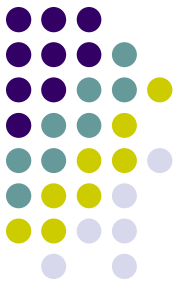


Q

Lenticonus

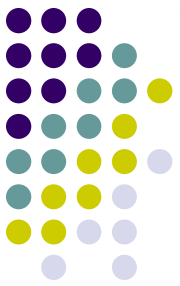
--Secondary to pathological process of central portion of location of path process



A

Lenticonus

2



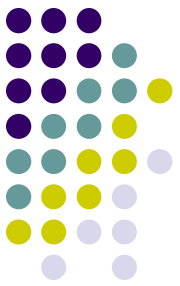
--Secondary to **ectasia** of central portion of **lens surface**

Q

Lenticonus

--Secondary to **ectasia** of central portion of **lens surface**

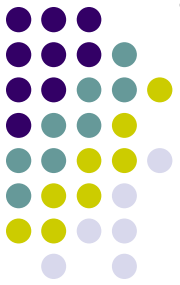
In one (unsurprising) word, what is the shape of the affected lens surface in lenticonus?



A

Lenticonus

4

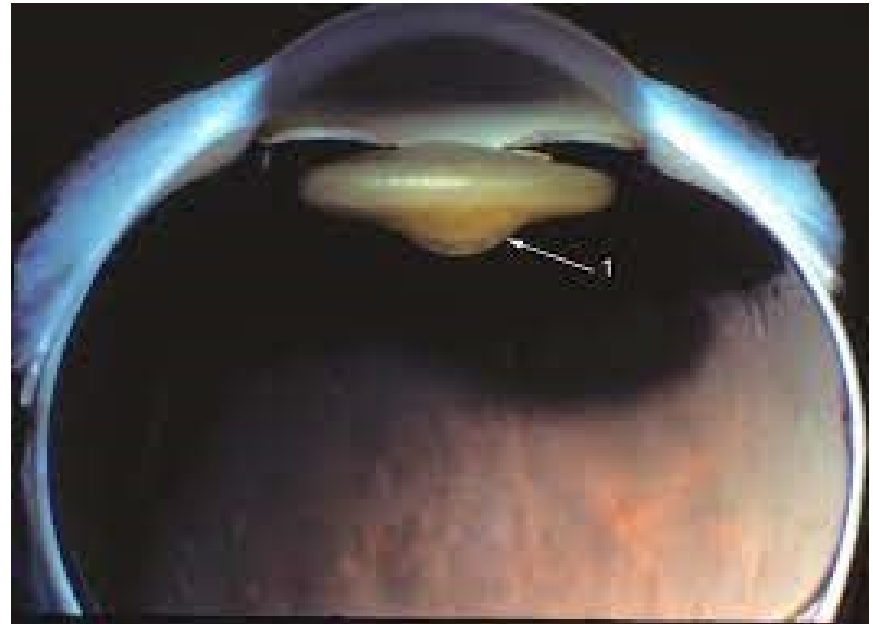
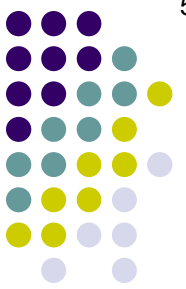


--Secondary to **ectasia** of central portion of **lens surface**

In one (unsurprising) word, what is the shape of the affected lens surface in lenticonus?

'Conical'

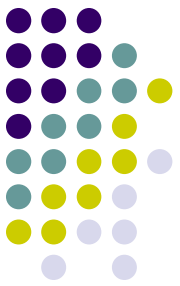
Lenticonus



The conical shape of a lenticonus lens

Q

Lenticonus



--Secondary to **ectasia** of central portion of **lens surface**

In one (unsurprising) word, what is the shape of the affected lens surface in lenticonus?

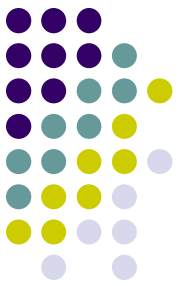
'Conical'

What is the classic two-word description of the clinical appearance of a lens with lenticonus?

A

Lenticonus

7



--Secondary to **ectasia** of central portion of **lens surface**

In one (unsurprising) word, what is the shape of the affected lens surface in lenticonus?

‘Conical’

What is the classic two-word description of the clinical appearance of a lens with lenticonus?

‘Oil droplet’

Q

Lenticonus



--Secondary to **ectasia** of central portion of **lens surface**

In one (unsurprising) word, what is the shape of the affected lens surface in lenticonus?

‘Conical’

What is the classic two-word description of the clinical appearance of a lens with lenticonus?

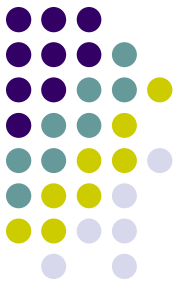
‘Oil droplet’

What simple exam maneuver is the surest way to detect the presence of an oil droplet cataract?

A

Lenticonus

9



--Secondary to **ectasia** of central portion of **lens surface**

In one (unsurprising) word, what is the shape of the affected lens surface in lenticonus?

‘Conical’

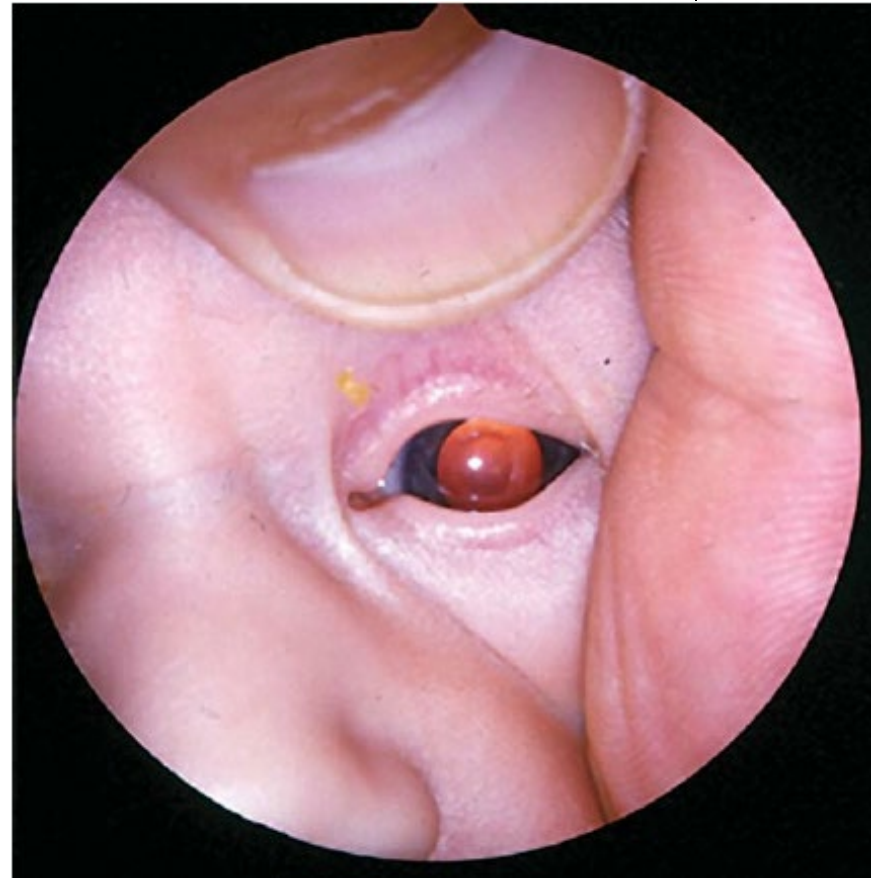
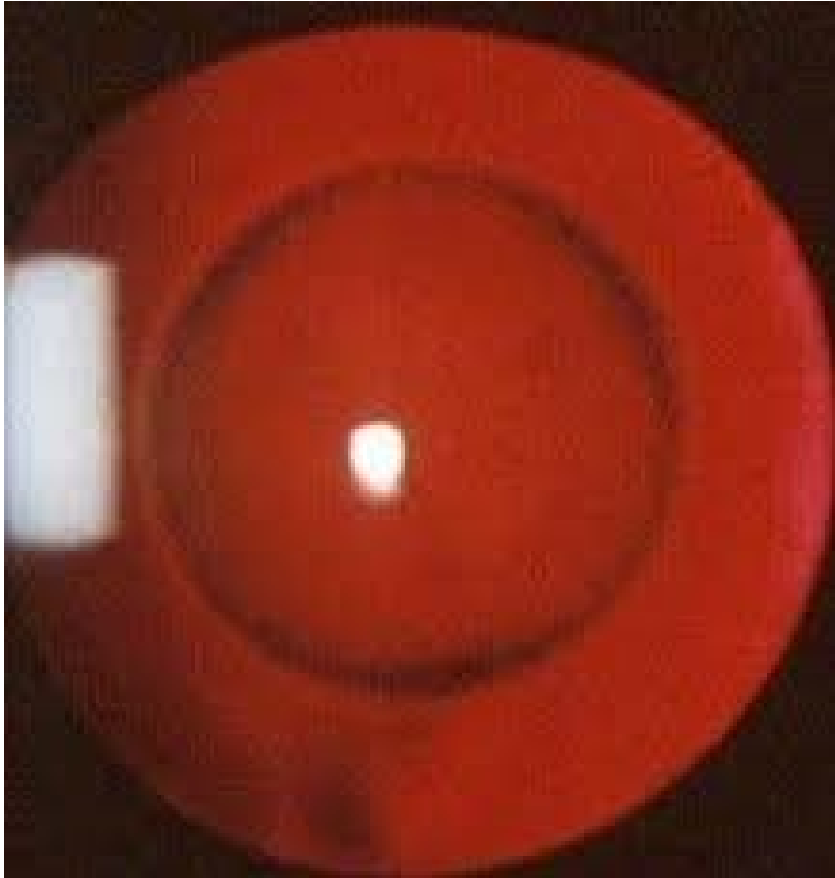
What is the classic two-word description of the clinical appearance of a lens with lenticonus?

‘Oil droplet’

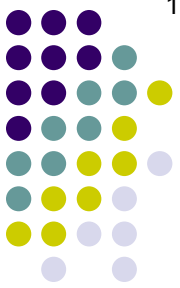
What simple exam maneuver is the surest way to detect the presence of an oil droplet cataract?

Retroillumination

Lenticonus



Oil-droplet lens change: Retroillumination



Q

Lenticonus

--Secondary to **ectasia** of central portion of **lens surface**

*In one (unusual) condition
in lenticonus*

'Conical'

*What is the condition
lens with lenticonus*

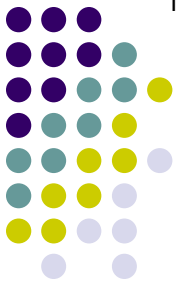
'Oil droplet'

*What simple test
an oil droplet
Retroillumination*

When you hear the term oil droplet applied to the appearance of the lens, three conditions should come to mind. Two are anterior and posterior lenticonus—what is the third?

A

Lenticonus



--Secondary to **ectasia** of central portion of **lens surface**

*In one (unusual) case of
in lenticonus
'Conical'*

When you hear the term oil droplet applied to the appearance of the lens, three conditions should come to mind. Two are anterior and posterior lenticonus—what is the third?

Galactosemia

*What is the cause of
lens with lenticonus?*

'Oil droplet'

*What simple test can identify
an oil droplet?*
Retroillumination



Q

Lenticonus

--Secondary to **ectasia** of central portion of **lens surface**

In one (unusual) condition, the lens can become lenticular in lenticonus.

'Conical'

What is the clinical appearance of the lens with lenticonus?

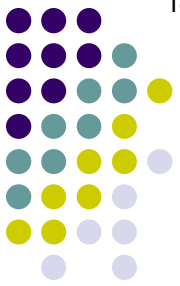
'Oil droplet'

What simple test can be used to identify an oil droplet?
Retroillumination

When you hear the term oil droplet applied to the appearance of the lens, three conditions should come to mind. Two are anterior and posterior lenticonus—what is the third?

Galactosemia

What is galactosemia?



--Secondary to **ectasia** of central portion of **lens surface**

When you hear the term oil droplet applied to the appearance of the lens, three conditions should come to mind. Two are anterior and posterior lenticonus—what is the third?

Galactosemia

What is galactosemia?

It is an **general term (four words)**

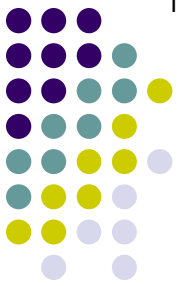
In one (unusual) condition, the lens can appear in lenticonus.

'Conical'

What is the cause of the oil droplet appearance of the lens with lenticonus?

'Oil droplet'

What simple test can be used to differentiate an oil droplet from a lenticonus?
Retroillumination



A

Lenticonus

--Secondary to **ectasia** of central portion of **lens surface**

In one (unusual) condition, the lens can become conical in lenticonus.

'Conical'

What is the cause of the conical lens with lenticonus?

'Oil droplet'

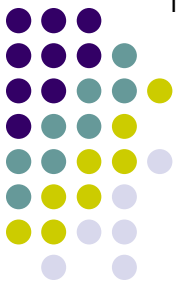
What simple test can identify an oil droplet?
Retroillumination

When you hear the term oil droplet applied to the appearance of the lens, three conditions should come to mind. Two are anterior and posterior lenticonus—what is the third?

Galactosemia

What is galactosemia?

It is an inborn error of metabolism



Q

Lenticonus

--Secondary to **ectasia** of central portion of **lens surface**

When you hear the term oil droplet applied to the appearance of the lens, three conditions should come to mind. Two are anterior and posterior lenticonus—what is the third?

Galactosemia

What is galactosemia?

It is an inborn error of metabolism in which **substance** cannot be converted to **diff substance**

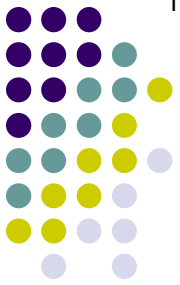
'Oil droplet'

*What simple
an oil droplet
Retroillumina*

A

Lenticonus

17



--Secondary to **ectasia** of central portion of **lens surface**

In one (unusual) condition, there is an oil droplet in lenticonus.

'Conical'

What is the cause of the oil droplet in the lens with lenticonus?

'Oil droplet'

What simple test can be used to identify an oil droplet in the lens?
Retroillumination

When you hear the term oil droplet applied to the appearance of the lens, three conditions should come to mind. Two are anterior and posterior lenticonus—what is the third?

Galactosemia

What is galactosemia?

It is an inborn error of metabolism in which galactose cannot be converted to glucose



Q

Lenticonus

--Secondary to **ectasia** of central portion of **lens surface**

In one (unusual) condition, there is an oil droplet in lenticonus.

'Conical'

What is the cause of an oil droplet in the lens with lenticonus?

'Oil droplet'

What simple test can detect an oil droplet in the lens?
Retroillumination

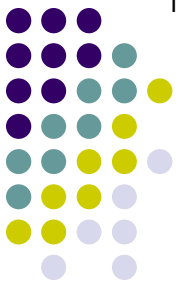
When you hear the term oil droplet applied to the appearance of the lens, three conditions should come to mind. Two are anterior and posterior lenticonus—what is the third?

Galactosemia

What is galactosemia?

It is an inborn error of metabolism in which galactose cannot be converted to glucose

How does this lead to an oil droplet lenticular appearance?



--Secondary to **ectasia** of central portion of **lens surface**

In one (unusual) case, in lenticonus

'Conical'

What is the cause of lens with lenticonus?

'Oil droplet'

What simple test can detect an oil droplet? Retroillumination

When you hear the term oil droplet applied to the appearance of the lens, three conditions should come to mind. Two are anterior and posterior lenticonus—what is the third?

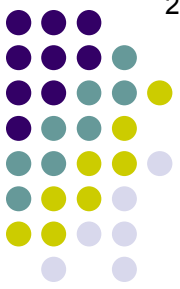
Galactosemia

What is galactosemia?

It is an inborn error of metabolism in which galactose cannot be converted to glucose

How does this lead to an oil droplet lenticular appearance?

An inert byproduct of galactose metabolism (**substance**) accumulates in lens fibers, creating an osmotic gradient that draws in aqueous. Swelling of the fibers damages them, leading to the changes that produce an oil droplet.



--Secondary to **ectasia** of central portion of **lens surface**

When you hear the term oil droplet applied to the appearance of the lens, three conditions should come to mind. Two are anterior and posterior lenticonus—what is the third?

Galactosemia

What is galactosemia?

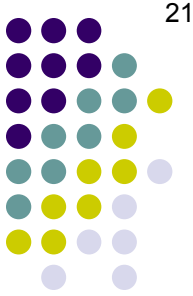
It is an inborn error of metabolism in which galactose cannot be converted to glucose

'Oil droplet'

How does this lead to an oil droplet lenticular appearance?

An inert byproduct of galactose metabolism (galactitol) accumulates in lens fibers, creating an osmotic gradient that draws in aqueous. Swelling of the fibers damages them, leading to the changes that produce an oil droplet.

Lenticonus



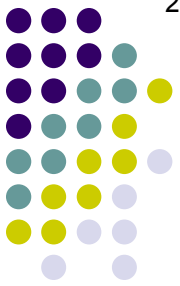
21



Galactosemia: Oil-droplet cataracts

A

Lenticonus



--Secondary to **ectasia** of central portion of **lens surface**

In one (unusual) condition, in lenticonus, the lens is 'Conical'

When you hear the term oil droplet applied to the appearance of the lens, three conditions should come to mind. Two are anterior and posterior lenticonus—what is the third?

Galactosemia

What is galactosemia?

What is the cause of lens with lenticonus?

It is an inborn error of metabolism in which galactose cannot be converted to glucose

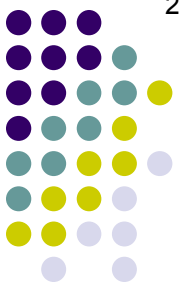
'Oil droplet'

How does this lead to an oil droplet lenticular appearance?

What simple explanation is there for an oil droplet appearance? Retroillumination

An inert byproduct of galactose metabolism (galactitol) accumulates in lens fibers, creating an osmotic gradient that draws in aqueous. Swelling of the fibers damages them, leading to the changes that produce an oil droplet.

So, there's a fundamental difference between the cause of the oil-droplet appearance in lenticonus (= altered central lenticular power) vs that of galactosemia (= the accumulation of galactitol and fluid in the central lens).



Q

Lenticonus

--Secondary to **ectasia** of central portion of **lens surface**

*In one (unusual) case of
in lenticonus 'Conical'*

When you hear the term oil droplet applied to the appearance of the lens, three conditions should come to mind. Two are anterior and posterior lenticonus—what is the third?

Galactosemia

What is galactosemia?

*What is the cause of
lens with lenticonus?*

It is an inborn error of metabolism in which galactose cannot be converted to glucose

'Oil droplet'

How does this lead to an oil droplet lenticular appearance?

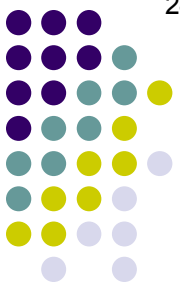
*What simple test can
an oil droplet
Retroillumination*

An inert byproduct of galactose metabolism (galactitol) accumulates in lens fibers, creating an osmotic gradient that draws in aqueous. Swelling of the fibers damages them, leading to the changes that produce an oil droplet.

So, there's a fundamental difference between the cause of the oil-droplet appearance in lenticonus (= altered central lenticular power) vs that of galactosemia (= the accumulation of galactitol and fluid in the central lens).

*What are the **systemic** manifestations of galactosemia?*

--?
--?
--?



--Secondary to **ectasia** of central portion of **lens surface**

*In one (unusual) case of
in lenticonus
'Conical'*

When you hear the term oil droplet applied to the appearance of the lens, three conditions should come to mind. Two are anterior and posterior lenticonus—what is the third?

Galactosemia

What is galactosemia?

*What is the cause of
lens with lenticonus?*

It is an inborn error of metabolism in which galactose cannot be converted to glucose

'Oil droplet'

How does this lead to an oil droplet lenticular appearance?

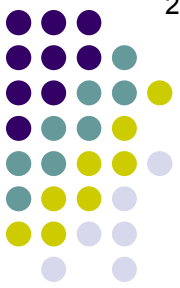
*What simple test can
an oil droplet
Retroillumination*

An inert byproduct of galactose metabolism (galactitol) accumulates in lens fibers, creating an osmotic gradient that draws in aqueous. Swelling of the fibers damages them, leading to the changes that produce an oil droplet.

So, there's a fundamental difference between the cause of the oil-droplet appearance in lenticonus (= altered central lenticular power) vs that of galactosemia (= the accumulation of galactitol and fluid in the central lens).

*What are the **systemic** manifestations of galactosemia?*

- Failure to thrive
- Hepatomegaly with jaundice
- Impaired cognitive development



Q

Lenticonus

--Secondary to **ectasia** of central portion of **lens surface**

In one (unusual) case of lenticonus 'Conical'

When you hear the term oil droplet applied to the appearance of the lens, three conditions should come to mind. Two are anterior and posterior lenticonus—what is the third?

Galactosemia

What is galactosemia?

What is the cause of lenticonus? It is an inborn error of metabolism in which galactose cannot be converted to glucose.

What is the long-term prognosis of galactosemia?

'Oil droplet'

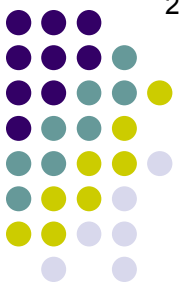
How does galactosemia lead to the oil droplet appearance?

What simple test can confirm the diagnosis of galactosemia? An inert byproduct of galactose metabolism (galactitol) accumulates in lens fibers, creating an osmotic gradient that draws in aqueous. Swelling of the fibers damages them, leading to the changes that produce an oil droplet.

So, there's a fundamental difference between the cause of the oil-droplet appearance in lenticonus (= altered central lenticular power) vs that of galactosemia (= the accumulation of galactitol and fluid in the central lens).

*What are the **systemic** manifestations of galactosemia?*

- Failure to thrive
- Hepatomegaly with jaundice
- Impaired cognitive development



--Secondary to **ectasia** of central portion of **lens surface**

In one (unusual) form of lenticular degeneration, the lens becomes conical.
In lenticular degeneration, the lens becomes conical.
 'Conical'

What is the cause of lenticular degeneration?
lens with lenticular degeneration.

'Oil droplet'

What simple test can be used to identify an oil droplet?
an oil droplet.
 Retroillumination

When you hear the term oil droplet applied to the appearance of the lens, three conditions should come to mind. Two are anterior and posterior lenticonus—what is the third?

Galactosemia

What is galactosemia?

It is an inborn error of metabolism in which galactose cannot be converted to glucose.

What is the long-term prognosis of galactosemia?

The severe form (which is also the most common) is uniformly fatal if not treated.

How does galactosemia affect the lens?

An inert byproduct of galactose metabolism (galactitol) accumulates in lens fibers, creating an osmotic gradient that draws in aqueous. Swelling of the fibers damages them, leading to the changes that produce an oil droplet.

So, there's a fundamental difference between the cause of the oil-droplet appearance in lenticonus (= altered central lenticular power) vs that of galactosemia (= the accumulation of galactitol and fluid in the central lens).

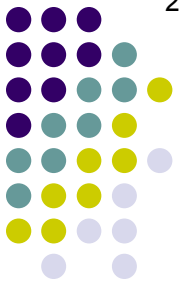
*What are the **systemic** manifestations of galactosemia?*

- Failure to thrive
- Hepatomegaly with jaundice
- Impaired cognitive development

Q

Lenticonus

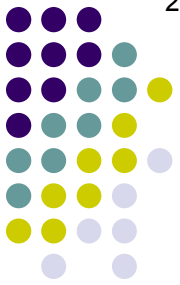
- Secondary to **ectasia** of central portion of **lens surface**
- Typical clinical course:
 - Early: Manifests as **refractive issue**

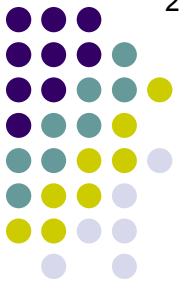


A

Lenticonus

- Secondary to **ectasia** of central portion of **lens surface**
- Typical clinical course:
 - Early: Manifests as **myopia**





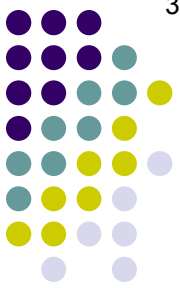
Q

Lenticonus

- Secondary to **ectasia** of central portion of **lens surface**
- Typical clinical course:
 - Early: Manifests as **myopia**
 - Next: Manifests as **different refractive issue**

A

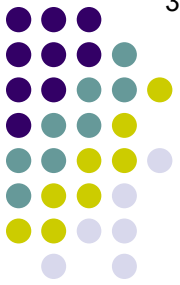
Lenticonus



- Secondary to **ectasia** of central portion of **lens surface**
- Typical clinical course:
 - Early: Manifests as **myopia**
 - Next: Manifests as **irregular astigmatism**

Q

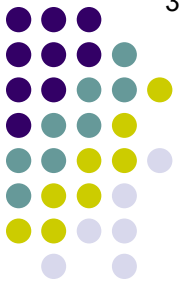
Lenticonus



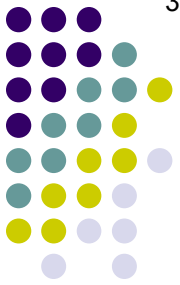
- Secondary to **ectasia** of central portion of **lens surface**
- Typical clinical course:
 - Early: Manifests as **myopia**
 - Next: Manifests as **irregular astigmatism**
 - Later: The **lens layer** opacifies

A

Lenticonus



- Secondary to **ectasia** of central portion of **lens surface**
- Typical clinical course:
 - Early: Manifests as **myopia**
 - Next: Manifests as **irregular astigmatism**
 - Later: The **cortex** opacifies



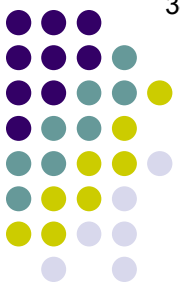
Q

Lenticonus

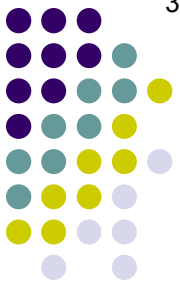
- Secondary to **ectasia** of central portion of **lens surface**
- Typical clinical course:
 - Early: Manifests as **myopia**
 - Next: Manifests as **irregular astigmatism**
 - Later: The **cortex** opacifies
 - At age **# - #** years: Capsule → **two words**

A

Lenticonus



- Secondary to **ectasia** of central portion of **lens surface**
- Typical clinical course:
 - Early: Manifests as **myopia**
 - Next: Manifests as **irregular astigmatism**
 - Later: The **cortex** opacifies
 - At age **4-5** years: Capsule **ruptures** → **total opacification**

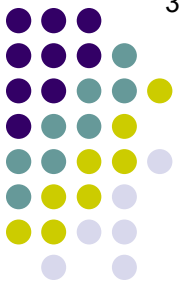


Q

Lenticonus

- Secondary to **ectasia** of central portion of **lens surface**
- Typical clinical course:
 - Early: Manifests as **myopia**
 - Next: Manifests as **irregular astigmatism**
 - Later: The **cortex** opacifies
 - At age **4-5** years: Capsule **ruptures** → **total opacification**

Lenticonus comes in two (very) basic forms—what are they?



A

Lenticonus

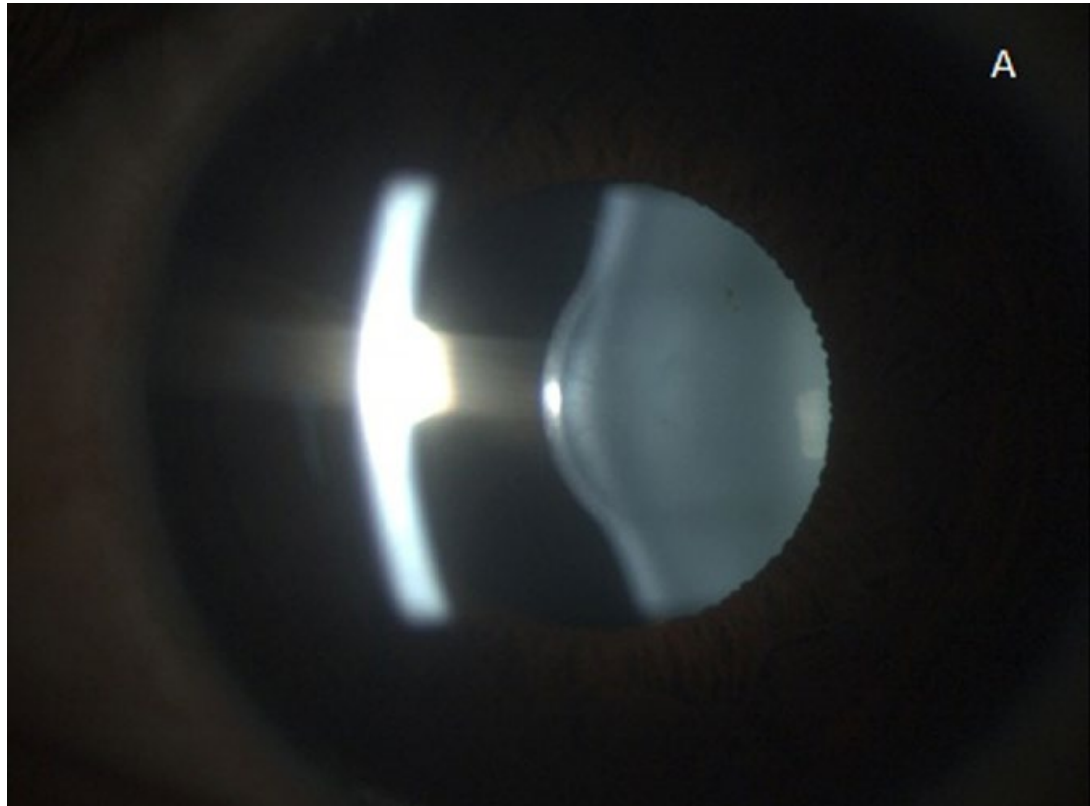
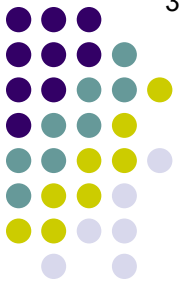
- Secondary to **ectasia** of central portion of **lens surface**
- Typical clinical course:
 - Early: Manifests as **myopia**
 - Next: Manifests as **irregular astigmatism**
 - Later: The **cortex** opacifies
 - At age **4-5** years: Capsule **ruptures** → **total opacification**

Lenticonus comes in two (very) basic forms—what are they?

Anterior lenticonus and **posterior** lenticonus, referring to involvement of the anterior and posterior capsules respectively

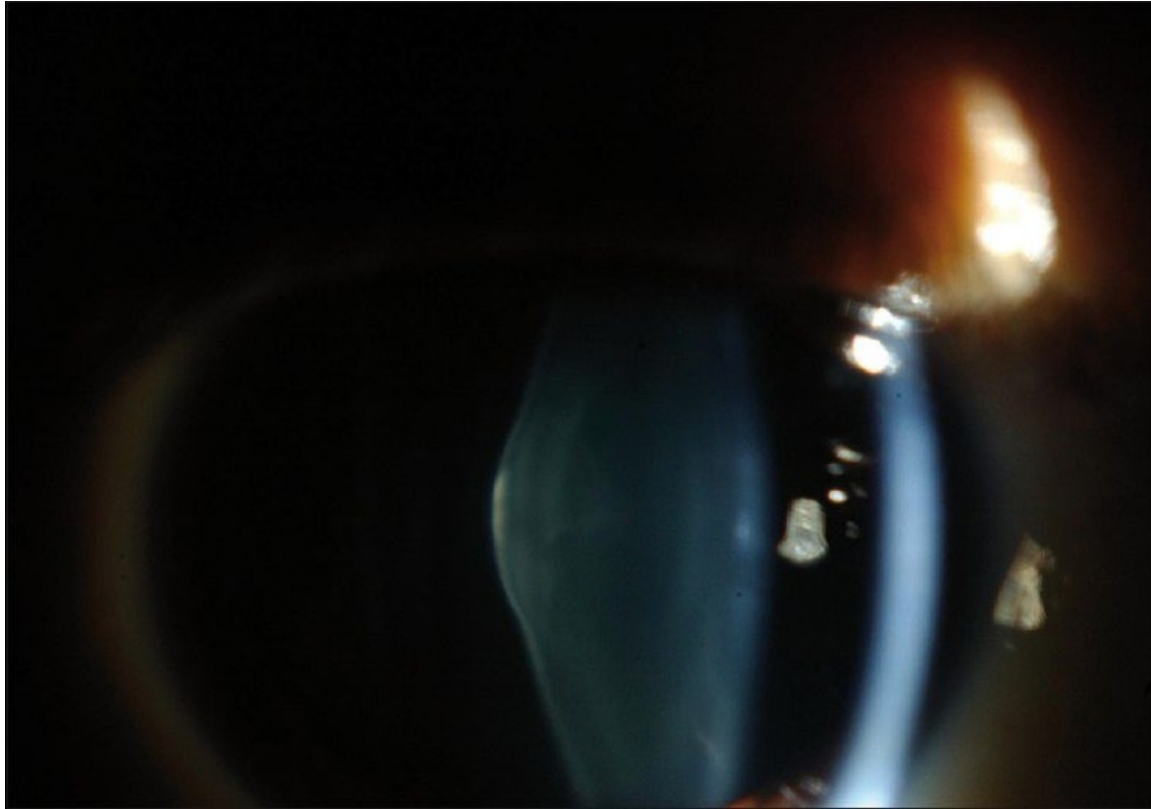
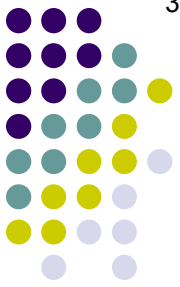
Lenticonus

37



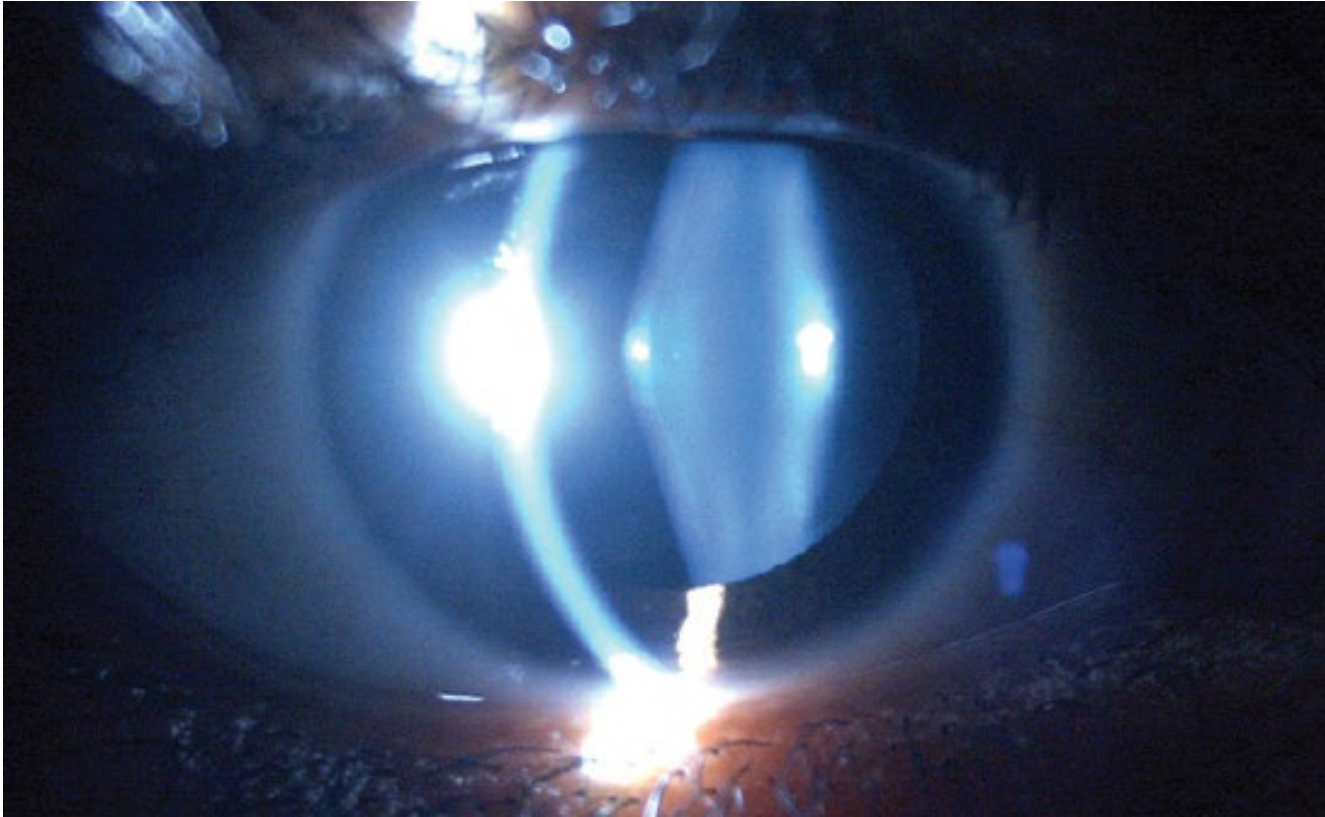
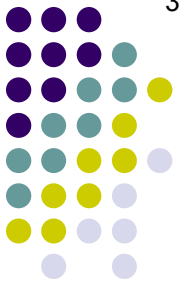
Anterior lenticonus

Lenticonus

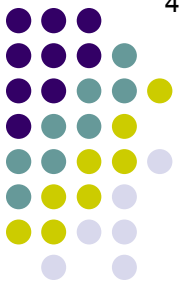


Posterior lenticonus

Lenticonus



Just for fun: An eye with both anterior *and* posterior lenticonus

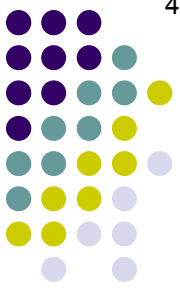


Q

Lenticonus

- Secondary to **ectasia** of central portion of **lens surface**
- Typical clinical course:
 - Early: Manifests as **myopia**
 - Next: Manifests as **irregular astigmatism**
 - Later: The **cortex** opacifies
 - At age **4-5** years: Capsule **ruptures** → **total opacification**

	Usually unilateral vs usually bilateral		
<i>Anterior lenticonus</i>	?		
<i>Posterior lenticonus</i>	?		

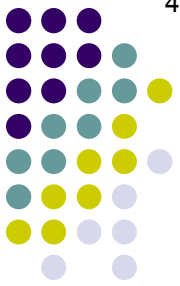


A

Lenticonus

- Secondary to **ectasia** of central portion of **lens surface**
- Typical clinical course:
 - Early: Manifests as **myopia**
 - Next: Manifests as **irregular astigmatism**
 - Later: The **cortex** opacifies
 - At age **4-5** years: Capsule **ruptures** → **total opacification**

	Usually unilateral vs usually bilateral		
<i>Anterior lenticonus</i>	Usually bilateral		
<i>Posterior lenticonus</i>	90% unilateral		

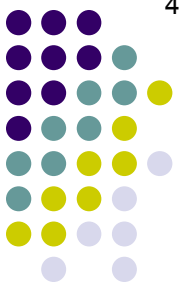


Q

Lenticonus

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	Usually unilateral vs usually bilateral	More common vs less common	
<i>Anterior lenticonus</i>	Usually bilateral	?	
<i>Posterior lenticonus</i>	90% unilateral	?	

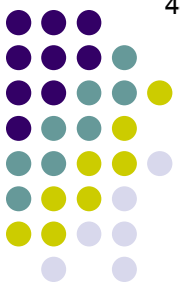


A

Lenticonus

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 - At age **4-5** years: Capsule **ruptures** → **total opacification**

	Usually unilateral vs usually bilateral	More common vs less common	
<i>Anterior lenticonus</i>	Usually bilateral	Less common	
<i>Posterior lenticonus</i>	90% unilateral	More common (but still rare)	

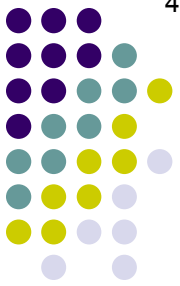


Q

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 - At age **4-5** years: Capsule **ruptures** → **total opacification**

	Usually unilateral vs usually bilateral	More common vs less common	Usually sporadic vs usually syndromic
<i>Anterior lenticonus</i>	Usually bilateral	Less common	?
<i>Posterior lenticonus</i>	90% unilateral	More common (but still rare)	?

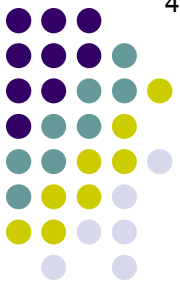


A

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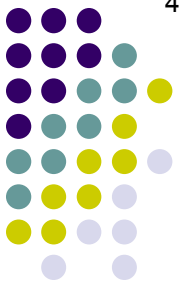
	Usually unilateral vs usually bilateral	More common vs less common	Usually sporadic vs usually syndromic
<i>Anterior lenticonus</i>	Usually bilateral	Less common	Usually syndromic
<i>Posterior lenticonus</i>	90% unilateral	More common (but still rare)	Usually sporadic (unless bilateral)



Lenticonus

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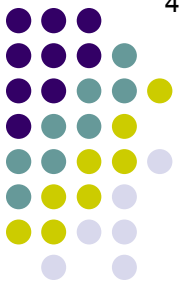
	Usually unilateral vs	More common	Usually sporadic vs
	Note that these factoids 'go together,' ie, syndromic conditions usually affect both eyes...		
Anterior lenticonus	Usually bilateral	Less common	Usually syndromic
Posterior lenticonus	90% unilateral	More common (but still rare)	Usually sporadic (unless bilateral)



Lenticonus

- Secondary to **ectasia** of central portion of **lens surface**
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 - Early: Manifests as **myopia**
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 - At age **4-5** years: Capsule **ruptures** → **total opacification**

	Usually unilateral vs usually bilateral	More common vs less common	Usually sporadic vs usually syndromic
Anterior lenticonus	usually bilateral	usually bilateral	usually sporadic
...and likewise, these two go together as well (sporadic conditions usually affect only one)			
Posterior lenticonus	99% unilateral	More common (but still rare)	Usually sporadic (unless bilateral)



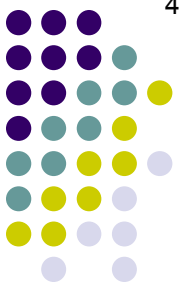
Q

Lenticonus

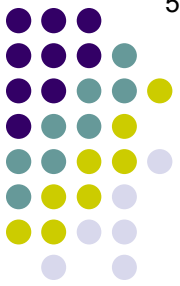
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 - At age **4-5** years: Capsule **ruptures** → **total opacification**
- unilateral vs bilateral** cases are associated with two of the **two words** *syndromes*

A

Lenticonus



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 - At age **4-5** years: Capsule **ruptures** → **total opacification**
- Bilateral** cases are associated with two of the ***familial oculorenal*** syndromes



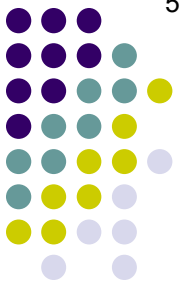
Q

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- Bilateral** cases are associated with two of the ***familial oculorenal*** syndromes
 (**eponym** syndrome; **different eponym** syndrome)

A

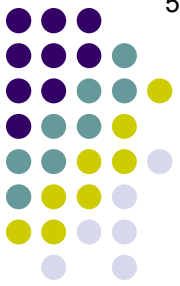
Lenticonus



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- Bilateral** cases are associated with two of the ***familial oculorenal*** syndromes (**Alport** syndrome; **Lowe** syndrome)

Q

Lenticonus

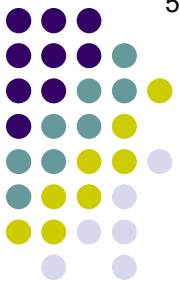


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- Bilateral** cases are associated with two of the **familial oculorenal** syndromes
(**Alport** syndrome; **Lowe** syndrome)

	Lenticonus location				
Alport Syndrome	?				
Lowe Syndrome	?				

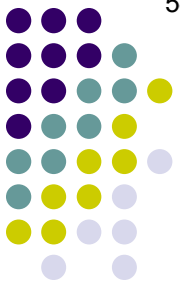
A

Lenticonus



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- Bilateral** cases are associated with two of the **familial oculorenal** syndromes
(**Alport** syndrome; **Lowe** syndrome)

	Lenticonus location				
Alport Syndrome	Anterior				
Lowe Syndrome	Posterior				

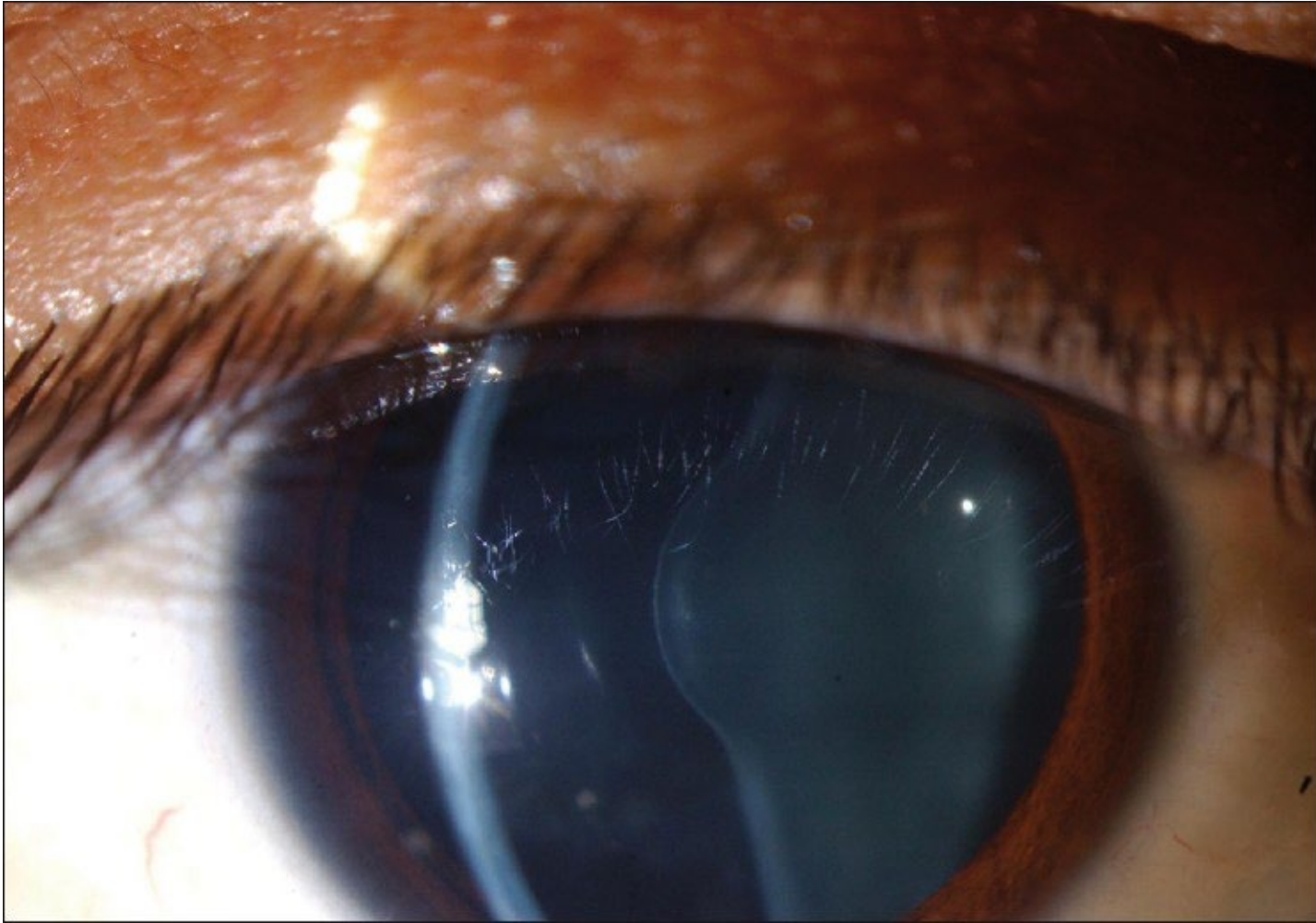


Lenticonus

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- Bilateral** cases are associated with two of the **familial oculorenal** syndromes
(**Alport** syndrome; **Lowe** syndrome)

A lport syndrome Anterior lenticonus					
Alport Syndrome	Anterior				
Lowe Syndrome	Posterior				

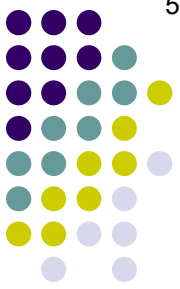
Lenticonus



Anterior lenticonus in Alport syndrome

Q

Lenticonus



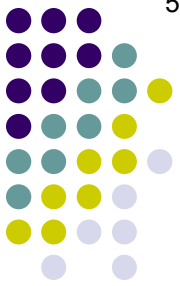
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How prevalent is lenticonus in Alport and Lowe syndromes?

	Lenticonus location				
Alport Syndrome	Anterior				
Lowe Syndrome	Posterior				

A

Lenticonus

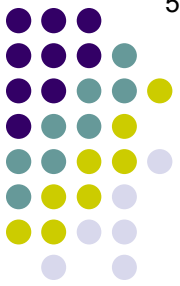


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How prevalent is lenticonus in Alport and Lowe syndromes?

Anterior lenticonus is not a major component of Alport syndrome, being present in only 25% of cases

	Lenticonus location				
Alport Syndrome	Anterior				
Lowe Syndrome	Posterior				



A

Lenticonus

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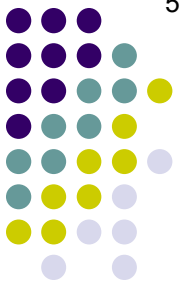
How prevalent is lenticonus in Alport and Lowe syndromes?

Anterior lenticonus is not a major component of Alport syndrome, being present in only 25% of cases. In contrast, posterior lenticonus is a **defining feature** of Lowe syndrome.

	Lenticonus location				
Alport Syndrome	Anterior				
Lowe Syndrome	Posterior				

Q

Lenticonus

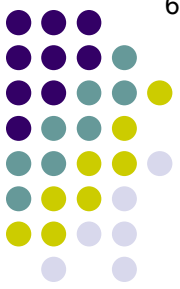


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- Bilateral** cases are associated with two of the **familial oculorenal** syndromes
(**Alport** syndrome; **Lowe** syndrome)

	Lenticonus location	Inheritance			
Alport Syndrome	Anterior	?			
Lowe Syndrome	Posterior	?			

A

Lenticonus

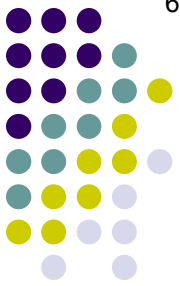


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(**Alport** syndrome; **Lowe** syndrome)

	Lenticonus location	Inheritance			
Alport Syndrome	Anterior	Most are X-linked			
Lowe Syndrome	Posterior	X-linked			

Q

Lenticonus

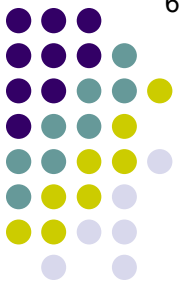


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	Lenticonus location	Inheritance	Classic presentation (nonocular)		
Alport Syndrome	Anterior	Most are X-linked	?		
Lowe Syndrome	Posterior	X-linked	?		

Q/A

Lenticonus

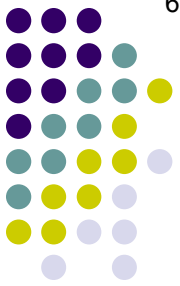


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 (**Alport** syndrome; **Lowe** syndrome)

	Lenticonus location	Inheritance	Classic presentation (nonocular)		
Alport Syndrome	Anterior	Most are X-linked	Hematuria in childhood vs infancy		
Lowe Syndrome	Posterior	X-linked	Hematuria in childhood vs infancy		

A

Lenticonus

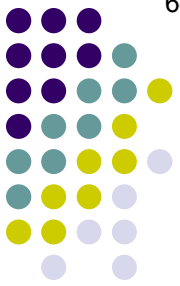


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	Lenticonus location	Inheritance	Classic presentation (nonocular)		
Alport Syndrome	Anterior	Most are X-linked	Hematuria in childhood		
Lowe Syndrome	Posterior	X-linked	Hematuria in infancy		

Q

Lenticonus

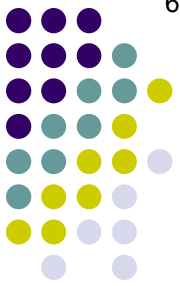


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	Lenticonus location	Inheritance	Classic presentation (nonocular)	Associated findings	
Alport Syndrome	Anterior	Most are X-linked	Hematuria in childhood	?	
Lowe Syndrome	Posterior	X-linked	Hematuria in infancy	?	

A

Lenticonus

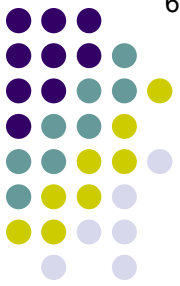


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- Bilateral** cases are associated with two of the **familial oculorenal** syndromes
 (**Alport** syndrome; **Lowe** syndrome)

	Lenticonus location	Inheritance	Classic presentation (nonocular)	Associated findings	
Alport Syndrome	Anterior	Most are X-linked	Hematuria in childhood	High-f nerve deafness; nephritis	
Lowe Syndrome	Posterior	X-linked	Hematuria in infancy	MR; rickets; hypotonia	

Q

Lenticonus

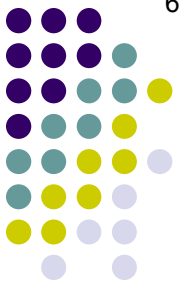


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(**Alport** syndrome; **Lowe** syndrome)

	Lenticonus location	Inheritance	Classic presentation (nonocular)	Associated findings	Another lens finding
Alport Syndrome	Anterior	Most are X-linked	Hematuria in childhood	High-f nerve deafness; nephritis	?
Lowe Syndrome	Posterior	X-linked	Hematuria in infancy	MR; rickets; hypotonia	?

A

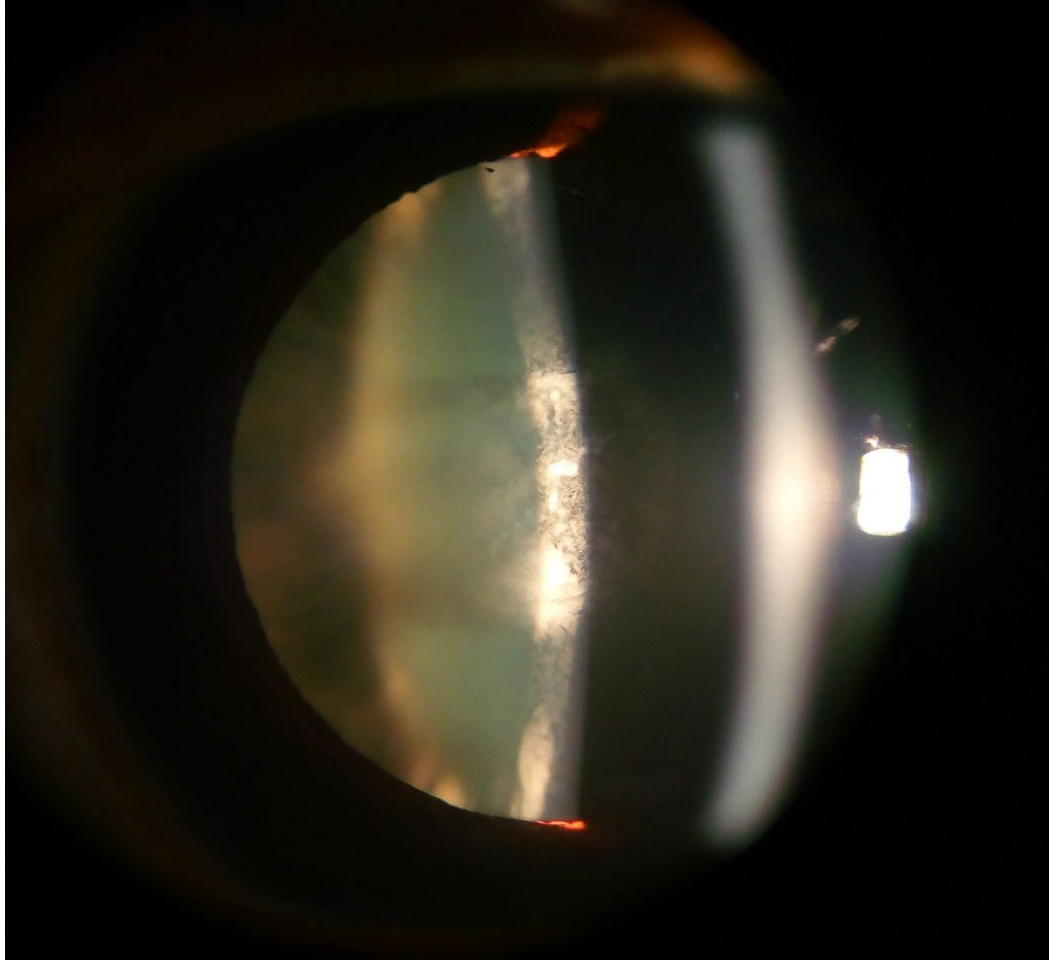
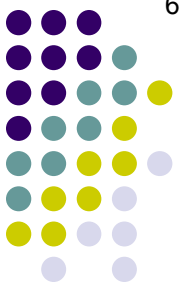
Lenticonus



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- Bilateral** cases are associated with two of the **familial oculorenal syndromes** (**Alport** syndrome; **Lowe** syndrome)

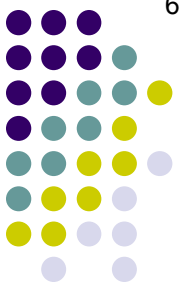
	Lenticonus location	Inheritance	Classic presentation (nonocular)	Associated findings	Another lens finding
Alport Syndrome	Anterior	Most are X-linked	Hematuria in childhood	High-f nerve deafness; nephritis	Anterior subcapsular cataract
Lowe Syndrome	Posterior	X-linked	Hematuria in infancy	MR; rickets; hypotonia	Microspherophakia

Lenticonus



Anterior subcapsular cataract

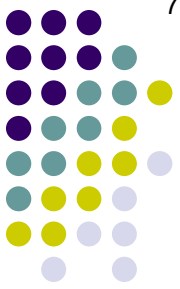
Lenticonus



Microspherophakia

Q

Lenticonus



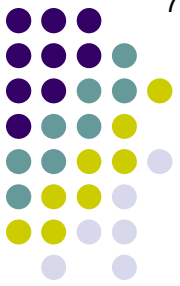
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- Bilateral** cases are associated with two of the **familial oculorenal syndromes** (**Alport** syndrome; **Lowe** syndrome, and the...)

	Lenticonus location	Inheritance	Classic presentation (nonocular)	Associated findings	Another lens finding
Alport Syndrome	Anterior	Most are X-linked	Hematuria in childhood	High-f nerve deafness; nephritis	Microspherophakia
Lowe Syndrome	Posterior	X-linked	Hematuria in infancy	MR; rickets; hypotonia	Microspherophakia

For completeness' sake: *There are four more familial oculorenal syndromes mentioned in the BCSC books. All are the same type of disorder. What type are they?*

A

Lenticonus



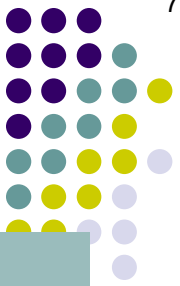
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- Bilateral** cases are associated with two of the **familial oculorenal syndromes** (**Alport** syndrome; **Lowe** syndrome, and the...**ciliopathies**)

	Lenticonus location	Inheritance	Classic presentation (nonocular)	Associated findings	Another lens finding
Alport Syndrome	Anterior	Most are X-linked	Hematuria in childhood	High-f nerve deafness; nephritis	Microspherophakia
Lowe Syndrome	Posterior	X-linked	Hematuria in infancy	MR; rickets; hypotonia	Microspherophakia

For completeness' sake: *There are four more familial oculorenal syndromes mentioned in the BCSC books. All are the same type of disorder. What type are they?* **Ciliopathy**

Q

Lenticonus



What is a ciliopathy?

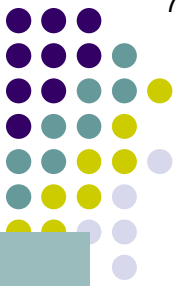
- Later: The cortex opacifies
- At age 4-5 years: Capsule ruptures → total opacification
- Bilateral cases are associated with two of the familial oculorenal syndromes (Alport syndrome; Lowe syndrome, and the...ciliopathies)

	Lenticonus location	Inheritance	Classic presentation (nonocular)	Associated findings	Another lens finding
Alport Syndrome	Anterior	Most are X-linked	Hematuria in childhood	High-f nerve deafness; nephritis	Microspherophakia
Lowe Syndrome	Posterior	X-linked	Hematuria in infancy	MR; rickets; hypotonia	Microspherophakia

For completeness' sake: There are **four more** familial oculorenal syndromes mentioned in the BCSC books. All are the same type of disorder. What type are they? **Ciliopathy**

A

Lenticonus



What is a ciliopathy?

An inherited condition marked by abnormal structure and/or function of cilia

--Later: The cortex opacifies

--At age 4-5 years: Capsule ruptures → total opacification

--Bilateral cases are associated with two of the familial oculorenal syndromes (Alport syndrome; Lowe syndrome, and the...ciliopathies)

	Lenticonus location	Inheritance	Classic presentation (nonocular)	Associated findings	Another lens finding
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For completeness' sake: There are **four more** familial oculorenal syndromes mentioned in the BCSC books. All are the same type of disorder. What type are they? **Ciliopathy**

Q

Lenticonus

What is a ciliopathy?

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Cilia are ubiquitous organelles. That said, ciliopathies primarily affect three organs--what are they?

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The eyes, brain and kidneys

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Cilia are ubiquitous

The eyes

The eyes??!! Which part of the eye contains cilia wiggling about?

are they?

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The eyes

The eyes??!! Which part of the eye contains cilia wiggling about?

None. Remember, cilia come in two basic flavors: Motile, and nonmotile. It is the **nonmotile** type which is ubiquitous in the eye.

Later: The cornea opacifies

--At age 4-5 years: Capsule ruptures → total opacification

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The eyes, brain

What are the four ciliopathies emphasized in the BCSC books?

- Note: You have heard of at least several of these, but may not have thought of them as a group, ie, as all being members of the *oculorenal syndrome* family. It's important that you make this connection!
-
-
-

--what are they?

--Bilateral cases are associated with two of the **familial oculorenal** syndromes (Alport syndrome; Lowe syndrome, and the...**ciliopathies**)

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--Joubert syndrome

--Alström syndrome

--Bardet-Biedl syndrome

--Senior-Løken syndrome

--what are they?

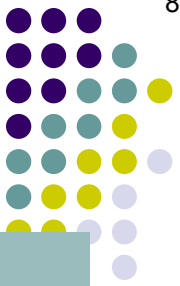
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Do you have a mnemonic for remembering the ciliopathies?

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Do you have a mnemonic for remembering the ciliopathies?
Indeed I do--picture a cilia that
JABS you in the eye

--what are they?

--Bilateral (Alport syndrome; Lowe syndrome, and the...ciliopathies)

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Cilio-pathies	★	Next we will compare/contrast the ciliopathies with Alport and Lowe syndromes				★

Q

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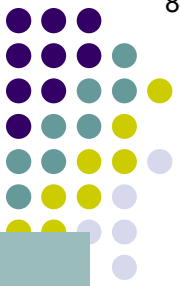
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Cilio-pathies	?	Are lenticonus or other lens findings a feature of the ciliopathies?			?

A

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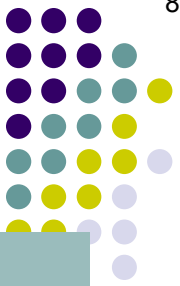
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Cilio-pathies	None ←	No--unlike Alport and Lowe syndromes, ciliopathies are <i>not</i> associated with lens abnormalities			→ None

Q

Lenticonus



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The eyes, brain

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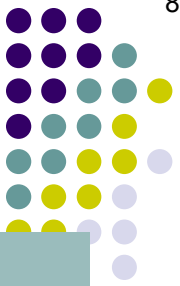
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Cilio-pathies	None	?	In what manner are the ciliopathies inherited?		None

A

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Cilio-pathies	None	AR ←	Unlike the X-linked Alport and Lowe syndromes, ciliopathies are inherited AR		None

Q

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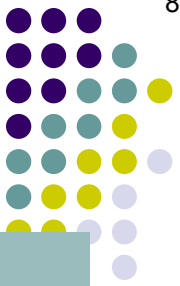
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Cilio-pathies	None	AR	Hematuria?	Is hematuria a feature of the ciliopathies?	

A

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Cilio-pathies	None	AR	No hematuria	Also unlike Alport/Lowe syndromes, ciliopathy pts don't have hematuria (but they do have renal failure)	

Q

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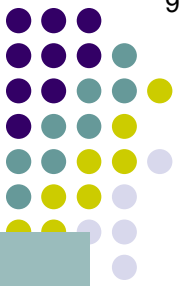
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Lowe Syndrome	Posterior	X-linked	Hematuria in infancy		Microspherophakia
Cilio-pathies	None	AR	No hematuria	?	None

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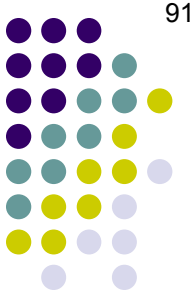
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Cilio-pathies	None	AR	No hematuria	Pigmentary retinopathy with flat ERG	None

Lenticonus



Pigmentary retinopathy in Bardet-Biedl syndrome

Q

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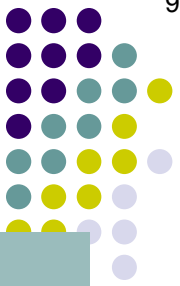
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Cilio-pathies	None	AR	No hematuria	Pigmentary retinopathy with flat ERG	None

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Lowe Syndrome	Pos	<i>'Pigmentary retinopathy with flat ERG in an infant' sounds like what disease?</i> Leber's congenital amaurosis. Therefore, before a child is diagnosed with LCA, one must consider the diagnosis of a ciliopathy.			
Cilio-pathies	None	AR	No hematuria	Pigmentary retinopathy with flat ERG	None

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Leber's congenital amaurosis is an age-related variant of...

two words

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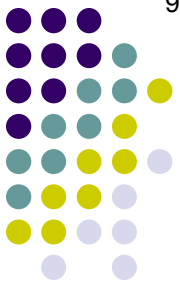
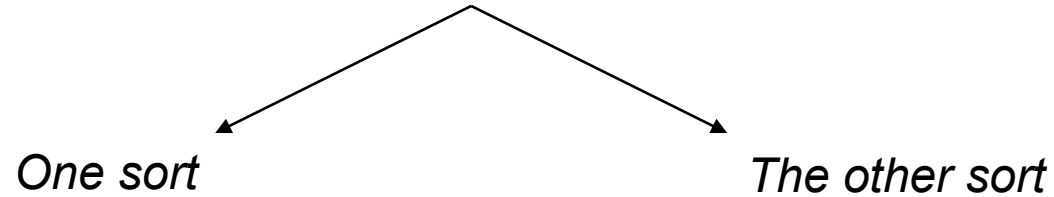
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Lenticonus

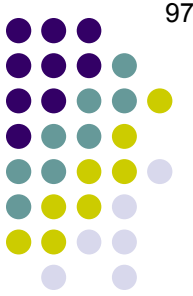
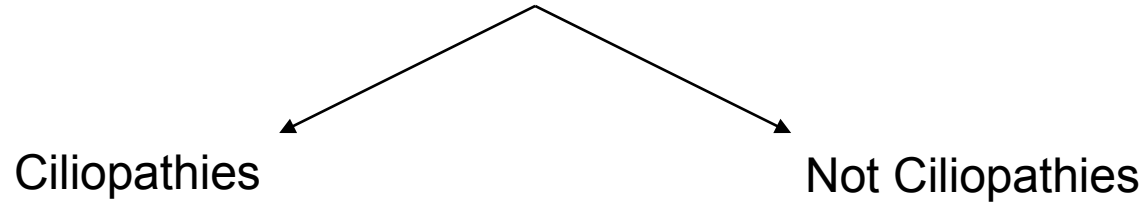
Familial Oculorenal Syndromes *tl;dr*

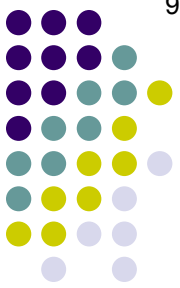
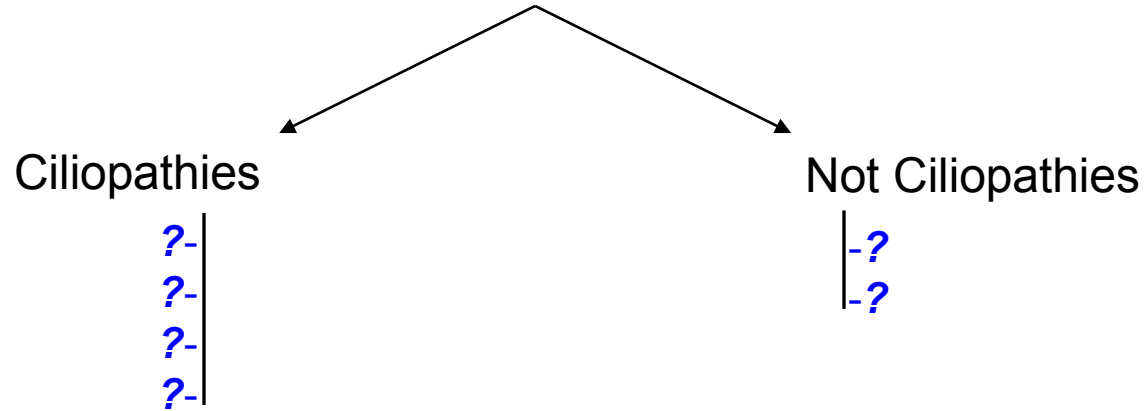


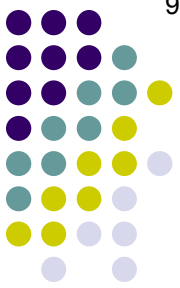
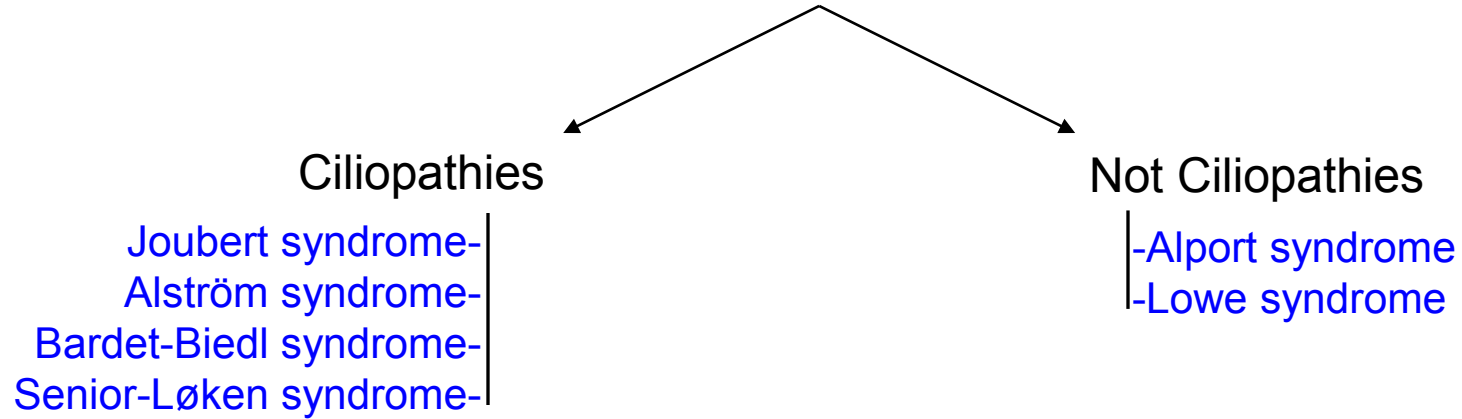
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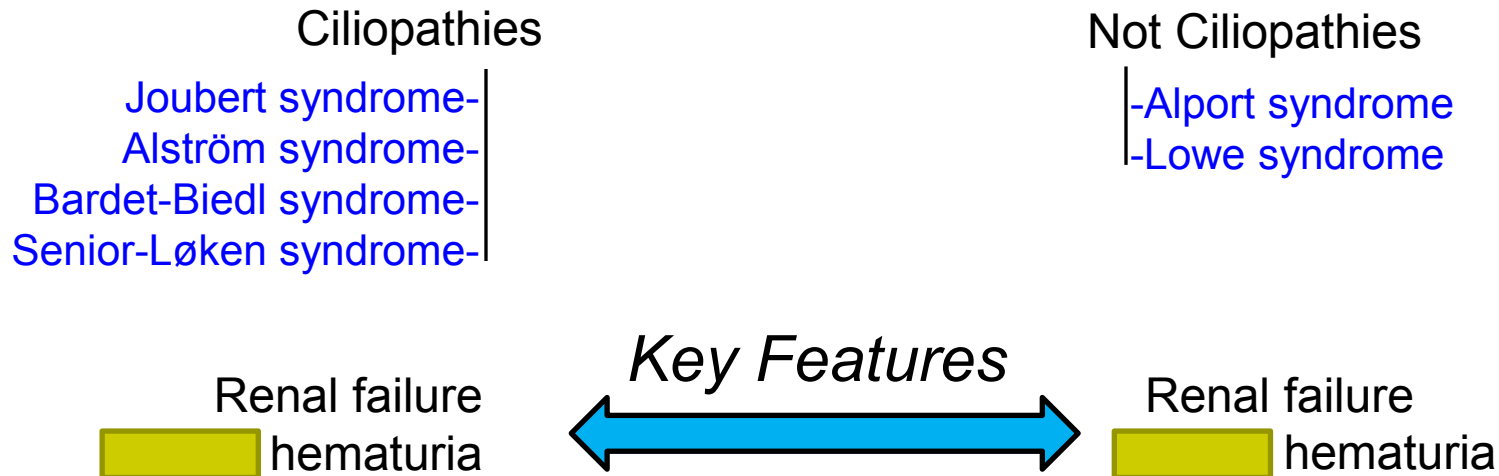
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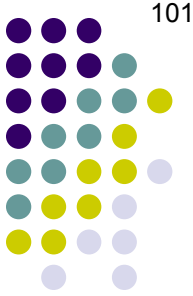
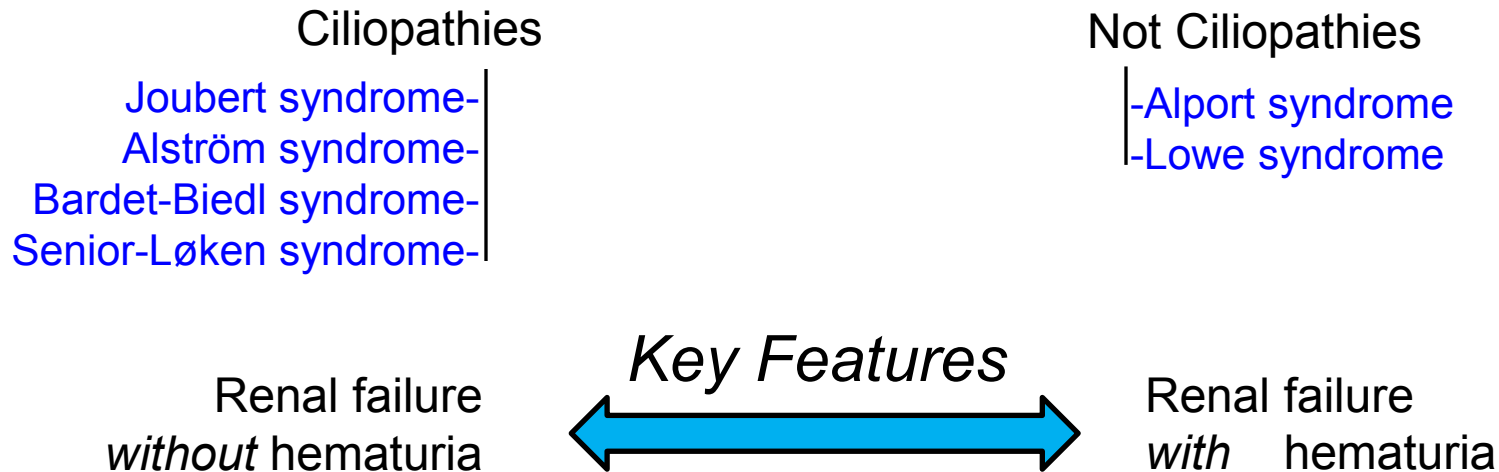
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Lenticonus

Familial Oculorenal Syndromes *tl;dr*





Q

Lenticonus

Familial Oculorenal Syndromes *tl;dr*

Ciliopathies

Joubert syndrome-
Alström syndrome-
Bardet-Biedl syndrome-
Senior-Løken syndrome-

Not Ciliopathies

-Alport syndrome
-Lowe syndrome

Key Features

Renal failure
without hematuria

Classic eye finding:



Renal failure
with hematuria

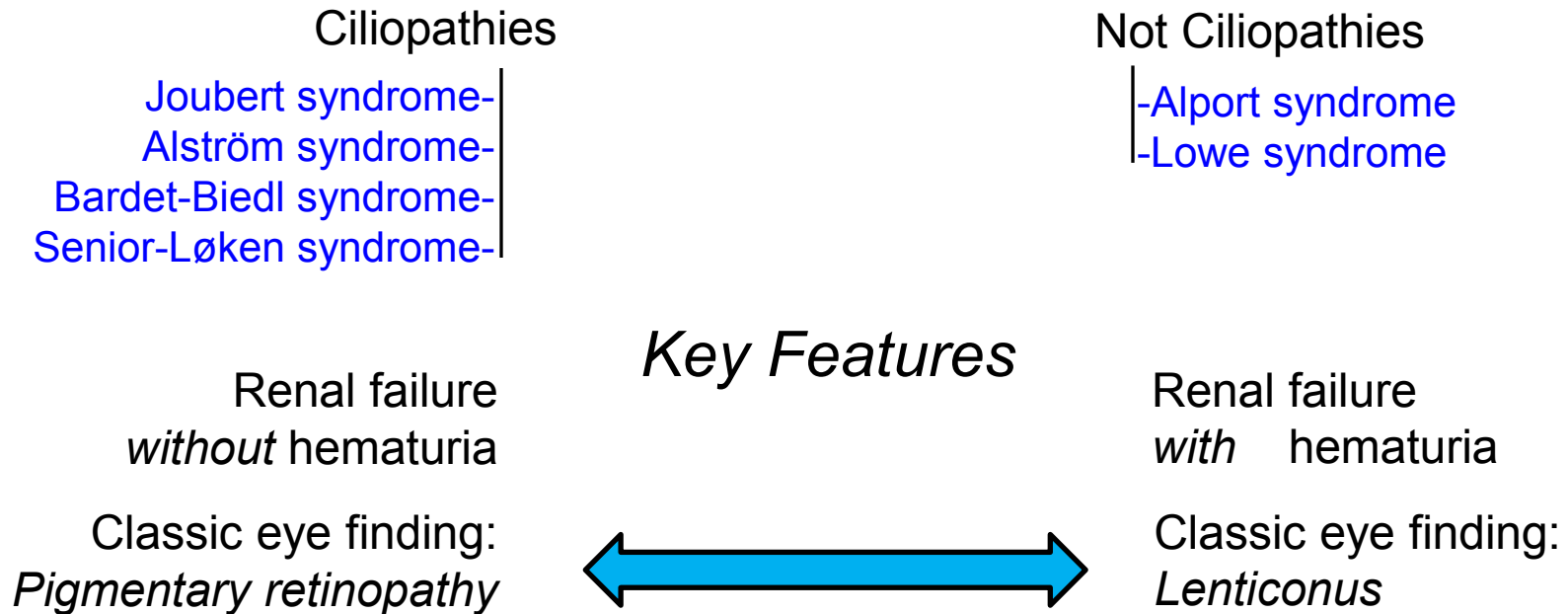
Classic eye finding:

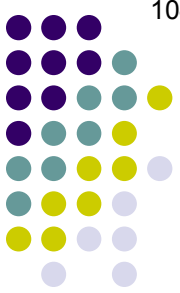
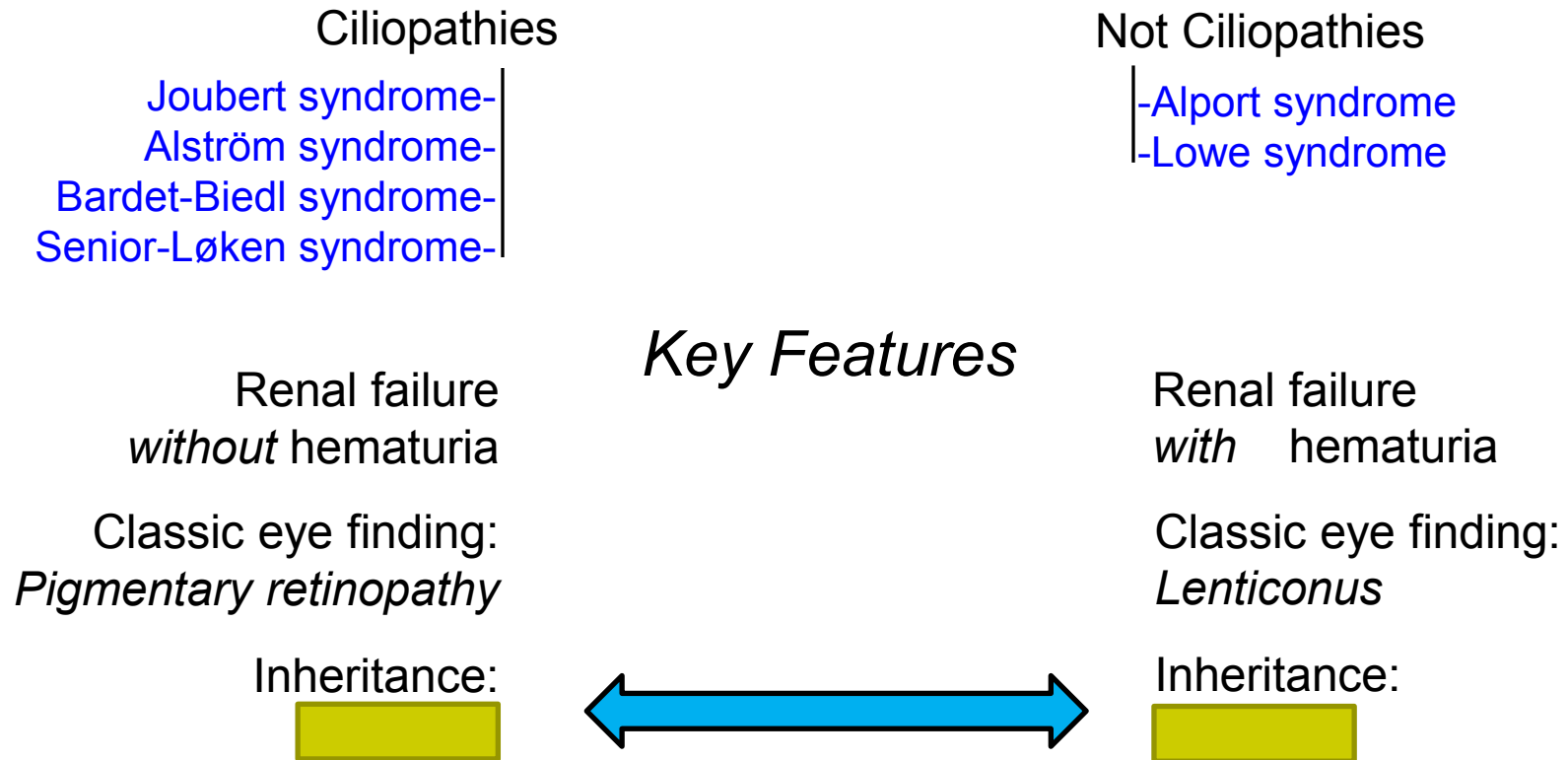


A

Lenticonus

Familial Oculorenal Syndromes *tl;dr*



Familial Oculorenal Syndromes *tl;dr*

A

Lenticonus

Familial Oculorenal Syndromes *tl;dr*

Ciliopathies

- Joubert syndrome-
- Alström syndrome-
- Bardet-Biedl syndrome-
- Senior-Løken syndrome-

Not Ciliopathies

- Alport syndrome
- Lowes syndrome

Key Features

Renal failure
without hematuria

Classic eye finding:
Pigmentary retinopathy

Inheritance:
AR

Renal failure
with hematuria

Classic eye finding:
Lenticonus

Inheritance:
X-linked

