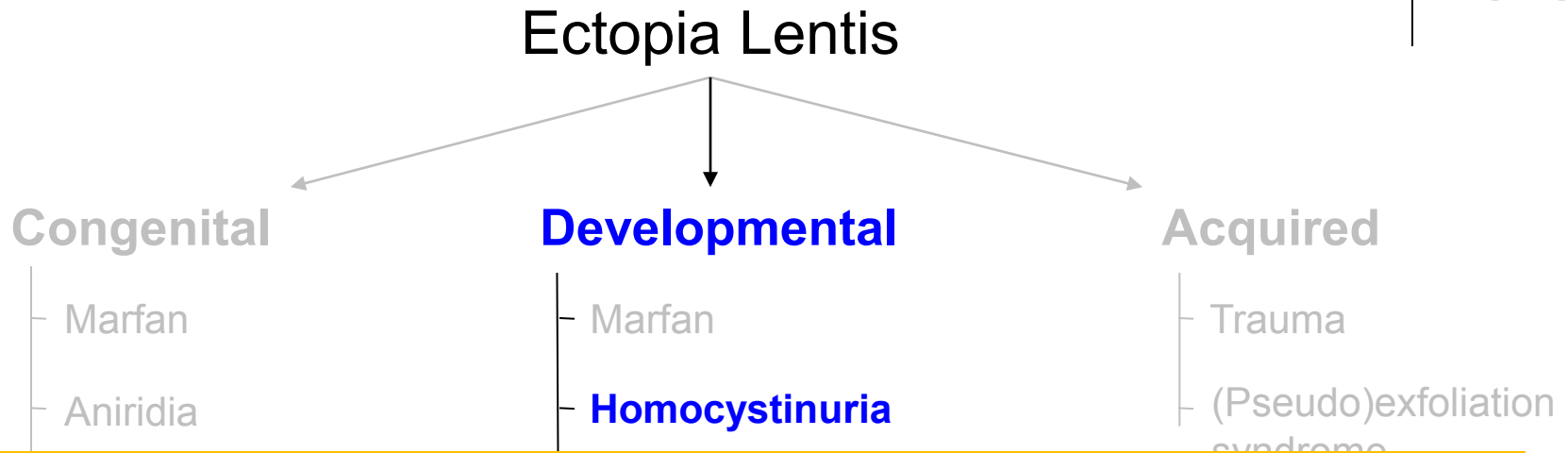
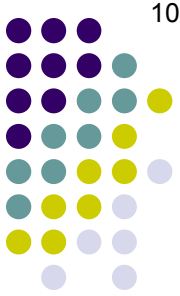
Because faulty methionine metabolism manifests as excess homocysteine, which spills into the urine—hence, homocystinuria

*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**



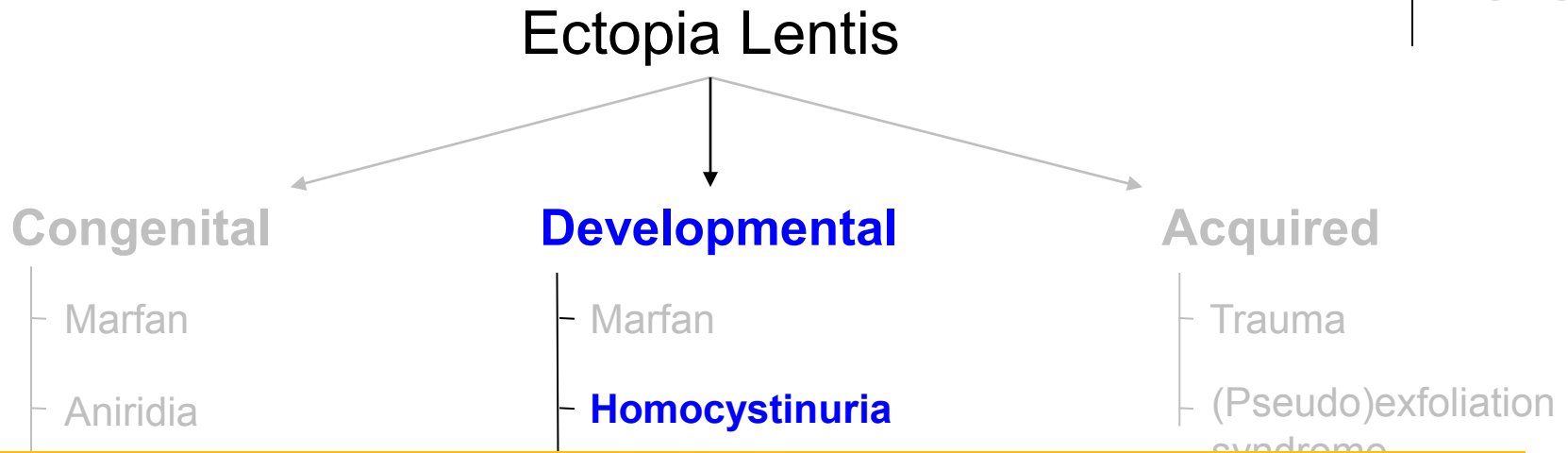
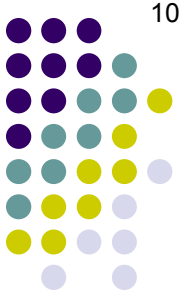
*Homocystinuria is due to faulty metabolism for what amino acid? (hint: it's not cysteine)*  
Methionine

*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)*  
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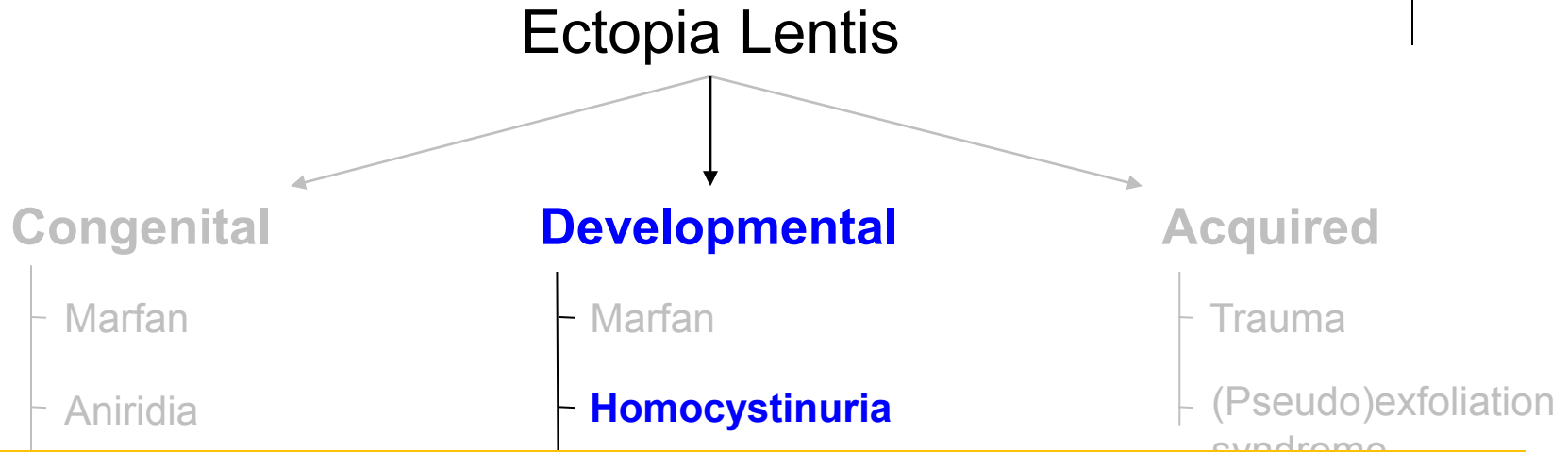
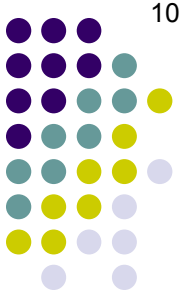
*Then why isn't it called 'methioninuria'?*

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 **12**, 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)*  
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Indeed there are. A diet low in methionine (and high in cysteine), along with supplemental vitamin B<sub>6</sub>, can significantly reduce the rate of ectopia (and systemic probs as well)

- The cardiovascular
- The musculoskeletal
- The CNS

## Ectopia Lentis

*Marfan syndrome*

*Homocystinuria*



*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.*

# Q

## Ectopia Lentis

*Marfan syndrome*

*Homocystinuria*

(Start here, work down the list) → ↑ risk thromboembolism

Lens subluxes *up*

Lens subluxes *down*

Tall

Thin habitus

Fair-haired

Zonules *stretched*

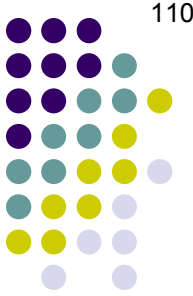
Zonules *broken*

Associated with  
microspherophakia

Ectopia usually bilateral  
and symmetric

Chest-wall deformities

*Divvy up the characteristics.  
Some go both ways.*



*Marfan syndrome*

*Homocystinuria*

↑ risk **thromboembolism**

*Do these simultaneously*

- Lens subluxes *up*
- Lens subluxes *down*

Tall

Thin habitus

Fair-haired

Zonules *stretched*

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Associated with  
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Ectopia usually bilateral  
and symmetric

Chest-wall deformities



# Q/A

## Ectopia Lentis

*Marfan syndrome*

*Homocystinuria*

↑ risk *thromboembolism*

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# Q/A

## Ectopia Lentis

*Marfan syndrome*

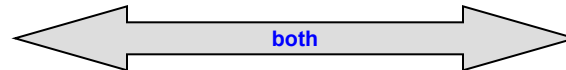
*Homocystinuria*

↑ risk *thromboembolism*

Lens subluxes *up*

Lens subluxes *down*

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Tall

Thin habitus

Fair-haired

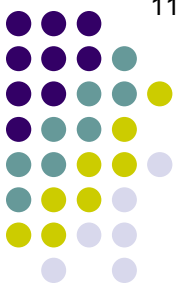
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# Q/A

## Ectopia Lentis

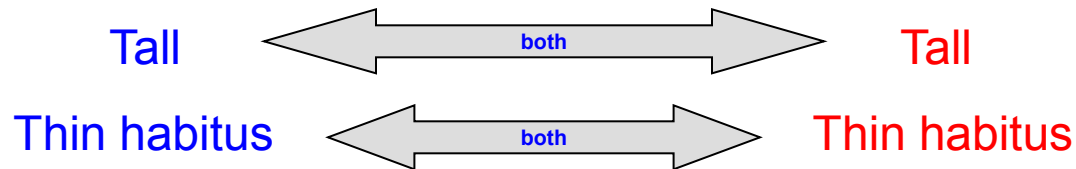
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*Homocystinuria*

↑ risk *thromboembolism*

Lens subluxes *up*

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Fair-haired

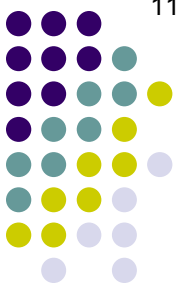
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Lens subluxes **down**

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Tall

Thin habitus

Thin habitus

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# Q/A

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*Homocystinuria*

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Lens subluxes **up**

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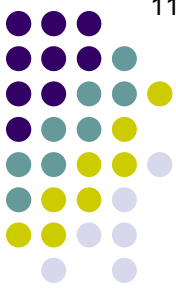
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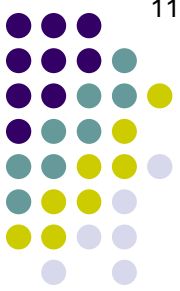
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Chest-wall deformities





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*Homocystinuria*

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Lens subluxes **down**

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Tall

Thin habitus

Thin habitus

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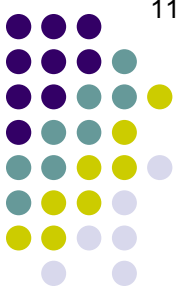
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# A

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↑ risk **thromboembolism**

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Ectopia usually bilateral  
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Chest-wall deformities

Chest-wall deformities

both

both

both

both

both

# Ectopia Lentis

*Marfan syndrome*

*Homocystinuria*

Lens subluxes **up**

↑ risk **thromboembolism**

Lens subluxes **down**

Tall  
Thin habitus

**Note the Key Differences!**

both

both

Tall  
Thin habitus

Zonules **stretched**

**Fair-haired**  
Zonules **broken**

Associated with  
microspherophakia

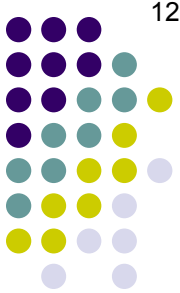
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Chest-wall deformities

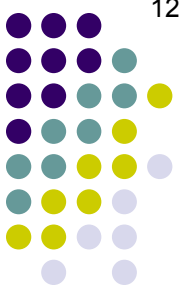
Chest-wall deformities



Q

## Ectopia Lentis

121



### Ectopia Lentis

#### Congenital

- Marfan
- Aniridia
- Ectopia lentis et pupillae

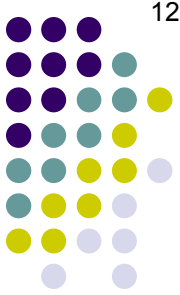
#### Developmental

- Marfan
- Homocystinuria
- **Microspherophakia**

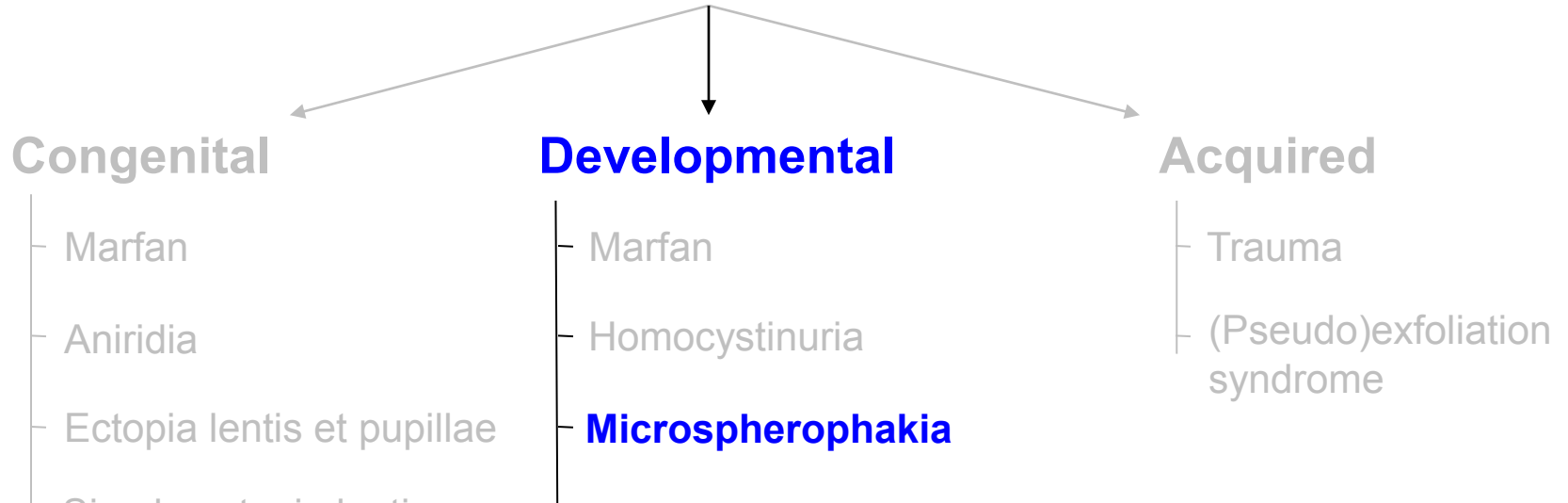
#### Acquired

- Trauma
- (Pseudo)exfoliation syndrome

*In a few words, how would you describe the shape of a microspherophakic lens?*

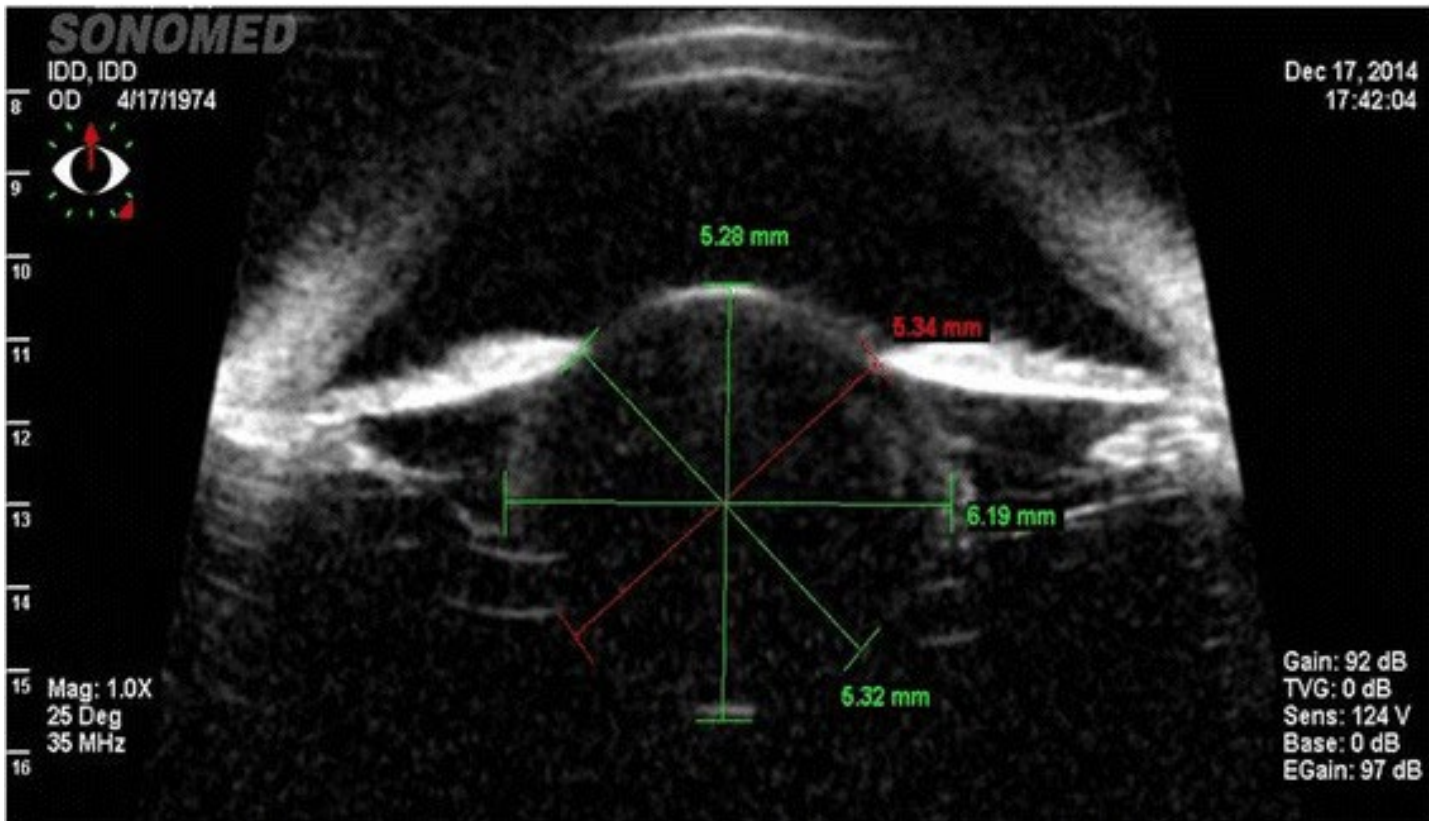


## Ectopia Lentis

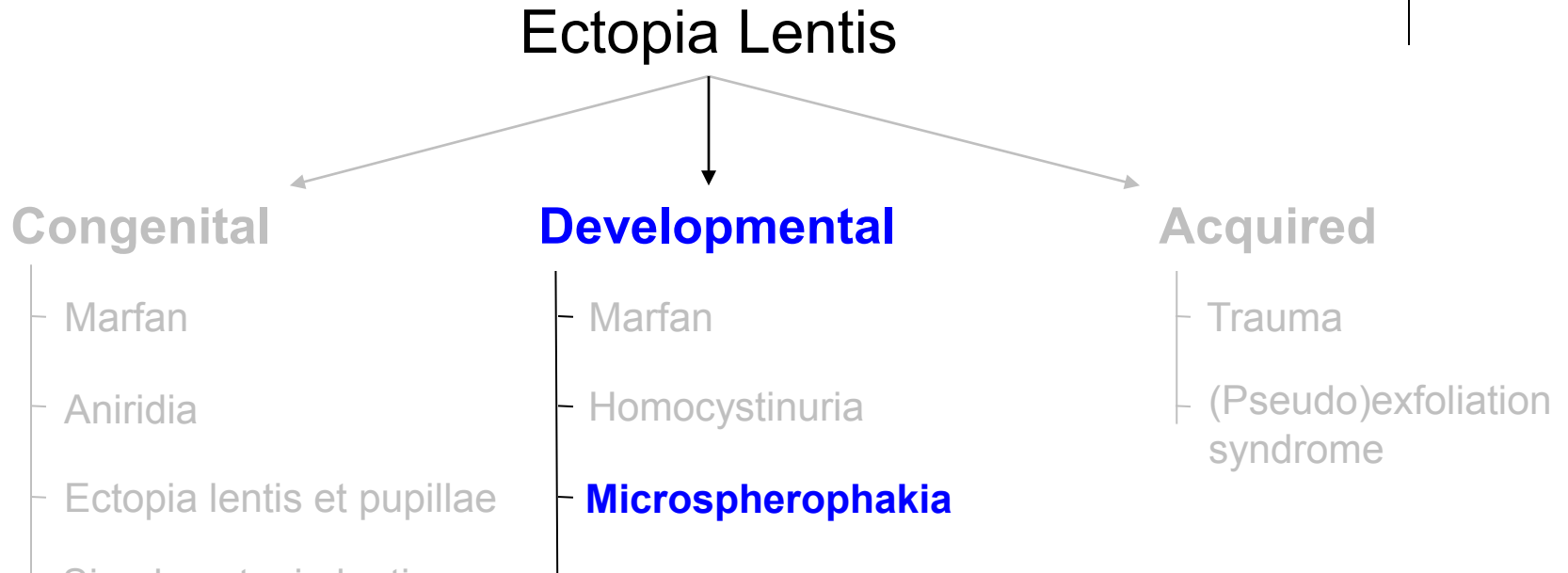


*In a few words, how would you describe the shape of a microspherophakic lens?*  
The name says it all: the lens is small ('micro') and round ('sphero')

## Ectopia Lentis



Microspherophakia. Note the small size, extreme curvature of the lens

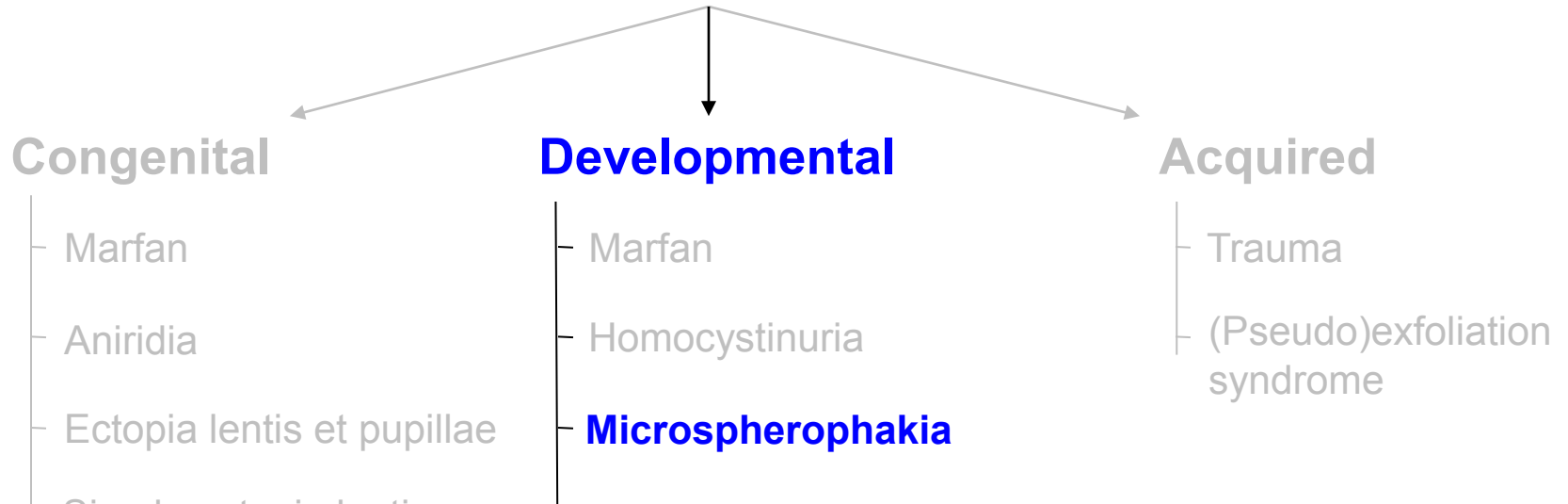


*In a few words, how would you describe the shape of a microspherophakic lens?*  
The name says it all: the lens is small ('micro') and round ('sphero')

*What common slit-lamp observation owes to the lens' small size?*



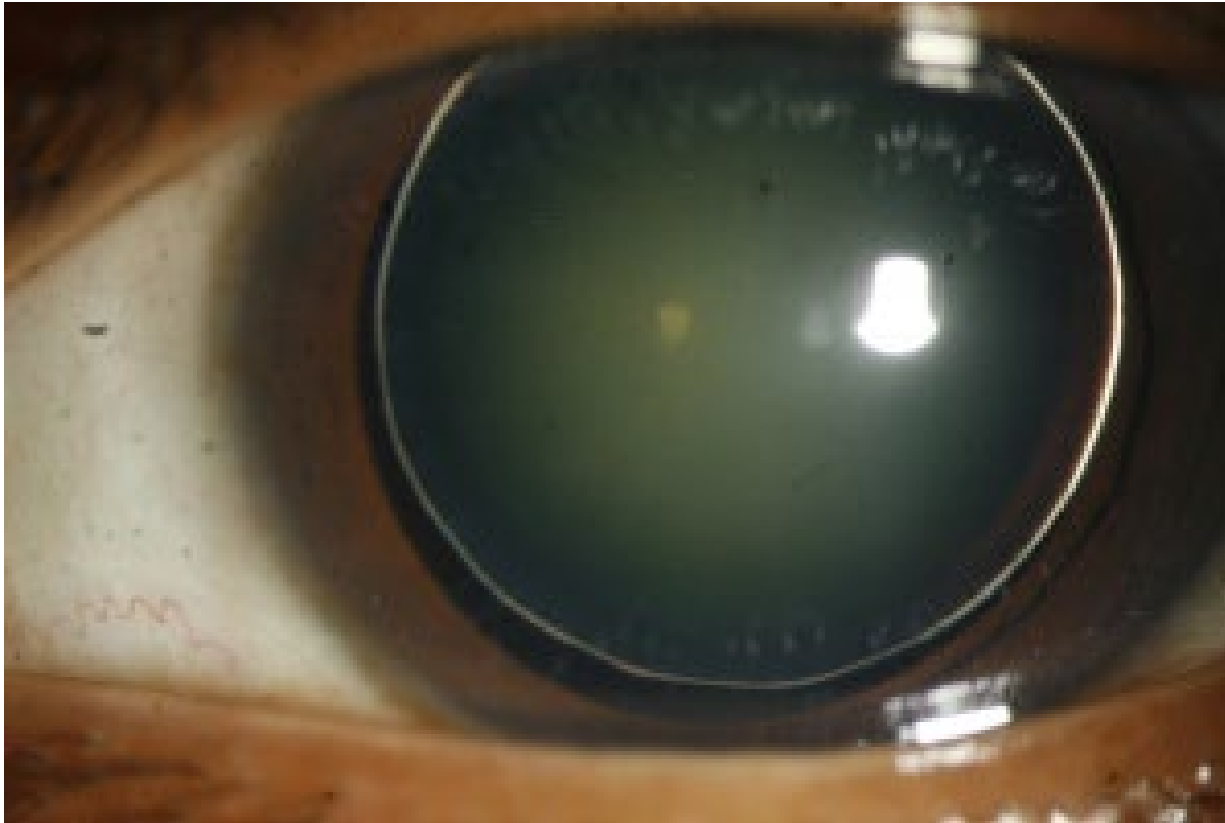
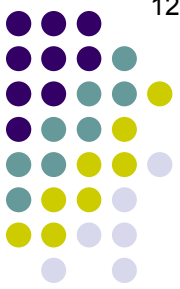
## Ectopia Lentis



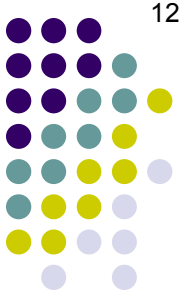
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The name says it all: the lens is small ('micro') and round ('sphero')

*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

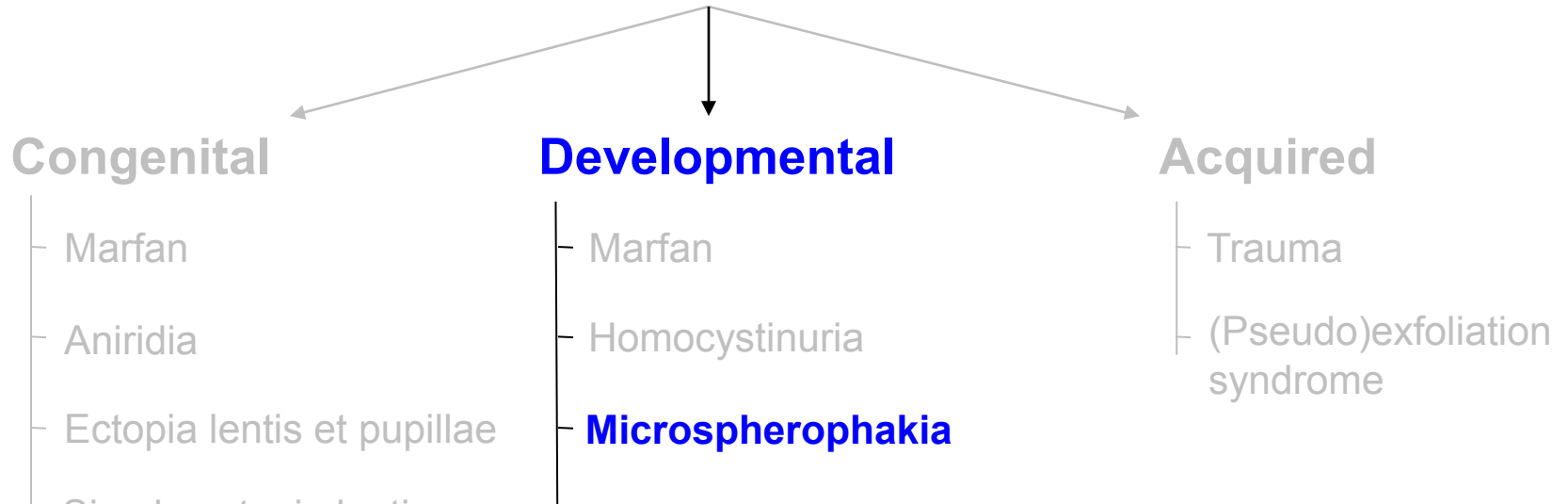
## Ectopia Lentis



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

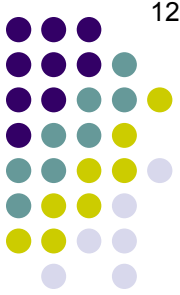


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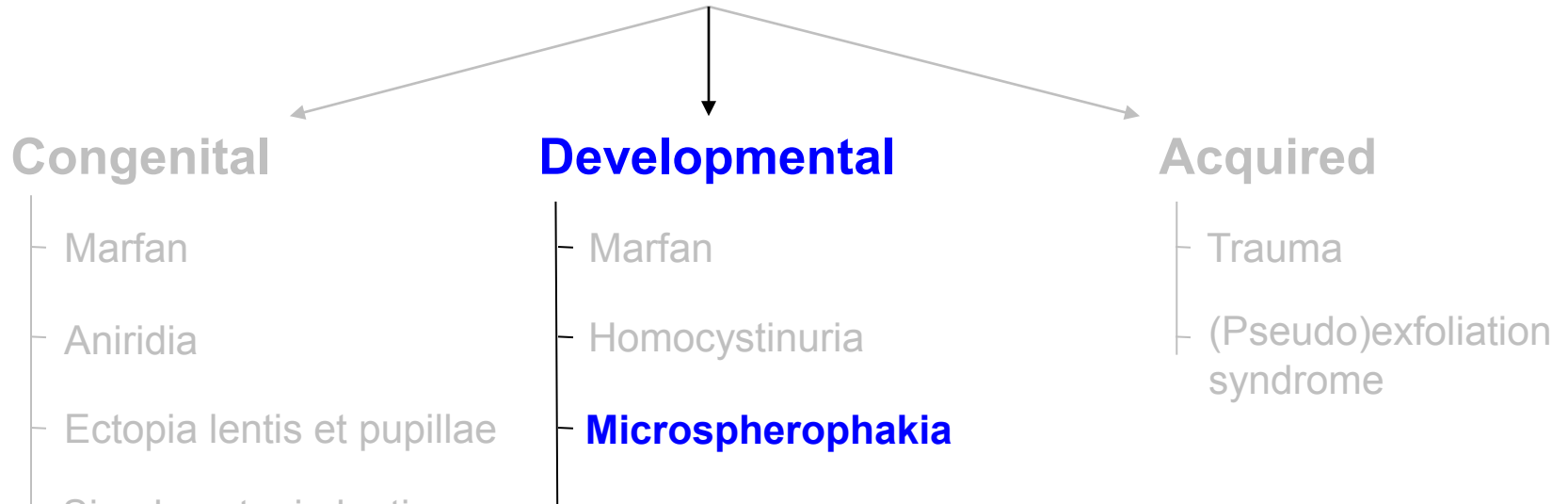


*In a few words, how would you describe the shape of a microspherophakic lens?*  
The name says it all: the lens is small ('micro') and round ('sphero')

*Pts with microspherophakia are almost always high myopes. Why?*

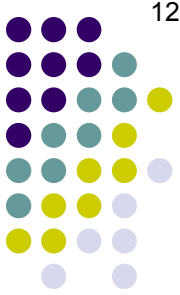


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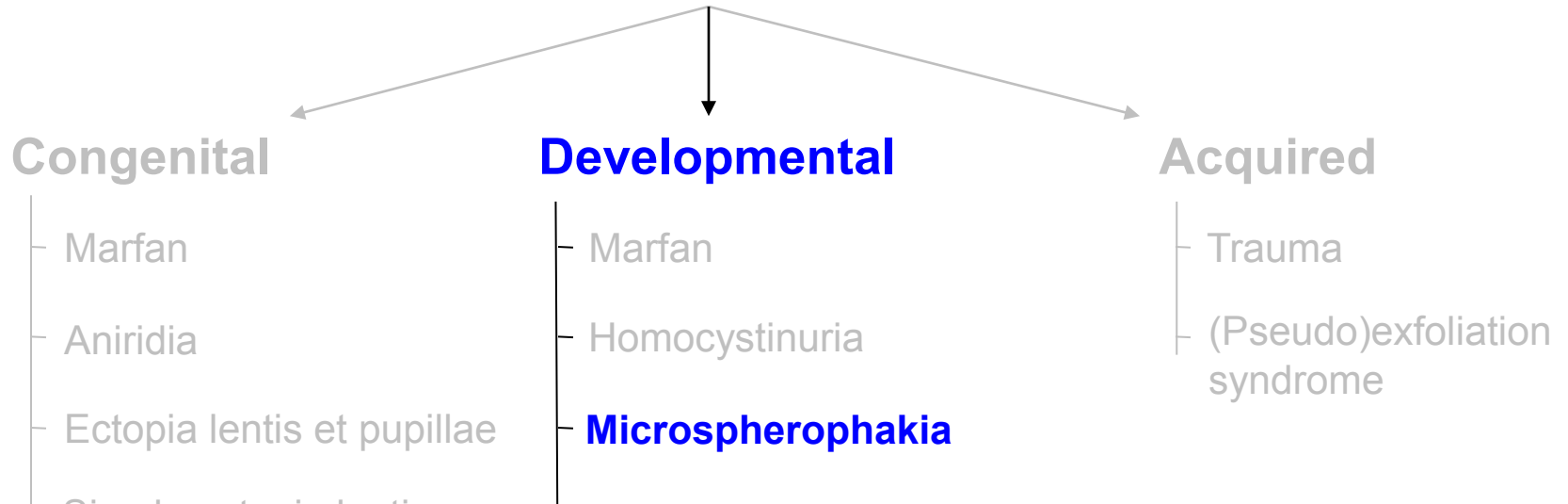


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Because the lens is small, it has a short radius of curvature.

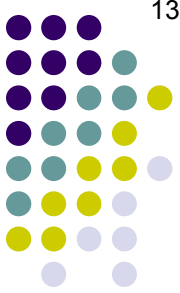


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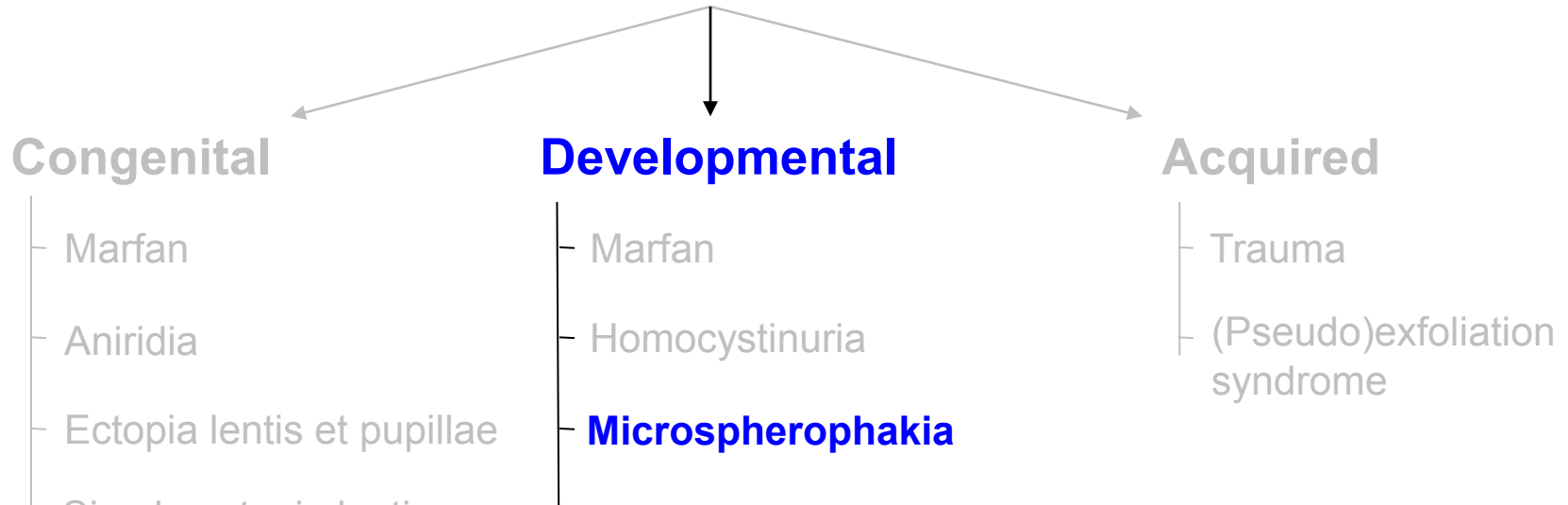


*In a few words, how would you describe the shape of a microspherophakic lens?*  
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*Pts with microspherophakia are almost always high myopes. Why?*  
Because the lens is small, it has a short radius of curvature. Further, because it is spherical, it is more curved than is a normal lens.



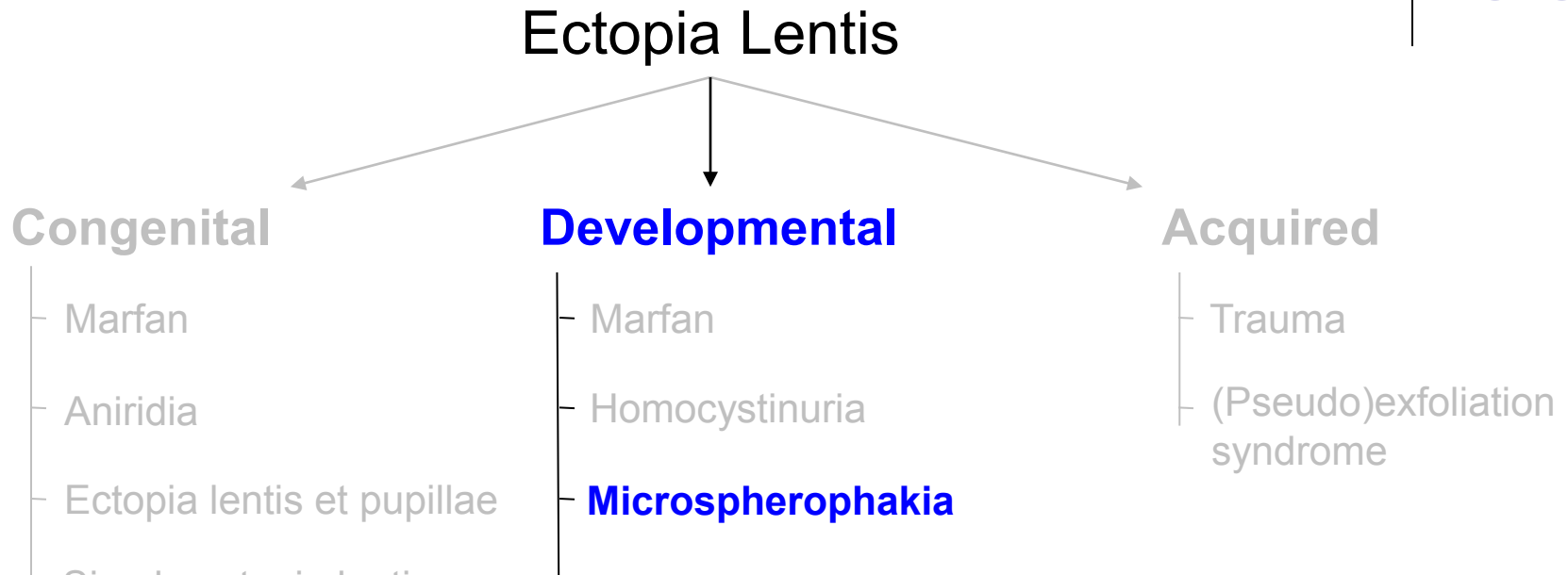
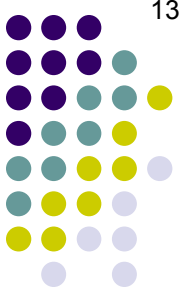
## Ectopia Lentis



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*Pts with microspherophakia are almost always high myopes. Why?*

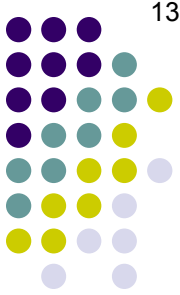
Because the lens is small, it has a short radius of curvature. Further, because it is spherical, it is more curved than is a normal lens. These two factors give the m'spheric lens vastly more converging power than a normal lens possesses.



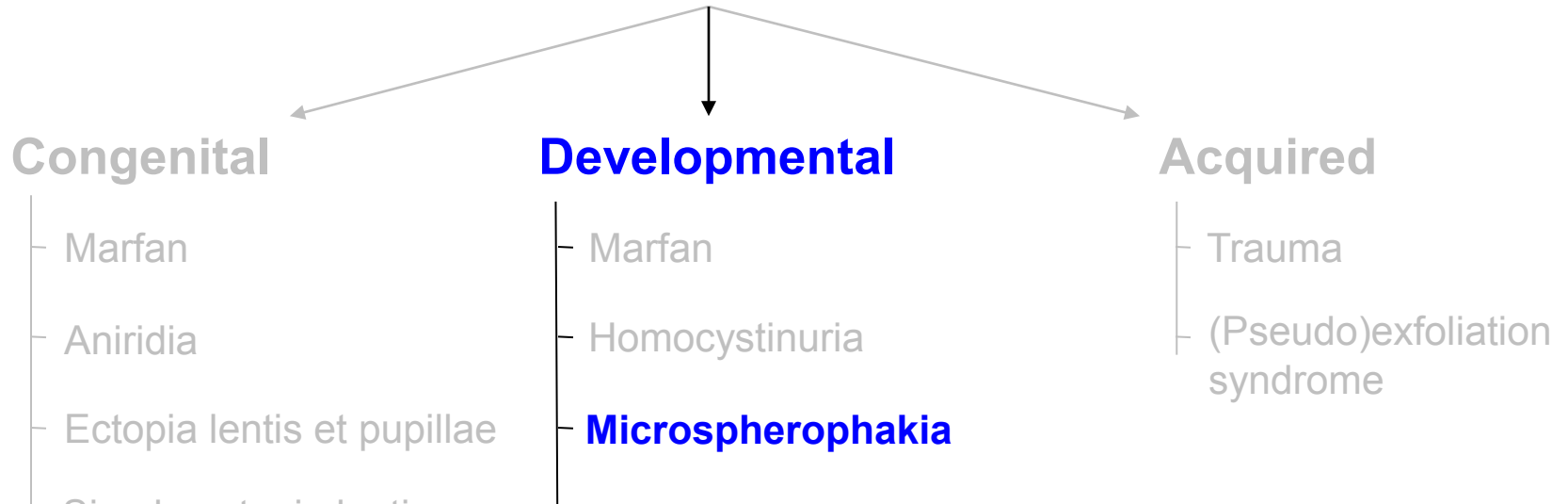
*In a few words, how would you describe the shape of a microspherophakic lens?*  
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*How does this differ from 'run of the mill' high myopia?*

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## Ectopia Lentis

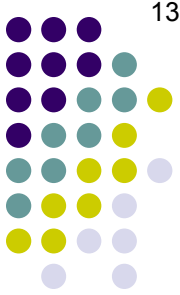


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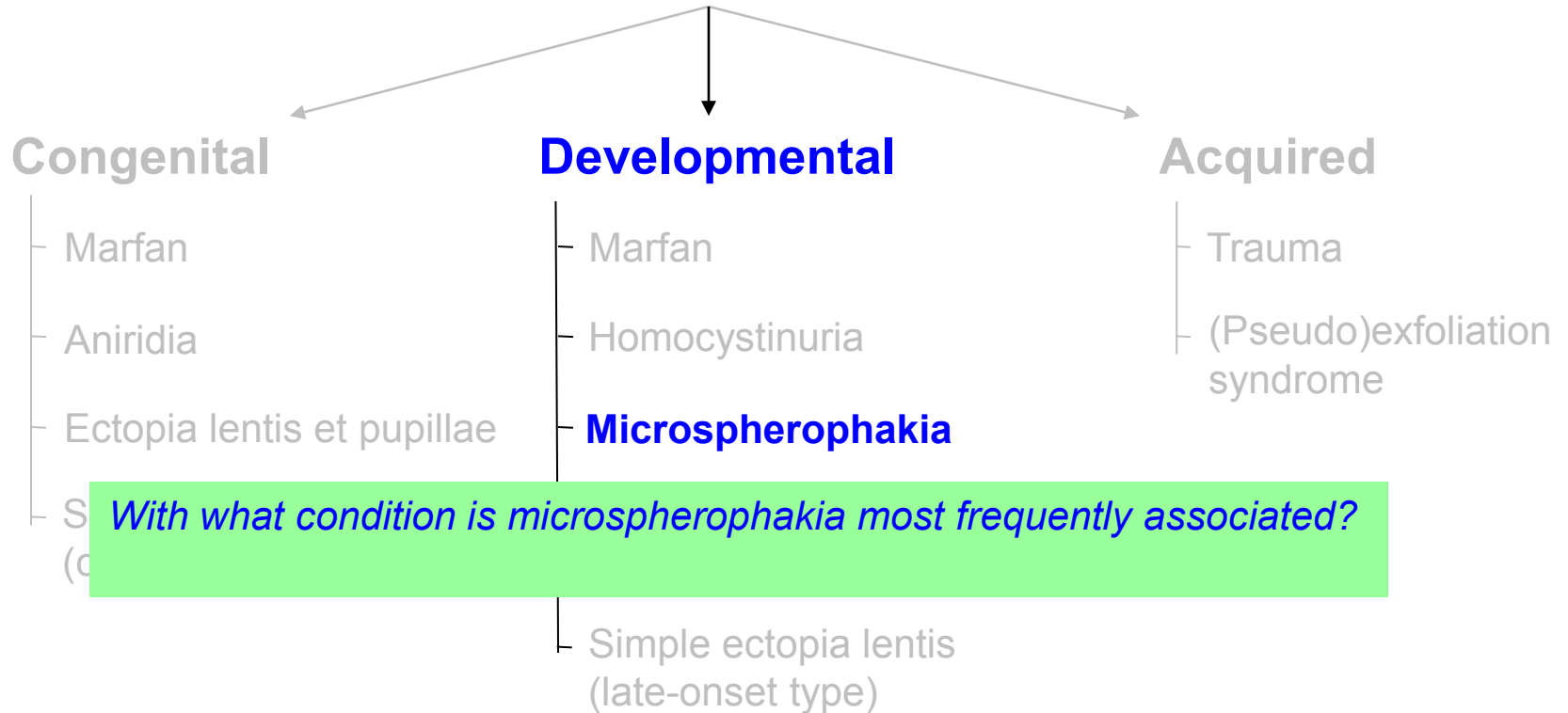
*How does this differ from 'run of the mill' high myopia?*

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.



## Ectopia Lentis



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## Ectopia Lentis

134



### Ectopia Lentis

#### Congenital

- Marfan
- Aniridia
- Ectopia lentis et pupillae
- S
- (c

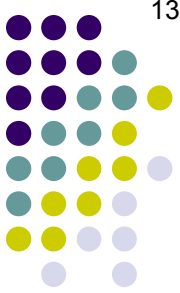
#### Developmental

- Marfan
- Homocystinuria
- **Microspherophakia**
- Simple ectopia lentis (late-onset type)

#### Acquired

- Trauma
- (Pseudo)exfoliation syndrome

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



## Ectopia Lentis

### Congenital

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

### Developmental

- Marfan
- Homocystinuria
- **Microspherophakia**

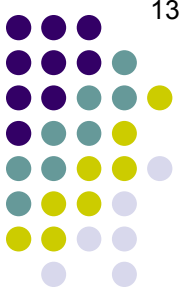
*With what condition is microspherophakia most frequently associated?*

**Weill-Marchesani syndrome**

### Acquired

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



### Ectopia Lentis

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

#### Developmental

- Marfan
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**Weill-Marchesani syndrome**

What are the findings in Weill-Marchesani?

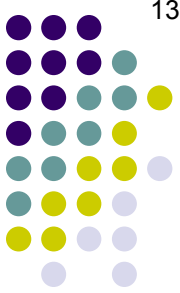
Patients with Weill-Marchesani have:

stature

A

## Ectopia Lentis

137



### Ectopia Lentis

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

#### Developmental

- Marfan
- Homocystinuria
- **Microspherophakia**

*With what condition is microspherophakia most frequently associated?*

**Weill-Marchesani syndrome**

#### Acquired

- Trauma
- (Pseudo)exfoliation syndrome

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

## Ectopia Lentis



Weill-Marchesani syndrome: Short stature



### Ectopia Lentis

#### Congenital

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

#### Developmental

- Marfan
- Homocystinuria
- **Microspherophakia**

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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

fingers

A

## Ectopia Lentis

140



### Ectopia Lentis

#### Congenital

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

#### Developmental

- Marfan
- Homocystinuria
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- (Pseudo)exfoliation syndrome

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**Weill-Marchesani syndrome**

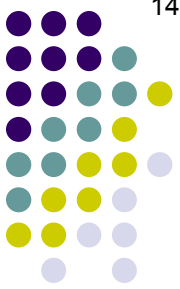
What are the findings in Weill-Marchesani?

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
- Simple ectopia lentis

#### Developmental

- Marfan
- Homocystinuria
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What are the findings in Weill-Marchesani?

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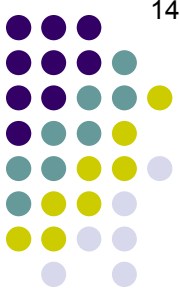
...**short** fingers

joints

A

## Ectopia Lentis

143



### Ectopia Lentis

#### Congenital

- Marfan
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- Ectopia lentis et pupillae
- Simple ectopia lentis

#### Developmental

- Marfan
- Homocystinuria
- **Microspherophakia**

#### Acquired

- Trauma
- (Pseudo)exfoliation syndrome

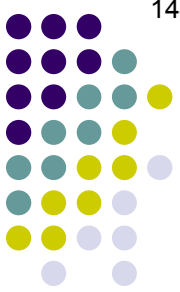
*With what condition is microspherophakia most frequently associated?*

**Weill-Marchesani syndrome**

*What are the findings in Weill-Marchesani?*

Patients with Weill-Marchesani have:

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



### Ectopia Lentis

#### Congenital

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

#### 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?

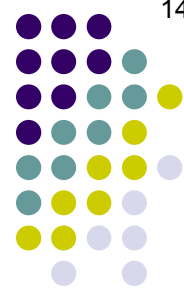
Patients with Weill-Marchesani have:

...**short** stature

...**short** fingers

...**stiff** joints

(Think of it as the opposite of  syndrome)



# Ectopia Lentis

## Ectopia Lentis

### Congenital

**Marfan**

Aniridia

Ectopia lentis et pupillae

### Developmental

**Marfan**

Homocystinuria

**Microspherophakia**

*With what condition is microspherophakia most frequently associated?*

**Weill-Marchesani syndrome**

### Acquired

Trauma

(Pseudo)exfoliation syndrome

*What are the findings in Weill-Marchesani?*

Patients with Weill-Marchesani have:

...**short** stature

(**Tall** stature)

...**short** fingers

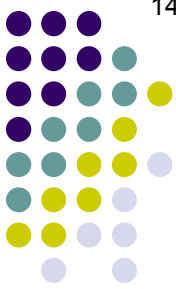
(**Long** fingers)

...**stiff** joints

(**Lax** joints)

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

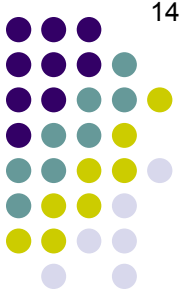
## Ectopia Lentis



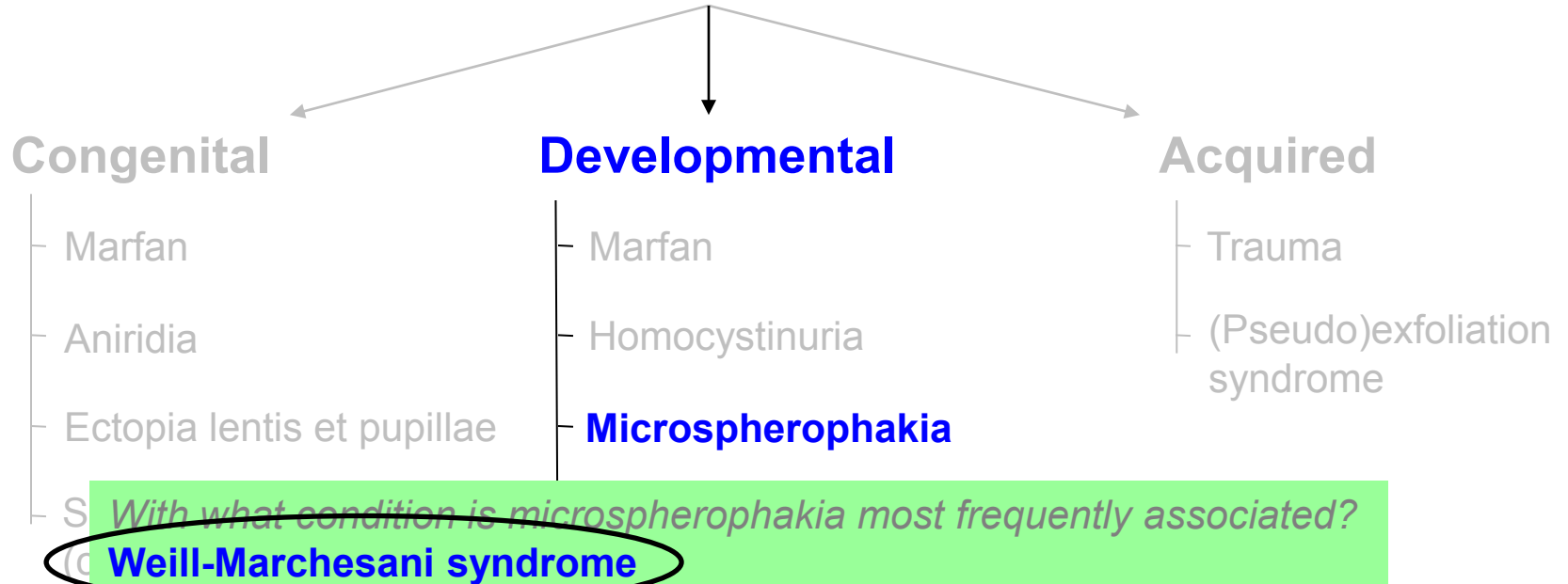
Weill-Marchesani syndrome



Marfan syndrome



## Ectopia Lentis

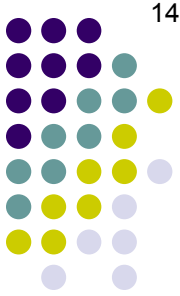


*Weill-Marchesani is strongly associated with microspherophakia. With what conditions is microspherophakia **occasionally** associated?*

A

# Ectopia Lentis

148



## Ectopia Lentis

### Congenital

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

### Developmental

- Marfan
- Homocystinuria
- **Microspherophakia**

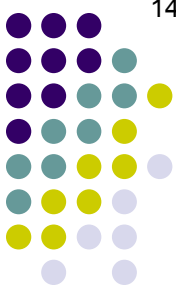
### Acquired

- 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

## Ectopia Lentis

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

**Ruby** = Rubella

**L**owe syndrome

**A**lport syndrome

**M**arfan syndrome

**P**eters anomaly

Ectopia lentis et pupillae

**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 **rub**ella, **L**owe syndrome, **A**lport syndrome, **M**arfan syndrome and **P**eters anomaly

Q

## Ectopia Lentis

150



### Ectopia Lentis

Congenital

**Developmental**

Acquired

Marfan

Marfan

Trauma

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

syndromes

We  
microspherophakia occasionally associated?

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



A

# Ectopia Lentis

## Ectopia Lentis

Congenital

Marfan

**Developmental**

Marfan

Acquired

Trauma

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

exfoliation  
e

We  
 microspherophakia **occasionally** associated?  
 Congenital **rub**ella, **Lowe syndrome**, **A**lport syndrome, Marfan syndrome and **P**eters anomaly

Q

## Ectopia Lentis

152



### Ectopia Lentis

Congenital

**Developmental**

Acquired

Marfan

Marfan

Trauma

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

*What is their classic (nonocular) presenting sign?*

We  
microspherophakia **occasionally** associated?  
Congenital **rub**ella, **Lowe syndrome**, **A**lport syndrome, Marfan syndrome and **P**eters anomaly



A

# Ectopia Lentis

## Ectopia Lentis

Congenital

Marfan

**Developmental**

Marfan

Acquired

Trauma

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

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

We  
 microspherophakia **occasionally** associated?  
 Congenital **rub**ella, **Lowe syndrome**, **A**lport syndrome, Marfan syndrome and **P**eters anomaly



## Ectopia Lentis

### Congenital

Marfan

### Developmental

Marfan

### Acquired

Trauma

*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?*

We  
microspherophakia **occasionally** associated?  
Congenital **rub**ella, **L**owe syndrome, **A**lport syndrome, **M**arfan syndrome and **P**eters anomaly



A

# Ectopia Lentis

## Ectopia Lentis

### Congenital

Marfan

### Developmental

Marfan

### Acquired

Trauma

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

Familial oculorenal syndromes

*What is their classic (nonocular) presenting sign?*

Hematuria

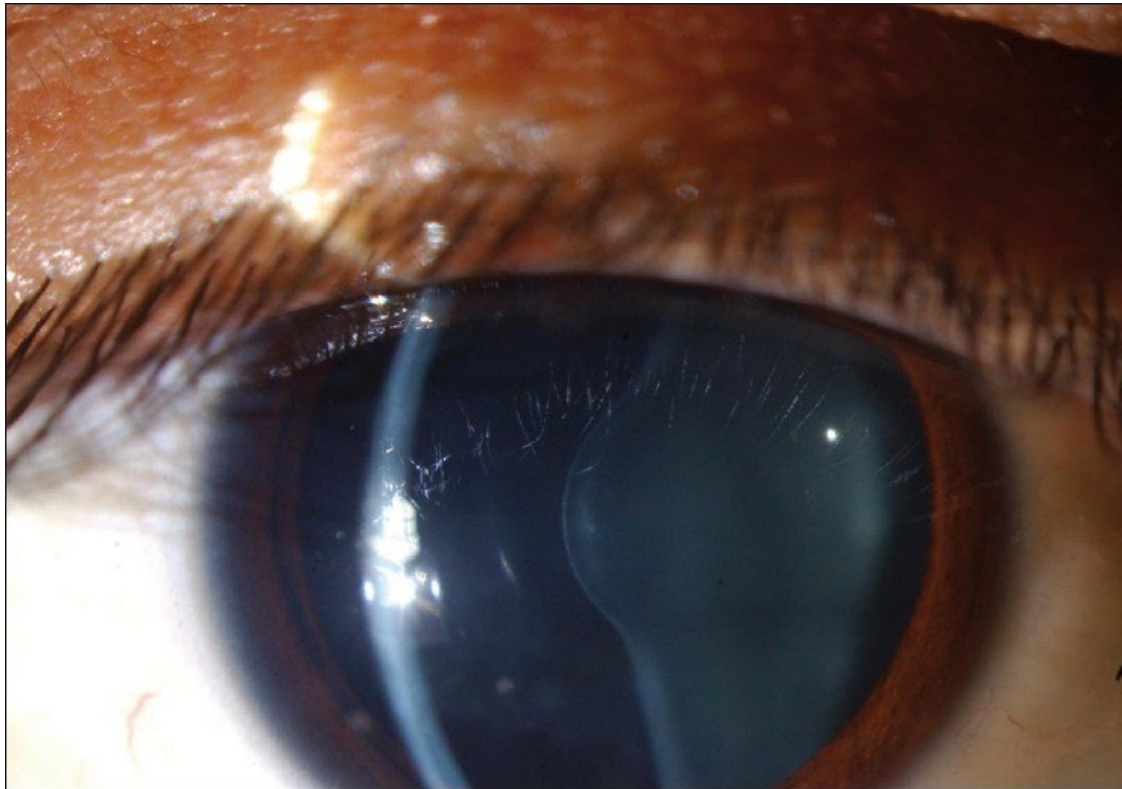
*Microspherophakia is **not** the classic lens finding in the oculorenal syndromes (and should not be the first one out of your mouth if pimped about them). What is?*

**Lenticonus**

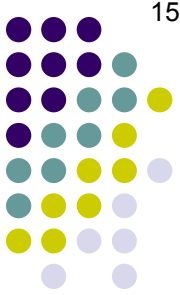
*We microspherophakia **occasionally** associated?*

Congenital **rubella**, **L**owe syndrome, **A**lport syndrome, Marfan syndrome and **P**eters anomaly

## Ectopia Lentis



Anterior lenticonus in Alport syndrome



## Ectopia Lentis

### Congenital

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

### 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 **rub**ella, **L**owe syndrome, **A**lport syndrome, **M**arfan syndrome and **P**eters anomaly

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

A form of



A

# Ectopia Lentis

## Ectopia Lentis

### Congenital

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- Aniridia
- Ectopia lentis et pupillae
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### Developmental

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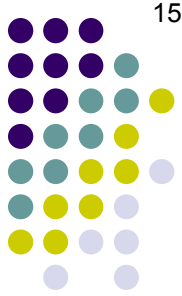
Congenital **rub**ella, **L**owe syndrome, **A**lport syndrome, **M**arfan syndrome and **P**eters anomaly

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

Q

## Ectopia Lentis

159



### Ectopia Lentis

#### Congenital

- Marfan
- Aniridia
- Ectopia lentis et pupillae
- Sturge-Weaver

#### Developmental

- Marfan
- Homocystinuria
- **Microspherophakia**

#### Acquired

- Trauma
- (Pseudo)exfoliation syndrome

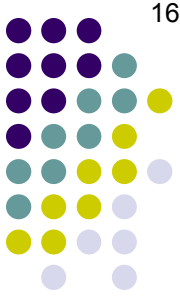
*In terms of the fundamental embryological disorder involved, what sort of condition is associated?*

Weill-Marchesani  
microspherophakia  
Congenital

conditions is

and **Peters anomaly**

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



A

# Ectopia Lentis

## Ectopia Lentis

### Congenital

- Marfan
- Aniridia
- Ectopia lentis et pupillae
- S
- (C)

### Developmental

- Marfan
- Homocystinuria
- **Microspherophakia**

### Acquired

- Trauma
- (Pseudo)exfoliation syndrome

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

associated?

Weill-Marchesani  
microspherophakia  
Congenital

conditions is

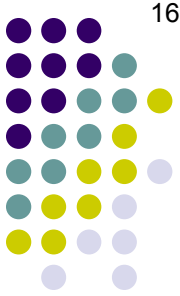
and **Peters anomaly**

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

Q

## Ectopia Lentis

161



### Ectopia Lentis

#### Congenital

- Marfan
- Aniridia
- Ectopia lentis et pupillae
- S

#### Developmental

- Marfan
- Homocystinuria
- **Microspherophakia**

#### Acquired

- Trauma
- (Pseudo)exfoliation syndrome

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

**We** A neurocristopathy

What is a neurocristopathy?

associated?

conditions is

and **Peters anomaly**

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



A

# Ectopia Lentis

## Ectopia Lentis

### Congenital

- Marfan
- Aniridia
- Ectopia lentis et pupillae
- Sturge-Weaver

### Developmental

- Marfan
- Homocystinuria
- Microspherophakia**

### Acquired

- Trauma
- (Pseudo)exfoliation syndrome

*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

*associated?*

*conditions is*

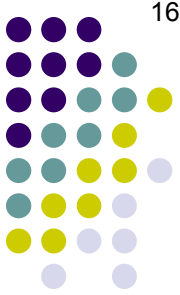
and **Peters anomaly**

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

Q

# Ectopia Lentis

163



## Ectopia Lentis

### Congenital

- Marfan
- Aniridia
- Ectopia lentis et pupillae

### Developmental

- Marfan
- Homocystinuria
- **Microspherophakia**

### Acquired

- Trauma
- (Pseudo)exfoliation syndrome

*How does Peters anomaly present?*

*associated?*

*conditions is*

and **Peters anomaly**



# Ectopia Lentis

## Ectopia Lentis

### Congenital

- Marfan
- Aniridia
- Ectopia lentis et pupillae

### Developmental

- Marfan
- Homocystinuria
- **Microspherophakia**

### Acquired

- Trauma
- (Pseudo)exfoliation syndrome

*How does Peters anomaly present?*

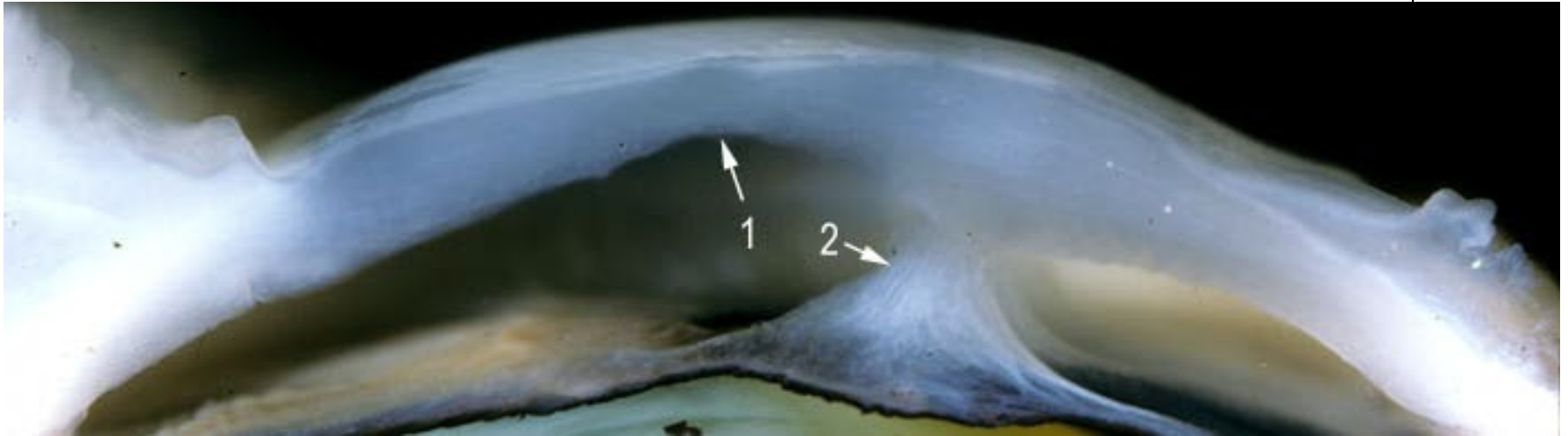
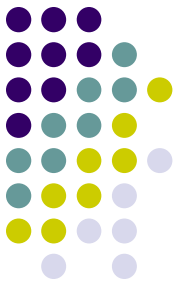
With a defect of the posterior central cornea, including the absence of Descemet's and subjacent endothelium. Adhesions extending from the iris to the posterior corneal defect are often present.

associated?

conditions is

and **Peters anomaly**

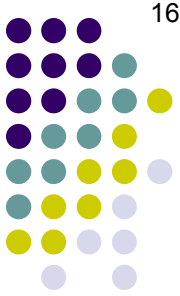
# Ectopia Lentis



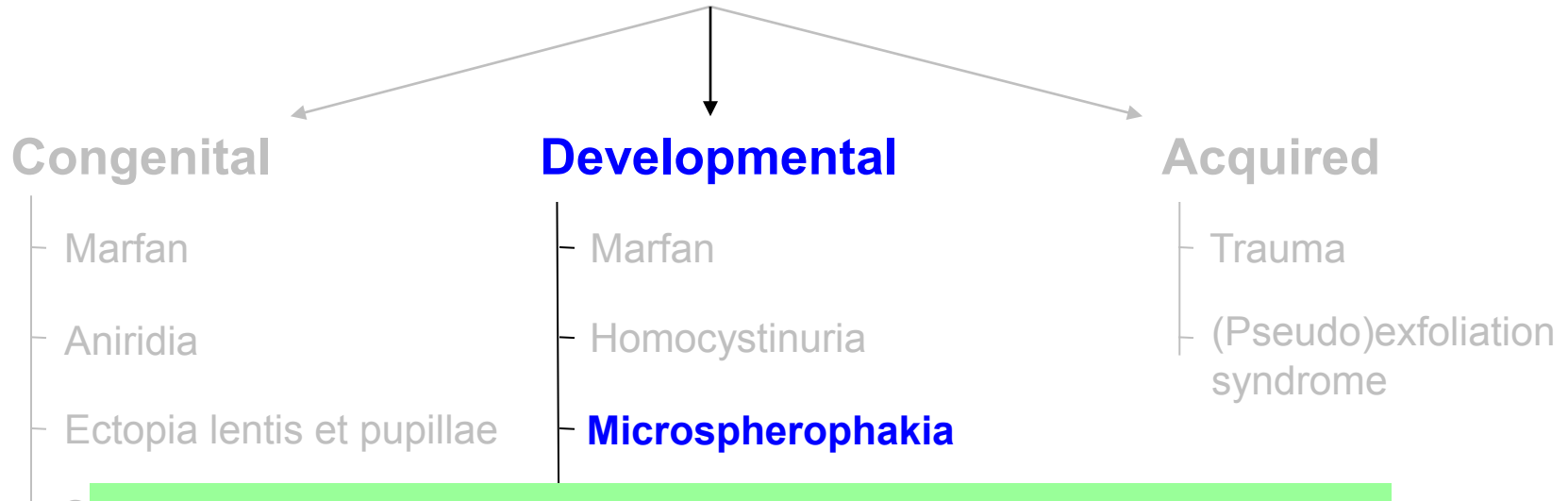
1. Defect of the posterior central cornea, including the absence of Descemet's and subjacent endothelium

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

Peters anomaly



## Ectopia Lentis



*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?*

associated?

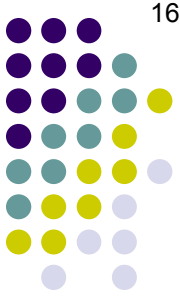
conditions is

and **Peters anomaly**

# A

## Ectopia Lentis

167



### 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.

associated?

conditions is

and **Peters anomaly**

## Ectopia Lentis



Peters anomaly: Hazy cornea

# Ectopia Lentis



## Ectopia Lentis

### Congenital

- Marfan
- Aniridia

### Developmental

- Marfan

### Acquired

- Trauma

Note: There are two  
S's  
and two  
E's

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

S  
T  
U  
M  
Peters anomaly  
E  
D

of Descemet's and subja  
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.

and **Peters anomaly**



A

# Ectopia Lentis

## Ectopia Lentis

### Congenital

- Marfan
- Aniridia

### Developmental

- Marfan

### Acquired

- Trauma

Note: There are two  
S's  
and two  
E's

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

**S**clerocornea; **S**tromal dystrophy (CHSD)

**T**rauma (eg, forcep injury)

**U**lcer

**M**ucopolysaccharidosis

**P**eters anomaly

**E**ndothelial dystrophy (CHED); **E**levated IOP (congenital glaucoma)

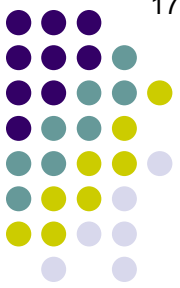
**D**ermoid of the cornea

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.

and **Peters anomaly**



## Ectopia Lentis

*Why is the term 'aniridia' technically a misnomer?*

### Congenital

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

### Acquired

- Trauma
- (pseudo)exfoliation  
syndrome



## Ectopia Lentis

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

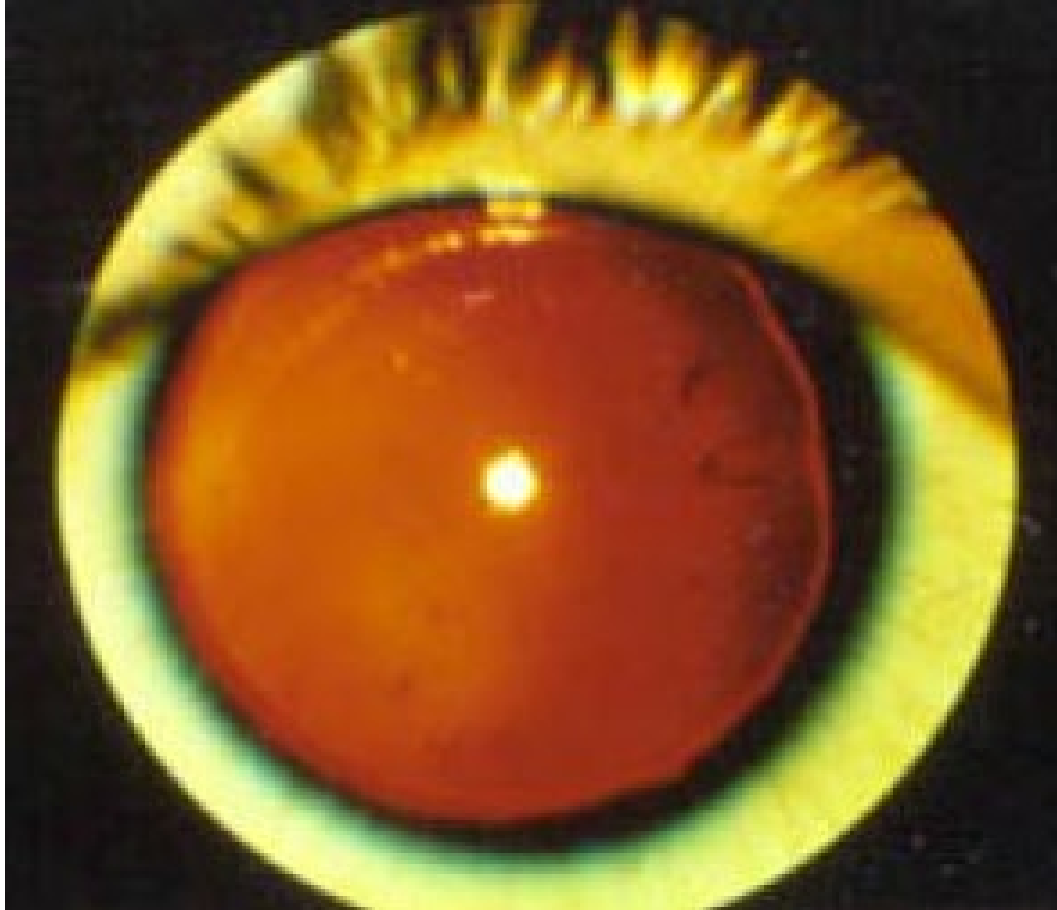
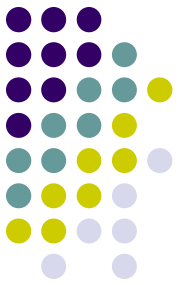
### Congenital

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

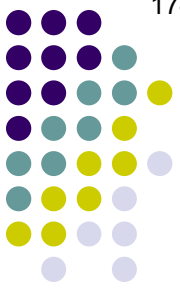
### Acquired

- Trauma
- (pseudo)exfoliation  
syndrome

## Ectopia Lentis



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

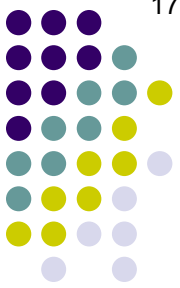
*Is aniridia usually unilateral, or bilateral?*

### Congenital

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

### Acquired

- Trauma
- (pseudo)exfoliation syndrome



## Ectopia Lentis

*Why is the term 'aniridia' technically a misnomer?*

Because a rudimentary iris root is always present

*Is aniridia usually unilateral, or bilateral?*

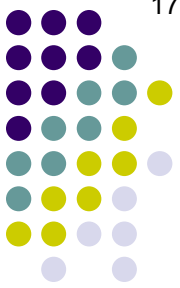
It is almost always bilateral

### Congenital

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

### Acquired

- Trauma
- (pseudo)exfoliation  
syndrome



## 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

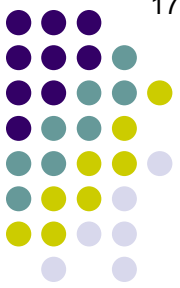
*Is nystagmus commonly associated with aniridia?*

## Congenital

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

## Acquired

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- (pseudo)exfoliation syndrome



## 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?*

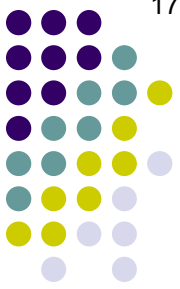
Yes

## Congenital

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

## Acquired

- Trauma
- (pseudo)exfoliation syndrome



## 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** present?*

Yes

*Is this a sensory or a motor nystagmus?*

## Congenital

Marfan

**Aniridia**

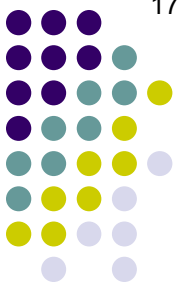
Ectopia lentis

Simple ectopia  
(congenital type)

## Acquired

Trauma

(pseudo)exfoliation  
syndrome



## Ectopia Lentis

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*Is aniridia usually unilateral, or bilateral?*

It is almost always bilateral

*Is **nystagmus** present?*

Yes

*Is this a sensory or a motor nystagmus?*

Sensory

## Congenital

Marfan

**Aniridia**

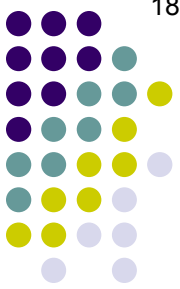
Ectopia lentis

Simple ectopia  
(congenital type)

## Acquired

Trauma

(pseudo)exfoliation  
syndrome



## 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** present?  
Yes

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

## Congenital

Marfan

**Aniridia**

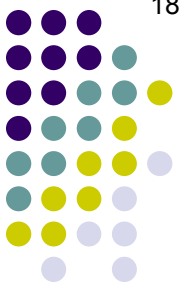
What anatomic abnormalities are responsible for the poor vision in aniridia?  
(hint: It's not the lack of an iris)

(congenital type)

## Acquired

Trauma

(pseudo)exfoliation  
syndrome



A

## 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** present? Is this a sensory or a motor nystagmus?

**Sensory**

### Congenital

Marfan

**Aniridia**

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

(congenital type)

### Acquired

Trauma

(pseudo)exfoliation  
syndrome



## 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?*

### Congenital

Marfan

**Aniridia**

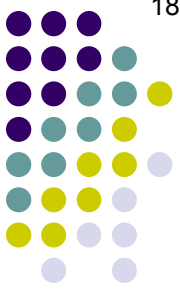
Ectopia lentis

Simple ectopia  
(congenital type)

### Acquired

Trauma

(pseudo)exfoliation  
syndrome



A

## Ectopia Lentis

### Ectopia Lentis

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*Is nystagmus commonly associated with aniridia?*

Yes

*With what developmental 'complex' is aniridia associated?*

The *WAGR* complex

### Congenital

Marfan

**Aniridia**

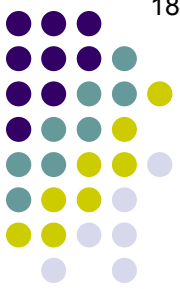
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*Is nystagmus commonly associated with aniridia?*

Yes

*With what developmental complex is it associated?*

**The WAGR complex**

**WAGR complex** consists of:

**W**

**A**niridia

**G**

**R**

## Congenital

Marfan

**Aniridia**

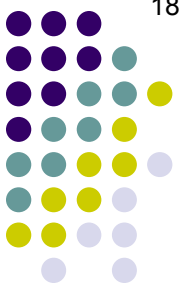
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*Is nystagmus commonly associated with aniridia?*

Yes

*With what developmental anomalies is aniridia associated?*

**The WAGR complex**

**WAGR complex** consists of:

**W**ilms tumor

**A**niridia

**G**enitourinary abnormalities

**R**etardation

### Congenital

Marfan

**Aniridia**

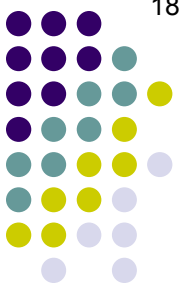
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Marfan

### Aniridia

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Ectopia lentis

**WAGR complex** consists of:

*With what developmental anomalies is it associated?*

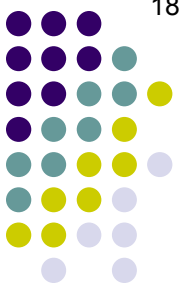
**Wilms tumor**

Simple ectopia

**Aniridia**

(congenital type)

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



## Ectopia Lentis

*Why is the term 'aniridia' technically a misnomer?*

Because a rudimentary iris root is always present

### Congenital

*Is aniridia usually unilateral, or bilateral?*

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Marfan

### Aniridia

*Is nystagmus commonly associated with aniridia?*

Yes

Ectopia lentis

**WAGR complex** consists of:

*With what developmental anomalies is it associated?*

**Wilms tumor**

Simple ectopia

**The WAGR complex**

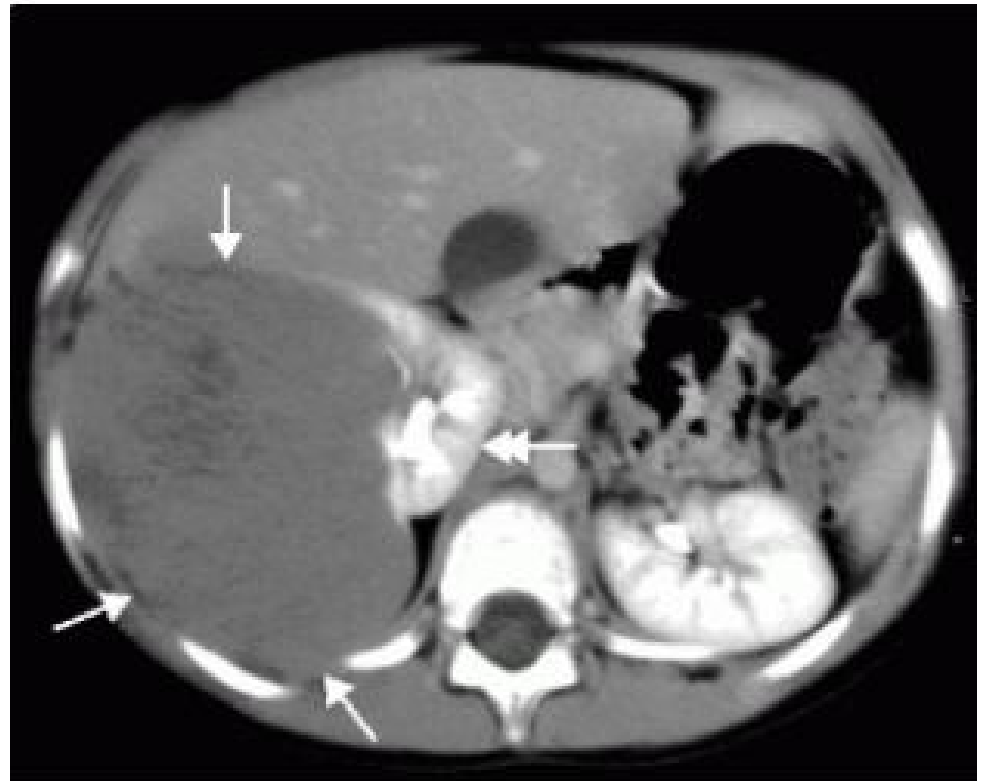
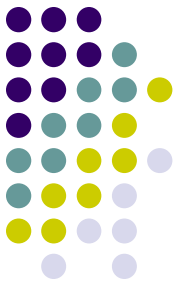
**Aniridia**

(congenital type)

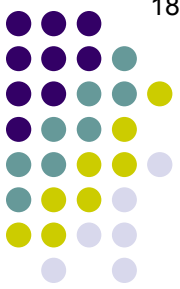
*What is the noneponymous name for Wilms tumor (ie, what sort of tumor is it)?*

**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

## Congenital

Marfan

## Aniridia

Ectopia lentis

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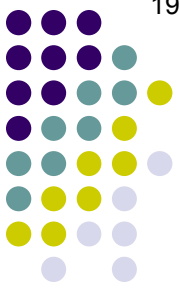
The WAGR complex

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

## Acquired

Trauma

(pseudo)exfoliation  
syndrome



### Ectopia Lentis

*Why is the term 'aniridia' technically a misnomer?*

Because a rudimentary iris root is always present

### Congenital

*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

Marfan

**Aniridia**

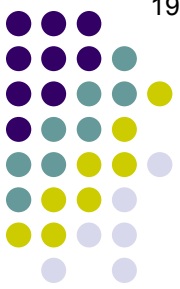
Ectopia lentis

Simple ectopia  
(congenital type)

quired

trauma

(pseudo)exfoliation  
syndrome



## Ectopia Lentis

*Why is the term 'aniridia' technically a misnomer?*

Because a rudimentary iris root is always present

## Congenital

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It is almost always bilateral

Marfan

**Aniridia**

*Is nystagmus commonly associated with aniridia?*

Yes

Ectopia lentis

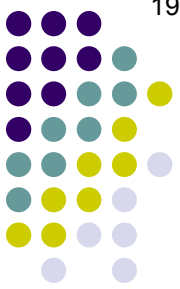
*With what developmental 'complex' is aniridia associated?*

The *WAGR* complex

Simple ectopia  
(congenital type)

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

No, only those in which the genetic mutation is sporadic



# 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?*

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*With what developmental 'complex' is aniridia associated?*

The WAGR complex

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No, only those in which the genetic mutation is sporadic

## Congenital

Marfan

### Aniridia

Ectopia lentis

Simple ectopia  
(congenital type)

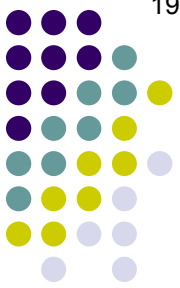
## Acquired

Trauma

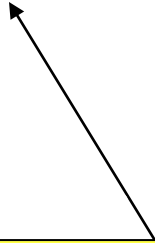
(pseudo)exfoliation  
syndrome

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

## Ectopia Lentis

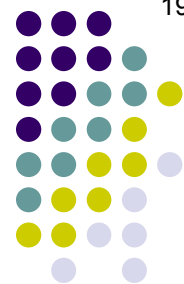


**Nystagmus**



*We mentioned that aniridia is associated with nystagmus...*

*One final point regarding aniridia:*



# Q

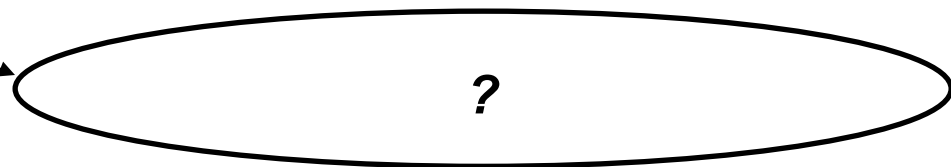
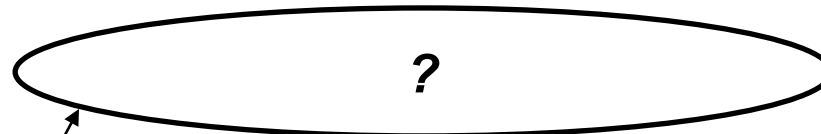
## Ectopia Lentis

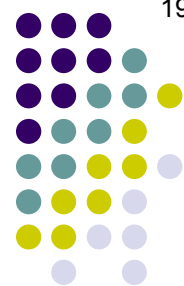
Nystagmus

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

*(Hints forthcoming)*

*One final point  
regarding aniridia:*





# Q/A

## Ectopia Lentis

Nystagmus

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

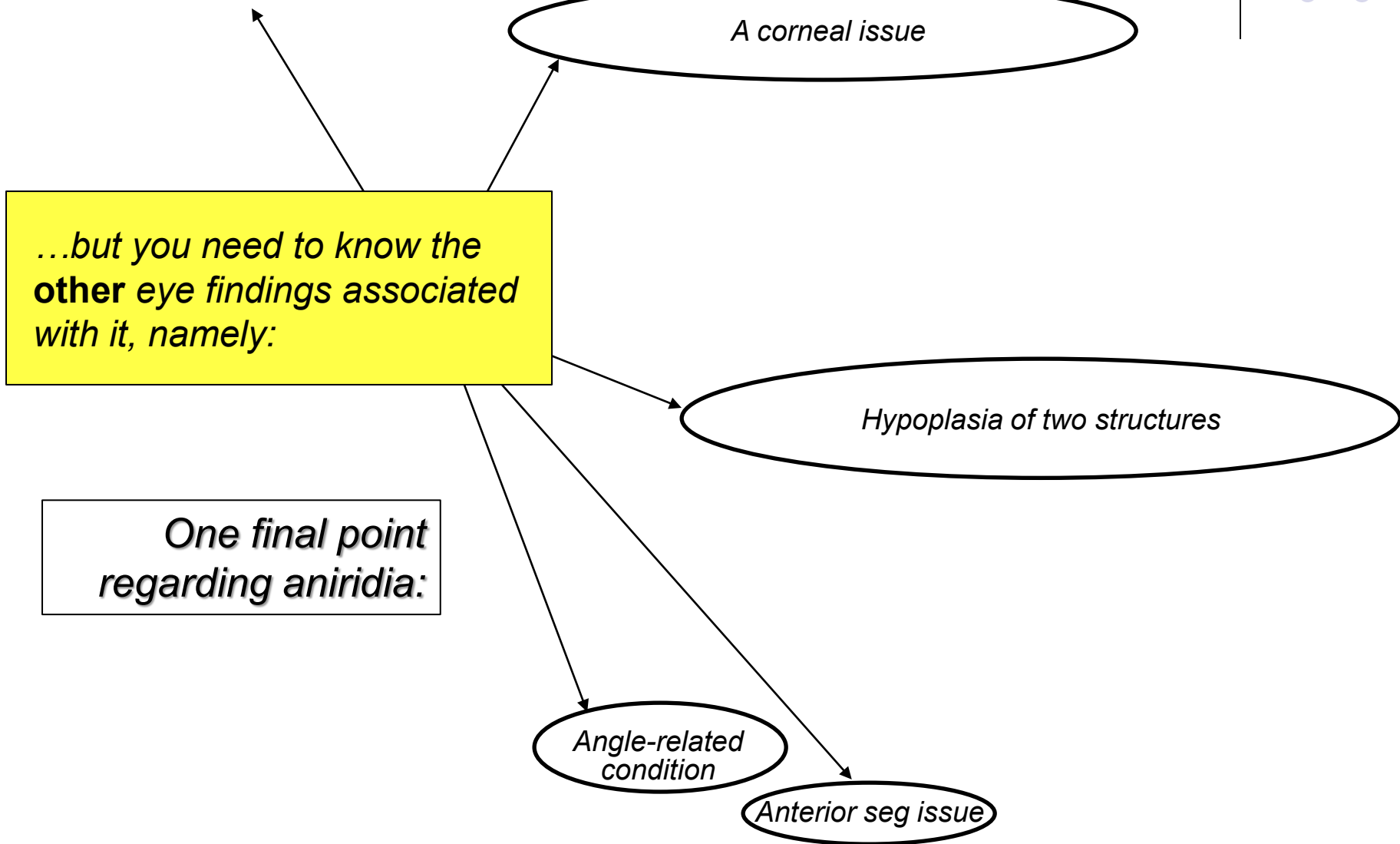
*A corneal issue*

*Hypoplasia of two structures*

*One final point regarding aniridia:*

*Angle-related condition*

*Anterior seg issue*





## Ectopia Lentis

# A

Nystagmus

limbal stem cell deficiency

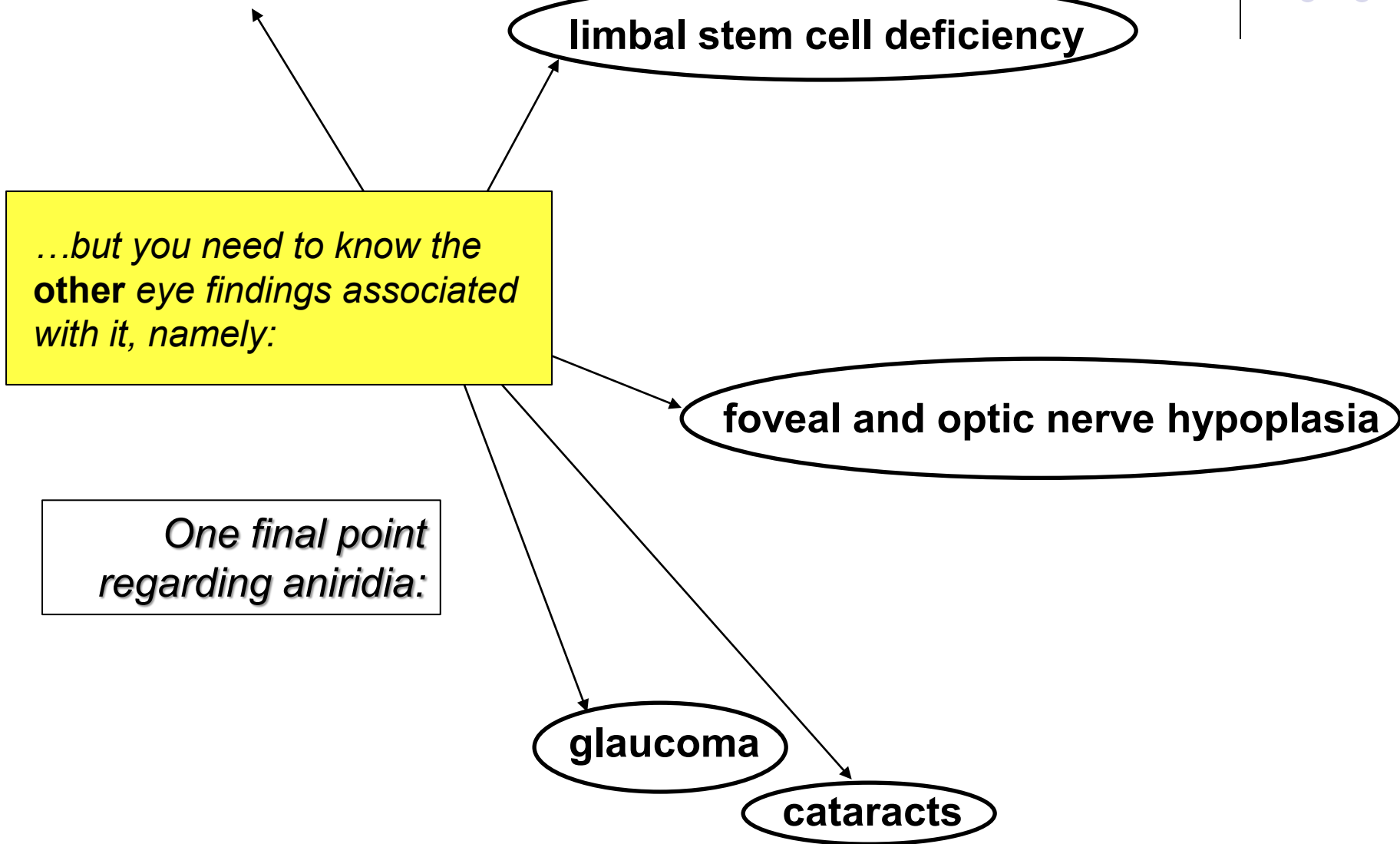
*...but you need to know the other eye findings associated with it, namely:*

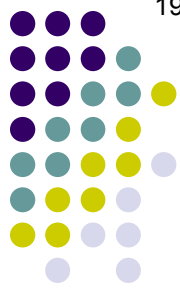
foveal and optic nerve hypoplasia

*One final point regarding aniridia:*

glaucoma

cataracts





## Ectopia Lentis

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

*One final point  
regarding aniridia:*

glaucoma

cataracts



Q

## 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

# A

*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)**

congenital glaucoma

**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?*

### Congenital

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

congenital glaucoma

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



# A

## 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

### Congenital

Marfan

Aniridia

Ectopia lentis et pupillae

**Simple ectopia lentis  
(congenital type)**

congenital glaucoma

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



Q

## 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  
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congenital glaucoma

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## Ectopia Lentis

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Bilateral

### Congenital

Marfan

Aniridia

Ectopia lentis et pupillae

**Simple ectopia lentis  
(congenital type)**

congenital glaucoma

**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?*

### Congenital

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

congenital glaucoma

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



# A

## Ectopia Lentis

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*In what direction are the lenses displaced?*

Superotemporal

### Congenital

Marfan

Aniridia

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(congenital type)**

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**Simple ectopia lentis  
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## Ectopia Lentis

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*How common is it?*

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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?*

### Congenital

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

congenital glaucoma

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



# A

## Ectopia Lentis

*What is simple ectopia lentis?*

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*How common is it?*

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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

### Congenital

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

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

Q

*What is ectopia lentis et pupillae?*

## Congenital

Marfan

Aniridia

**Ectopia lentis et pupillae**

Simple ectopia lentis  
(congenital type)

Microspherophakia

Buphthalmos 2° to  
congenital glaucoma

Simple ectopia lentis  
(late-onset type)

syndrome

tion

**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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Marfan

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syndrome

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*What is ectopia lentis et pupillae?*

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*How common is it?*

It is very rare

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

--?

--?

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

--They are very miotic vs dilated and dilate well vs poorly

--?

## Congenital

Marfan

Aniridia

**Ectopia lentis et pupillae**

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

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Marfan

Aniridia

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Simple ectopia lentis  
(congenital type)

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Buphthalmos 2° to  
congenital glaucoma

Simple ectopia lentis  
(late-onset type)

syndrome

tion

## Ectopia Lentis

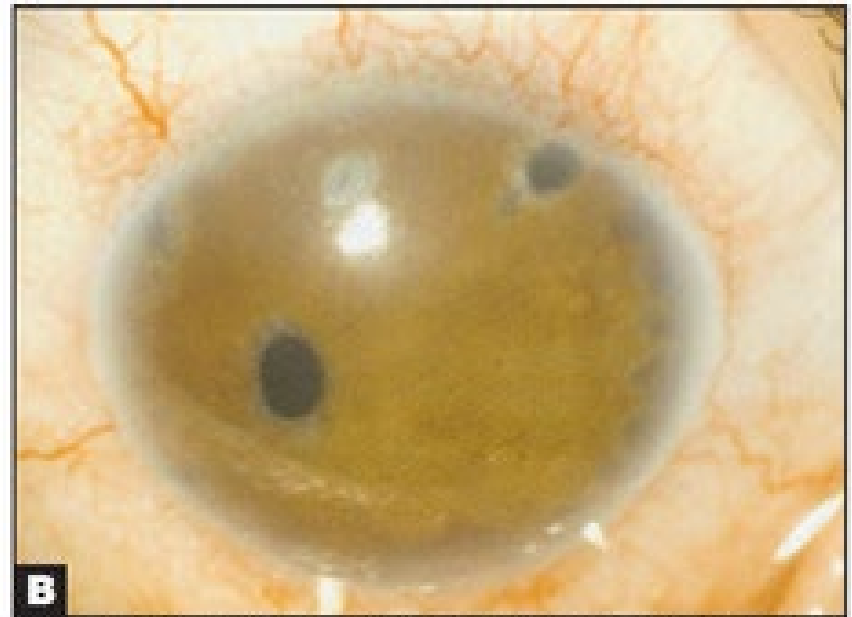
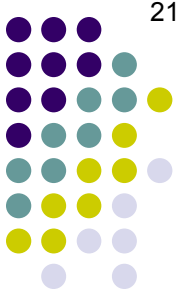


Figure 1. Preoperative view of both eyes in a patient with bilateral ectopia lentis et pupillae syndrome showing several lesions on the iris due to previous laser therapy. (A) Right eye. (B) Left eye.

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?*

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Bilateral

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

--They are very miotic, and dilate poorly

--They are 'something -like' in shape

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Marfan

Aniridia

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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

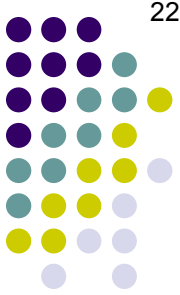
Buphthalmos 2° to  
congenital glaucoma

Simple ectopia lentis  
(late-onset type)

syndrome

tion

## Ectopia Lentis



*Ectopia lentis et pupillae*: Bilateral inferonasal displacement of slit(ish) pupils

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?*

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

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--They are slit-like in shape

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

## Congenital

Marfan

Aniridia

**Ectopia lentis et pupillae**

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--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 [redacted], lenses [redacted] ion

## Congenital

Marfan

Aniridia

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Simple ectopia lentis  
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*The pupils typically have two further abnormalities—what are they?*

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--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)

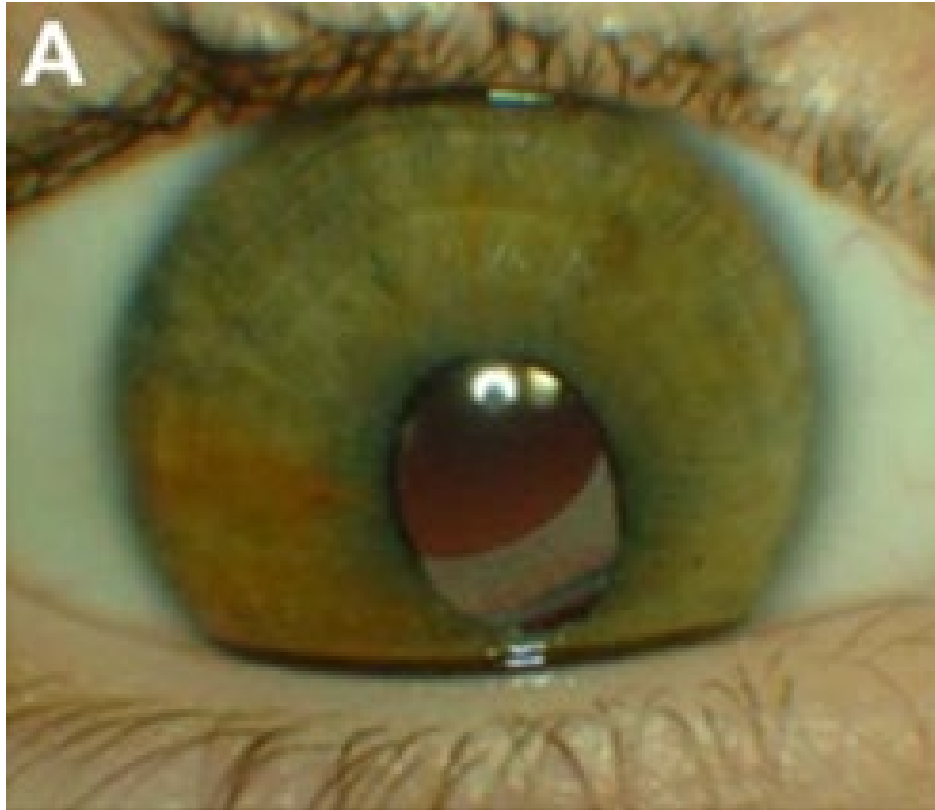
Microspherophakia

Buphthalmos 2° to  
congenital glaucoma

Simple ectopia lentis  
(late-onset type)

Werner's syndrome

## Ectopia Lentis



Ectopia lentis et pupillae: Pupil displaced inferonasal; lens, superotemporal

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 and lenses are displaced in opposite directions—why?*

--They are

--They are

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

## Congenital

Marfan

Aniridia

**Ectopia lentis et pupillae**

Simple ectopia lentis  
(congenital type)

In opposite directions—pupils inferotemporal, lenses superonasal

Microspherophakia

Buphthalmos 2° to  
congenital glaucoma

Simple ectopia lentis  
(late-onset type)

tion

A

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*How common is it?*

It is very rare

*Is it unilateral, or bilateral?*

Bilateral

*The pupils and lenses are displaced in opposite directions, aren't they?*

--They are

--They are

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**

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*What is ectopia lentis et pupillae?*

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

## Congenital

Marfan

Aniridia

**Ectopia lentis et pupillae**

Simple ectopia lentis  
(congenital type)

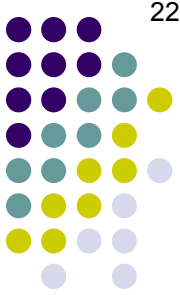
***In what direction are the pupils and lenses displaced?***

In opposite directions—pupils inferotemporal, lenses superonasal  
syndrome

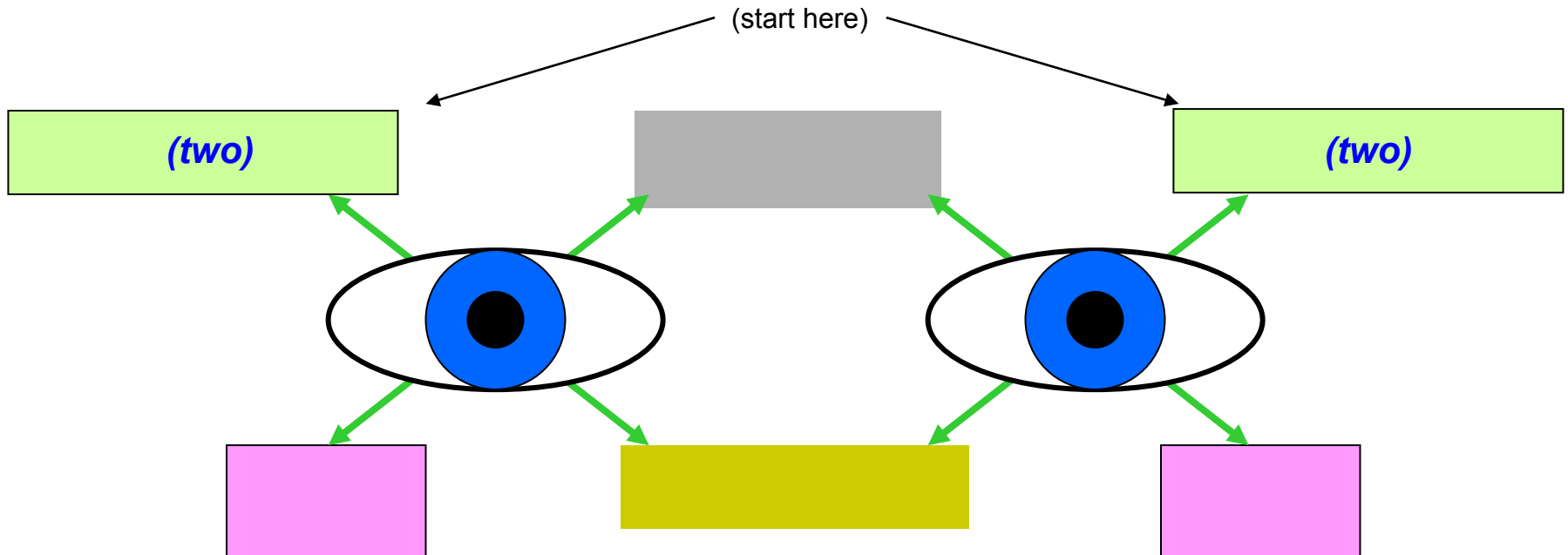
Microspherophakia

Buphthalmos 2° to  
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Simple ectopia lentis  
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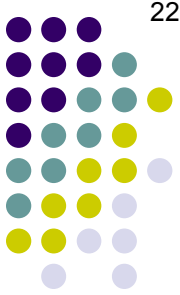
Lens subluxation: Which conditions are associated with displacement in which directions?



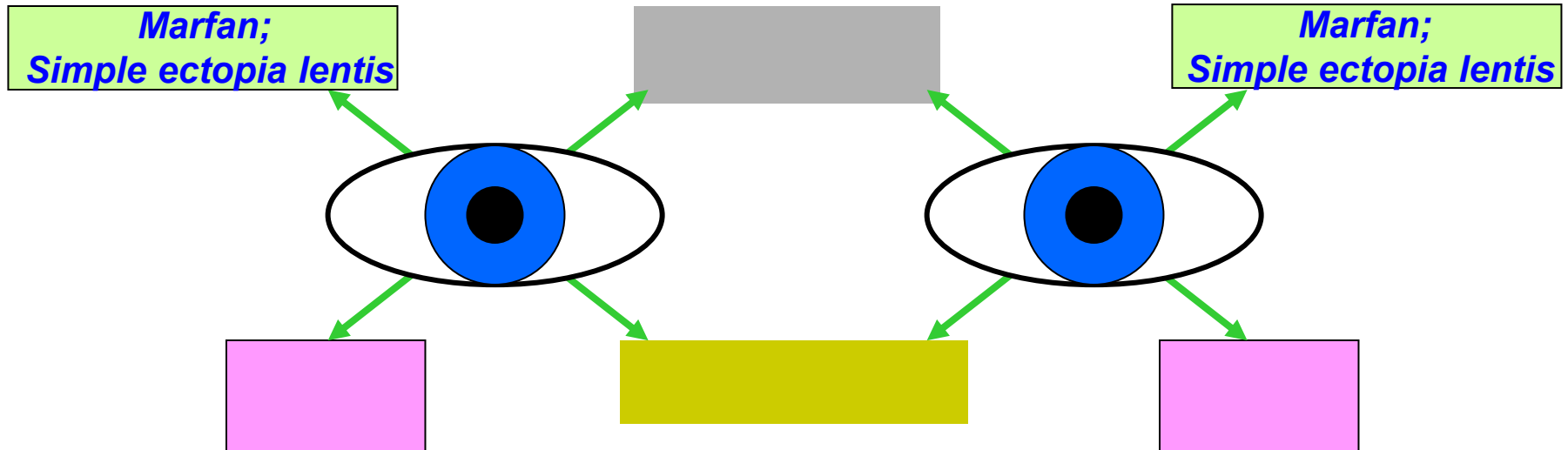
# A

## Ectopia Lentis

229



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



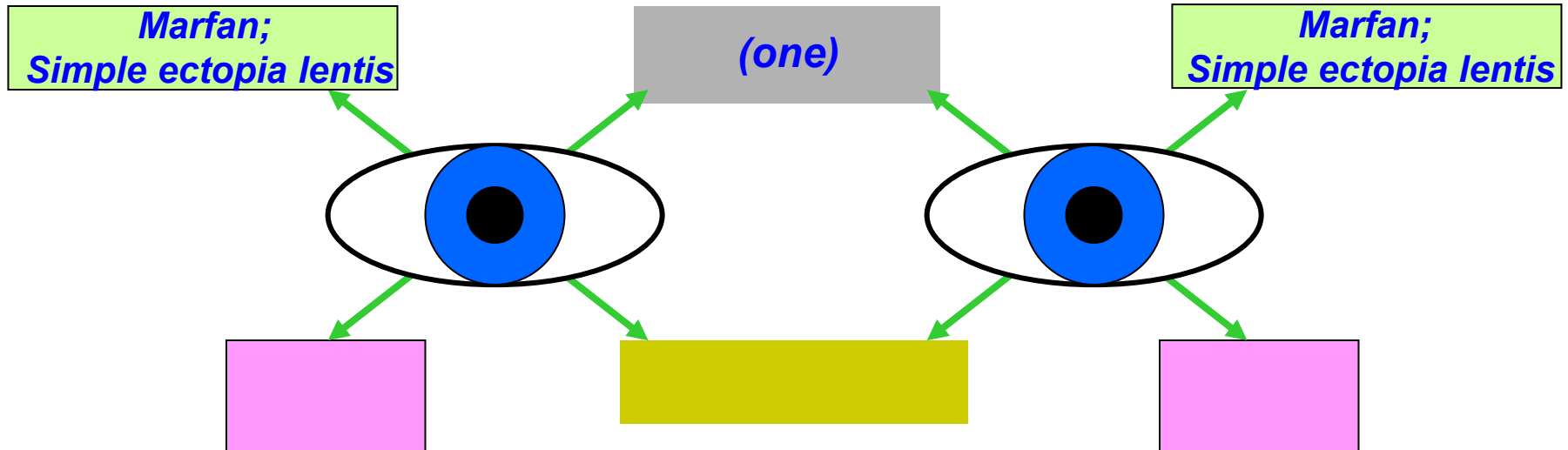
Q

## Ectopia Lentis

230



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



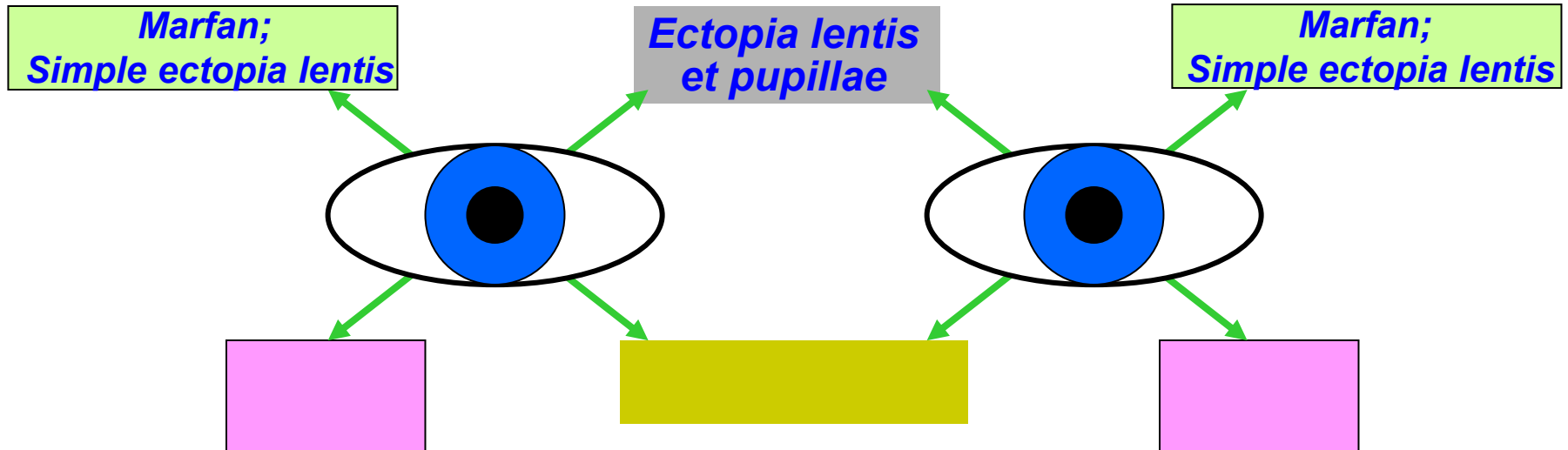
# A

## Ectopia Lentis

231



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



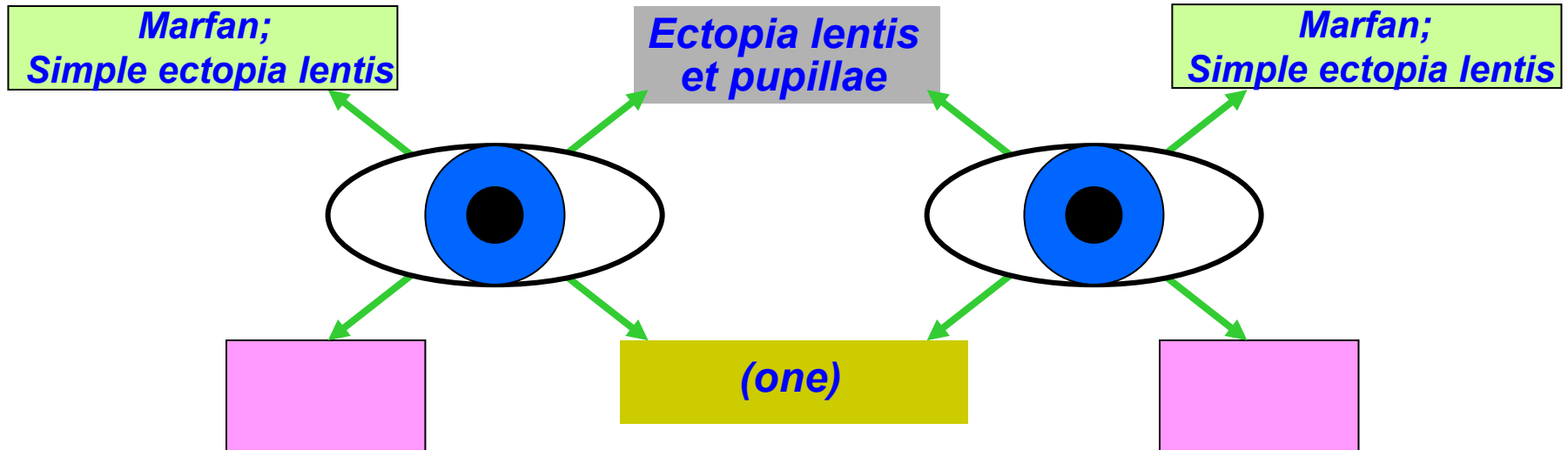
Q

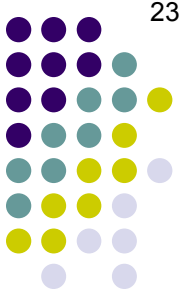
## Ectopia Lentis

232



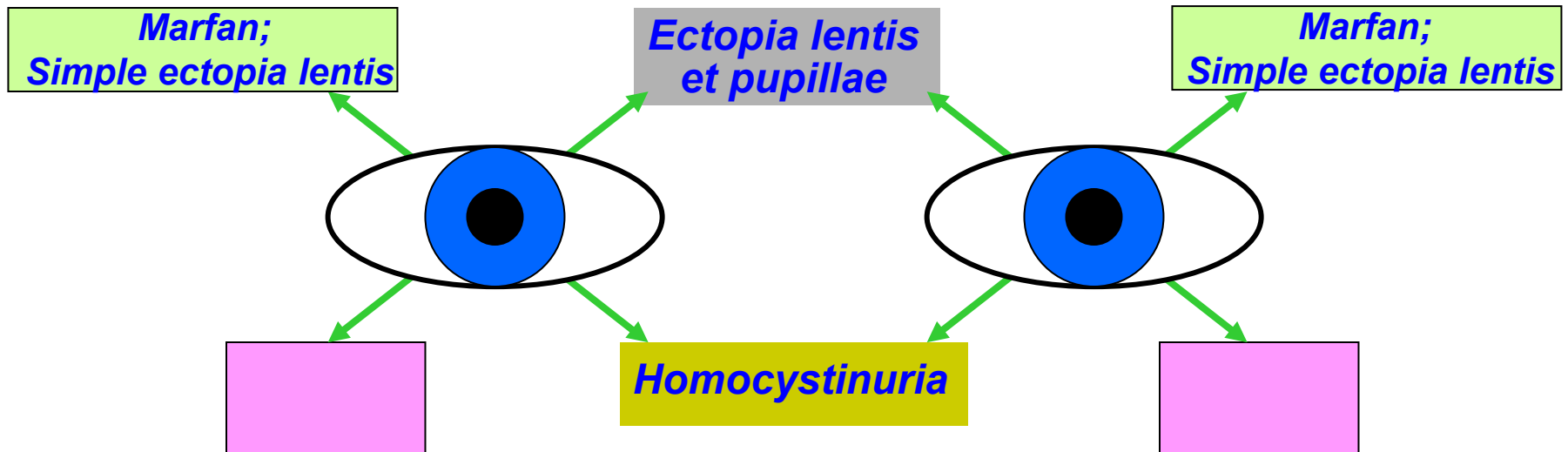
Lens subluxation: Which conditions are associated with displacement in which directions?



**A**

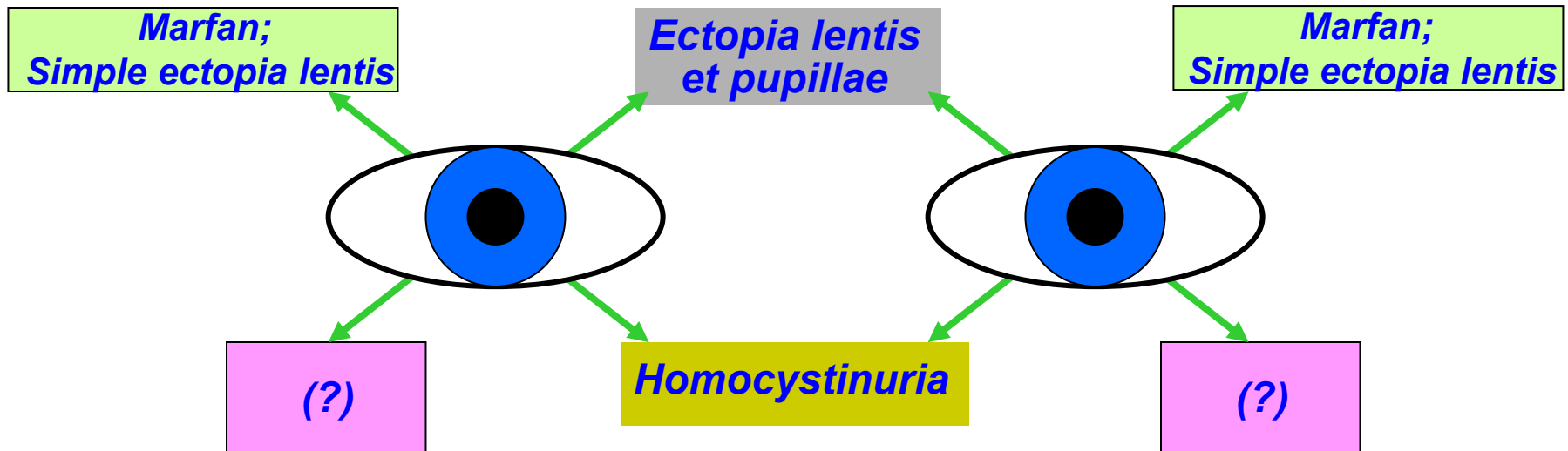
## Ectopia Lentis

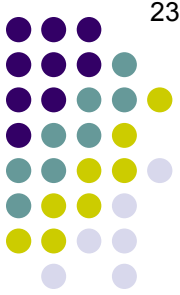
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Lens subluxation: Which conditions are associated with displacement in which directions?



**A**

## Ectopia Lentis

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

