

Q

Lenticonus: Wadda ya know?

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



A

Lenticonus: Wadda ya know?

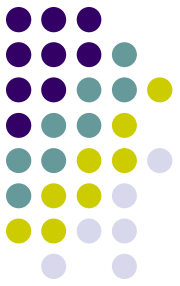
--Secondary to **ectasia** of central portion of **lens surface**



Q

Lenticonus: Wadda ya know?

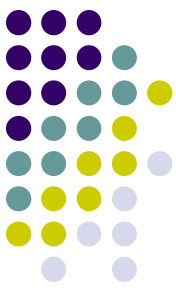
- Secondary to **ectasia** of central portion of **lens surface**
- Typical clinical course:
 - Early: Manifests as **refractive issue**

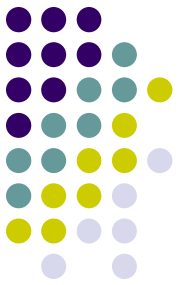


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Lenticonus: Wadda ya know?

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





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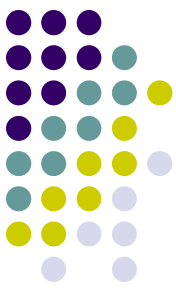
Lenticonus: Wadda ya know?

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

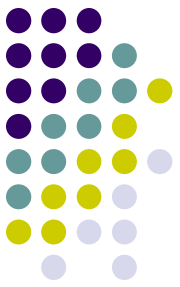
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- Secondary to **ectasia** of central portion of **lens surface**
- Typical clinical course:
 - Early: Manifests as **myopia**
 - Next: Manifests as **irregular astigmatism**



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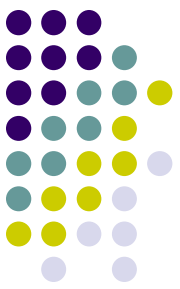
Lenticonus: Wadda ya know?

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

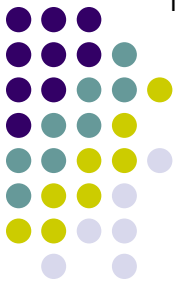
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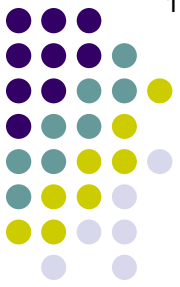
- Secondary to **ectasia** of central portion of **lens surface**
- Typical clinical course:
 - Early: Manifests as **myopia**
 - Next: Manifests as **irregular astigmatism**
 - Later: The **cortex** opacifies



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

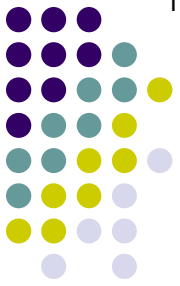


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	Usually unilateral vs usually bilateral		
<i>Anterior lenticonus</i>			
<i>Posterior lenticonus</i>			

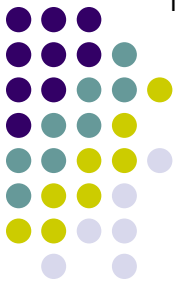


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	Usually unilateral vs usually bilateral		
<i>Anterior lenticonus</i>	Usually bilateral		
<i>Posterior lenticonus</i>	90% unilateral		

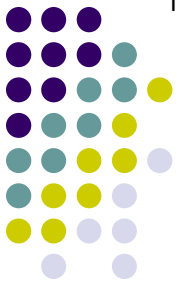


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	Usually unilateral vs usually bilateral	More common vs less common	
Anterior lenticonus	Usually bilateral		
Posterior lenticonus	90% unilateral		

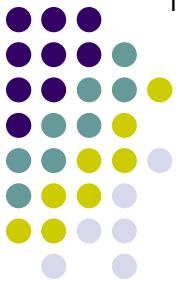


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	Usually unilateral vs usually bilateral	More common vs less common	
<i>Anterior lenticonus</i>	Usually bilateral	Less common	
<i>Posterior lenticonus</i>	90% unilateral	More common (but still rare)	

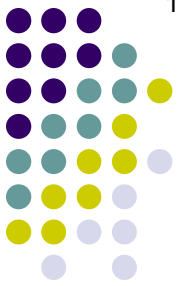


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 - Early: Manifests as **myopia**
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	Usually unilateral vs usually bilateral	More common vs less common	Usually sporadic vs usually syndromic
Anterior lenticonus	Usually bilateral	Less common	
Posterior lenticonus	90% unilateral	More common (but still rare)	

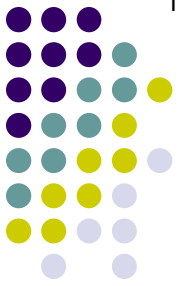


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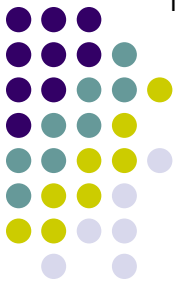
	Usually unilateral vs usually bilateral	More common vs less common	Usually sporadic vs usually syndromic
<i>Anterior lenticonus</i>	Usually bilateral	Less common	Usually syndromic
<i>Posterior lenticonus</i>	90% unilateral	More common (but still rare)	Usually sporadic (unless bilateral)



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 - Early: Manifests as **myopia**
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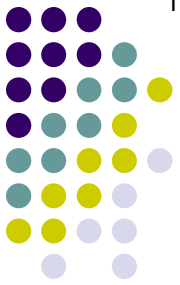
	Usually unilateral vs	More common	Usually sporadic vs
<p><i>Note that these factoids 'go together,' ie, syndromic conditions usually affect both eyes...</i></p>			
Anterior lenticonus	Usually bilateral	Less common	Usually syndromic
Posterior lenticonus	90% unilateral	More common (but still rare)	Usually sporadic (unless bilateral)



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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	less common	usually syndromic
<i>...and likewise, these two go together as well (sporadic conditions usually affect only one)</i>			
Posterior lenticonus	usually unilateral	more common (but still rare)	usually sporadic (unless bilateral)



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 - Later: The **cortex** opacifies
 - At age **4-5** years: Capsule **ruptures** → **total opacification**
- **unilateral vs bilateral** cases are associated with two of the **two words** *syndromes*
 - (**eponym** syndrome; **different eponym** syndrome)

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



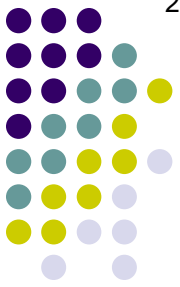
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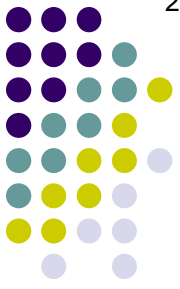
	Lenticonus location				
Alport Syndrome	?				
Lowe Syndrome	?				

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

	Lenticonus location				
Alport Syndrome	Anterior				
Lowe Syndrome	Posterior				

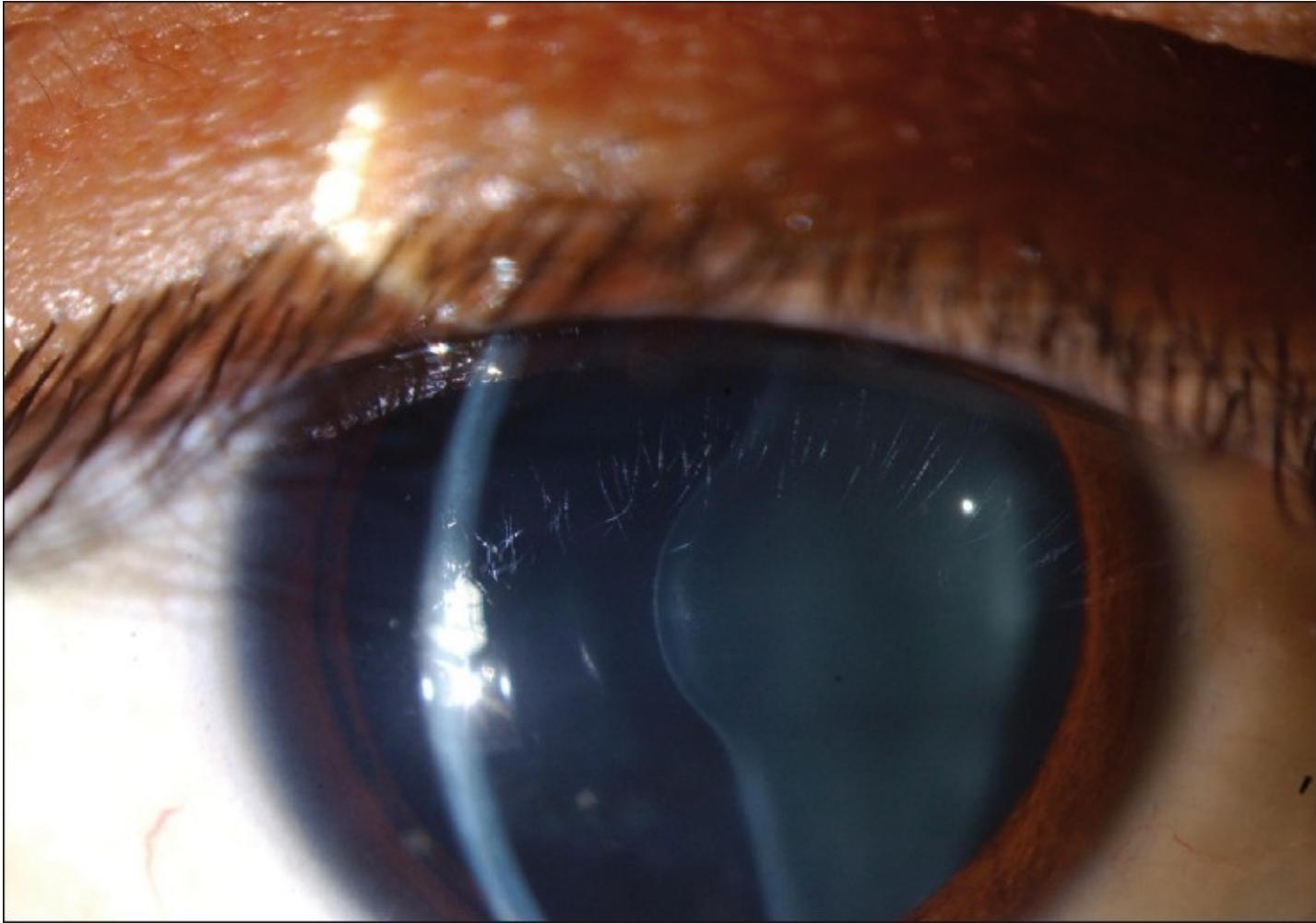


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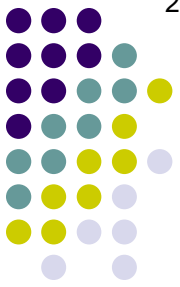
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A lport syndrome Anterior lenticonus					
Alport Syndrome	Anterior				
Lowe Syndrome	Posterior				

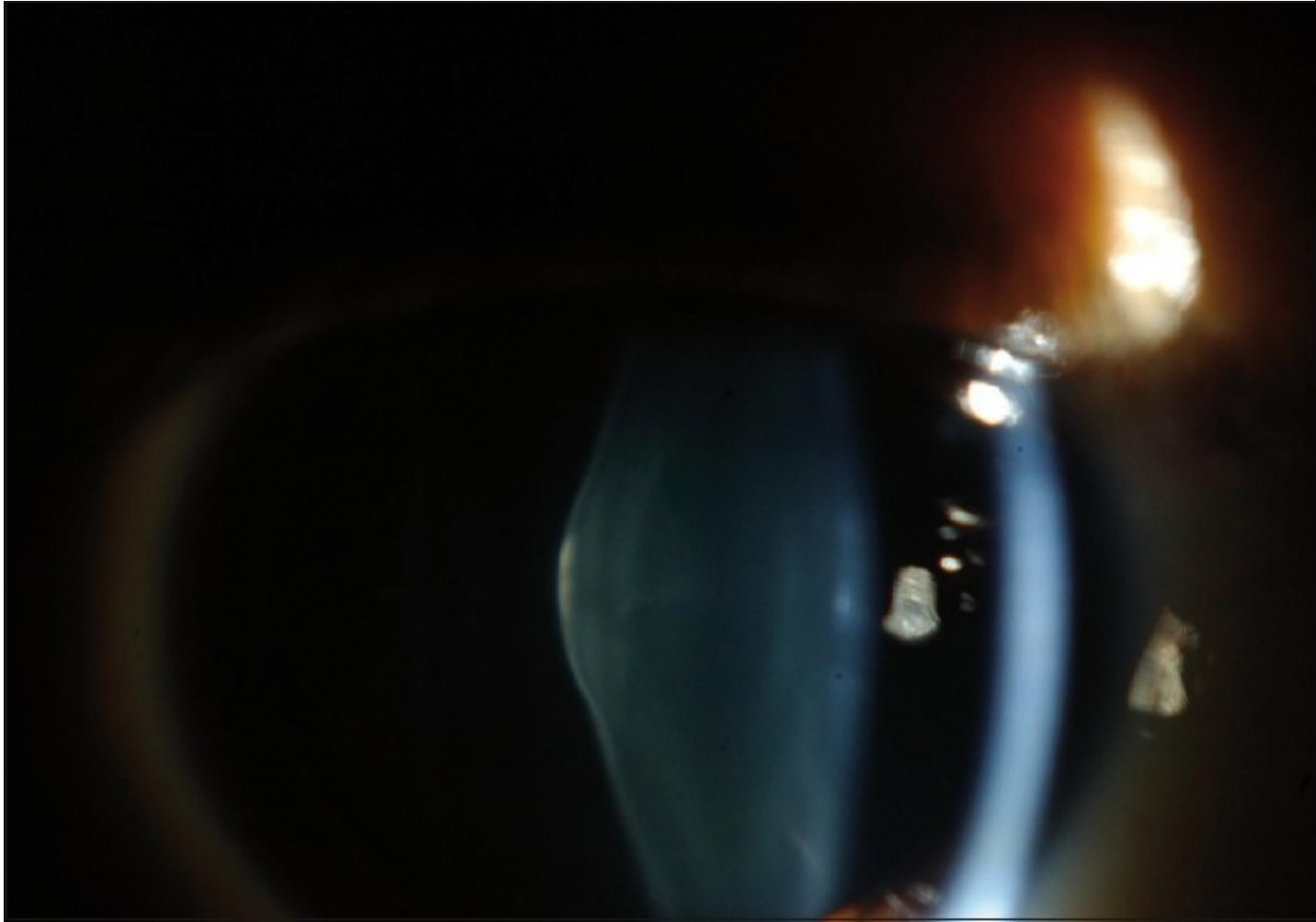
Lenticonus: Wadda ya know?



Anterior lenticonus in Alport syndrome

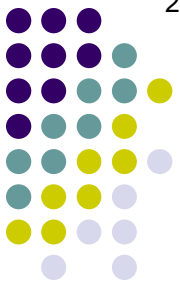


Lenticonus: Wadda ya know?



Posterior lenticonus





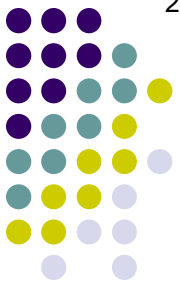
Q

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How prevalent is lenticonus in Alport and Lowe syndromes?

	Lenticonus location				
Alport Syndrome	Anterior				
Lowe Syndrome	Posterior				



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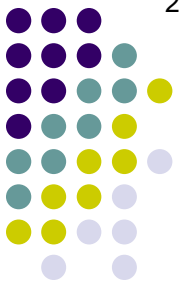
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How prevalent is lenticonus in Alport and Lowe syndromes?

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				

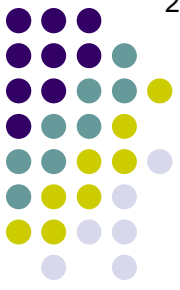


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- Bilateral** cases are associated with two of the **familial oculorenal syndromes** (**Alport** syndrome; **Lowe** syndrome)

	Lenticonus location	Inheritance			
Alport Syndrome	Anterior	?			
Lowe Syndrome	Posterior	?			



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



Q

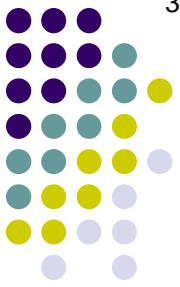
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	Lenticonus location	Inheritance	Classic presentation (nonocular)		
Alport Syndrome	Anterior	Most are X-linked	?		
Low e Syndrome	Posterior	X-linked	?		

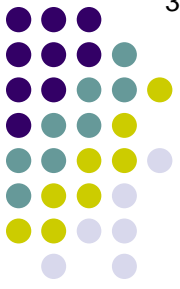
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	Lenticonus location	Inheritance	Classic presentation (nonocular)		
Alport Syndrome	Anterior	Most are X-linked	Hematuria in childhood vs infancy		
Lowe Syndrome	Posterior	X-linked	Hematuria in childhood vs infancy		



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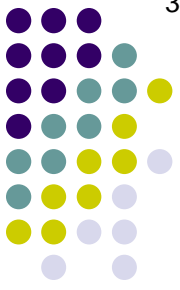


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	Lenticonus location	Inheritance	Classic presentation (nonocular)	Associated findings	
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Lowe Syndrome	Posterior	X-linked	Hematuria in infancy	?	

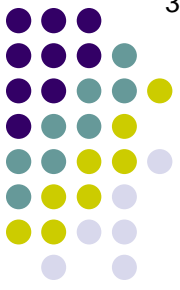


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	Lenticonus location	Inheritance	Classic presentation (nonocular)	Associated findings	
Alport Syndrome	Anterior	Most are X-linked	Hematuria in childhood	High- <i>f</i> nerve deafness; nephritis	
Lowe Syndrome	Posterior	X-linked	Hematuria in infancy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

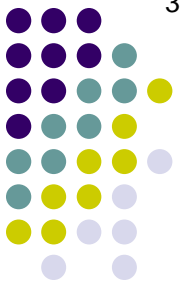


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	Lenticonus location	Inheritance	Classic presentation (nonocular)	Associated findings	Another lens finding
Alport Syndrome	Anterior	Most are X-linked	Hematuria in childhood	High- <i>f</i> nerve deafness; nephritis	?
Lowe Syndrome	Posterior	X-linked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



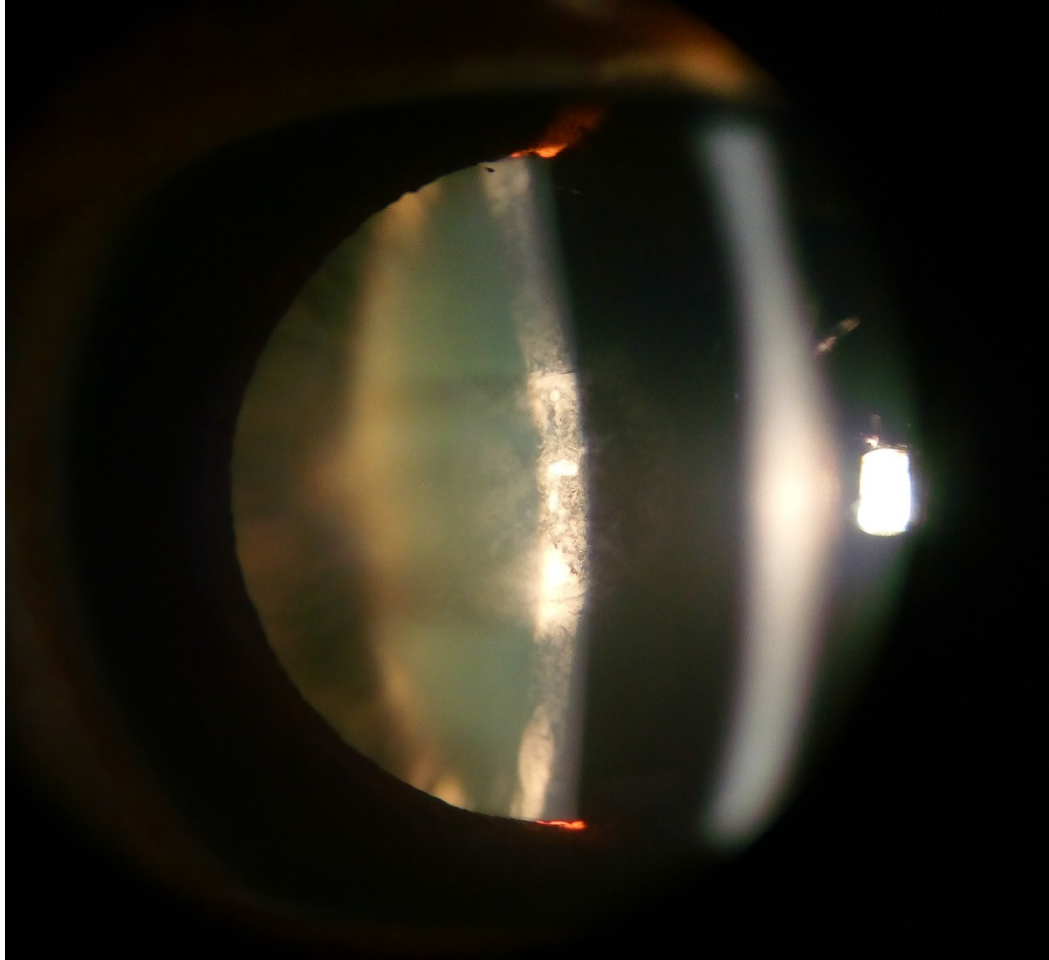
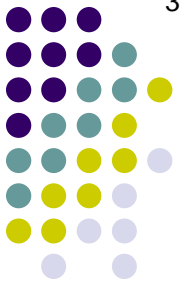
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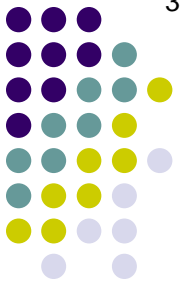
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Alport Syndrome	Anterior	Most are X-linked	Hematuria in childhood	High- <i>f</i> nerve deafness; nephritis	Anterior subcapsular cataract
Lowe Syndrome	Posterior	X-linked	Hematuria in infancy	MR; rickets; hypotonia	Microspherophakia

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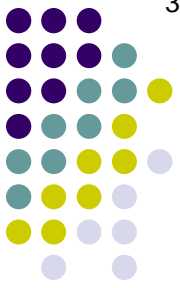


Anterior subcapsular cataract

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Microspherophakia



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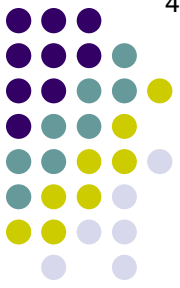
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- Bilateral** cases are associated with two of the **familial oculorenal syndromes** (**Alport** syndrome; **Lowe** syndrome, and the...)

	Lenticonus location	Inheritance	Classic presentation (nonocular)	Associated findings	Another lens finding
<i>Alport Syndrome</i>	Anterior	Most are X-linked	Hematuria in childhood	High- <i>f</i> nerve deafness; nephritis	Microspherophakia
<i>Lowe Syndrome</i>	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: Wadda ya know?

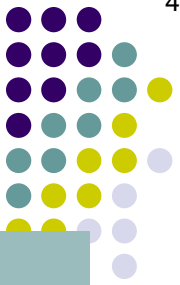
- Secondary to **ectasia** of central portion of **lens surface**
- Typical clinical course:
 - Early: Manifests as **myopia**
 - Next: Manifests as **irregular astigmatism**
 - Later: The **cortex** opacifies
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Q

Lenticonus: Wadda ya know?



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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What is a ciliopathy?

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Cilia are ubiquitous

The eyes

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

Where are they?

Later: The cortex opacifies

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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 cortex opacifies

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What is a ciliopathy?

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Cilia are ubiquitous
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!
-
-
-

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

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

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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

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

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Cilio-pathies	None ←	No--unlike Alport and Low			→ None

Q

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Cilio-pathies	None	? ←	<i>In what manner are the ciliopathies inherited?</i>		None

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

Q

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Cilio-pathies	None	AR	Hematuria?	<i>Is hematuria a feature of the ciliopathies?</i>	

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Lowe Syndrome	Posterior	X-linked	Hematuria in infancy	MR; rickets; hypotonia	Microspherophakia
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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Low e Syndrome	Posterior	X-linked	Hematuria in infancy		Microspherophakia
Cilio-pathies	None	AR	No hematuria	?	None

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Low e Syndrome	Posterior	X-linked	Hematuria in infancy		Microspherophakia
Cilio-pathies	None	AR	No hematuria	Pigmentary retinopathy with flat ERG	None

Lenticonus: Wadda ya know?



Pigmentary retinopathy in Bardet-Biedl syndrome

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Lowe Syndrome	Pos	'Pigmentary retinopathy with flat ERG in an infant' sounds like what disease?			
Cilio-pathies	None	AR	No hematuria	Pigmentary retinopathy with flat ERG	None

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Low e Syndrome	Pos	'Pigmentary retinopathy with flat ERG in an infant' sounds like what disease? 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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Alport	Anterior	Most are	Hematuria in	High-f nerve deafness:	Microspherokakia
Lowe Syndrome	Posterior				
Cilio-pathies	None	AR	No hematuria	Pigmentary retinopathy with flat ERG	None

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

'Pigmentary retinopathy with flat ERG in an infant' sounds like what disease? Leber's congenital amaurosis therefore, before a child is diagnosed with LCA, one must consider the diagnosis of a ciliopathy.

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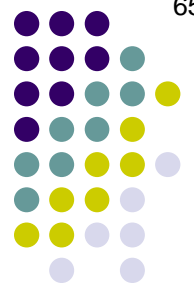
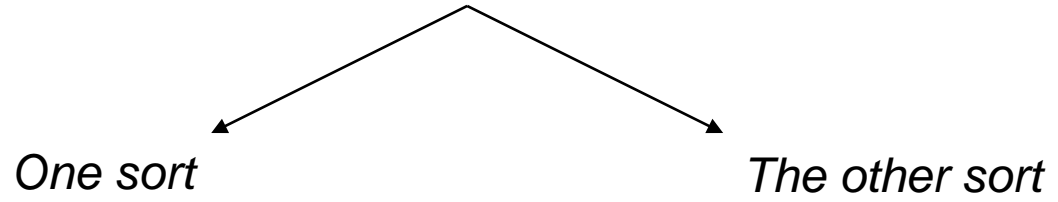
Leber's congenital amaurosis is an age-related variant of...retinitis pigmentosa

'Pigmentary retinopathy with flat ERG in an infant' sounds like what disease? Leber's congenital amaurosis

Therefore, before a child is diagnosed with LCA, one must consider the diagnosis of a ciliopathy.

Lenticonus: Wadda ya know?

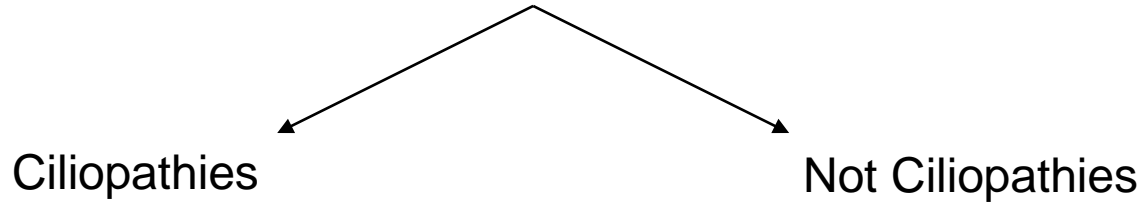
Familial Oculorenal Syndromes *tl;dr*

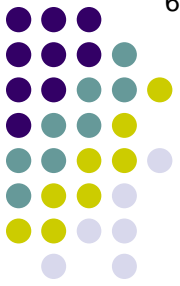


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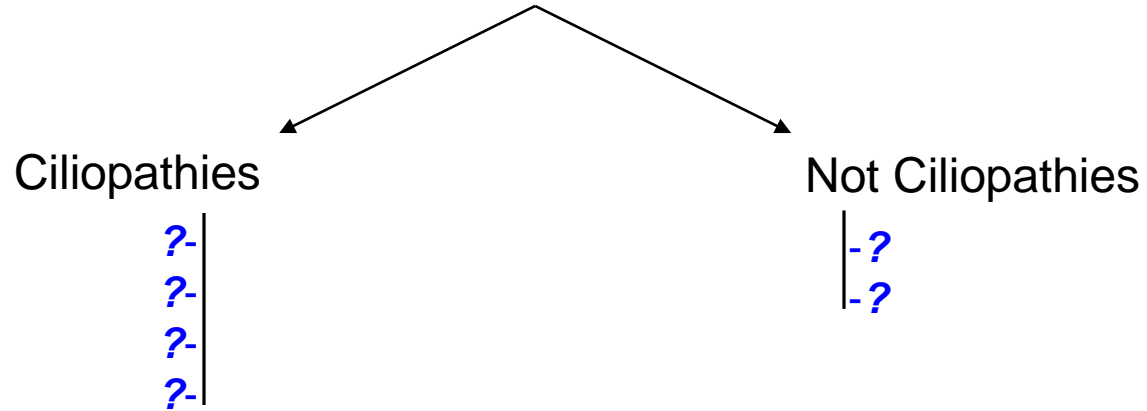




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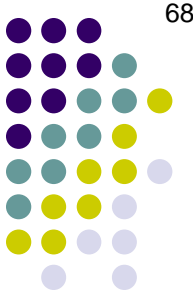
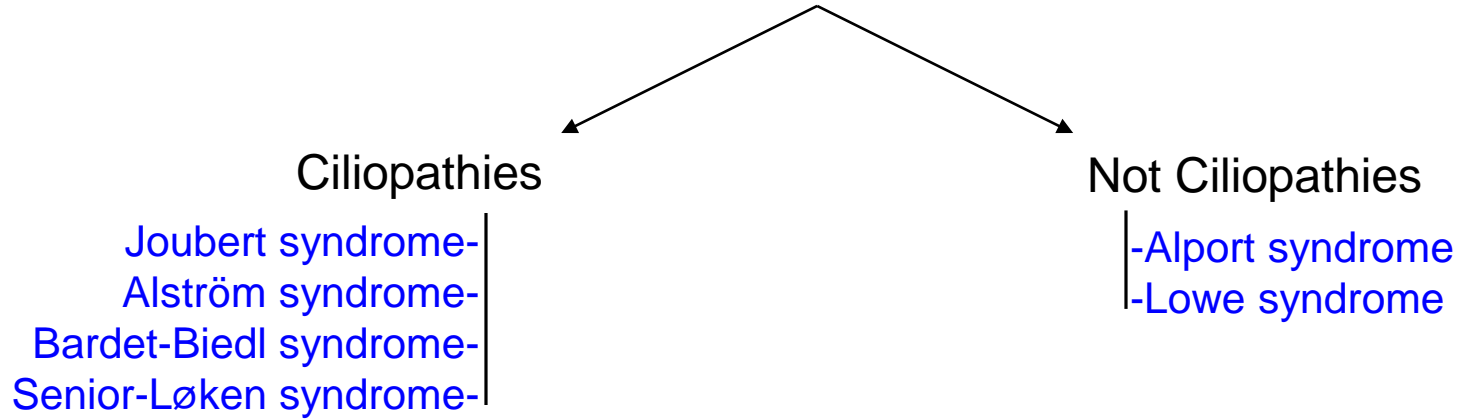
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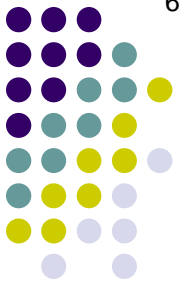


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Lenticonus: Wadda ya know?

Familial Oculorenal Syndromes *tl;dr*

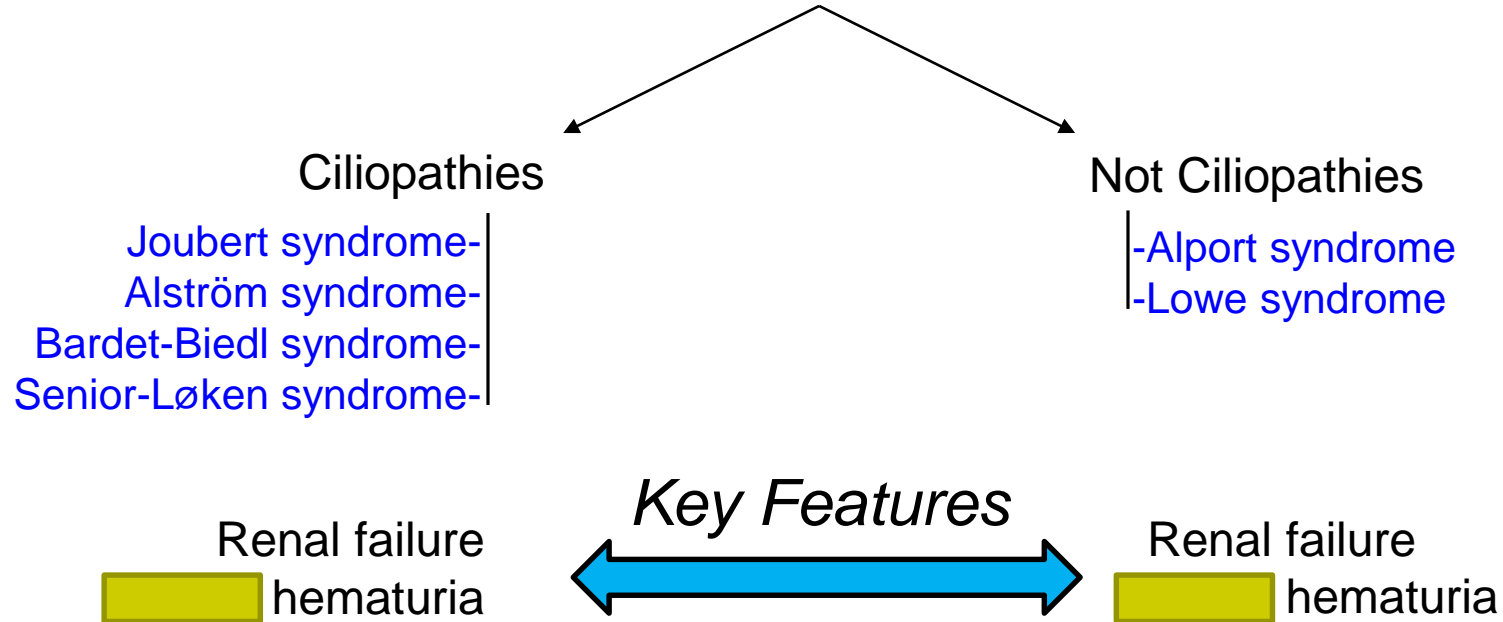


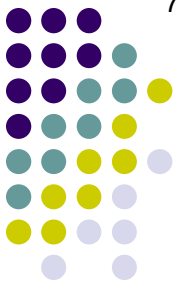


Q

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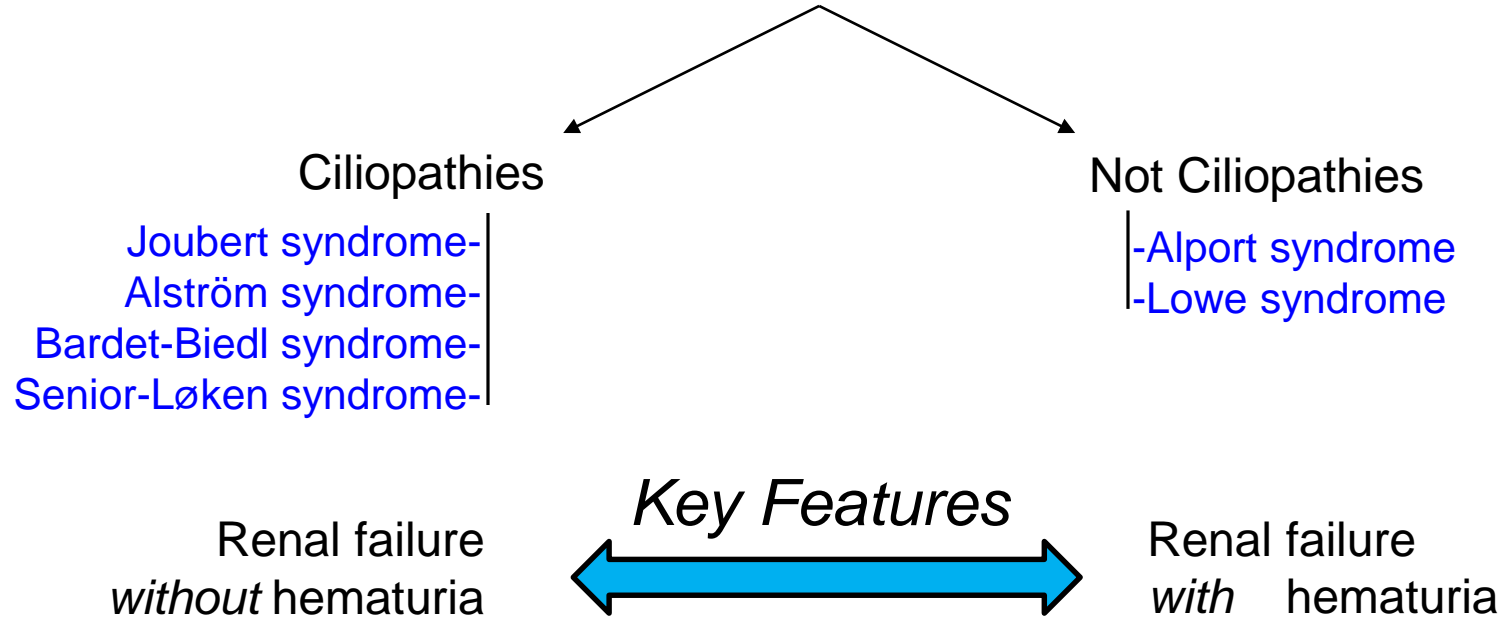


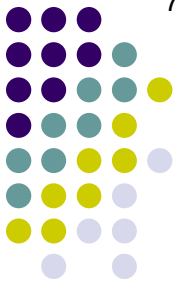


A

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Q

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Ciliopathies

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

Not Ciliopathies

-Alport syndrome
-Lowe syndrome

Key Features

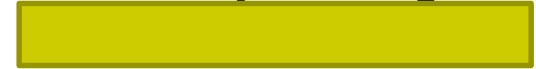
Renal failure
without hematuria

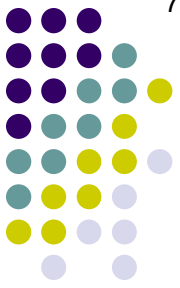
Classic eye finding:



Renal failure
with hematuria

Classic eye finding:

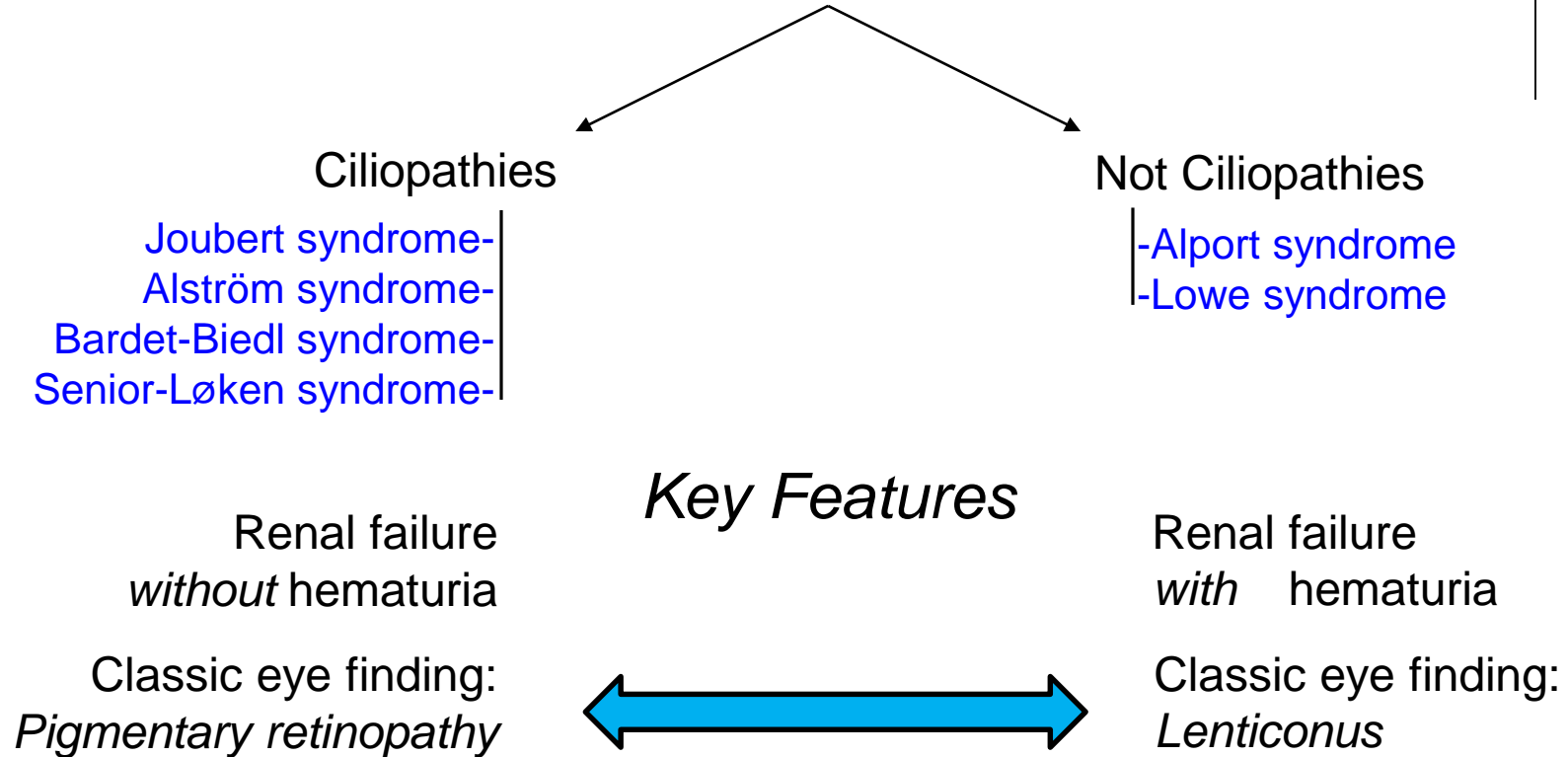


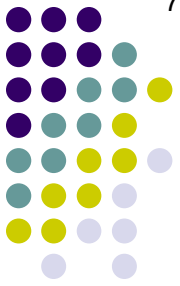


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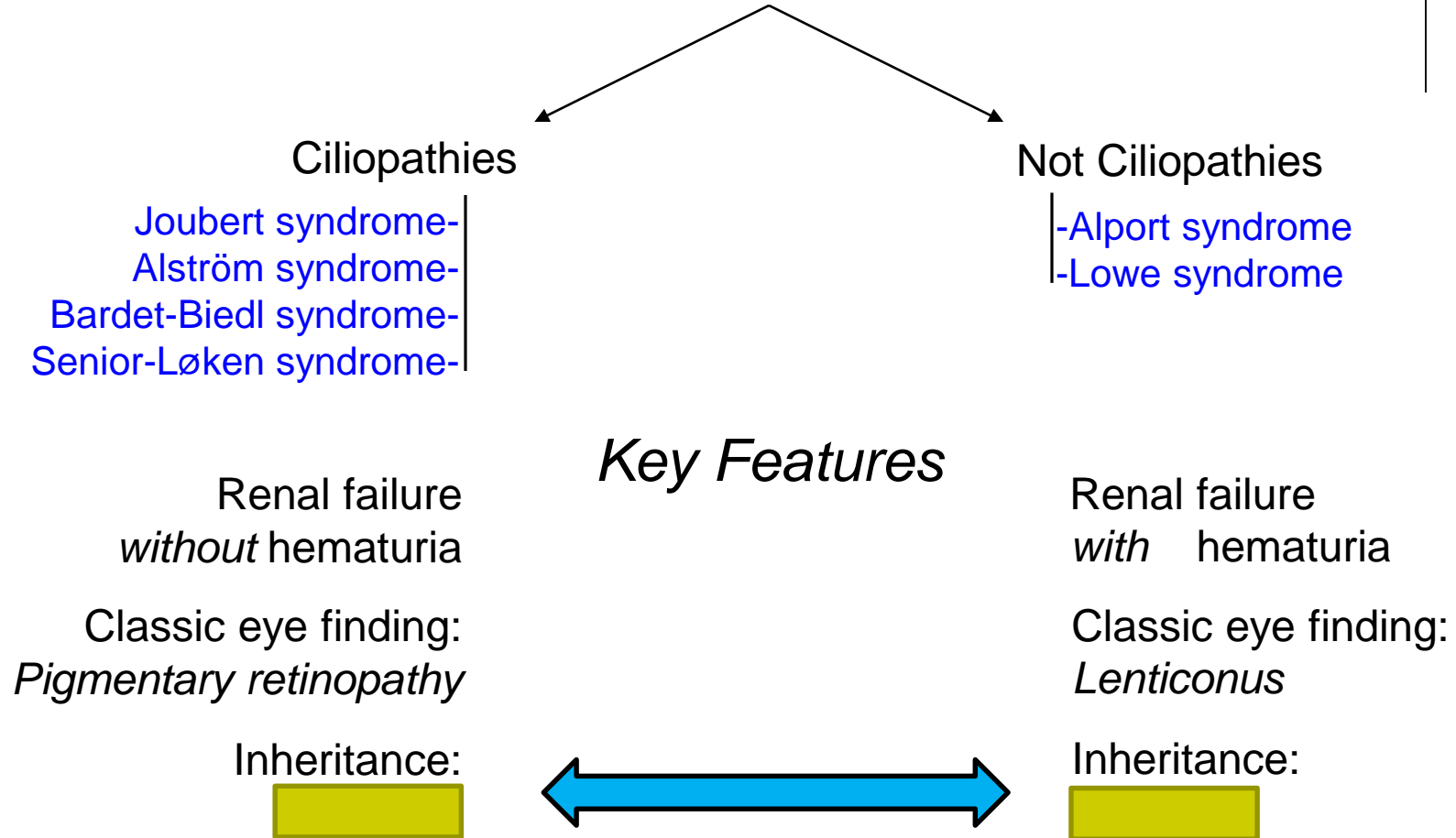


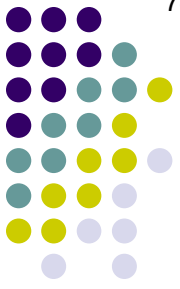


Q

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A

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