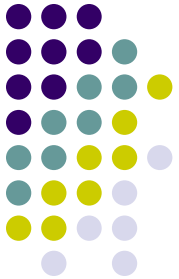
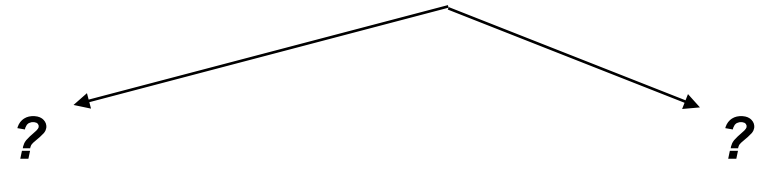
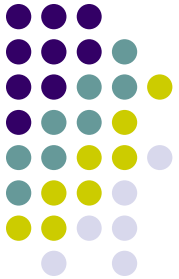
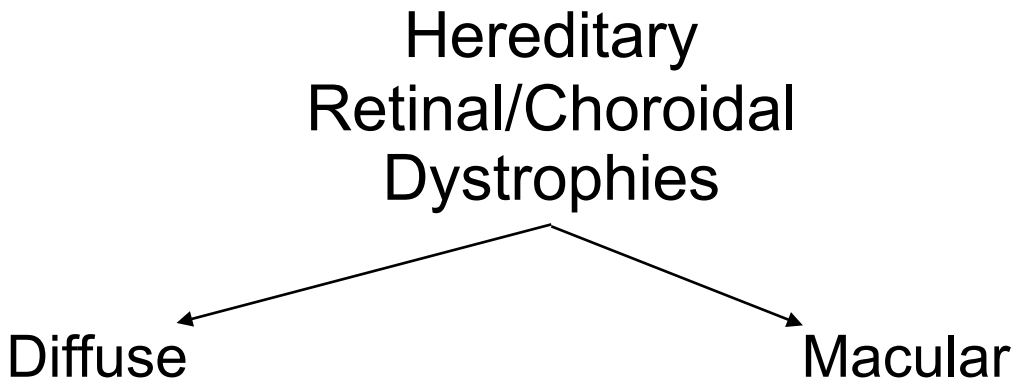


Hereditary Retinal/Choroidal Dystrophies



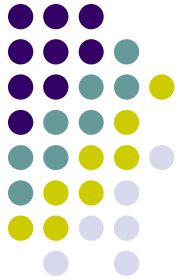
The Retina *book divvies the hereditary retinal/choroidal dystrophies into two groups—what are they?*





The Retina *book divvies the hereditary retinal/choroidal dystrophies into two groups—what are they?*

Those that affect the entire retina vs those affecting the macula only

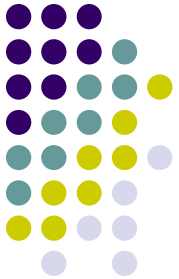


Hereditary Retinal/Choroidal Dystrophies

Diffuse **Macular**

So when you think about the retinal/choroidal dystrophies, the first thing to recall is that they come in two basic flavors—*macular* and *diffuse*

No question—proceed when ready



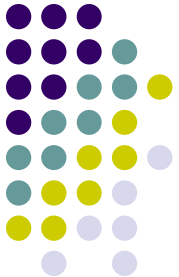
Hereditary Retinal/Choroidal Dystrophies

Diffuse **Macular**

Spoiler alert: There is **one** retinal dystrophy (and a highly testable one at that) which doesn't fit neatly into the *diffuse* vs *macular* system. (We'll get to it at the end of the set.)

?

No question—proceed when ready



Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular

- *One of two with* macular *in its name* — ?
- *This one is the best* — ?
- *The star of the mac dystrophies* — ?
- *A group of non-lookalike conditions* — ?
- *Looks like GA* — ?
- *The other with* macular *in its name* — ?

What are the hereditary macular dystrophies covered in the Retina *book?*

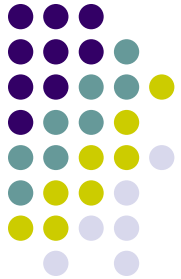
Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular

What are the hereditary macular dystrophies covered in the Retina *book?*

- *One of two with* macular *in its name* — Sorsby macular dystrophy
- *This one is the best* — Best dz
- *The star of the mac dystrophies* — Stargardt
- *A group of non-lookalike conditions* — Pattern dystrophies
- *Looks like GA* — Central areolar choroidal dystrophy
- *The other with* macular *in its name* — North Carolina macular dystrophy



Hereditary Retinal/Choroidal Dystrophies

- Diffuse
- Macular
  - **Sorsby macular dystrophy**
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy
  - North Carolina macular dystrophy

What is the classic (and unfortunate) manifestation of Sorsby's?

Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular

- **Sorsby macular dystrophy**
- Best dz
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What is the classic (and unfortunate) manifestation of Sorsby's?

Bilateral CNVMs occurring at around age #

Hereditary Retinal/Choroidal Dystrophies

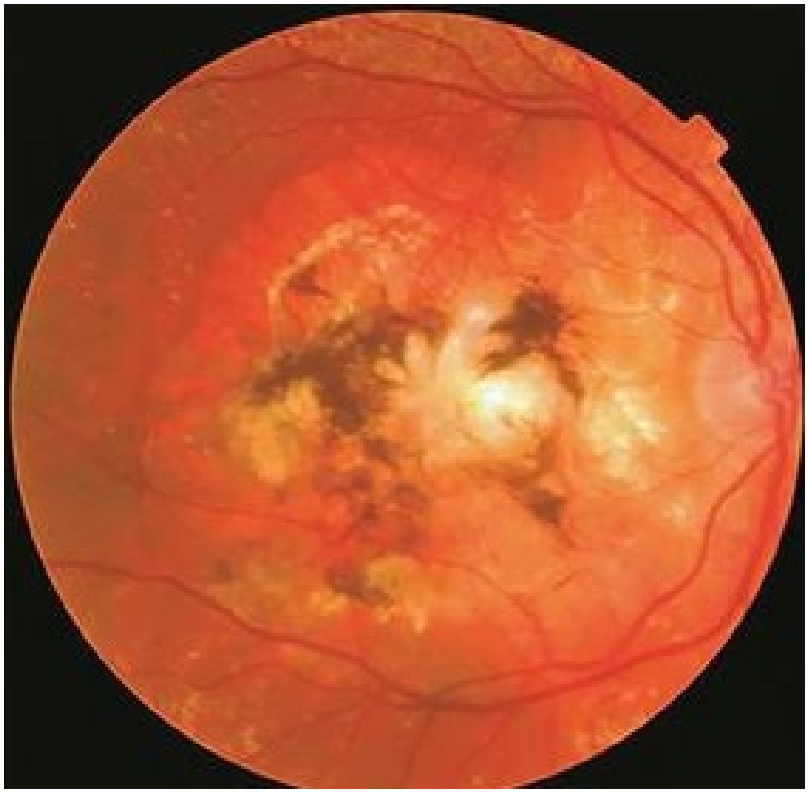
Diffuse

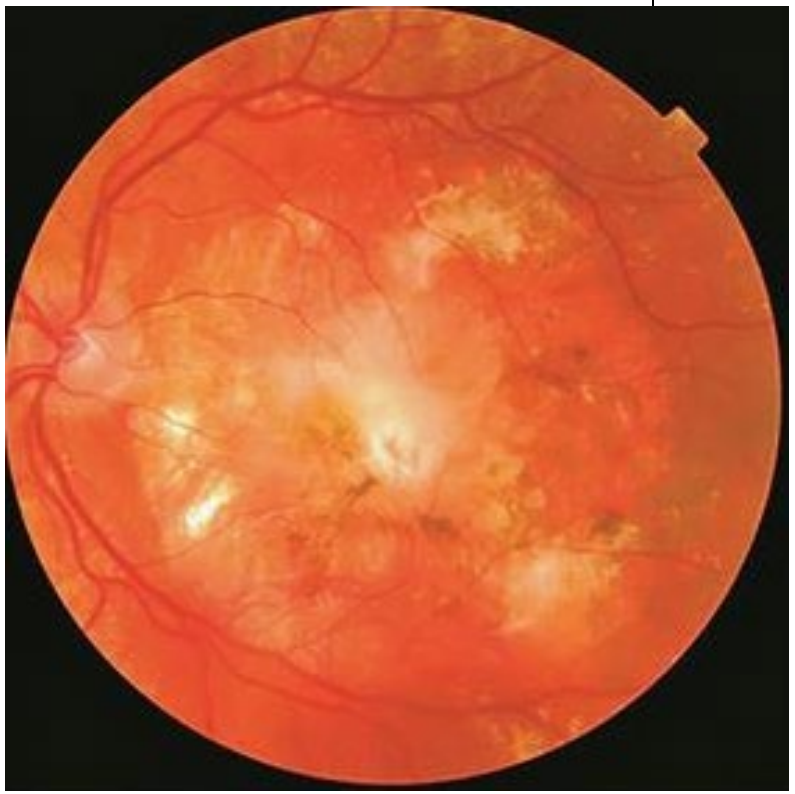
Macular

- **Sorsby macular dystrophy**
- Best dz
- Stargardt
- Pattern dystrophies
- Central areolar choroidal dystrophy
- North Carolina macular dystrophy

What is the classic (and unfortunate) manifestation of Sorsby's?

Bilateral CNVMs occurring at around age 40





Sorsby: Active CNVM OD, already (disciform) scarred OS

Hereditary Retinal/Choroidal Dystrophies

- Diffuse
- Macular
  - Sorsby macular dystrophy
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How does Best dz present?

Hereditary Retinal/Choroidal Dystrophies

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How does Best dz present?
With a vitelliform lesion (or lesions) during life period
(the condition is aka *Best vitelliform dystrophy*)

Hereditary Retinal/Choroidal Dystrophies

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Hereditary Retinal/Choroidal Dystrophies

Diffuse

How does Best dz present?

With a **vitelliform** lesion (or lesions) during childhood (the condition is aka *Best vitelliform dystrophy*)

What does vitelliform *mean?*

Macular

- Sorsby macular dystrophy
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Hereditary Retinal/Choroidal Dystrophies

Diffuse

How does Best dz present?
With a **vitelliform** lesion (or lesions) during childhood
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What does vitelliform *mean?*
It means 'egg-yolk-like'

Macular

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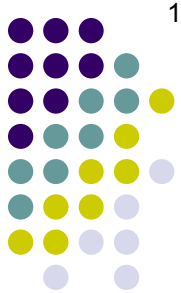

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Because they're egg-yolk-like, what can you infer about a vitelliform lesion's…
…color?

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Because they're egg-yolk-like, what can you infer about a vitelliform lesion's…
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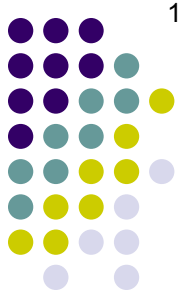

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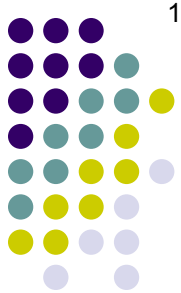

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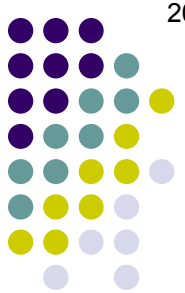
Because they're egg-yolk-like, what can you infer about a vitelliform lesion's…
…color? Yellow
…shape? **Round**



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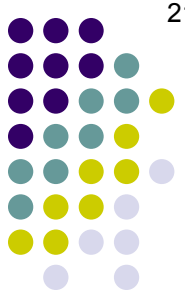

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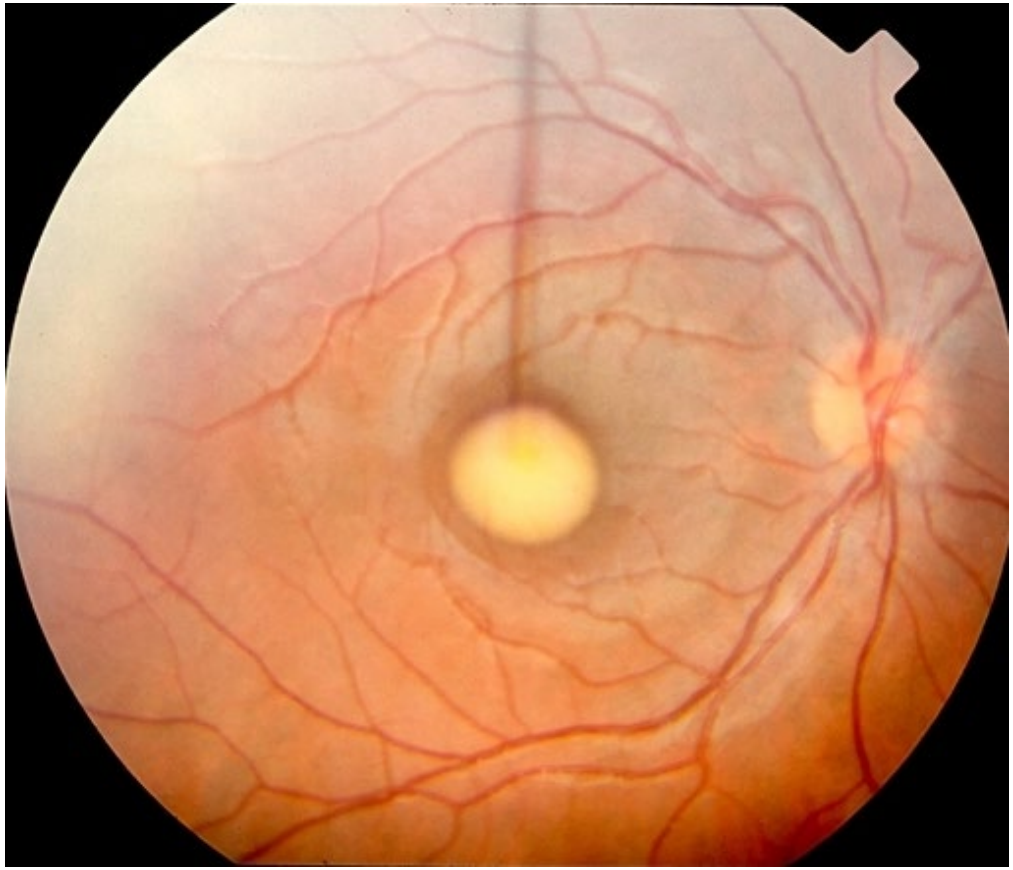

Best dz: Vitelliform lesion

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How does Best dz present?
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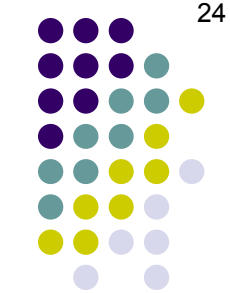

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The electro-oculogram (EOG)

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RPE function

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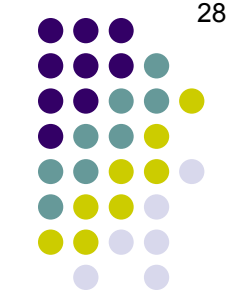
RPE function

The results of EOG testing are reported in terms of a ratio. What is its eponymous name?



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The **Arden ratio**

Hereditary Retinal/Choroidal Dystrophies

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Other than its name, what makes Stargardt's the 'star of the macular dystrophies'?

Hereditary Retinal/Choroidal Dystrophies

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Other than its name, what makes Stargardt's the 'star of the macular dystrophies'?

It is the most common

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It is the most common

What is the classic finding on DFE?

Hereditary Retinal/Choroidal Dystrophies

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Other than its name, what makes Stargardt's the 'star of the macular dystrophies'?
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What is the classic finding on DFE?
[redacted] (that means [redacted]–shaped) lesions in the macula

Hereditary Retinal/Choroidal Dystrophies

- Diffuse
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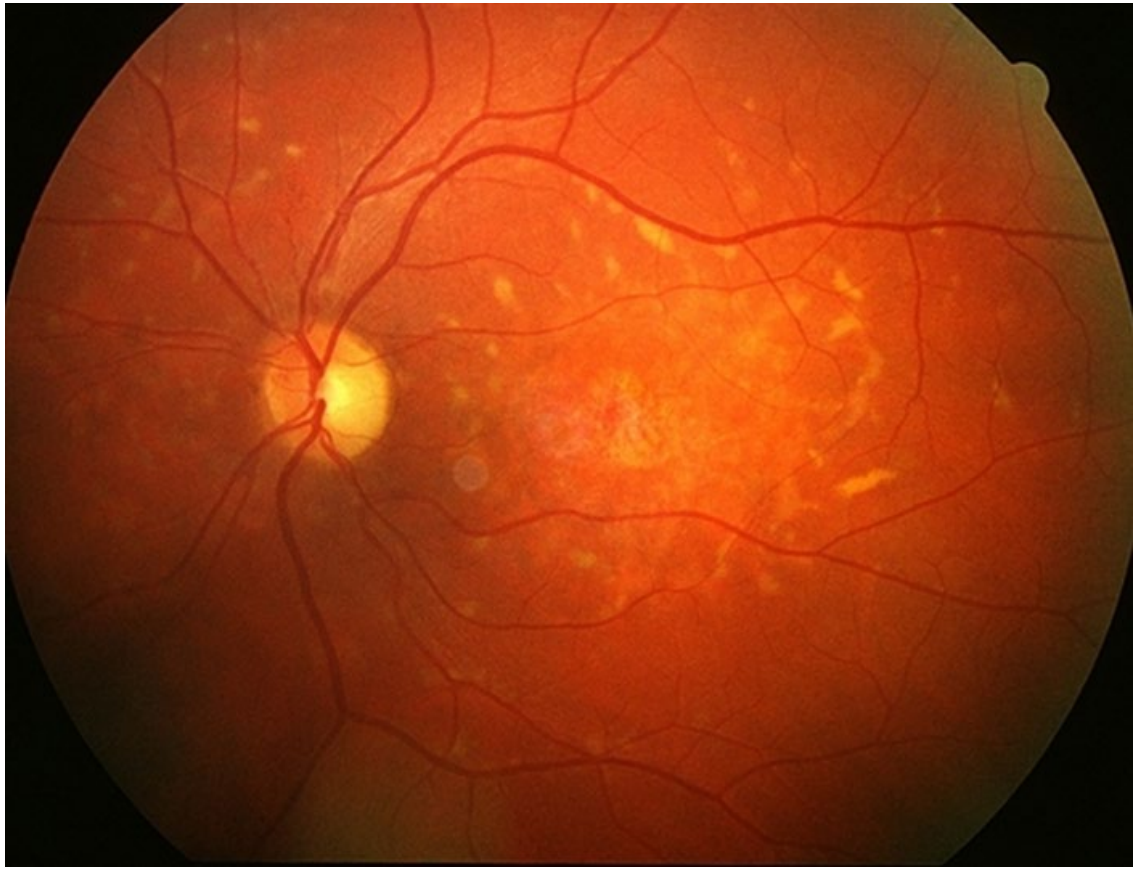
It is the most common

What is the classic finding on DFE?
Pisciform (that means fish –shaped) lesions in the macula



Stargardt: RPE-level pisciform lesions

Hereditary Retinal/Choroidal Dystrophies

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What is the classic finding on fluorescein angiography?

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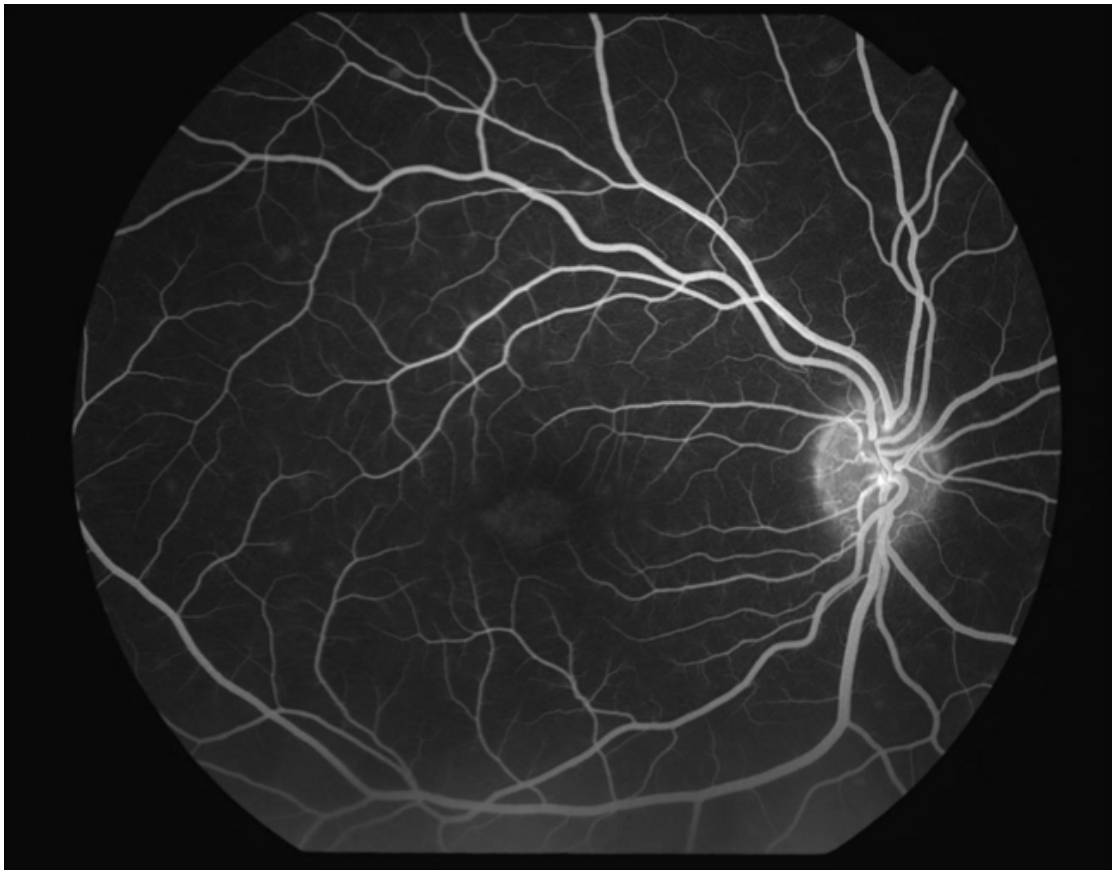
What is the classic finding on DFE?
Pisciform (that means fish –shaped) lesions in the macula

What is the classic finding on fluorescein angiography?
'Dark choroid'



Stargardt—*dark choroid* appearance on FA

Hereditary Retinal/Choroidal Dystrophies

Diffuse

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Pisciform (that means fish –shaped) lesions in the macula

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'Dark choroid'

What is the classic finding on autofluorescence imaging?

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A ring of perifoveal hypo *vs* hyper-autofluorescence surrounding a central foveal area of hypo *vs* hyper-autofluorescence

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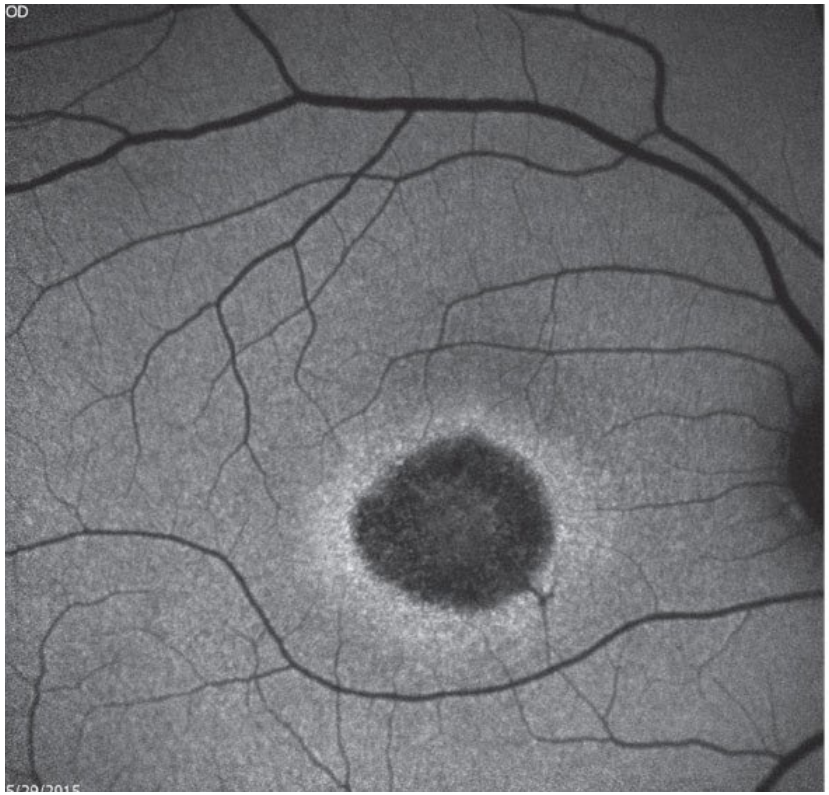
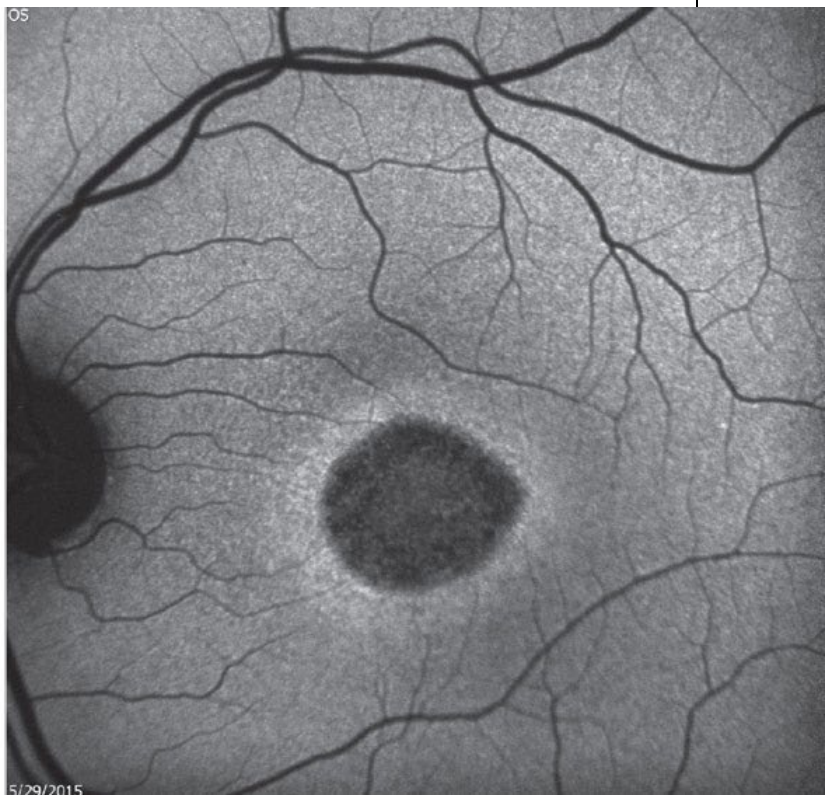
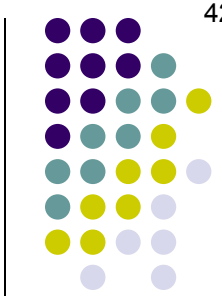




Stargardt—hyper/hypofluorescence on FAF



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Briefly, what is a pattern dystrophy*?*

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Briefly, what is a pattern dystrophy*?*
An inherited macular dystrophy that has a characteristic appearance (ie, a particular 'pattern')

Hereditary Retinal/Choroidal Dystrophies

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An inherited macular dystrophy that has a characteristic appearance (ie, a particular 'pattern')

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--***?***
--***?***
--***?***
--***?***

With apologies, the mnemonic is…

Hereditary Retinal/Choroidal Dystrophies

- Diffuse
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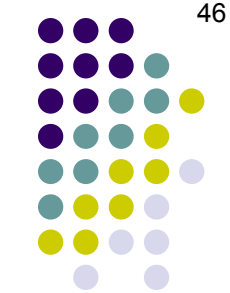

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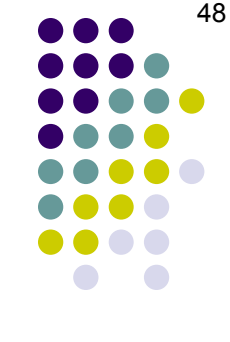
Are pattern dystrophies associated with severe vision loss?



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Generally no—vision is only mildly affected

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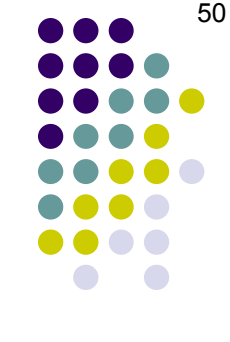

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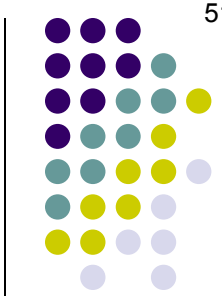
Generally no—they usually show up in middle adulthood



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The *Retina* book says essentially nothing about butterfly dystrophy, but does show a couple of pics

Are pattern dystrophies associated with severe vision loss?
Generally no—vision is only slightly affected

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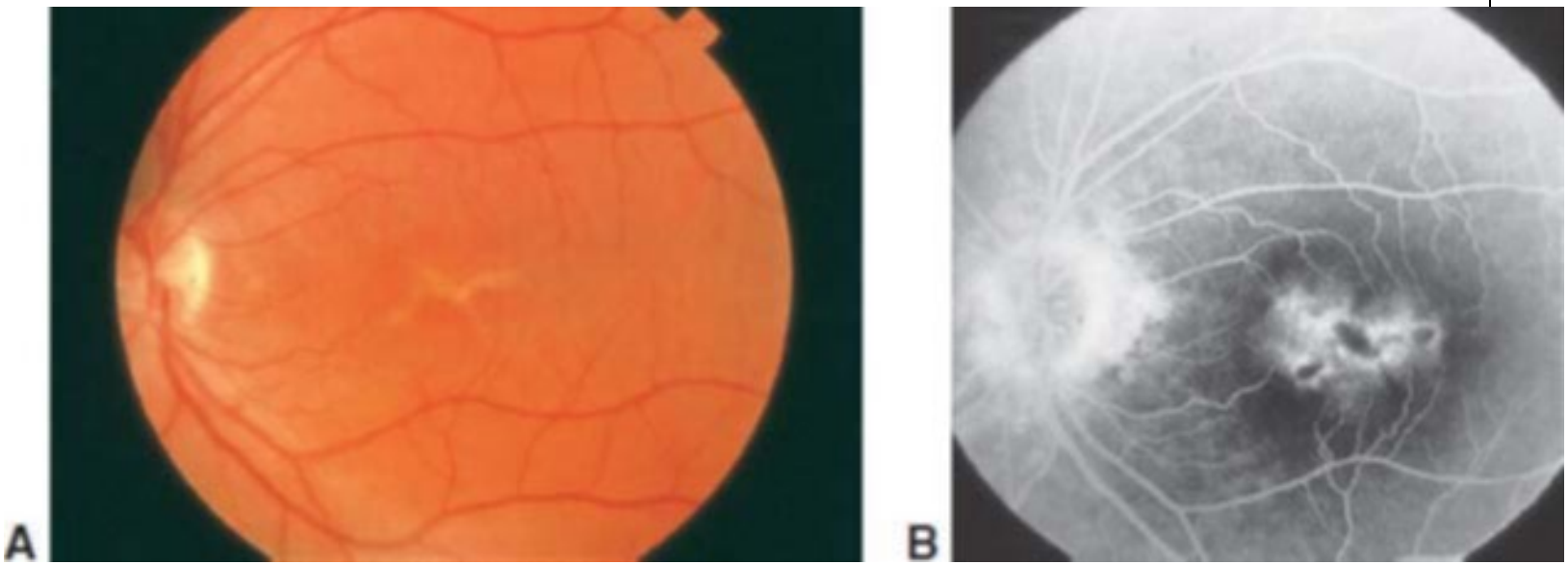
Generally no—they usually show up in middle adulthood



A, Fundus photo from a 56-year-old woman shows a typical yellow macular pigment pattern. B, FA shows blocked fluorescence of the pigment lesion itself and a rim of hyperfluorescence from surrounding RPE atrophy

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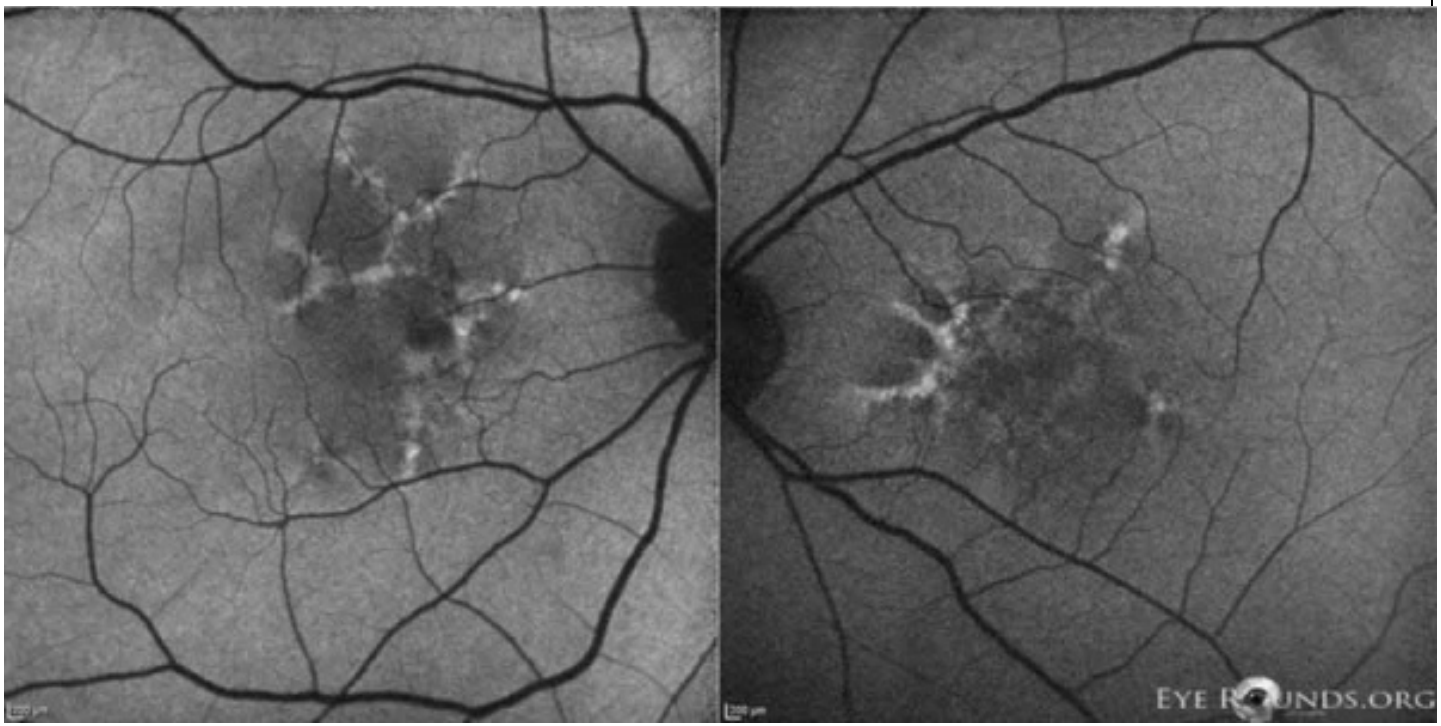

FAF demonstrates a butterfly-shaped pattern in the macula OU. Hypofluorescent areas corresponded to pigment clumping seen on fundoscopy.

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Yes, although they tend to be somewhat smaller vs larger

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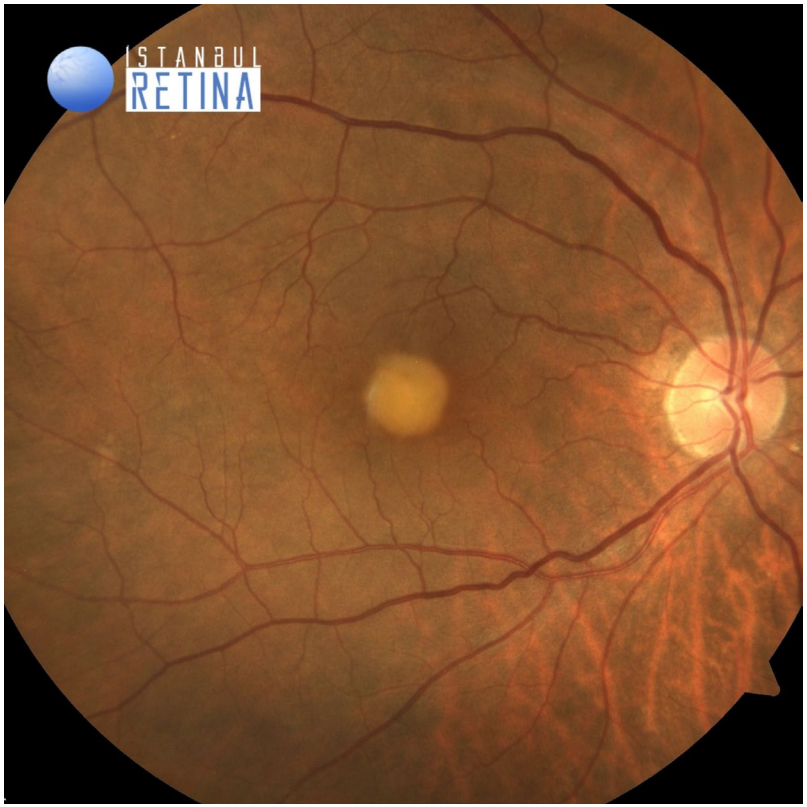
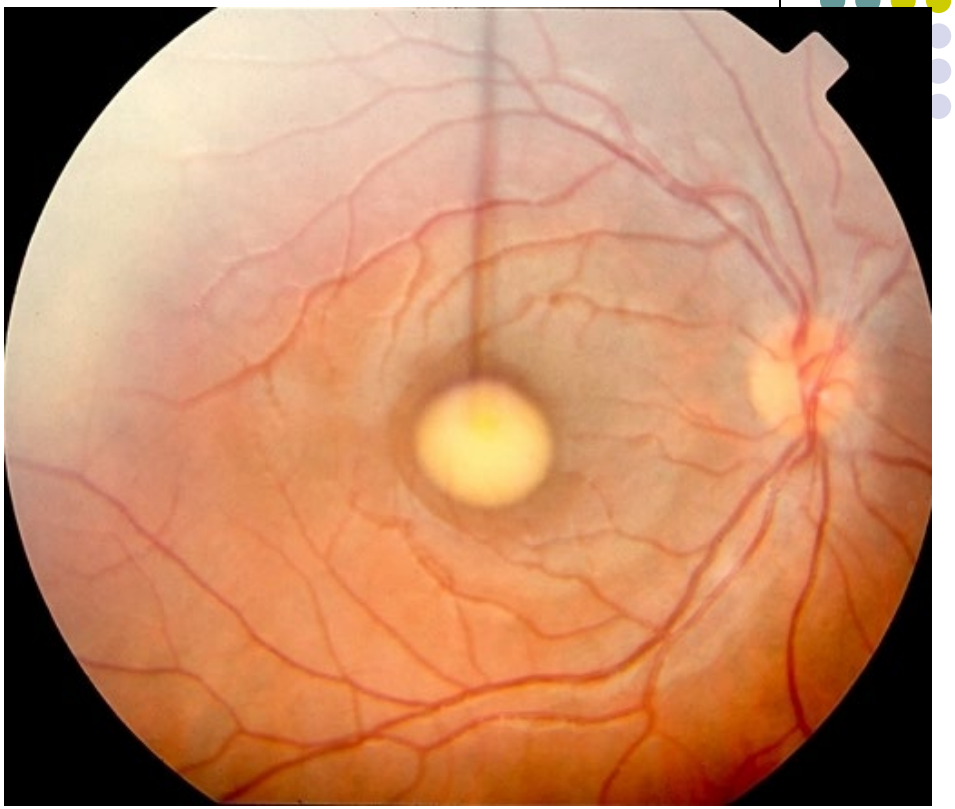


Typical vitelliform lesion of AOFVD



Best dz lesion for comparison

(Compare each lesion to the size of its corresponding ONH)

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Do Best dz and AOFVD share a common genetic heritage?

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You'd think so, but no—they're genetically unrelated

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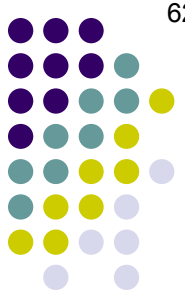
You'd think so, but no—they're genetically unrelated

Is the EOG abnormal in AOFVD, like it is in Best dz?
Again, you'd think so, but no—EOG is normal in AOFVD



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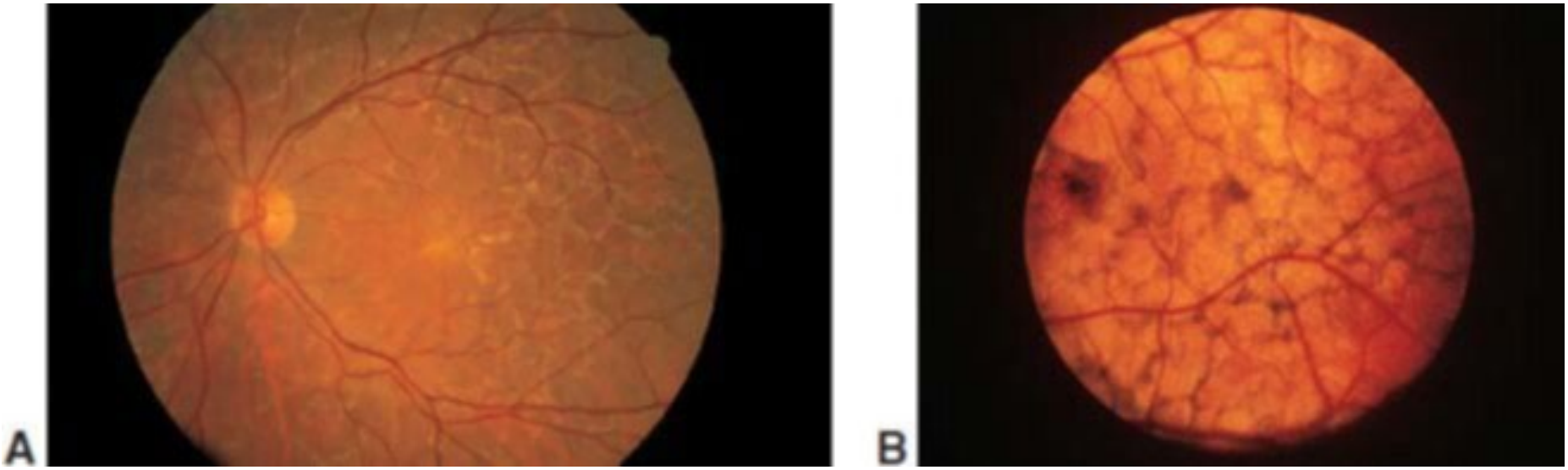
Likewise, the *Retina* book says essentially nothing about reticular dystrophy or fundus pulverulentus (but does show a couple of pics of reticular)

Do the macular 'patterns' appear early in life?
Generally no—they usually show up in middle adulthood



Fundus photos show 2 examples of reticular-type pattern dystrophy, characterized by a “fishnet” pattern of yellowish-orange (A) or brown (B) pigment deposition in the posterior fundus

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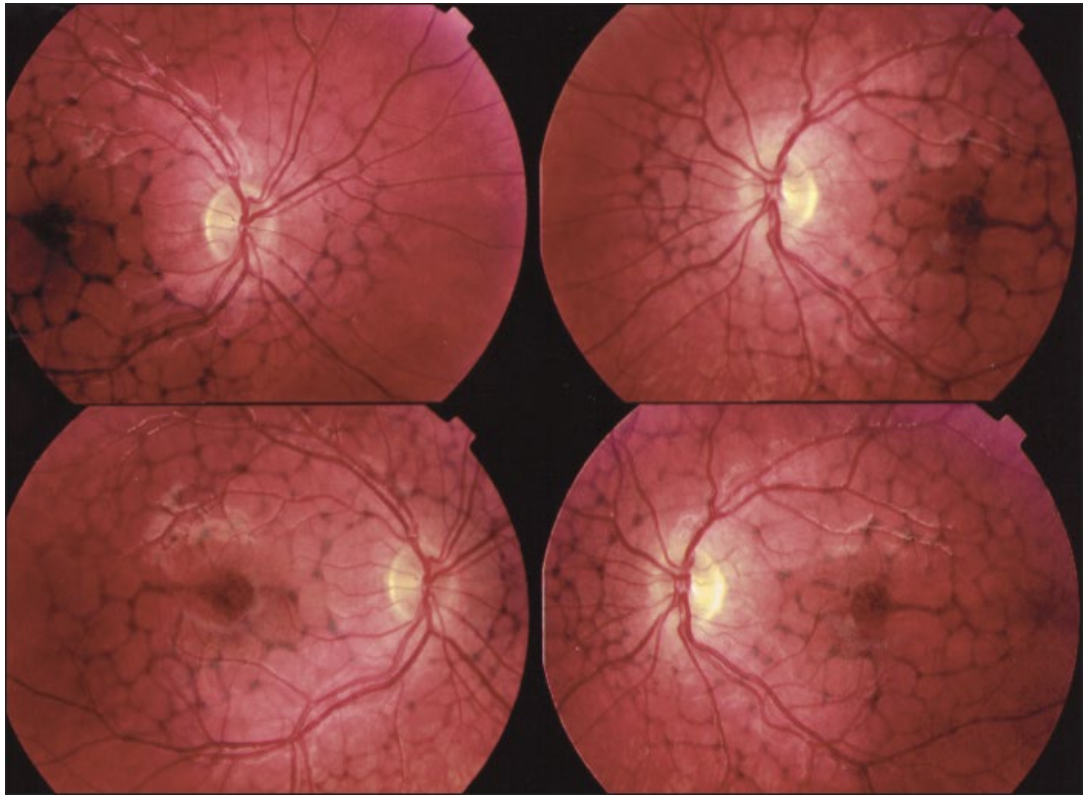

Symmetrical and reticular pattern of accumulated pigment in the fovea that radiates from the center to the periphery like a “fishnet with knots.”

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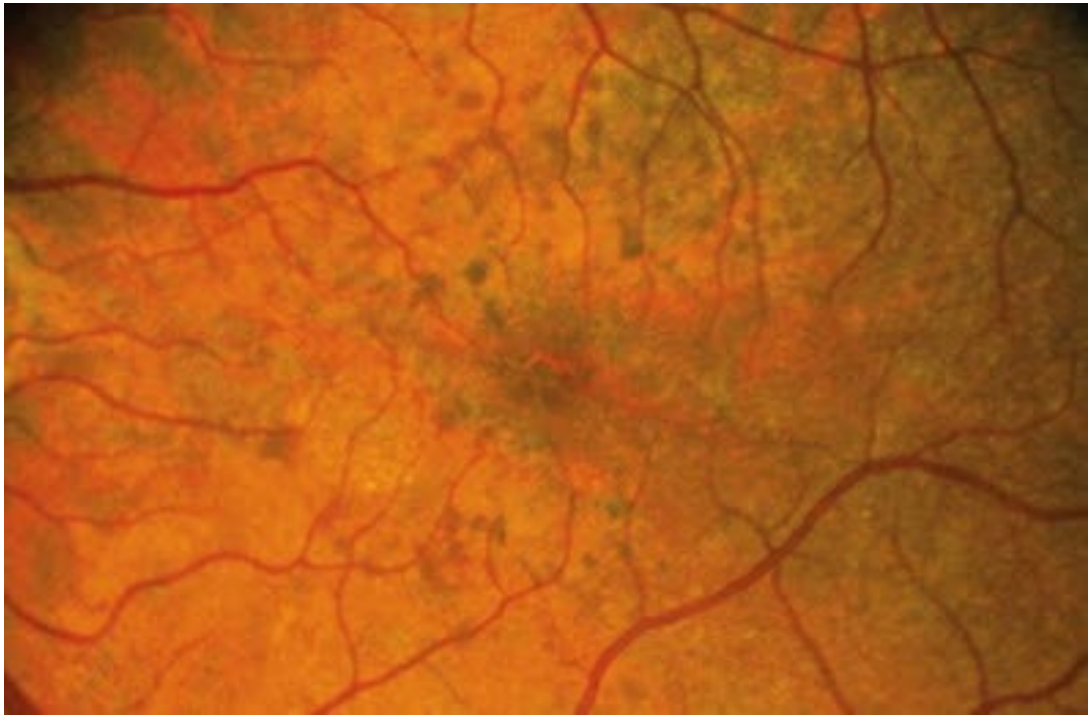

A pic of fundus pulverulentus, just because

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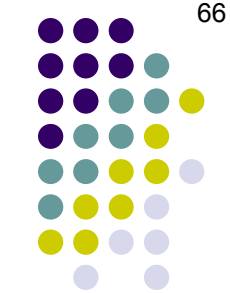

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How does central areolar choroidal dystrophy *present?*

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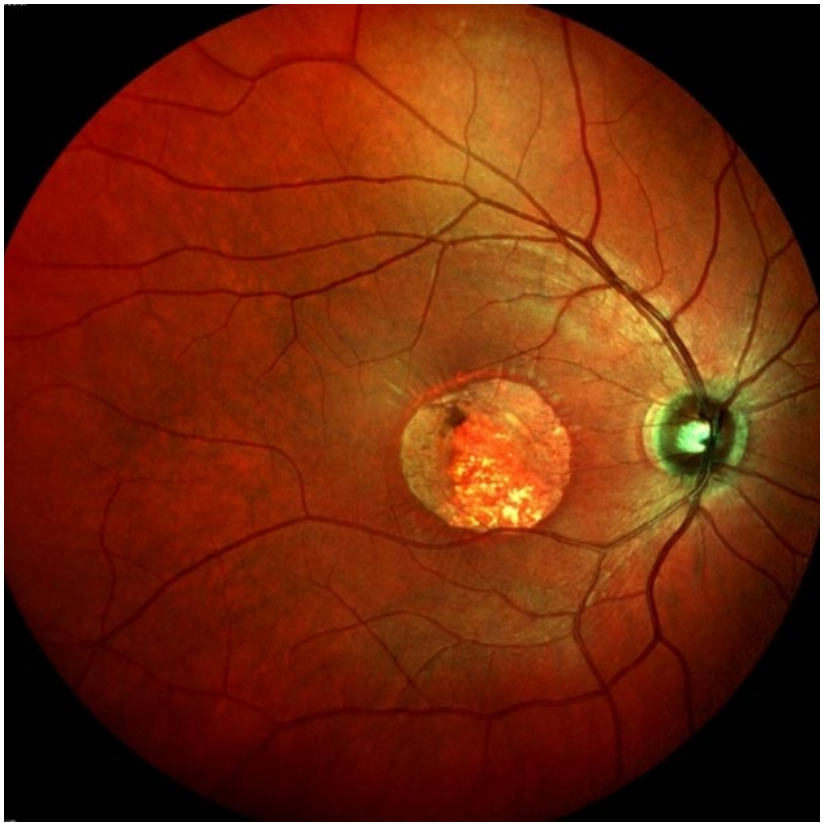
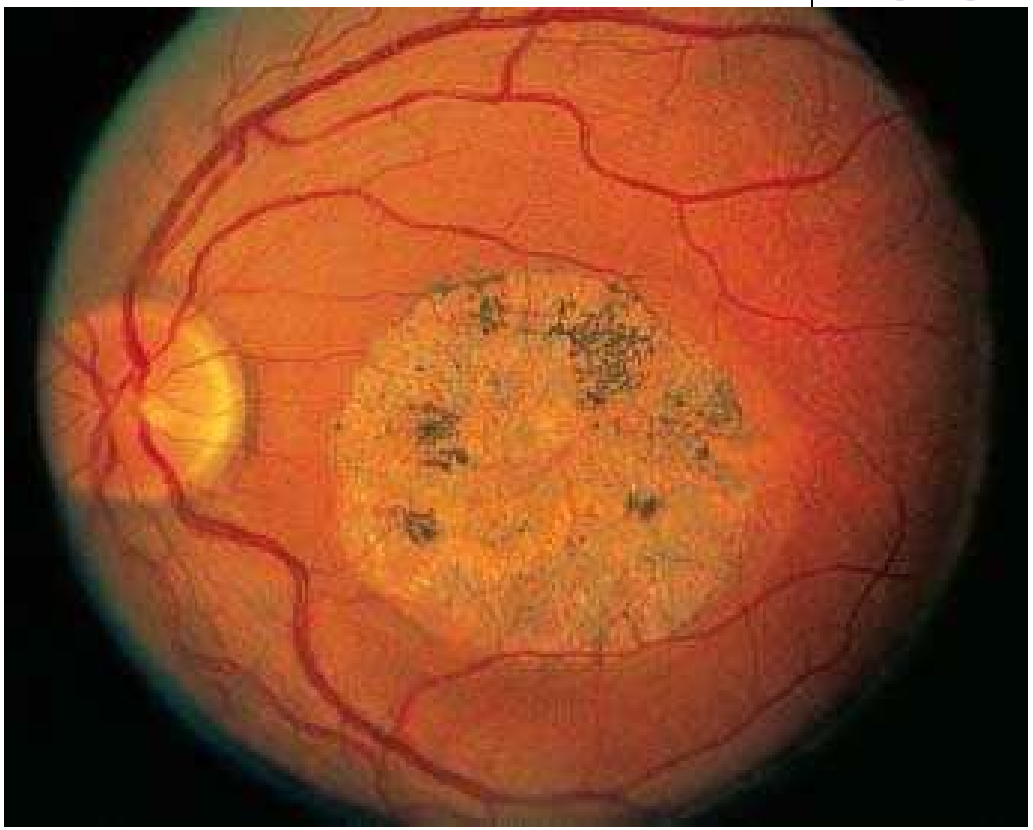
How does central areolar choroidal dystrophy *present?*
With well-circumscribed, bilateral and symmetric areas of choroidal and retinal atrophy





Central areolar choroidal dystrophy (two different pts)

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How does central areolar choroidal dystrophy present?

With **well-circumscribed, bilateral and symmetric areas of choroidal and retinal atrophy**

'Well-circumscribed, bilateral and symmetric areas of choroidal and retinal atrophy'—that sounds like geographic atrophy (GA), one of the advanced forms of ARMD. The pics look like it too.

No question—proceed when ready

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it is, sort of. What we now call *geographic atrophy* was formerly known as *central areolar atrophy*.

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Is it common, or rare?

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Quite rare

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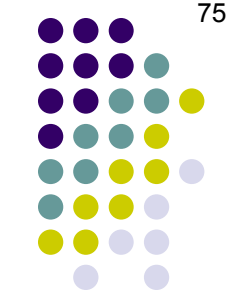
At what age are the changes first discernable?



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With well-circumscribed, bilateral and symmetric areas of choroidal and retinal atrophy

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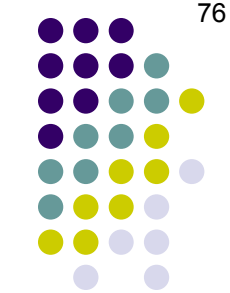
Quite rare

At what age are the changes first discernable?
Between 20 and 40 or so



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Is the visual prognosis good, like in the pattern dystrophies?

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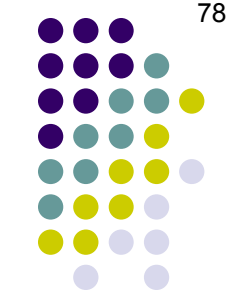
Unfortunately no—much like GA, pts experience severe vision loss, often as early as in their 40s



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At what
Betweer

So if you see a pt 50+ with bilateral lesions of this sort, how do you determine whether it's GA 2ndry to ARMD vs central areolar choroidal dystrophy (CACD)?
--

--

--

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--Age of onset of visual deterioration (it will be much earlier in [redacted])
--

--

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So if you see a pt 50+ with bilateral lesions of this sort, how do you determine whether it's GA 2ndry to ARMD vs central areolar choroidal dystrophy (CACD)?
--Age of onset of visual deterioration (it will be much earlier in CACD)
--

--

Is the visual prognosis good, like in the pattern dystrophies?
Unfortunately no—much like GA, pts experience severe vision loss, often as early as in their 40s

Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular
- Sorsby macular dystrophy
- Best dz
- Stargardt
- Pattern dystrophies
- **Central areolar choroidal dystrophy**
- North Carolina macular dystrophy

How does central areolar choroidal dystrophy *present?*
With well-circumscribed, bilateral and symmetric areas of choroidal and retinal atrophy

Is it com
Quite ra

At what
Betweer

So if you see a pt 50+ with bilateral lesions of this sort, how do you determine whether it's GA 2ndry to ARMD vs central areolar choroidal dystrophy (CACD)?
--Age of onset of visual deterioration (it will be much earlier in CACD)
--Family hx (will be stronger v weaker in pts with CACD because it is inherited in fashion)
--

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Hereditary Retinal/Choroidal Dystrophies

- Diffuse
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  - Best dz
  - Stargardt
  - Pattern dystrophies
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--Family hx (will be stronger in pts with CACD because it is inherited in AD fashion)
--

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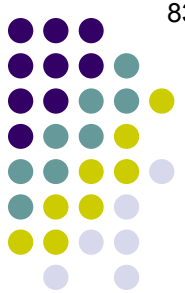

Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular

- Sorsby macular dystrophy
- Best dz
- Stargardt
- Pattern dystrophies
- **Central areolar choroidal dystrophy**
- North Carolina macular dystrophy

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With well-circumscribed, bilateral and symmetric areas of choroidal and retinal atrophy

Is it com
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At what
Betweer

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--Age of onset of visual deterioration (it will be much earlier in CACD)
--Family hx (will be stronger in pts with CACD because it is inherited in AD fashion)
--The lack of [redacted] in the macula of CACD pts

Is the visual prognosis good, like in the pattern dystrophies?
Unfortunately no—much like GA, pts experience severe vision loss, often as early as in their 40s

Hereditary Retinal/Choroidal Dystrophies

- Diffuse
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - **Central areolar choroidal dystrophy**
  - North Carolina macular dystrophy

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With well-circumscribed, bilateral and symmetric areas of choroidal and retinal atrophy

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At what
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So if you see a pt 50+ with bilateral lesions of this sort, how do you determine whether it's GA 2ndry to ARMD vs central areolar choroidal dystrophy (CACD)?
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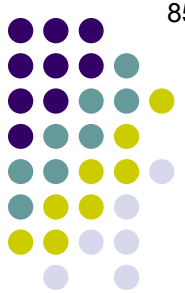

Hereditary Retinal/Choroidal Dystrophies

- Diffuse
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy
  - **North Carolina macular dystrophy**

North Carolina macular dystrophy is called (by the Retina *book) an "atypical" macular dystrophy. What makes it atypical?*

Hereditary Retinal/Choroidal Dystrophies

- Diffuse
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy
  - **North Carolina macular dystrophy**

North Carolina macular dystrophy is called (by the Retina *book) an "atypical" macular dystrophy. What makes it atypical?*
For one thing, it's nonprogressive

Hereditary Retinal/Choroidal Dystrophies

- Diffuse
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy
  - **North Carolina macular dystrophy**

North Carolina macular dystrophy is called (by the Retina *book) an "atypical" macular dystrophy. What makes it atypical?*
For one thing, it's nonprogressive. In fact, the *Retina* book equivocates over whether it's a 'dystrophy' at all, referring to it as a "stationary maldevelopment of the macula."

Hereditary Retinal/Choroidal Dystrophies

- Diffuse
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy
  - **North Carolina macular dystrophy**

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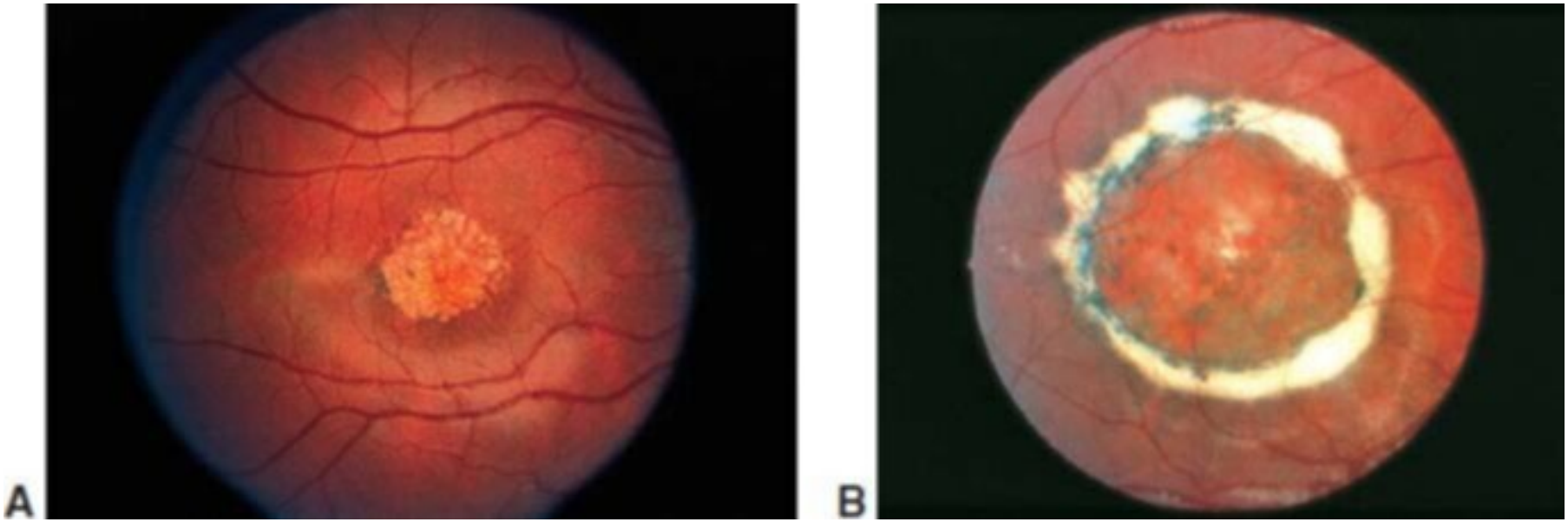
What does NCMD look like?

Hereditary Retinal/Choroidal Dystrophies

- Diffuse
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy
  - **North Carolina macular dystrophy**

North Carolina macular dystrophy is called (by the Retina *book) an "atypical" macular dystrophy. What makes it atypical?*
For one thing, it's nonprogressive. In fact, the *Retina* book equivocates over whether it's a 'dystrophy' at all, referring to it as a "stationary maldevelopment of the macula."

What does NCMD look like?
It has a great many appearances, and thus can't easily be described (for what it's worth, the pics used in the *Retina* book are on the next slide)



Clinical variations in North Carolina macular dystrophy.
A, Fundus of a 7-year-old patient with a cluster of peculiar yellowish-white atrophic lesions in the macula.
B, Example of a severe, almost colobomatous, macular defect.

North Carolina macular dystrophy

Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular
- Sorsby macular dystrophy
- Best dz
- Stargardt
- Pattern dystrophies
- Central areolar choroidal dystrophy
- **North Carolina macular dystrophy**

North Carolina macular dystrophy is called (by the Retina *book) an "atypical" macular dystrophy. What makes it atypical?*
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What does NCMD look like?
It has a great many appearances, and thus can't easily be described (for what it's worth, the pics used in the *Retina* book are on the next slide)

Do NCMD pts tend to have good vision, or poor?

Hereditary Retinal/Choroidal Dystrophies

- Diffuse
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy
  - **North Carolina macular dystrophy**

North Carolina macular dystrophy is called (by the Retina *book) an "atypical" macular dystrophy. What makes it atypical?*
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What does NCMD look like?
It has a great many appearances, and thus can't easily be described (for what it's worth, the pics used in the *Retina* book are on the next slide)

Do NCMD pts tend to have good vision, or poor?
Good, often surprisingly so given the appearance of their macula

Hereditary Retinal/Choroidal Dystrophies

- Diffuse
  - ?
  - ?
  - ?
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy
  - North Carolina macular dystrophy

There are three subtypes of diffuse dystrophies—what are they?

Hereditary Retinal/Choroidal Dystrophies

- Diffuse
  - Rod-dominant
  - Cone-dominant
  - Choroidal
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy
  - North Carolina macular dystrophy

There are three subtypes of diffuse dystrophies—what are they?

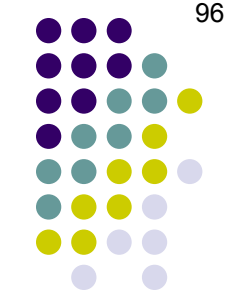
Hereditary Retinal/Choroidal Dystrophies

- **Diffuse**
  - **Rod-dominant**
  - **Choroidal**
  - **Cone-dominant**
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy
  - North Carolina macular dystrophy

There are three subtypes of diffuse dystrophies—what are they?

When considering the diffuse dystrophies, note that they are broken down into three natural groupings—*rod-dominant, cone-dominant,* and *choroidal*

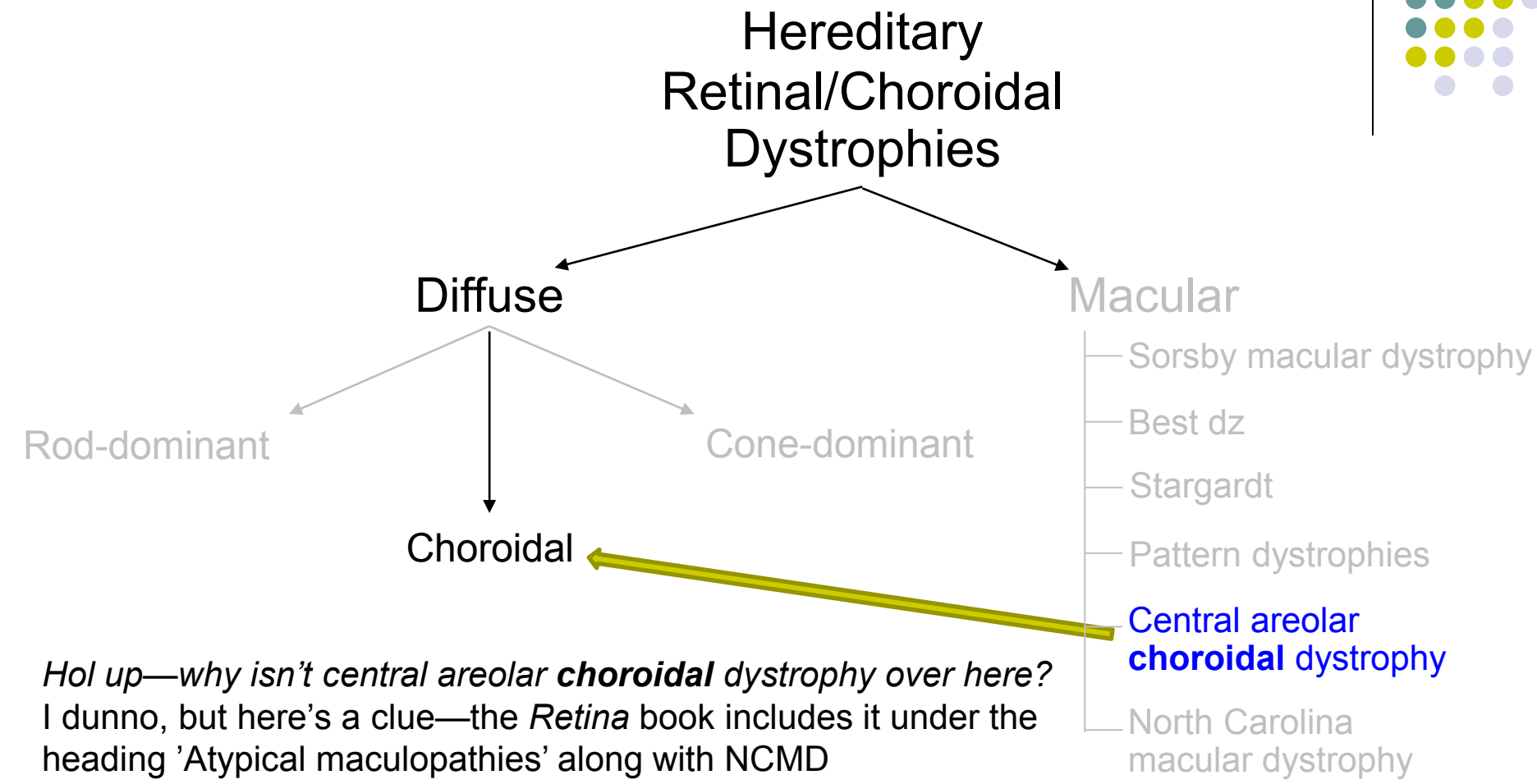
No question—proceed when ready



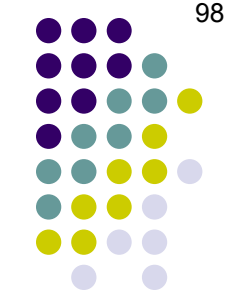
Hereditary Retinal/Choroidal Dystrophies

- Diffuse
  - Rod-dominant
  - Cone-dominant
  - Choroidal
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar **choroidal** dystrophy
  - North Carolina macular dystrophy

*Hol up—why isn't central areolar **choroidal** dystrophy over here?*



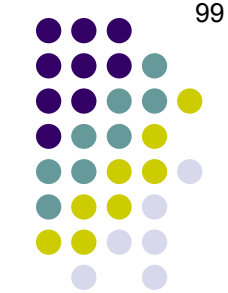
*Hol up—why isn't central areolar **choroidal** dystrophy over here?*
I dunno, but here's a clue—the *Retina* book includes it under the heading 'Atypical maculopathies' along with NCMD



Hereditary Retinal/Choroidal Dystrophies

- Diffuse
  - Rod-dominant
    - *?*
  - Choroidal
  - Cone-dominant
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy
  - North Carolina macular dystrophy

What is the catch-all name for the rod-dominant dystrophies?



Hereditary Retinal/Choroidal Dystrophies

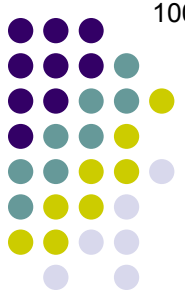
Diffuse

- Rod-dominant
  - Rod-cone
- Choroidal
- Cone-dominant

Macular

- Sorsby macular dystrophy
- Best dz
- Stargardt
- Pattern dystrophies
- Central areolar choroidal dystrophy
- North Carolina macular dystrophy

What is the catch-all name for the rod-dominant dystrophies?
The ‘rod-cone dystrophies’

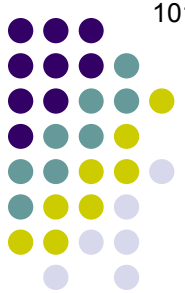


Hereditary Retinal/Choroidal Dystrophies

- Diffuse
  - Rod-dominant
    - Rod-cone abb.
  - Cone-dominant
  - Choroidal
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy
  - North Carolina macular dystrophy

What is the catch-all name for the rod-dominant dystrophies?
The 'rod-cone dystrophies'

And what is the catch-all name for the rod-cone dystrophies?

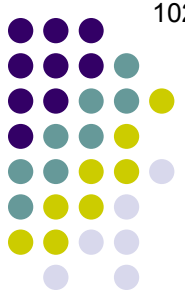


Hereditary Retinal/Choroidal Dystrophies

- Diffuse
  - Rod-dominant
    - Rod-cone (RP)
  - Choroidal
  - Cone-dominant
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy
  - North Carolina macular dystrophy

What is the catch-all name for the rod-dominant dystrophies?
The 'rod-cone dystrophies'

And what is the catch-all name for the rod-cone dystrophies?
Retinitis pigmentosa (RP)



Hereditary Retinal/Choroidal Dystrophies

- Diffuse
  - Rod-dominant
    - Rod-cone (RP)
  - Choroidal
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Pattern dystrophies
  - Central areolar choroidal dystrophy
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***For more on RP, see slide-set* R38**

What is the catch-all name for the rod-dominant dystrophies?
The 'rod-cone dystrophies'

And what is the catch-all name for the rod-cone dystrophies?
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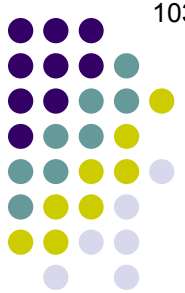
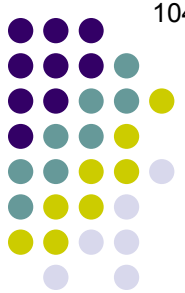


Hereditary Retinal/Choroidal Dystrophies

- Diffuse
  - Rod-dominant
    - Rod-cone (RP)
  - Choroidal
  - Cone-dominant
    - ?
    - ?
    - ?
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy
  - North Carolina macular dystrophy

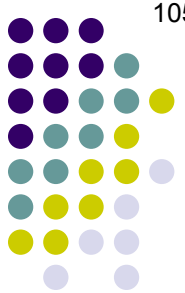
What are the three cone-dominant conditions highlighted in the Retina *book?*



Hereditary Retinal/Choroidal Dystrophies

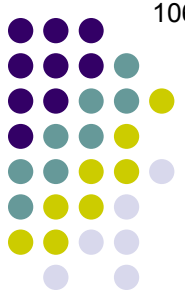
- Diffuse
  - Rod-dominant
    - Rod-cone (RP)
  - Choroidal
  - Cone-dominant
    - Cone/cone-rod
    - LCA
    - Enhanced *S*-cone dz
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy
  - North Carolina macular dystrophy

What are the three cone-dominant conditions highlighted in the Retina *book?*



Hereditary Retinal/Choroidal Dystrophies

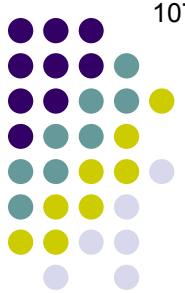
- Diffuse
  - Rod-dominant
    - **Rod-cone (RP)**
    - **?**
  - *Type of 'blindness' produced*
  - Cone-dominant
    - **Cone/cone-rod**
    - **?**
    - Enhanced *S*-cone dz
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy
  - North Carolina macular dystrophy



Hereditary Retinal/Choroidal Dystrophies

- Diffuse
  - Rod-dominant
    - **Rod-cone (RP)**
  - Cone-dominant
    - **Cone/cone-rod**
    - Enhanced *S*-cone dz
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy
  - North Carolina macular dystrophy

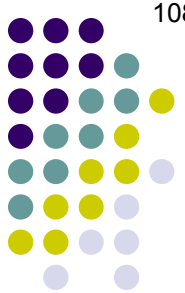
Nyctalopia	*Type of 'blindness' produced*	Hemeralopia



Hereditary Retinal/Choroidal Dystrophies

- Diffuse
  - Rod-dominant
    - **Rod-cone (RP)**
  - Cone-dominant
    - **Cone/cone-rod**
  - **Nyctalopia**
  - *Type of 'blindness' produced*
  - **Hemeralopia**
  - Enhanced -cone dz
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy
  - North Carolina macular dystrophy

What is…
--nyctalopia?
--hemeralopia?

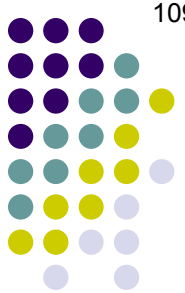


Hereditary Retinal/Choroidal Dystrophies

- Diffuse
  - Rod-dominant
    - **Rod-cone (RP)**
  - Cone-dominant
    - **Cone/cone-rod**
    - Enhanced S-cone dz
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy
  - North Carolina macular dystrophy

Nyctalopia	*Type of 'blindness' produced*	**Hemeralopia**

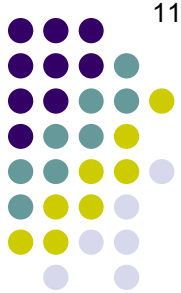
What is…
--nyctalopia? Night blindness
--hemeralopia? Day blindness



Hereditary Retinal/Choroidal Dystrophies

- Diffuse
  - Rod-dominant
    - **Rod-cone (RP)**
  - Cone-dominant
    - **Cone/cone-rod**
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy
  - North Carolina macular dystrophy

Nyctalopia	*Type of 'blindness' produced*	Hemeralopia
?	*Chief visual defect(s)*	*?*



Hereditary Retinal/Choroidal Dystrophies

- Diffuse
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    - **Rod-cone (RP)**
  - Cone-dominant
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- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
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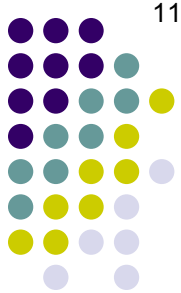
  - North Carolina macular dystrophy

Nyctalopia	*Type of 'blindness' produced*	Hemeralopia
VF loss	*Chief visual defect(s)*	Acuity, and color loss



Hereditary Retinal/Choroidal Dystrophies

Diffuse

Rod-dominant

- **Rod-cone (RP)**

Cone-dominant

- **Cone/cone-rod**

Nyctalopia	*Type of 'blindness' produced*	Hemeralopia
VF loss	*Chief visual defect(s)*	Acuity, and color loss

This makes sense, as rods dominate the retinal periphery

Macular

- Sorsby macular dystrophy
- Best dz
- Stargardt
- Pattern dystrophies
- Central areolar choroidal dystrophy
- North Carolina macular dystrophy

No question—proceed when ready

Hereditary Retinal/Choroidal Dystrophies

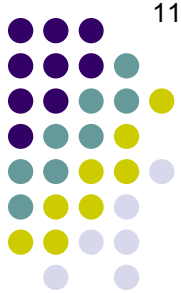
- Diffuse
  - Rod-dominant
    - **Rod-cone (RP)**
  - Cone-dominant
    - **Cone/cone-rod**
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy
  - North Carolina macular dystrophy

Nyctalopia	*Type of 'blindness' produced*	Hemeralopia
VF loss	*Chief visual defect(s)*	**Acuity, and color loss**

This makes sense, as rods dominate the retinal periphery

This makes sense too, as cones dominate the foveal center (and are responsible for color perception)

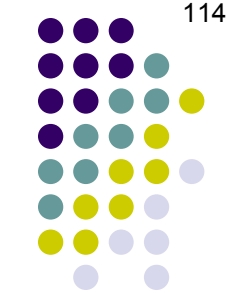
No question—proceed when ready



Hereditary Retinal/Choroidal Dystrophies

- Diffuse
  - Rod-dominant
    - **Rod-cone (RP)**
  - Cone-dominant
    - **Cone/cone-rod**
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy
  - North Carolina macular dystrophy

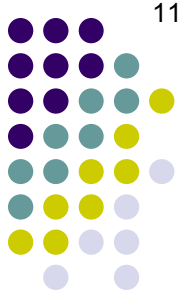
Nyctalopia	*Type of 'blindness' produced*	Hemeralopia
VF loss	*Chief visual defect(s)*	Acuity, and color loss
?	*c/o photophobia?*	***?***



Hereditary Retinal/Choroidal Dystrophies

- Diffuse
  - Rod-dominant
    - **Rod-cone (RP)**
  - Cone-dominant
    - **Cone/cone-rod**
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy
  - North Carolina macular dystrophy

Nyctalopia	*Type of 'blindness' produced*	Hemeralopia
VF loss	*Chief visual defect(s)*	Acuity, and color loss
No	*c/o photophobia?*	Yes



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  - North Carolina macular dystrophy

Nyctalopia	*Type of 'blindness' produced*	Hemeralopia
VF loss	*Chief visual defect(s)*	Acuity, and color loss
No	*c/o photophobia?*	**Yes**

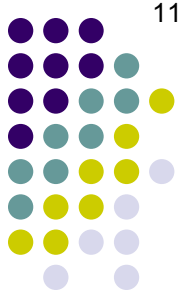
If all you have are rods, normal light levels will seem unbearably bright

No question—proceed when ready

Hereditary Retinal/Choroidal Dystrophies

- Diffuse
  - Rod-dominant
    - Rod-cone (RP)
  - Choroidal
  - Cone-dominant
    - Cone/cone-rod
    - LCA
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - ar
    strophy
  - na
    rophy

What does LCA *stand for in this context?*



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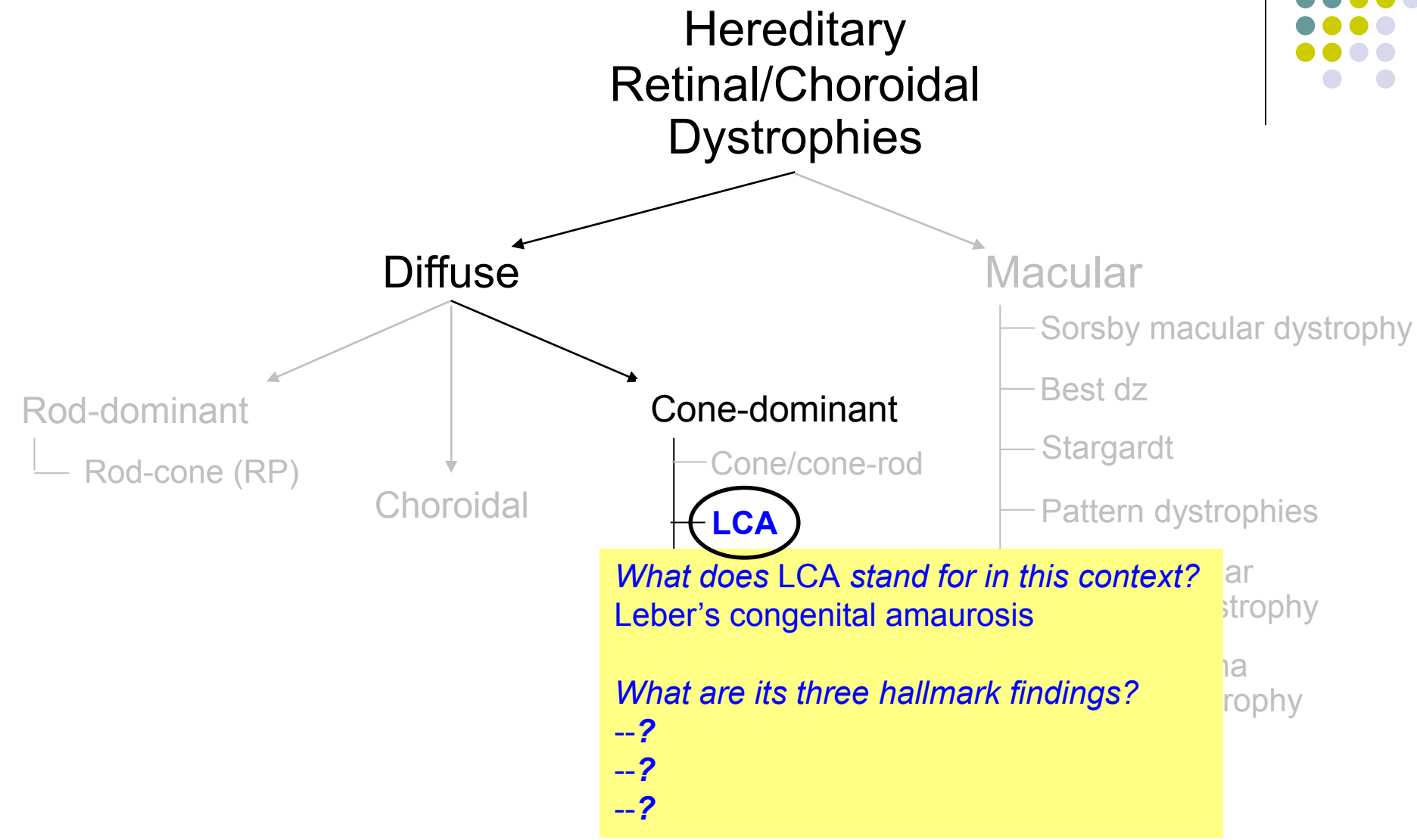
    rophy

What does LCA *stand for in this context?*
Leber's congenital amaurosis


Hereditary Retinal/Choroidal Dystrophies
Diffuse
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Sorsby macular dystrophy
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What does LCA stand for in this context?
Leber's congenital amaurosis
What are its three hallmark findings?
--?
--?
--?

Hereditary Retinal/Choroidal Dystrophies

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  - na
  - rophy

What does LCA *stand for in this context?*
Leber's congenital amaurosis

What are its three hallmark findings?
--Profoundly poor VA from birth
--*?*
--*?*

Hereditary Retinal/Choroidal Dystrophies

- Diffuse
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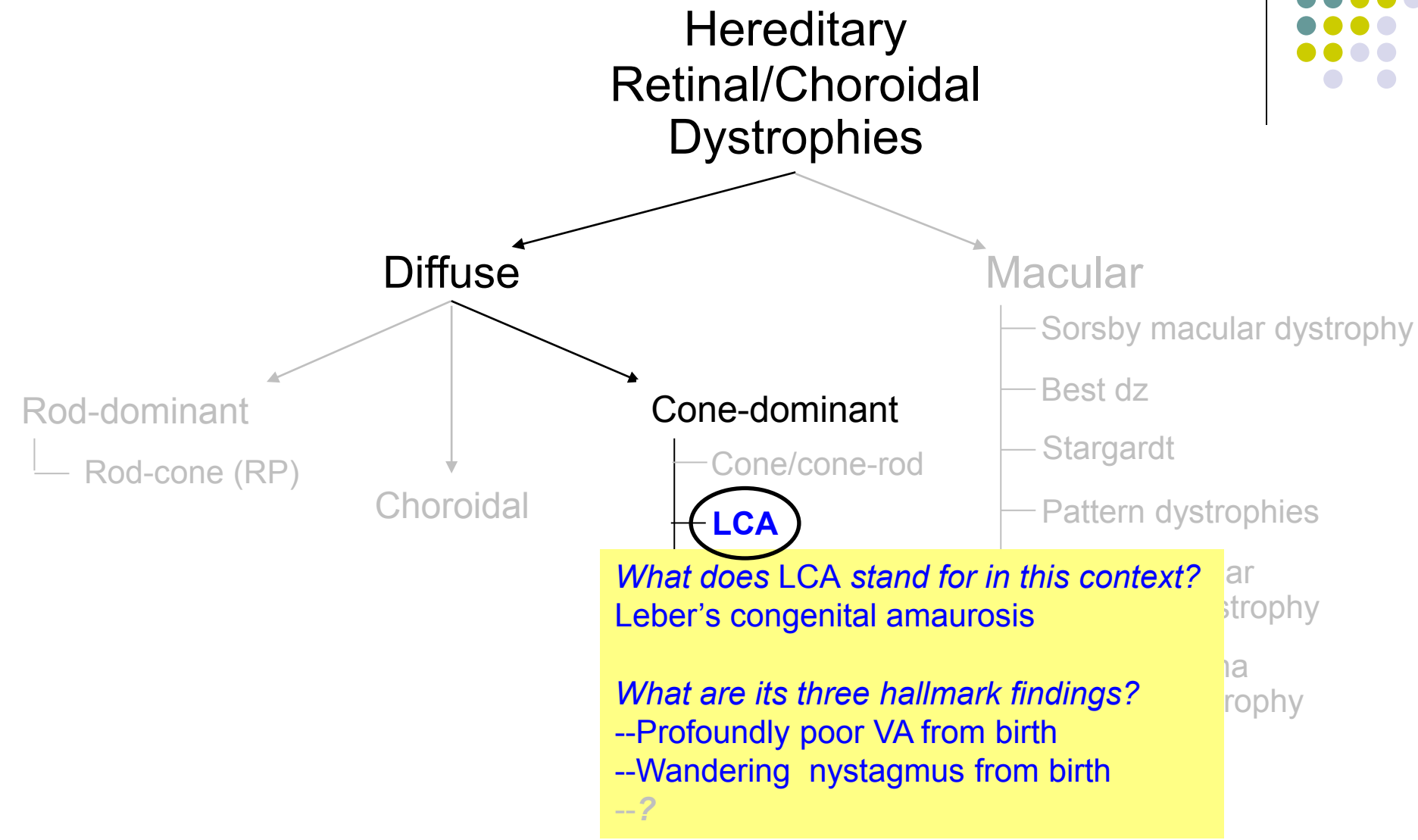
What does LCA *stand for in this context?*
Leber's congenital amaurosis

What are its three hallmark findings?
--Profoundly poor VA from birth
-- pattern nystagmus from birth
--*?*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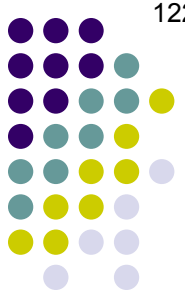
--Wandering nystagmus from birth
--?



Hereditary Retinal/Choroidal Dystrophies

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  - Cone-dominant
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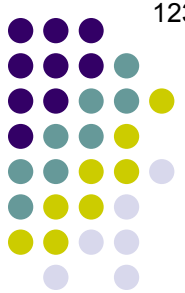
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - ar
  - trophy
  - na
  - rophy

What does LCA *stand for in this context?*
Leber's congenital amaurosis

What are its three hallmark findings?
--Profoundly poor VA from birth
--Wandering nystagmus from birth
--Pts engage in [two words] behavior



Hereditary Retinal/Choroidal Dystrophies

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What does LCA *stand for in this context?*
Leber's congenital amaurosis

What are its three hallmark findings?
--Profoundly poor VA from birth
--Wandering nystagmus from birth
--Pts engage in eye rubbing behavior

Hereditary Retinal/Choroidal Dystrophies

- Diffuse
  - Rod-dominant
    - Rod-cone (RP)
  - Choroidal
  - Cone-dominant
    - Cone/cone-rod
    - LCA
    - **Enhanced *S*-cone dz**
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy

By what eponymous name is enhanced *S*-cone dystrophy also *known?*

Hereditary Retinal/Choroidal Dystrophies

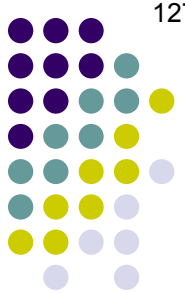
- Diffuse
  - Rod-dominant
    - Rod-cone (RP)
  - Choroidal
  - Cone-dominant
    - Cone/cone-rod
    - LCA
    - **Enhanced *S*-cone dz**
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy

By what eponymous name is enhanced *S*-cone dystrophy also *known?*
Goldmann-Favre syndrome

Hereditary Retinal/Choroidal Dystrophies

- Diffuse
  - Rod-dominant
    - Rod-cone (RP)
  - Choroidal
  - Cone-dominant
    - Cone/cone-rod
    - LCA
    - Enhanced *S*-cone dz
    - *Monochromatism*
    - *Dichromatism*
    - *Trichromatism*
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - Pattern dystrophies
  - Central areolar choroidal dystrophy
  - North Carolina macular dystrophy

Hol up—where's all those classic inherited color-blindness conditions…



Hereditary Retinal/Choroidal Dystrophies

- Diffuse
  - Rod-dominant
    - Rod-cone (RP)
    - *CSNB*
    - *Fundus albipunctatus*
    - *Oguchi dz*
  - Choroidal
  - Cone-dominant
    - Cone/cone-rod
    - LCA
    - Enhanced *S*-cone dz
    - *Monochromatism*
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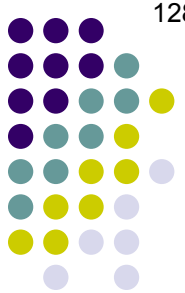
…and night-blindness conditions?

Hol up—where's all those classic inherited color-blindness conditions…



Hereditary Retinal/Choroidal Dystrophies

To be sure, these are hereditary diseases of the retina and/or choroid. However, they are **not** dystrophies—rather, they are *stationary* conditions, ie, they do not worsen over time. (One of them, *congenital stationary night blindness*, [CSNB], even has the word *stationary* in its name.)

Rod-dominant

- Rod-cone (RP
- *CSNB*
- *Fundus albipunctatus*
- *Oguchi dz*

…and night-blindness conditions?

- Enhanced *S*-cone dz
- *Monochromatism*
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- *Trichromatism*

- Central areolar choroidal dystrophy
- North Carolina macular dystrophy

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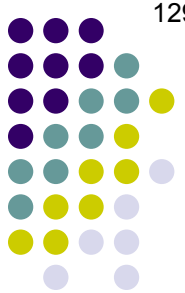

Hereditary Retinal/Choroidal Dz

Stationary Dz

Dystrophies

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Rod-dominant

Rod-cone (RP

CSNB

Fundus albipunctatus

Oguchi dz

…and night-blindness conditions?

Enhanced *S*-cone dz

Monochromatism

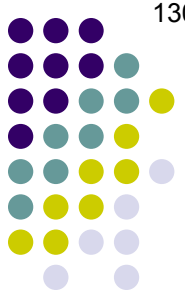
Dichromatism

Trichromatism

Central areolar choroidal dystrophy

North Carolina macular dystrophy

Hol up—where's all those classic inherited color-blindness conditions…



Hereditary Retinal/Choroidal Dz

Stationary Dz

Dystrophies

To be sure, these are hereditary diseases of the retina and/or choroid. However, they are **not** dystrophies—rather, they are *stationary* conditions, ie, they do not worsen over time. (One of them, *congenital stationary night*

For more on these conditions, see slide-set R9

conditions. It is on *this* branch that these conditions belong.

Rod-dc

Rod

CSNB

Fundus albipunctatus

Oguchi dz

…and night-blindness conditions?

Enhanced *S*-cone dz

Monochromatism

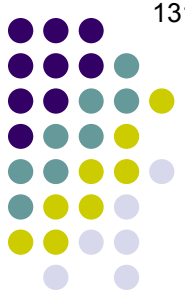
Dichromatism

Trichromatism

Central areolar choroidal dystrophy

North Carolina macular dystrophy

Hol up—where's all those classic inherited color-blindness conditions…



Hereditary Retinal/Choroidal Dz

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Rod-dominant

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Oguchi dz

…and night-blindness conditions?

(And no, I can't explain why the 'stationary maldevelopment of the macula' that is **NCMD** isn't listed over there with the other stationary conditions)

Monochromatism

?

North Carolina macular dystrophy

?

Dichromatism

Trichromatism

Hol up—where's all those classic inherited color-blindness conditions…

Hereditary Retinal/Choroidal Dystrophies

- Diffuse
  - Rod-dominant
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      - *Usher syndrome*
      - *Bardet-Biedl syndrome*
      - *Bassen-Kornzweig dz*
      - *Infantile Refum*
      - *(a number of others)*
    - Choroidal
  - Cone-dominant
    - Cone/cone-rod
    - LCA
    - Enhanced *S*-cone dz
- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
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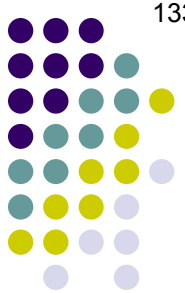
OK, but what about the RP-lookalike conditions? They're all inherited and progressive. Why aren't they listed here?



Hereditary Retinal/Choroidal Dystrophies

Again, these **are** hereditary diseases of the retina and/or choroid.
And unlike the 'stationary' conditions, these do progress.
However, they are **not** dystrophies—rather, they are *degenerations*.

strophy

Rod-dominant

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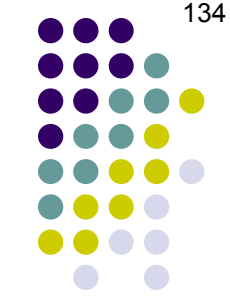

Hereditary Retinal/Choroidal Dz

Stationary Dz

Dystrophies

Degenerations

Again, these **are** hereditary diseases of the retina and/or choroid. And unlike the 'stationary' conditions, these do progress. However, they are **not** dystrophies—rather, they are *degenerations*. If we were to pull back focus again, we see that the *degenerations* form yet another separate branch on the organizational tree of 'hereditary retinal/choroidal conditions.'

strophy

Rod-dominant

- Rod-cone (RP)
- *Usher syndrome*
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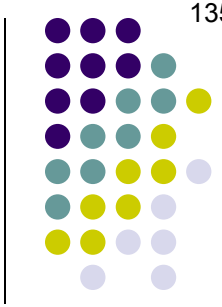

Hereditary Retinal/Choroidal Dz

Stationary Dz

Dystrophies

Degenerations

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strophy

Rod-d

Choroidal

***For more on these conditions, see slide-set* R5**

- ***Usher syndrome***
- ***Bardet-Biedl syndrome***
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- *(a number of others)*

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Pattern dystrophies

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OK, but what about the RP-lookalike *conditions? They're all inherited and progressive. Why aren't they listed here?*

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    - ?
    - ?
    - ?
  - Cone-dominant
    - Cone/cone-rod
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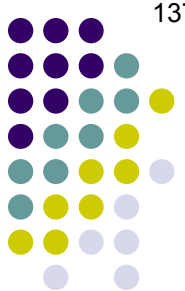
The Retina *book lists three choroidal dystrophies—what are they?*



Hereditary Retinal/Choroidal Dystrophies

- Diffuse
  - Rod-dominant
    - Rod-cone (RP)
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    - Bietti
    - Gyrate
    - Choroideremia
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The Retina *book lists three choroidal dystrophies—what are they?*

One word captures the dz process common to all—what is it?

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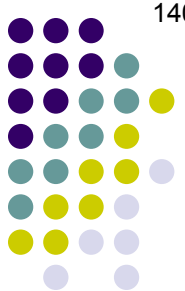
The Retina *book lists three choroidal dystrophies—what are they?*

One word captures the dz process common to all—what is it?
'Atrophy'



Hereditary Retinal/Choroidal Dystrophies

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    - Rod-cone (RP)
  - Choroidal
    - **Bietti**
    - **Gyrate**
    - **Choroideremia**
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  - Pattern dystrophies
  - Central areolar choroidal dystrophy
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The Retina *book lists three choroidal dystrophies—what are they?*

One word c
'Atrophy'

These conditions share a common feature on one retinal-imaging modality. What is the modality, and what is the feature?

Hereditary Retinal/Choroidal Dystrophies

- Diffuse
  - Rod-dominant
    - Rod-cone (RP)
  - Choroidal
    - **Bietti**
    - **Gyrate**
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The Retina *book lists three choroidal dystrophies—what are they?*

One word c

'Atrophy'

These conditions share a common feature on one retinal-imaging modality. What is the modality, and what is the feature?

The areas of atrophy are hyper- vs hypoautofluorescent on fundus autofluorescence (FAF)

Hereditary Retinal/Choroidal Dystrophies

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  - Rod-dominant
    - Rod-cone (RP)
  - Choroidal
    - **Bietti**
    - **Gyrate**
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The Retina *book lists three choroidal dystrophies—what are they?*

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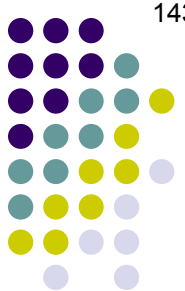
The areas of atrophy are hypoautofluorescent on fundus autofluorescence (FAF)



What is the full name of this condition?

Hereditary

macular dystrophy

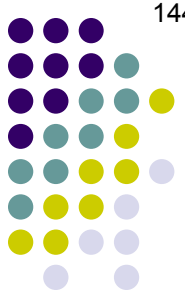
Rod-cone (RP)

Choroidal

- **Bietti**
- Gyrate
- Choroideremia

- LCA
- Enhanced *S*-cone dz

- Pattern dystrophies
- Central areolar choroidal dystrophy
- North Carolina macular dystrophy



What is the full name of this condition?
Bietti crystalline dystrophy

Hereditary

macular dystrophy

Choroidal

- **Bietti**
- Gyrate
- Choroideremia

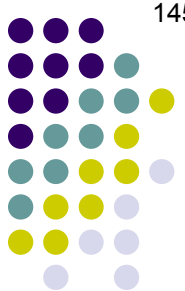
LCA

Enhanced *S*-cone dz

Pattern dystrophies

Central areolar choroidal dystrophy

North Carolina macular dystrophy



What is the full name of this condition?
Bietti **crystalline** dystrophy

Is it fair to guess that DFE reveals crystals of some sort?

Hereditary

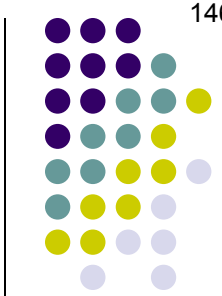
macular dystrophy

Choroidal

- **Bietti**
- Gyrate
- Choroideremia

- LCA
- Enhanced *S*-cone dz

- Pattern dystrophies
- Central areolar choroidal dystrophy
- North Carolina macular dystrophy



What is the full name of this condition?
Bietti **crystalline** dystrophy

Is it fair to guess that DFE reveals crystals of some sort?
Indeed it is—two colors crystals are present within the two words

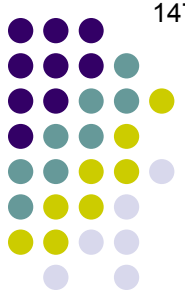
Hereditary

macular dystrophy

Choroidal
- **Bietti**
- Gyrate
- Choroideremia

- LCA
- Enhanced *S*-cone dz

- Pattern dystrophies
- Central areolar choroidal dystrophy
- North Carolina macular dystrophy



Hereditary

What is the full name of this condition?
Bietti **crystalline** dystrophy

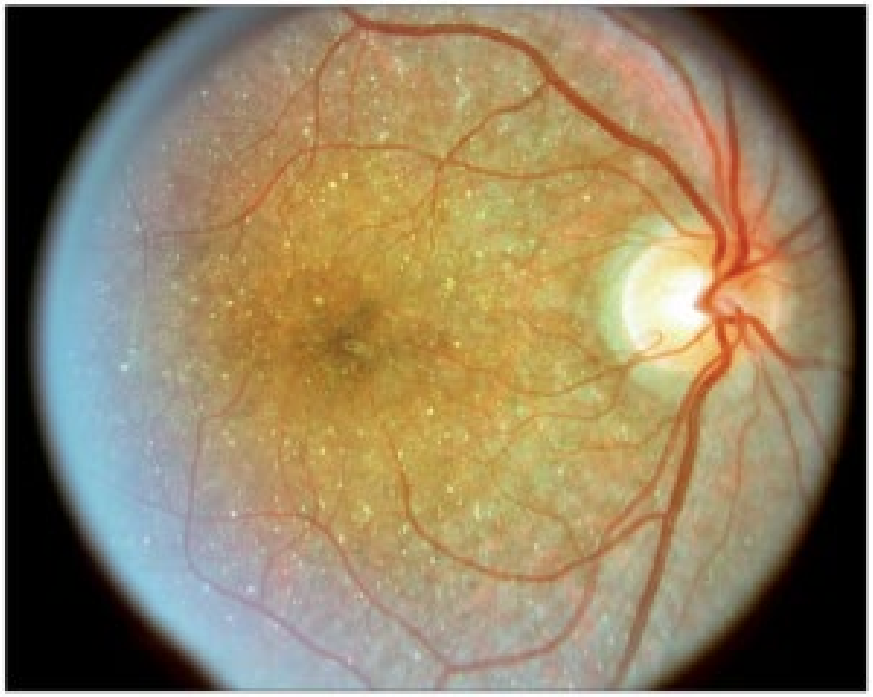
Is it fair to guess that DFE reveals crystals of some sort?
Indeed it is—**yellow**-**white** crystals are present within the posterior retina

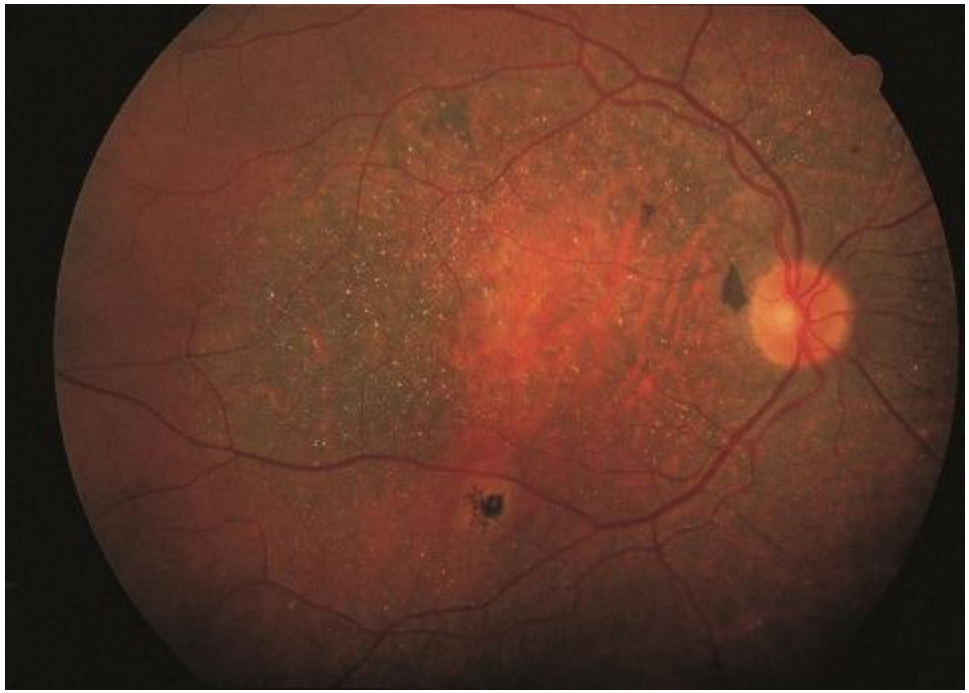
macular dystrophy

Choroidal
- **Bietti**
- Gyrate
- Choroideremia

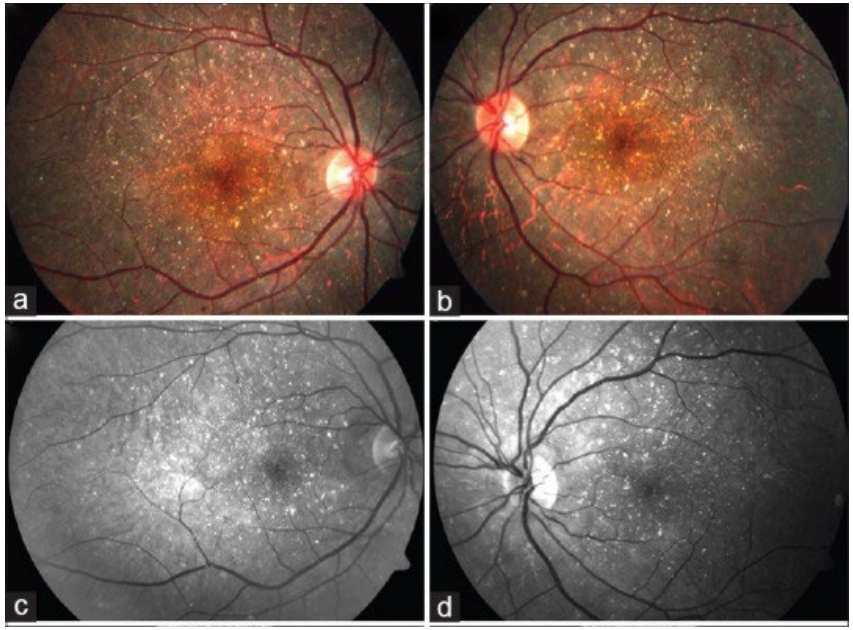
- LCA
- Enhanced *S*-cone dz

- Pattern dystrophies
- Central areolar choroidal dystrophy
- North Carolina macular dystrophy



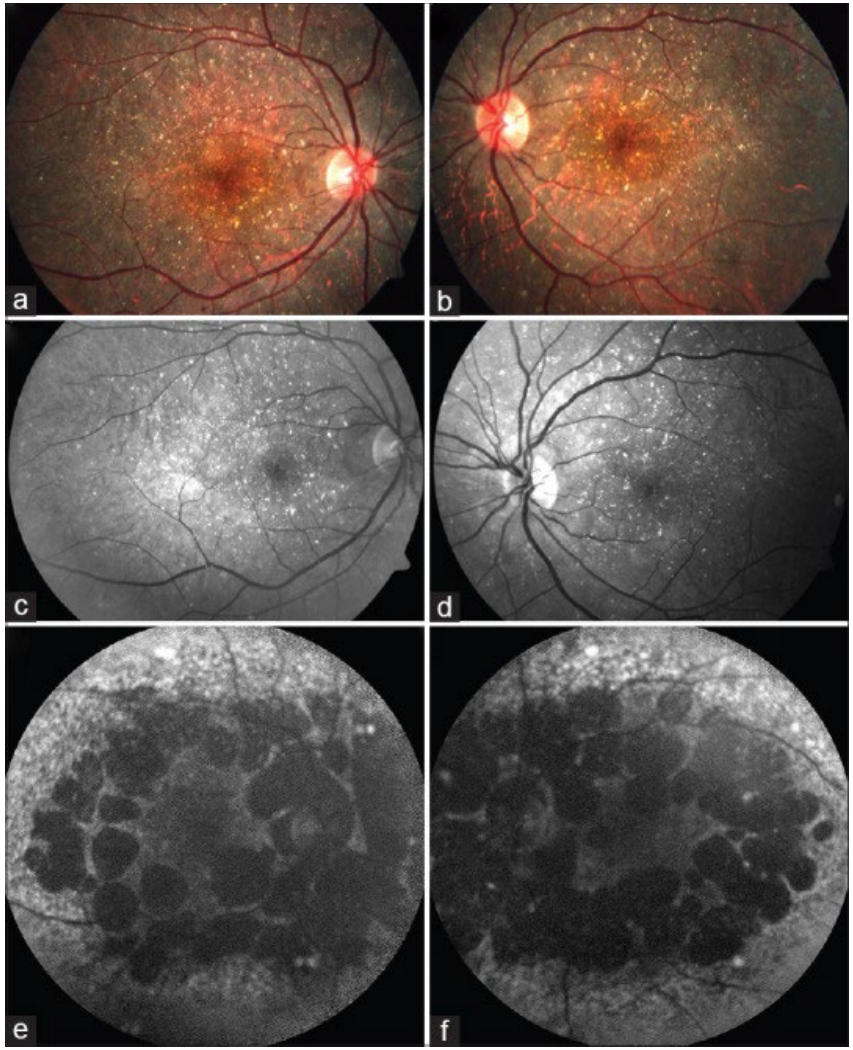


Bietti crystalline dystrophy



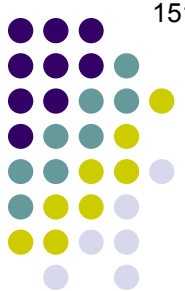
(*a* and *b*) FP of a patient with Bietti's crystalline dystrophy. (*c* and *d*) Red-free photographs

Bietti crystalline dystrophy



(*a* and *b*) FP of a patient with Bietti's crystalline dystrophy. (*c* and *d*) Red-free photographs (*e* and *f*) FAF shows well-defined areas of hypoautofluorescence encroaching on the fovea

Bietti crystalline dystrophy



What is the full name of this condition?
Bietti crystalline dystrophy

Is it fair to guess that DFE reveals crystals of some sort?
Indeed it is—**yellow**-**white** crystals are present within the posterior retina

Crystals are found in the anterior segment as well—where?

Hereditary

macular dystrophy

Rod-cone (RP)

Choroidal

Bietti

Gyrate

Choroideremia

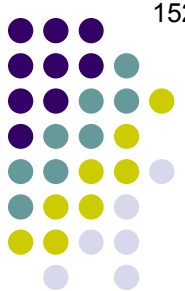
LCA

Enhanced *S*-cone dz

Pattern dystrophies

Central areolar choroidal dystrophy

North Carolina macular dystrophy



What is the full name of this condition?
Bietti crystalline dystrophy

Is it fair to guess that DFE reveals crystals of some sort?
Indeed it is—**yellow**-**white** crystals are present within the posterior retina

Crystals are found in the anterior segment as well—where?
In the structure

Hereditary

macular dystrophy

Choroidal

Bietti

Gyrate

Choroideremia

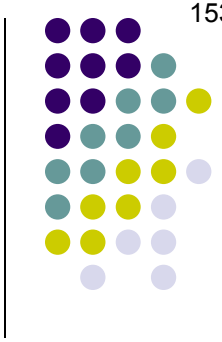
LCA

Enhanced *S*-cone dz

Pattern dystrophies

Central areolar choroidal dystrophy

North Carolina macular dystrophy



What is the full name of this condition?
Bietti crystalline dystrophy

Is it fair to guess that DFE reveals crystals of some sort?
Indeed it is—**yellow**-**white** crystals are present within the posterior retina

Crystals are found in the anterior segment as well—where?
In the cornea

Hereditary

macular dystrophy

Choroidal

Bietti

Gyrate

Choroideremia

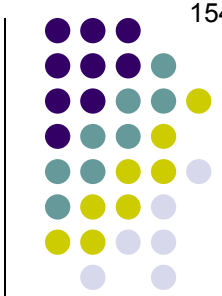
LCA

Enhanced *S*-cone dz

Pattern dystrophies

Central areolar choroidal dystrophy

North Carolina macular dystrophy



What is the full name of this condition?
Bietti crystalline dystrophy

Is it fair to guess that DFE reveals crystals of some sort?
Indeed it is—**yellow**-**white** crystals are present within the posterior retina

Crystals are found in the anterior segment as well—where?
In the cornea near the location

Hereditary

macular dystrophy

Choroidal

Bietti

Gyrate

Choroideremia

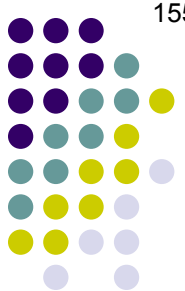
LCA

Enhanced *S*-cone dz

Pattern dystrophies

Central areolar choroidal dystrophy

North Carolina macular dystrophy



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Bietti crystalline dystrophy

Is it fair to guess that DFE reveals crystals of some sort?
Indeed it is—**yellow**-**white** crystals are present within the posterior retina

Crystals are found in the anterior segment as well—where?
In the cornea near the limbus

Hereditary

macular dystrophy

Choroidal

Bietti

Gyrate

Choroideremia

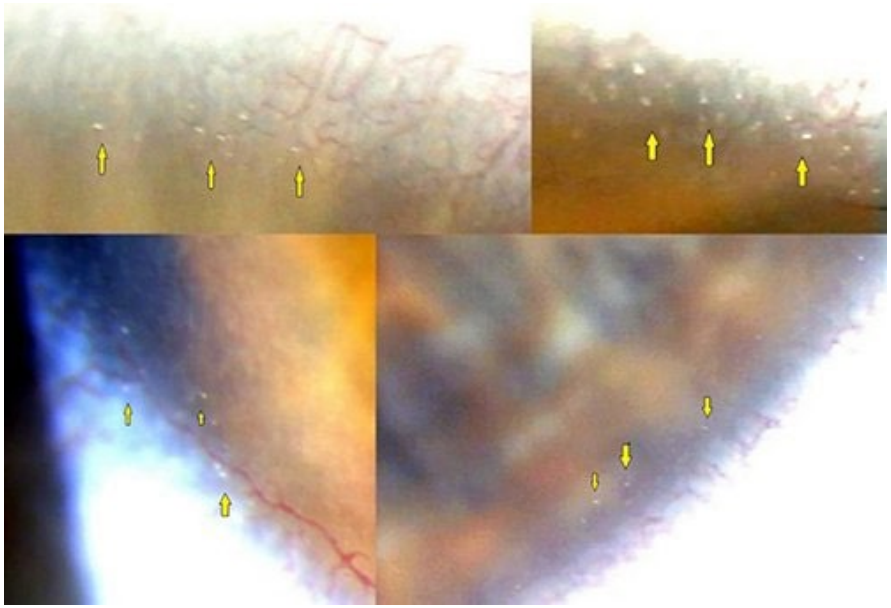
LCA

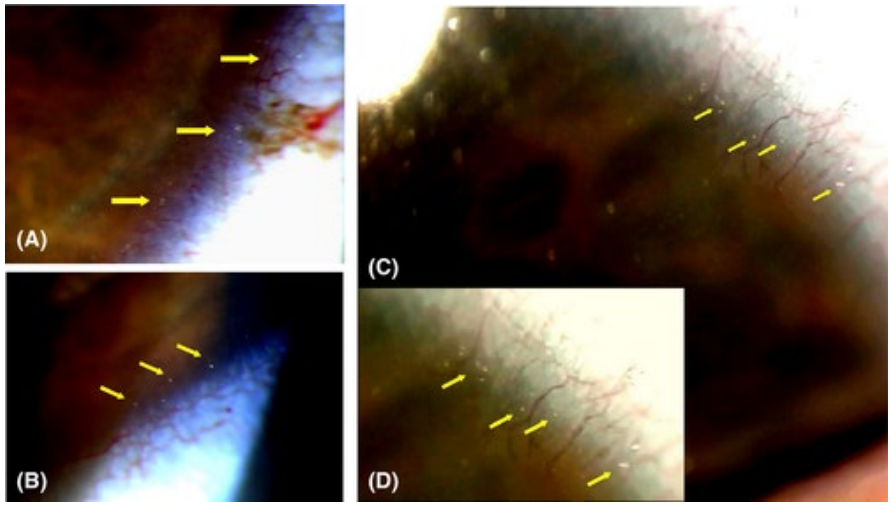
Enhanced *S*-cone dz

Pattern dystrophies

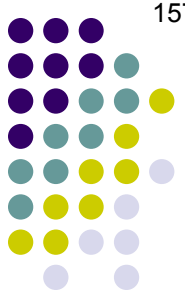
Central areolar choroidal dystrophy

North Carolina macular dystrophy





Bietti crystalline dystrophy: Cornea



What is the full name of this condition?
Bietti crystalline dystrophy

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Crystals are found in the anterior segment as well—where?
In the cornea near the limbus

How does Bietti present clinically?

Hereditary

macular dystrophy

Choroidal

Bietti

Gyrate

Choroideremia

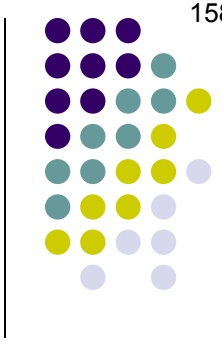
LCA

Enhanced *S*-cone dz

Pattern dystrophies

Central areolar choroidal dystrophy

North Carolina macular dystrophy



Hereditary

What is the full name of this condition?
Bietti crystalline dystrophy

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Crystals are found in the anterior segment as well—where?
In the cornea near the limbus

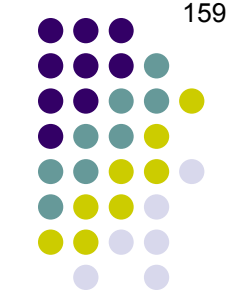
How does Bietti present clinically?
With visual symptom and location scotomas

macular dystrophy

Choroidal
- **Bietti**
- Gyrate
- Choroideremia

- LCA
- Enhanced *S*-cone dz

- Pattern dystrophies
- Central areolar choroidal dystrophy
- North Carolina macular dystrophy



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Bietti crystalline dystrophy

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Crystals are found in the anterior segment as well—where?
In the cornea near the limbus

How does Bietti present clinically?
With nyctalopia and paracentral scotomas

Hereditary

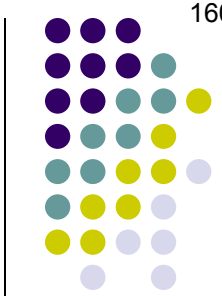
macular dystrophy

Choroidal

- **Bietti**
- Gyrate
- Choroideremia

- LCA
- Enhanced *S*-cone dz

- Pattern dystrophies
- Central areolar choroidal dystrophy
- North Carolina macular dystrophy



What is the full name of this condition?
Bietti crystalline dystrophy

Is it fair to guess that DFE reveals crystals of some sort?
Indeed it is—yellow-white crystals are present within the posterior retina

Crystals are found in the anterior segment as well—where?
In the cornea near the limbus

How does Bietti present clinically?
With nyctalopia and paracentral scotomas in life period years

Hereditary

macular dystrophy

Choroidal
- **Bietti**
- Gyrate
- Choroideremia

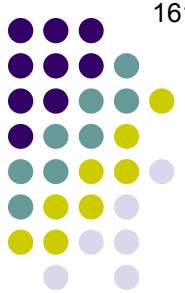
LCA

Enhanced *S*-cone dz

Pattern dystrophies

Central areolar choroidal dystrophy

North Carolina macular dystrophy



What is the full name of this condition?
Bietti crystalline dystrophy

Is it fair to guess that DFE reveals crystals of some sort?
Indeed it is—yellow-white crystals are present within the posterior retina

Crystals are found in the anterior segment as well—where?
In the cornea near the limbus

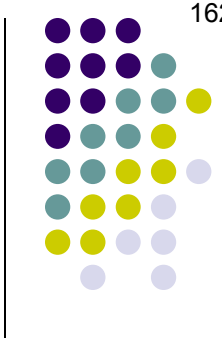
How does Bietti present clinically?
With nyctalopia and paracentral scotomas in teen/young adult years

Hereditary

Choroidal
- **Bietti**
- Gyrate
- Choroideremia

- LCA
- Enhanced *S*-cone dz

- Pattern dystrophies
- Central areolar choroidal dystrophy
- North Carolina macular dystrophy



Hereditary

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Crystals are found in the anterior segment as well—where?
In the cornea near the limbus

How does Bietti present clinically?
With nyctalopia and paracentral s[illegible] ult years

Is Bietti's common, or rare?

macular dystrophy

R[illegible]

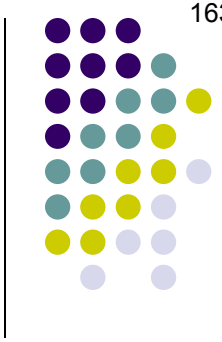
Rod-cone (RP)

Choroidal

- **Bietti**
- Gyrate
- Choroideremia

S-cone d2

- Pattern dystrophies
- Central areolar choroidal dystrophy
- North Carolina macular dystrophy



What is the full name of this condition?
Bietti crystalline dystrophy

Is it fair to guess that DFE reveals crystals of some sort?
Indeed it is—**yellow-white** crystals are present within the posterior retina

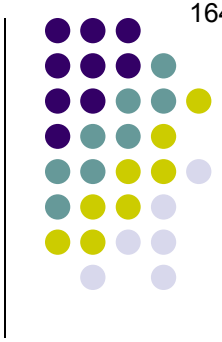
Crystals are found in the anterior segment as well—where?
In the cornea near the limbus

How does Bietti present clinically?
With nyctalopia and paracentral s… …ult years

Is Bietti's common, or rare?
Very rare

Choroidal
- **Bietti**
- Gyrate
- Choroideremia

- Pattern dystrophies
- Central areolar choroidal dystrophy
- North Carolina macular dystrophy



What is the full name of this condition?
Bietti crystalline dystrophy

Is it fair to guess that DFE reveals crystals of some sort?
Indeed it is—**yellow-white** crystals are present within the posterior retina

Crystals are found in the anterior segment as well—where?
In the cornea near the limbus

How does Bietti present clinically?
With nyctalopia and paracentral s[illegible] ult years

Is Bietti's common, or rare?
Very rare

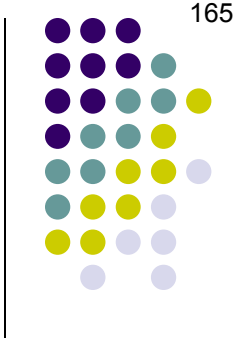
Is it treatable?

Choroidal
- **Bietti**
- Gyrate
- Choroideremia

macular dystrophy

rdt

- Pattern dystrophies
- Central areolar choroidal dystrophy
- North Carolina macular dystrophy



What is the full name of this condition?
Bietti crystalline dystrophy

Is it fair to guess that DFE reveals crystals of some sort?
Indeed it is—**yellow-white** crystals are present within the posterior retina

Crystals are found in the anterior segment as well—where?
In the cornea near the limbus

How does Bietti present clinically?
With nyctalopia and paracentral s[illegible] ult years

Is Bietti's common, or rare?
Very rare

Is it treatable?
No

Hereditary

macular dystrophy

Rod-cone (RP)

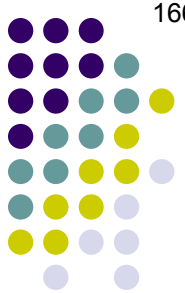
rdt

Choroidal

- **Bietti**
- Gyrate
- Choroideremia

S-cone d2

- Pattern dystrophies
- Central areolar choroidal dystrophy
- North Carolina macular dystrophy



What is the full name of this condition?

dystrophy

Rod-dor

Rod-

Bietti

Gyrate

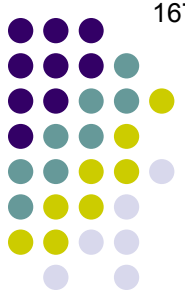
Choroideremia

LCA

Enhanced *S*-cone dz

Central areolar choroidal dystrophy

North Carolina macular dystrophy



What is the full name of this condition?
Gyrate atrophy

dystrophy

Rod-dor

Rod-

Bietti

Gyrate

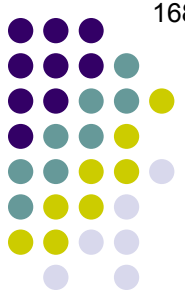
Choroideremia

LCA

Enhanced
S-cone dz

Central areolar
choroidal dystrophy

North Carolina
macular dystrophy



What is the full name of this condition?
Gyrate atrophy

How does it present on DFE?

dystrophy

Rod-dor

Rod-

Bietti

Gyrate

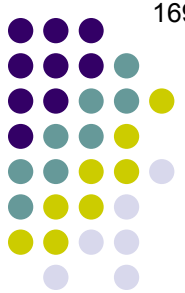
Choroideremia

LCA

Enhanced *S*-cone dz

Central areolar choroidal dystrophy

North Carolina macular dystrophy



What is the full name of this condition?
Gyrate atrophy

How does it present on DFE?
Early dz is marked by the presence of large peripheral classic description areas of RPE and choriocapillaris atrophy

dystrophy

Rod-dor

Rod-

Bietti

Gyrate

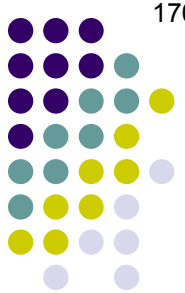
Choroideremia

LCA

Enhanced *S*-cone dz

Central areolar choroidal dystrophy

North Carolina macular dystrophy



What is the full name of this condition?
Gyrate atrophy

How does it present on DFE?
Early dz is marked by the presence of large peripheral ‘pavingstone’ areas of RPE and choriocapillaris atrophy

dystrophy

Rod-dor

Rod-

Bietti

Gyrate

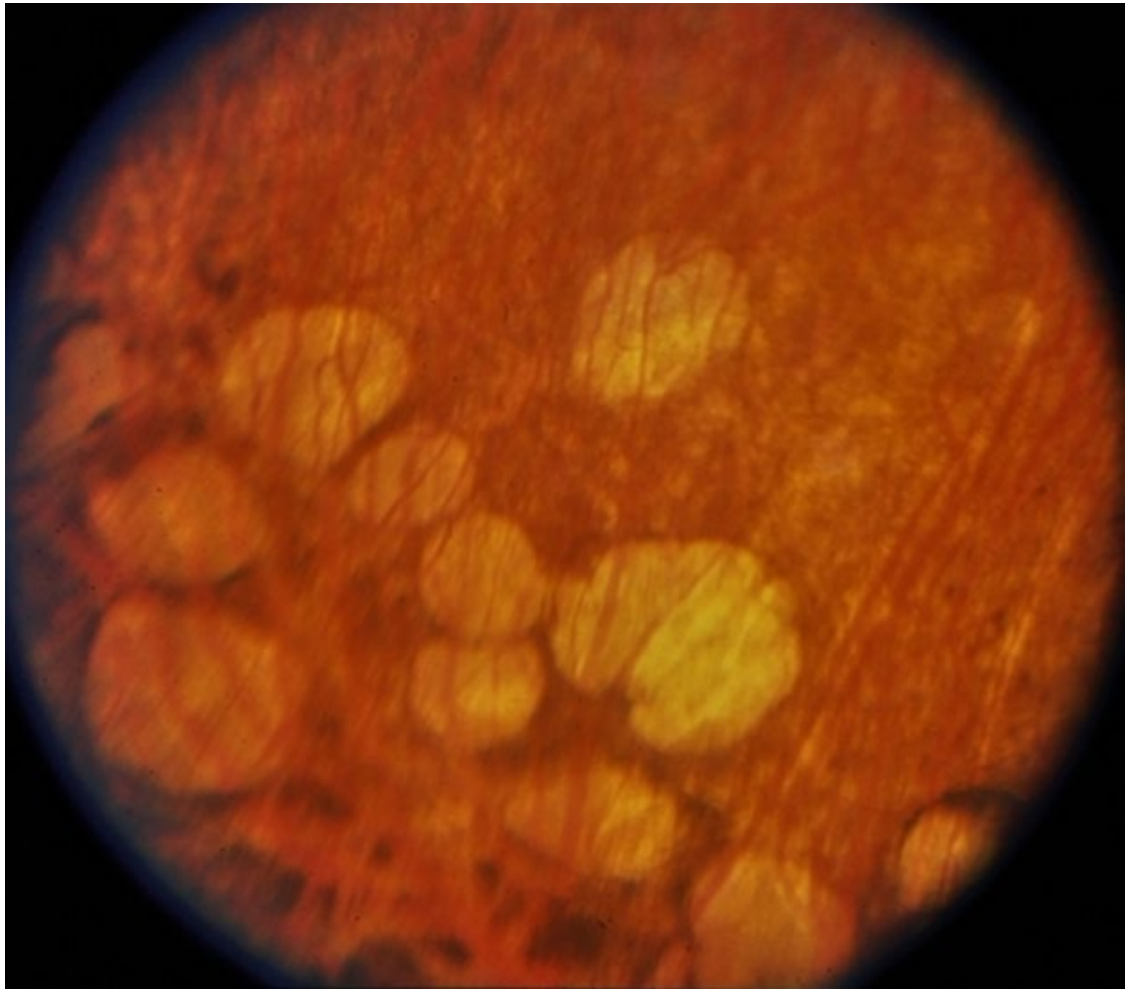
Choroideremia

LCA

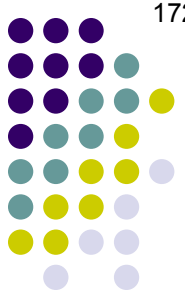
Enhanced *S*-cone dz

Central areolar choroidal dystrophy

North Carolina macular dystrophy



Gyrate atrophy: Pavingstones



What is the full name of this condition?
Gyrate atrophy

How does it present on DFE?
Early dz is marked by the presence of large peripheral ‘pavingstone’ areas of RPE and choriocapillaris atrophy. Over time these areas will coalesce, ultimately forming a scalloped perimeter around the posterior pole.

dystrophy

Rod-dor

Rod-

Bietti

Gyrate

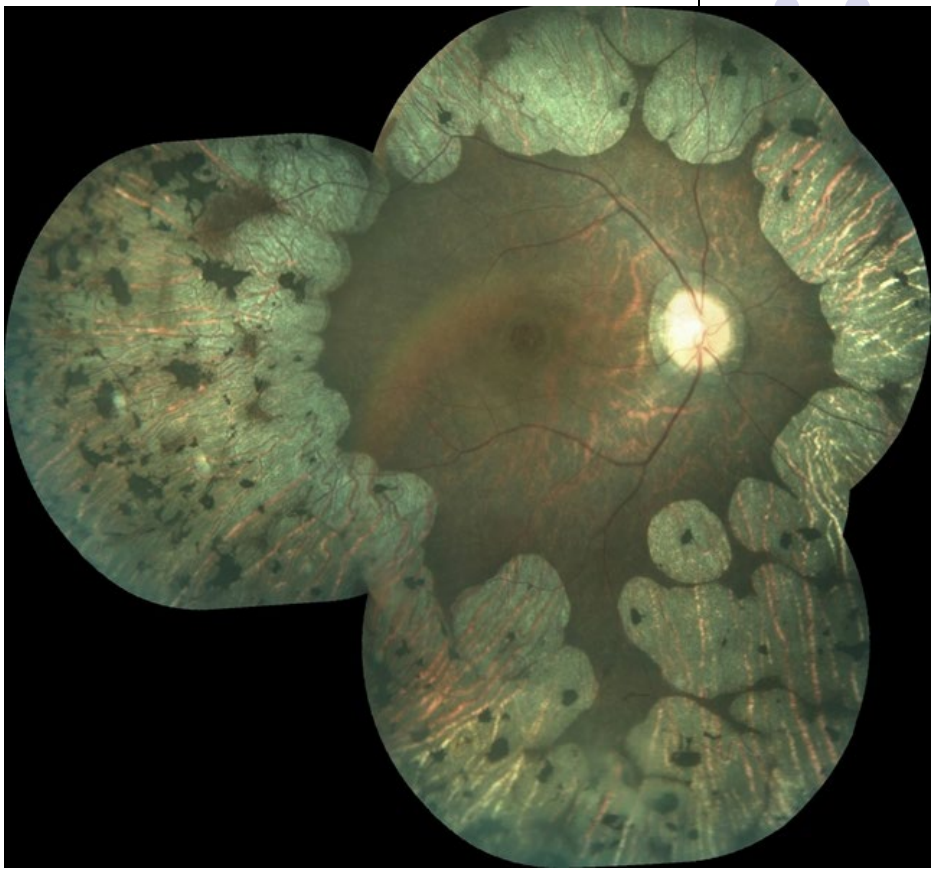
Choroideremia

LCA

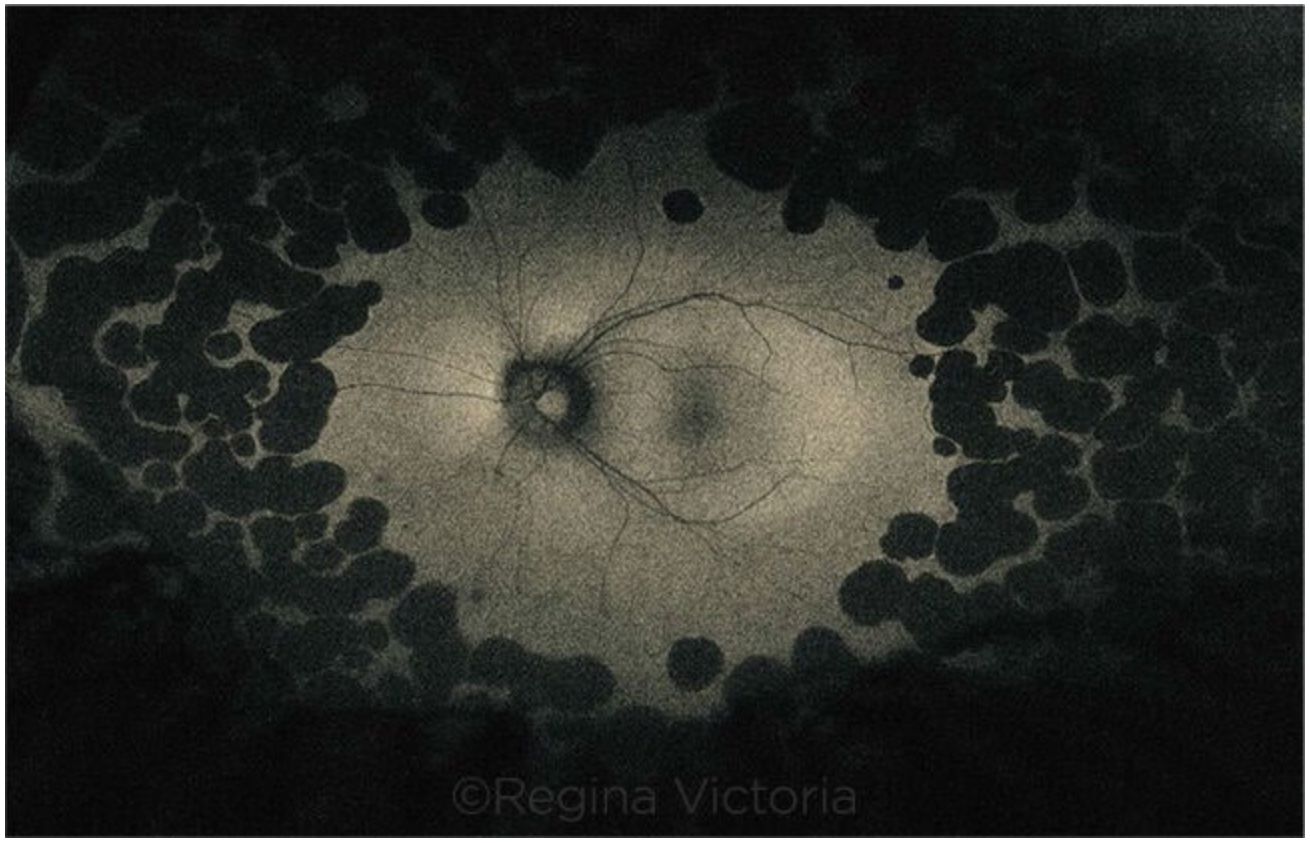
Enhanced *S*-cone dz

Central areolar choroidal dystrophy

North Carolina macular dystrophy

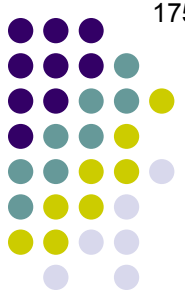


Gyrate atrophy: Scalloped areas



A 16-year-old myopic girl presented with a complaint of diminished vision, especially at night. FAF revealed hypoautofluorescent scalloped areas of choroidal atrophy in the retinal periphery.

Gyrate atrophy: FAF



What is the full name of this condition?
Gyrate atrophy

How does it present on DFE?
Early dz is marked by the presence of large peripheral ‘pavingstone’ areas of RPE and choriocapillaris atrophy. Over time these areas will coalesce, ultimately forming a scalloped perimeter around the posterior pole.

How does it present clinically?

dystrophy

Rod-dor

Rod-

Bietti

Gyrate

Choroideremia

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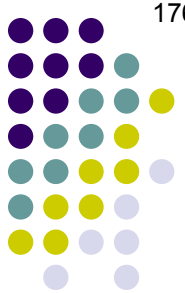
Central areolar choroidal dystrophy

North Carolina macular dystrophy



What is the full name of this condition?
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Early dz is marked by the presence of large peripheral ‘pavingstone’ areas of RPE and choriocapillaris atrophy. Over time these areas will coalesce, ultimately forming a scalloped perimeter around the posterior pole.

How does it present clinically?
Severe symptom commencing in life period

dystrophy

Rod-dor
Rod-

Bietti
Gyrate
Choroideremia

LCA
Enhanced *S*-cone dz

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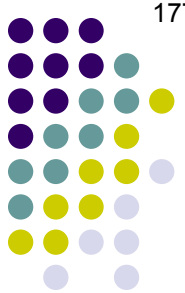

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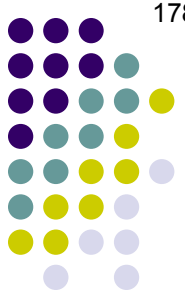

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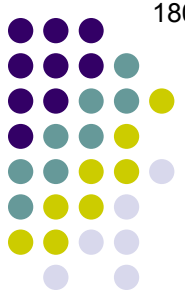

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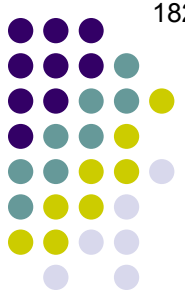

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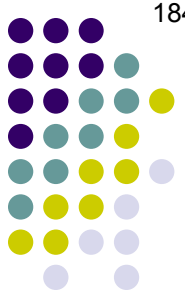

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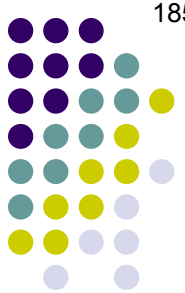
What lab test supports the dx?
Finding elevated levels of ornithine in the serum



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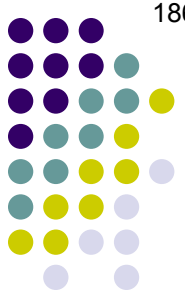

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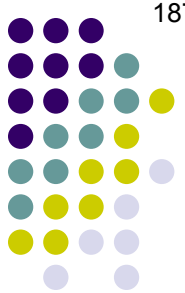

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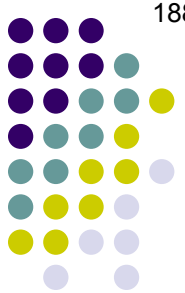

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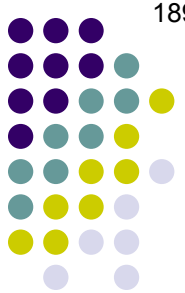

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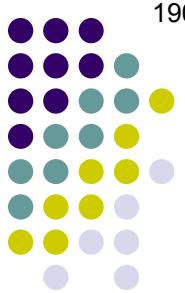

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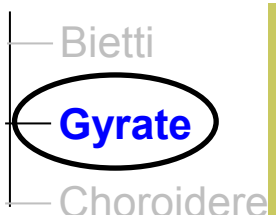

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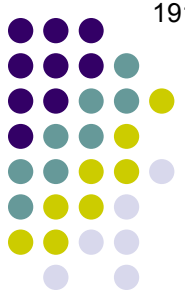

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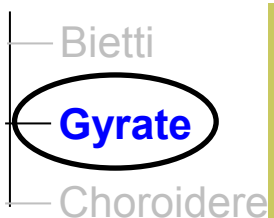

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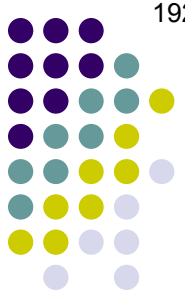

How does choroideremia present on DFE?

dystrophy

Rod-dor

Rod-cone (RP)

Choroidal

- Bietti
- Gyrate
- **Choroideremia**

- LCA
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- Pattern dystrophies
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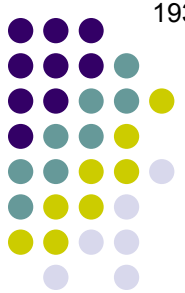

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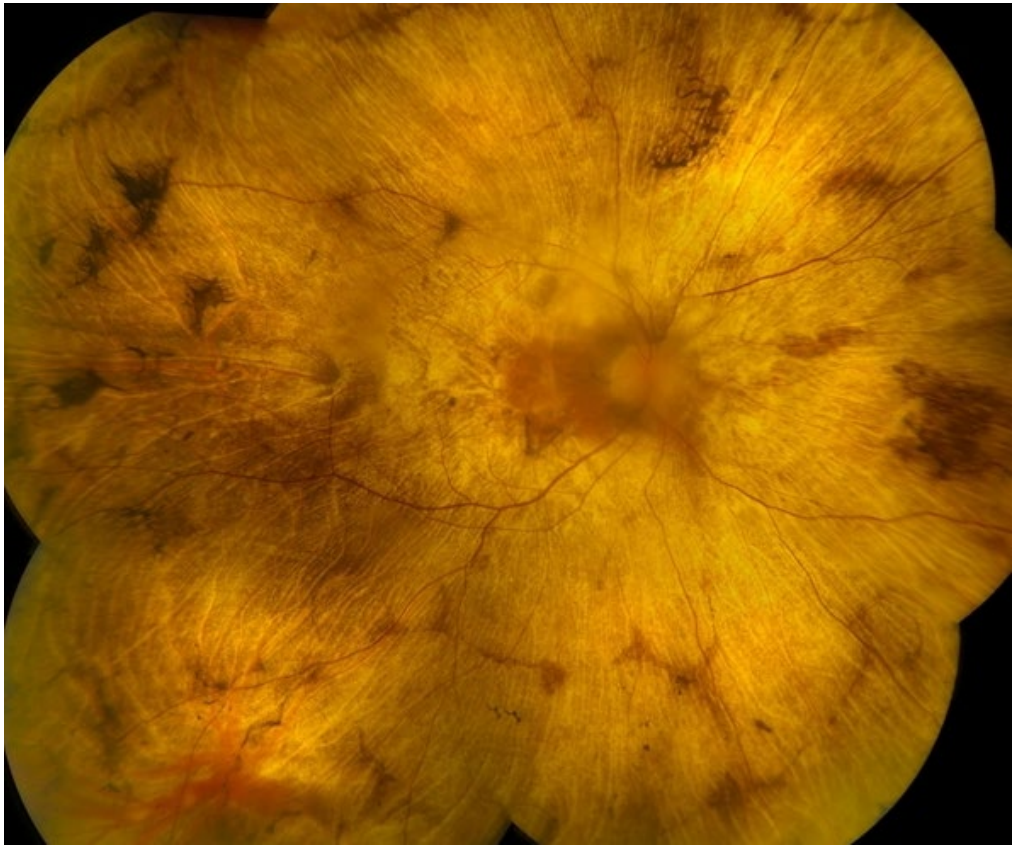
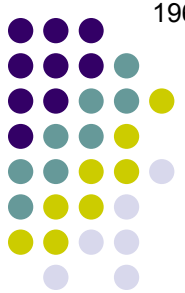


Choroideremia

FAF reveals the expected hypofluorescent lesions

Choroideremia



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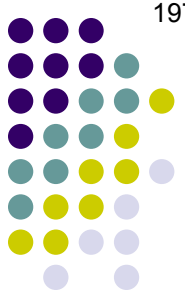

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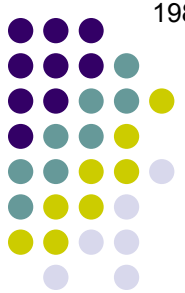

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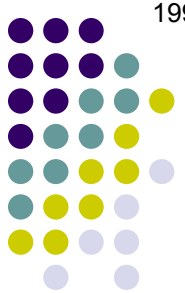

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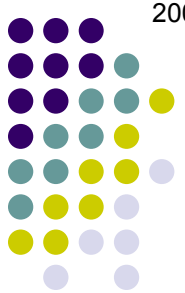

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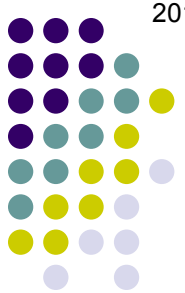

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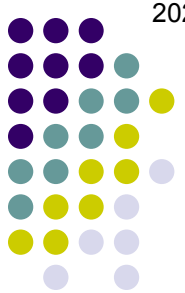

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The short answer is, you may not be able to. There is considerable phenotypical and clinical overlap among the choroidal dystrophies, to the extent that it is injudicious to treat any particular presentation as pathognomonic.

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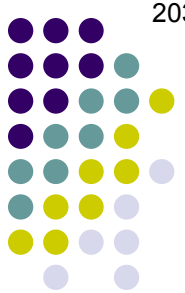

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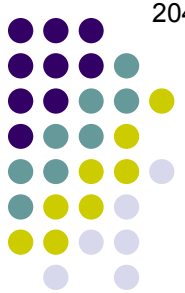

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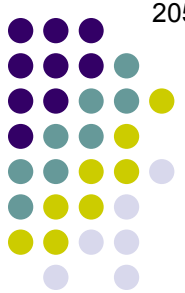
Pattern dystrophies
Central areolar choroidal dystrophy
North Carolina macular dystrophy



How does choroideremia present on DFE?
With pronounced atrophic changes of the RPE and choriocapillaris leading to profound depigmentation of much of the posterior pole, characteristically with a scalloped border

How does it present clinically?
Severe nyctalopia commencing in childhood, and progressive VF loss

What is the underlying pathophysiology?
A defect in the gene coding for the *geranylgeranyl transferase* enzyme

dystrophy

Rod-dor

Rod-cone (RP)

Choroidal

Bietti

Gyrate

Choroideremia

LCA

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Pattern dystrophies

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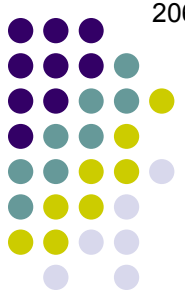

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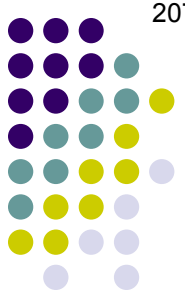

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dystrophy

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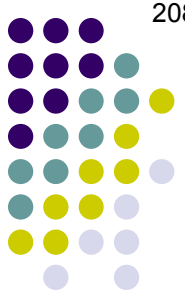
Pattern dystrophies

Central areolar choroidal dystrophy

North Carolina macular dystrophy



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dystrophy

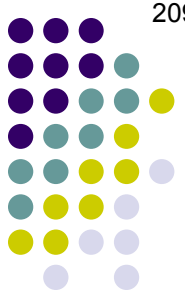
***For more on gyrate and choroideremia, see slide-set* R42**

Rod-
X-linked
Rod-cone (RP)

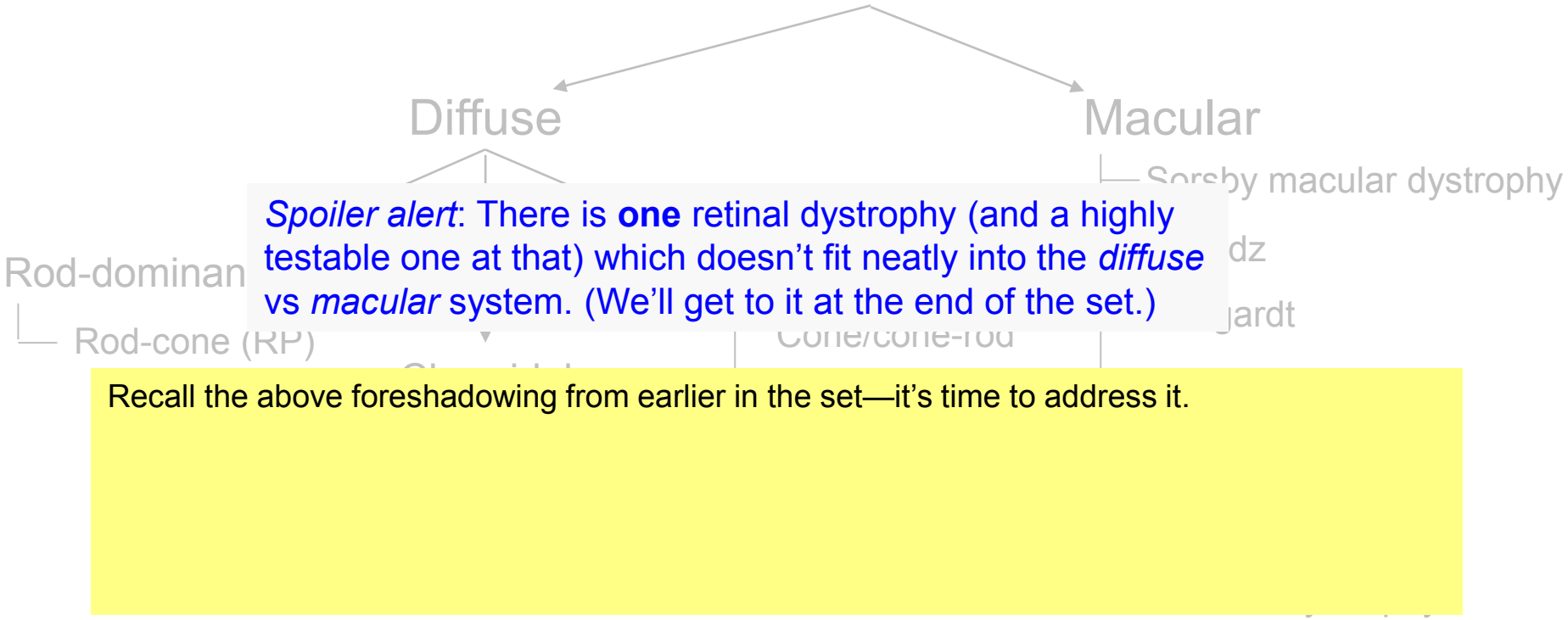
Choroidal
Bietti
Gyrate
Choroideremia

LCA
Enhanced *S*-cone dz

Pattern dystrophies
Central areolar choroidal dystrophy
North Carolina macular dystrophy



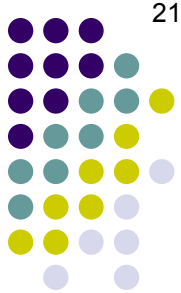
Hereditary Retinal/Choroidal Dystrophies



Spoiler alert: There is **one** retinal dystrophy (and a highly testable one at that) which doesn't fit neatly into the *diffuse* vs *macular* system. (We'll get to it at the end of the set.)

Recall the above foreshadowing from earlier in the set—it's time to address it.

No question—proceed when ready



Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular

Sorsby macular dystrophy

Rod-dominan

dz

Rod-cone (RP)

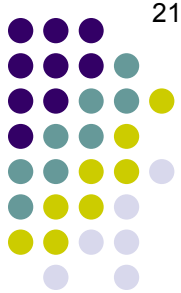
jardt

Cone/cone-rod

Spoiler alert: There is **one** retinal dystrophy (and a highly testable one at that) which doesn't fit neatly into the *diffuse* vs *macular* system. (We'll get to it at the end of the set.)

Recall the above foreshadowing from earlier in the set—it's time to address it. *What is this class of hereditary retinal dystrophy that can't be slotted into* a diffuse vs macular *system?*

?



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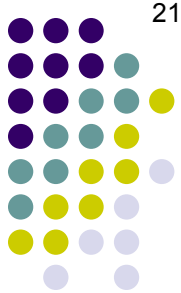
Recall the above foreshadowing from earlier in the set—it's time to address it. *What is this class of hereditary retinal dystrophy that can't be slotted into* a diffuse vs macular *system?*
It's one that requires an inner- vs outer-retinal divide, specifically, a dystrophy of the inner retina

Inner retinal dystrophy



Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular

Sorsby macular dystrophy

Rod-dominan

dz

Rod-cone (RP)

jardt

Cone/cone-rod

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It's one that requires an inner- vs outer-retinal divide, specifically, a dystrophy of the inner retina

What is the lone such condition discussed in the Retina *book?*

Inner retinal dystrophy

?

Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular

Sorsby macular dystrophy

Rod-dominan

dz

Rod-cone (RP)

jardt

Cone/cone-rod

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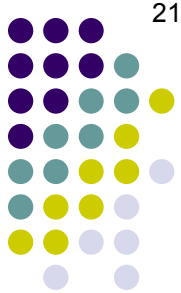
It's one that requires an inner- vs outer-retinal divide, specifically, a dystrophy of the inner retina

What is the lone such condition discussed in the Retina *book?*
X-linked retinoschisis

Inner retinal dystrophy

X-linked retinoschisis



Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular

Sorsby macular dystrophy

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Rod-dominan

Rod-cone (RP)

dz

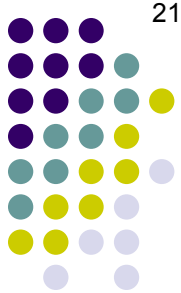
jardt

Cone/cone-rod

Note: There is some disagreement within the *BCSC* regarding how X-linked retinoschisis should be classified.

Inner retinal dystrophy

X-linked retinoschisis



Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular

Sorsby macular dystrophy

Spoiler alert: There is **one** retinal dystrophy (and a highly testable one at that) which doesn't fit neatly into the *diffuse* vs *macular* system. (We'll get to it at the end of the set.)

Rod-dominan

Rod-cone (RP)

dz

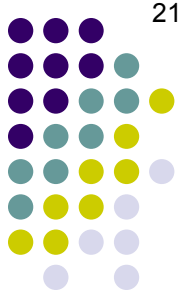
jardt

Cone/cone-rod

Note: There is some disagreement within the *BCSC* regarding how X-linked retinoschisis should be classified. Whereas the *Retina* book calls it an *inner retinal dystrophy…*

Inner retinal dystrophy (per the *Retina* book)

X-linked retinoschisis



Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular

Sorsby macular dystrophy

Spoiler alert: There is **one** retinal dystrophy (and a highly testable one at that) which doesn't fit neatly into the *diffuse* vs *macular* system. (We'll get to it at the end of the set.)

Rod-dominan

Rod-cone (RP)

dz

jardt

Cone/cone-rod

Note: There is some disagreement within the *BCSC* regarding how X-linked retinoschisis should be classified. Whereas the *Retina* book calls it an *inner retinal dystrophy*…the *Peds* book classifies it as a two words.

two words (per the *Peds* book)

X-linked retinoschisis

Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular

Sorsby macular dystrophy

Rod-dominan

dz

jardt

Rod-cone (RP)

Cone/cone-rod

Spoiler alert: There is **one** retinal dystrophy (and a highly testable one at that) which doesn't fit neatly into the *diffuse* vs *macular* system. (We'll get to it at the end of the set.)

Note: There is some disagreement within the *BCSC* regarding how X-linked retinoschisis should be classified. Whereas the *Retina* book calls it an *inner retinal dystrophy*…the *Peds* book classifies it as a *hereditary vitreoretinopathy**. Caveat emptor.

Hereditary vitreoretinopathy (per the *Peds* book)

X-linked retinoschisis

*As to *why* they do this, we will see shortly

By what other (very similar) name is X-linked ***retinoschisis*** *(XLR) known?*

Before we get any deeper—what does retinoschisis *refer to in this context?*

phy

Rod-

R

Inner retinal dystrophy

X-linked retinoschisis

By what other (very similar) name is X-linked ***retinoschisis*** *(XLR) known?*

Before we get any deeper—what does retinoschisis *refer to in this context?*
Splitting within the layers of the neurosensory retina

phy

Rod-

F

Inner retinal dystrophy

X-linked retinoschisis

By what other (very similar) name is X-linked retinoschisis (XLR) known?

phy

Rod-

R

Inner retinal dystrophy

X-linked retinoschisis

By what other (very similar) name is X-linked retinoschisis (XLR) known?
X-linked juvenile retinoschisis

phy

Rod-
└ R

Inner retinal dystrophy
└ X-linked retinoschisis

By what other (very similar) name is X-linked retinoschisis (XLR) known?

X-linked **juvenile** retinoschisis

What is implied—correctly—by the word juvenile *above?*

phy

Rod-

R

Inner retinal dystrophy

X-linked retinoschisis

By what other (very similar) name is X-linked retinoschisis (XLR) known?

X-linked **juvenile** retinoschisis

What is implied—correctly—by the word juvenile *above?*

That the condition manifests early in life (in fact, it is [redacted])

phy

Rod-

R

Inner retinal dystrophy

└ X-linked retinoschisis

By what other (very similar) name is X-linked retinoschisis (XLR) known?

X-linked **juvenile** retinoschisis

What is implied—correctly—by the word juvenile *above?*

That the condition manifests early in life (in fact, it is congenital)

phy

Rod-

└ R

Inner retinal dystrophy

└ X-linked retinoschisis

By what other (very similar) name is X-linked retinoschisis (XLR) known?
X-linked juvenile retinoschisis

How does it present on DFE?

phy

Rod-
└ R

Inner retinal dystrophy
└ X-linked retinoschisis

By what other (very similar) name is X-linked retinoschisis (XLR) known?
X-linked juvenile retinoschisis

How does it present on DFE?
With macular schisis in a [redacted] pattern, +/- peripheral schisis

phy

Rod-

R

Inner retinal dystrophy

X-linked retinoschisis

By what other (very similar) name is X-linked retinoschisis (XLR) known?
X-linked juvenile retinoschisis

How does it present on DFE?
With macular schisis in a radial pattern, +/- peripheral schisis

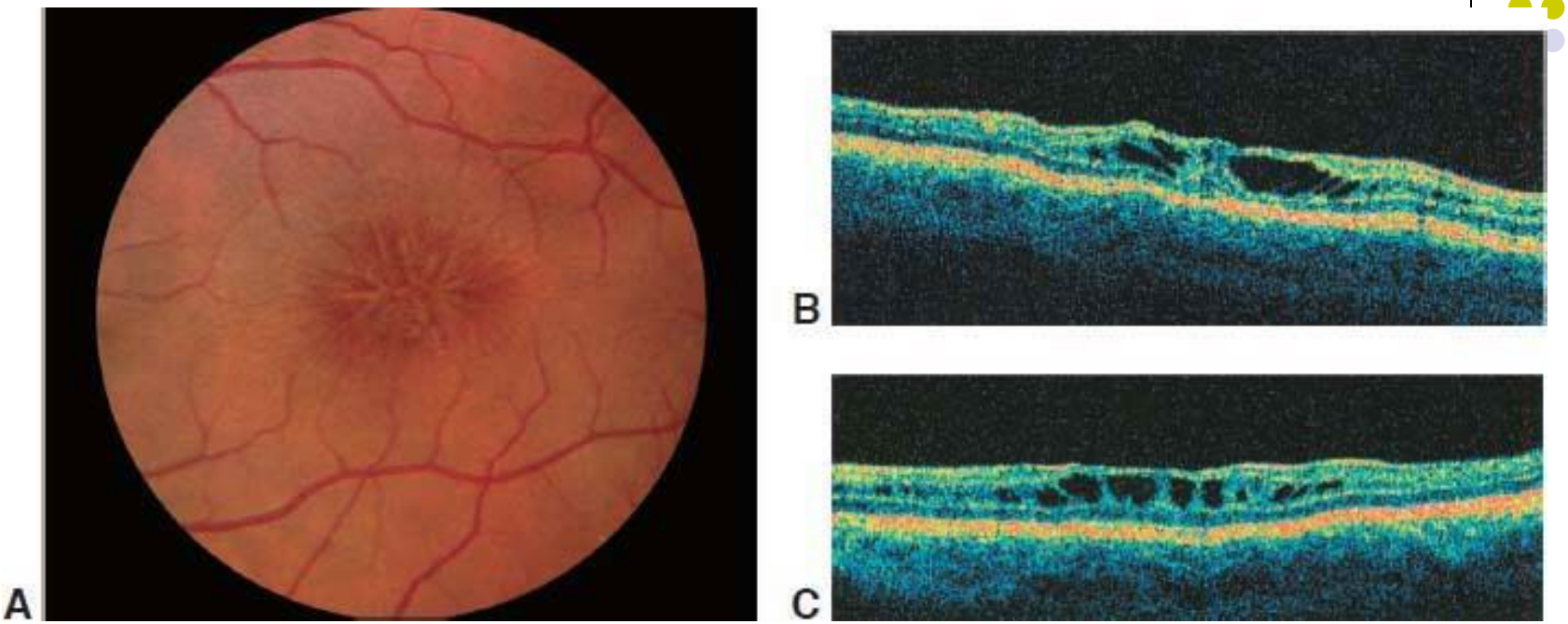
phy

Rod-

R

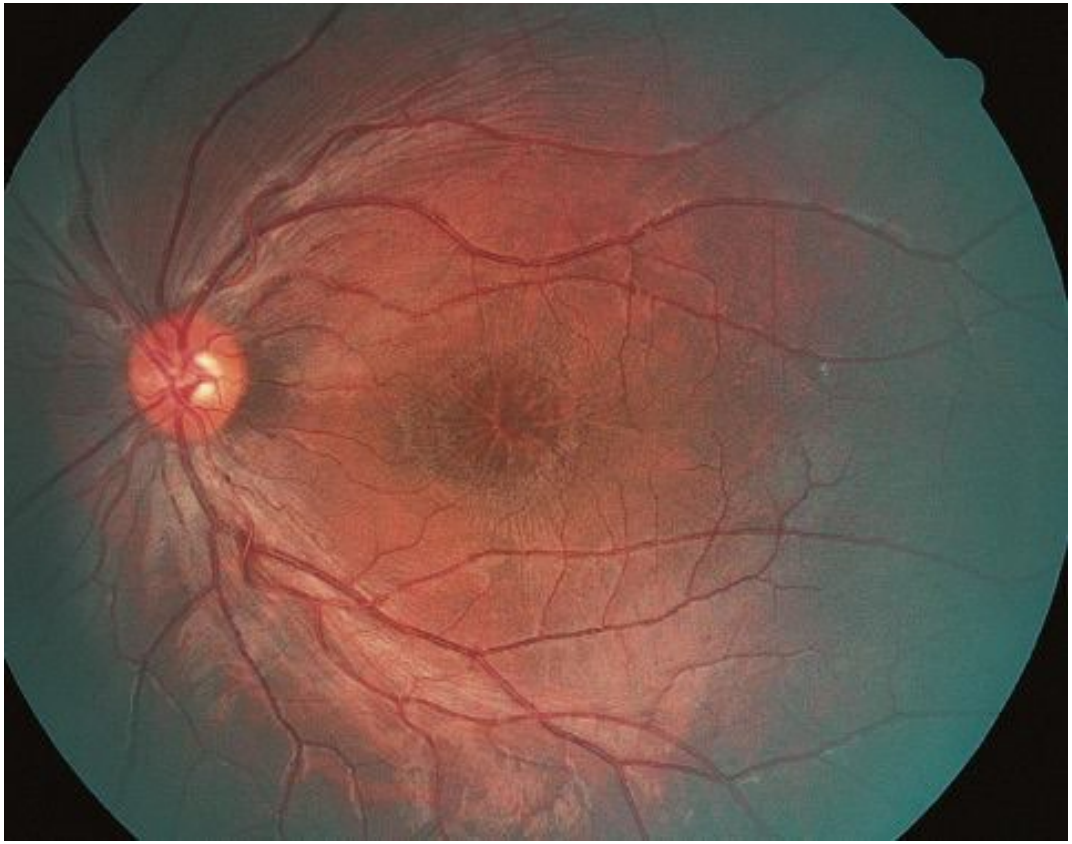
Inner retinal dystrophy

X-linked retinoschisis

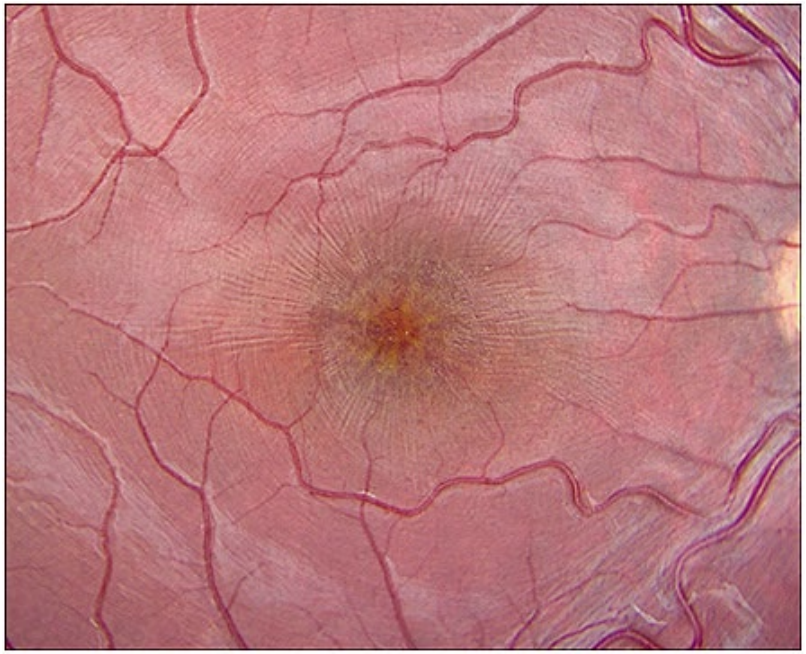


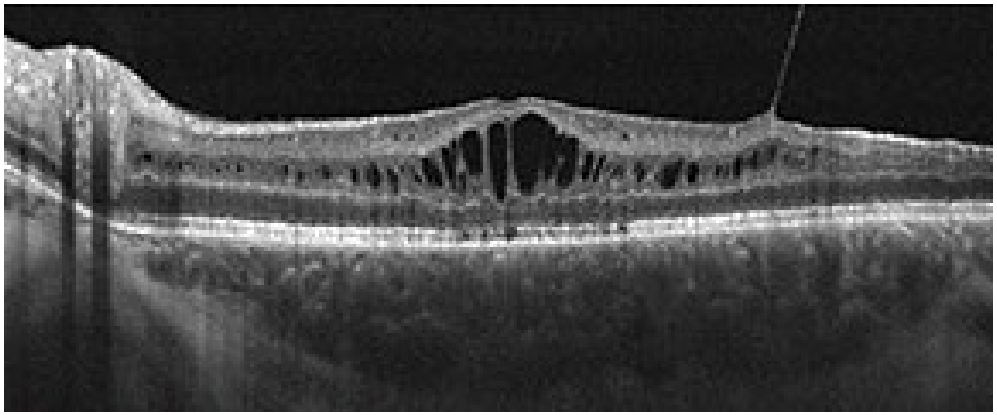
A, Color fundus photograph shows the characteristic pattern of macular schisis, a more consistent finding than peripheral changes. Vertical (B) and horizontal (C) OCT scans demonstrate schisis spaces in the middle layers of the macula.

X-linked retinoschisis



X-linked juvenile retinoschisis: Foveal cysts





X-linked juvenile retinoschisis: Foveal cysts

By what other (very similar) name is X-linked retinoschisis (XLR) known?
X-linked juvenile retinoschisis

How does it present on DFE?
With **macular schisis** in a radial pattern, +/- peripheral schisis

What layer(s) of the retina are involved in the schisis?

phy

Rod-
└ R

Inner retinal dystrophy
└ X-linked retinoschisis

By what other (very similar) name is X-linked retinoschisis (XLR) known?
X-linked juvenile retinoschisis

How does it present on DFE?
With **macular schisis** in a radial pattern, +/- peripheral schisis

What layer(s) of the retina are involved in the schisis?
Mainly the abb., but the abb. can be involved as well

phy

Rod-
R

Inner retinal dystrophy
X-linked retinoschisis

By what other (very similar) name is X-linked retinoschisis (XLR) known?
X-linked juvenile retinoschisis

How does it present on DFE?
With **macular schisis** in a radial pattern, +/- peripheral schisis

What layer(s) of the retina are involved in the schisis?
Mainly the NFL , but the OPL can be involved as well

phy

Rod-

R

Inner retinal dystrophy

X-linked retinoschisis

By what other (very similar) name is X-linked retinoschisis (XLR) known?
X-linked juvenile retinoschisis

How does it present on DFE?
With **macular schisis** in a radial pattern, +/- peripheral schisis

What layer(s) of the retina are involved in the schisis?
Mainly the **NFL** , but the **OPL** can be involved as well

What do these stand for in this context?
NFL: **?**
OPL: **?**

Rod-
R

phy

Inner retinal dystrophy
X-linked retinoschisis

By what other (very similar) name is X-linked retinoschisis (XLR) known?
X-linked juvenile retinoschisis

How does it present on DFE?
With **macular schisis** in a radial pattern, +/- peripheral schisis

What layer(s) of the retina are involved in the schisis?
Mainly the **NFL** , but the **OPL** can be involved as well

What do these stand for in this context?
NFL: Nerve fiber layer
OPL: Outer plexiform layer

Rod-
└ R

phy

Inner retinal dystrophy
└ X-linked retinoschisis

By what other (very similar) name is X-linked retinoschisis (XLR) known?
X-linked juvenile retinoschisis

How does it present on DFE?
With **macular schisis** in a radial pattern, +/- peripheral schisis

What layer(s) of the retina are involved in the schisis?
Mainly the NFL , but the OPL can be involved as well

What proportion of XLR pts manifest foveal schisis?

phy

Rod-
└ R

Inner retinal dystrophy
└ X-linked retinoschisis

By what other (very similar) name is X-linked retinoschisis (XLR) known?
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Mainly the NFL , but the OPL can be involved as well

What proportion of XLR pts manifest foveal schisis?
All of them, essentially

phy

Rod-

R

Inner retinal dystrophy

X-linked retinoschisis

By what other (very similar) name is X-linked retinoschisis (XLR) known?
X-linked juvenile retinoschisis

How does it present on DFE?
With **macular schisis** in a radial pattern, +/- **peripheral schisis**

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All of them, essentially

What proportion of pts manifest peripheral *schisis?*

Rod-
└ R

phy

Inner retinal dystrophy
└ X-linked retinoschisis

By what other (very similar) name is X-linked retinoschisis (XLR) known?
X-linked juvenile retinoschisis

How does it present on DFE?
With **macular schisis** in a radial pattern, +/- **peripheral schisis**

What layer(s) of the retina are involved in the schisis?
Mainly the NFL , but the OPL can be involved as well

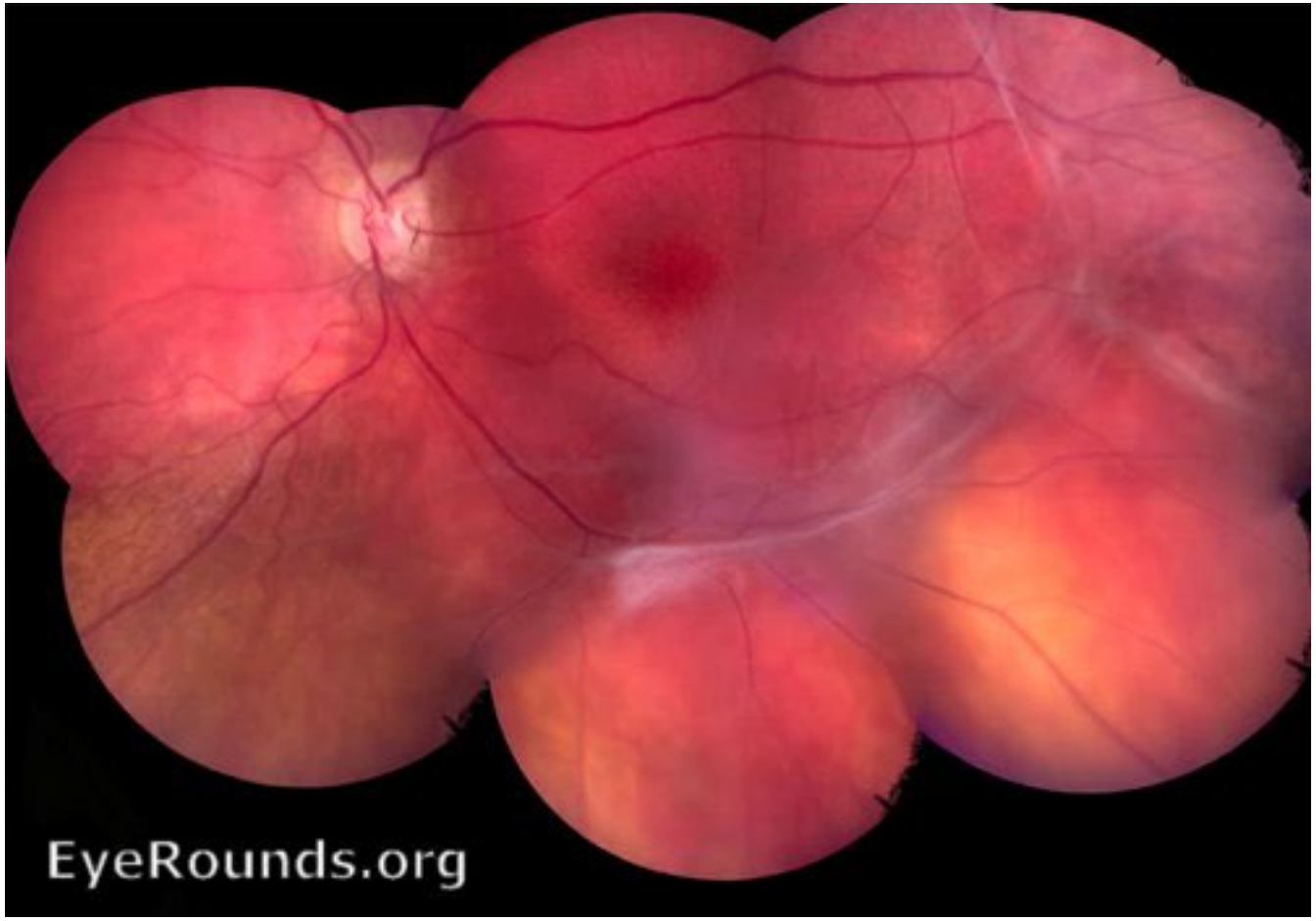
What proportion of XLR pts manifest foveal schisis?
All of them, essentially

What proportion of pts manifest peripheral *schisis?*
About half

phy

Rod-
└ R

Inner retinal dystrophy
└ X-linked retinoschisis



X-linked juvenile retinoschisis: Peripheral retinoschisis

By what other (very similar) name is X-linked retinoschisis (XLR) known?
X-linked juvenile retinoschisis

How does it present on DFE?
With **macular schisis** in a radial pattern, +/- **peripheral schisis**

What layer(s) of the
Mainly the NFL , but

What proportion of X
All of them, essential

What proportion of p
About half

Previous slide notwithstanding, photographic documentation of peripheral schisis is not practical. (It's too far out there.) What's the preferred method for monitoring the status of peripheral schisis?

Rod-
└ R

Inner retinal dystrophy
└ X-linked retinoschisis

By what other (very similar) name is X-linked retinoschisis (XLR) known?
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VF testing

Rod-
└ R

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VF testing

Does schisis result in a relative, or absolute scotoma?

Rod-
└ F

Inner retinal dystrophy
└ X-linked retinoschisis

By what other (very similar) name is X-linked retinoschisis (XLR) known?
X-linked juvenile retinoschisis

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VF testing

Does schisis result in a relative, or absolute scotoma?
Absolute

Rod-
└ F

ny

Inner retinal dystrophy
└ X-linked retinoschisis

By what other (very similar) name is X-linked retinoschisis (XLR) known?
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Does schisis result in a relative, or absolute scotoma?
Absolute

What HVF protocol is needed?

Rod-
└ R

ny

Inner retinal dystrophy
└ X-linked retinoschisis

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VF testing

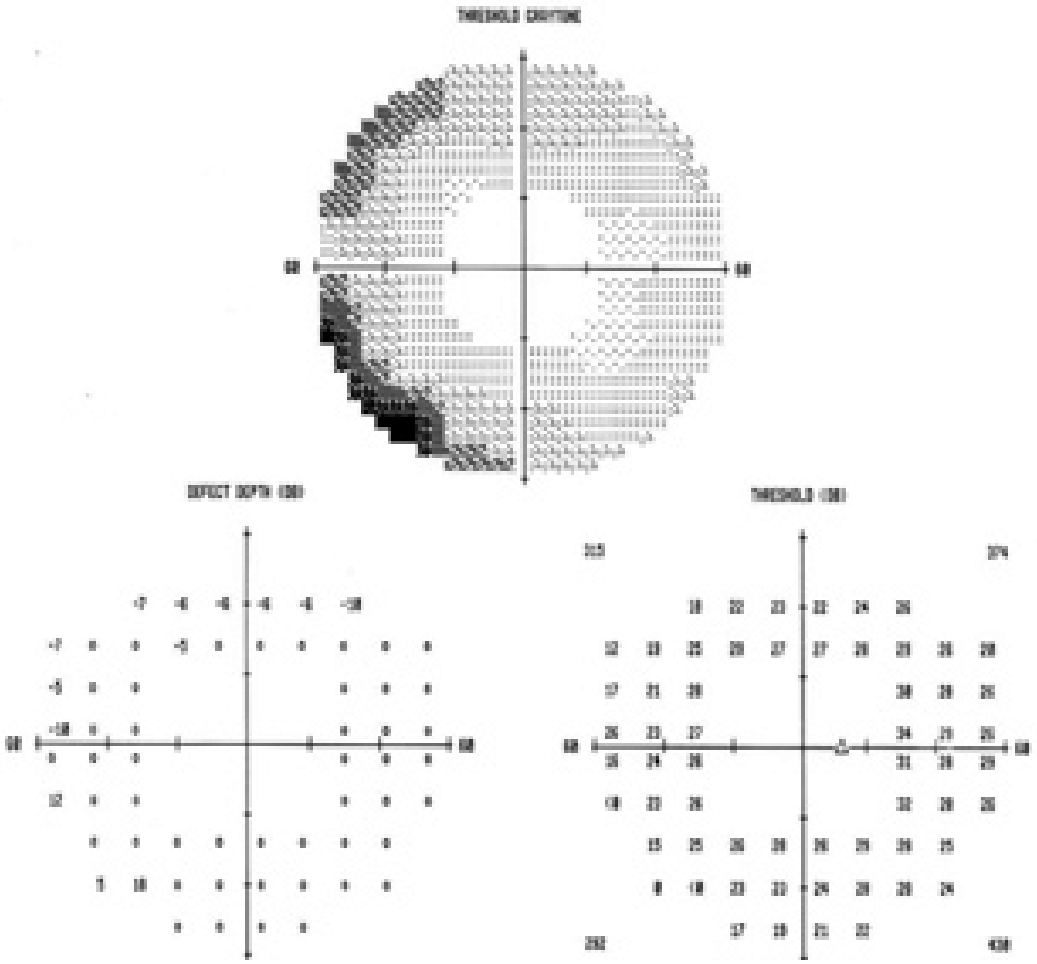
Does schisis result in a relative, or absolute scotoma?
Absolute

What HVF protocol is needed?
60-4 (I know, I've never seen one either)

Rod-
└ R

ny

Inner retinal dystrophy
└ X-linked retinoschisis



HVF 60-4, normal (I think)

By what other (very similar) name is X-linked retinoschisis (XLR) known?
X-linked juvenile retinoschisis

How does it present on DFE?
With **macular schisis** in a radial pattern, +/- **peripheral schisis**

What layer(s) of the
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All of

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About

Previous slide notwithstanding, photographic documentation of
ut there.) What's the
peripheral schisis?

cotoma?

In addition to OCT and HVF, there's a psychophysical test that's very useful in diagnosing XLR—what is it?

Rod-
R

ny

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In one sentence, what is it?

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In one sentence, what is it?
An [redacted] test that measures how [redacted] cells respond to a [redacted] stimulus

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The pt is dilated vs undilated, and usually light- vs dark--adapted

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What are the three main types of ERG?
Full-field (ffERG, aka German word ERG), **multifocal** (mfERG), and **pattern** (pERG)

ny

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What is the classic ERG finding in XLR?

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Inner retinal dystrophy
└ X-linked retinoschisis

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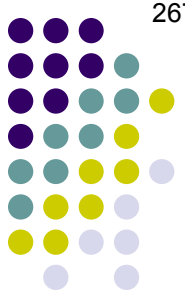
Rod-

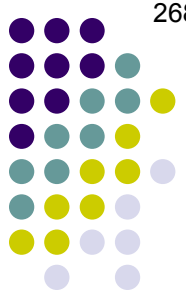
Before we answer this, let's get familiar with the* normal *ffERG

Inner retinal dystrophy
└ X-linked retinoschisis



Normal ffERG

The normal ffERG. Note:



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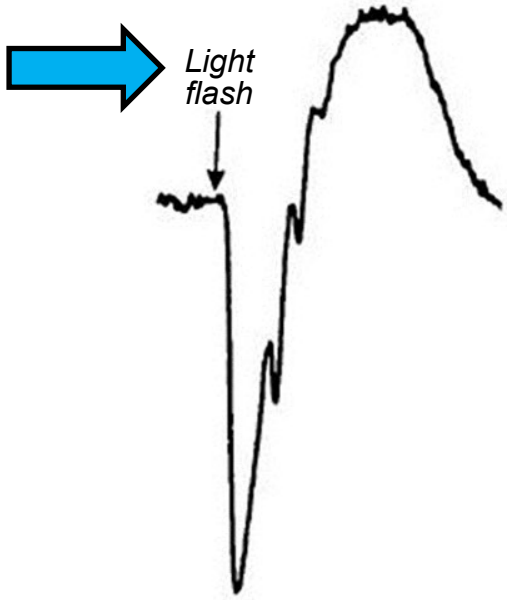
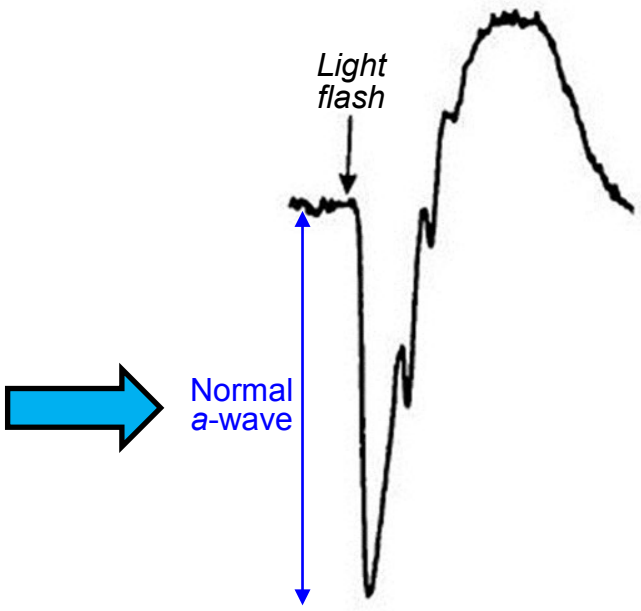


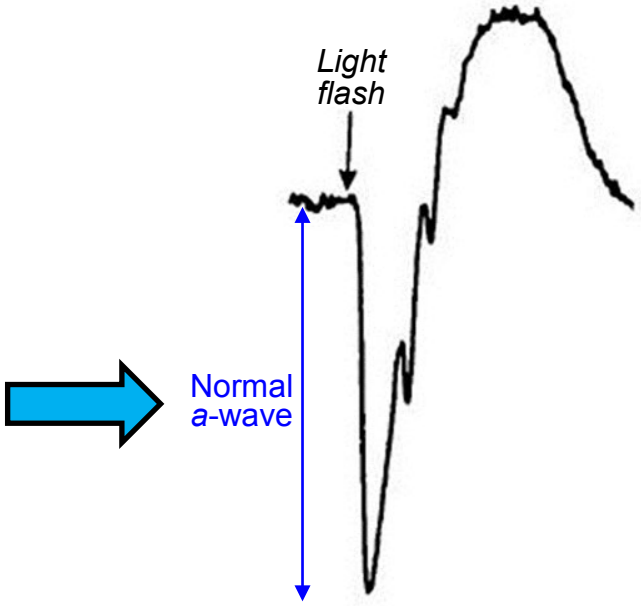
The normal ffERG. Note:
--The response commences with the stimulus flash

Normal ffERG



The normal ffERG. Note:
--The response commences with the stimulus flash
--The first deflection—the ***a*-wave**—is sharply downward.

Normal ffERG



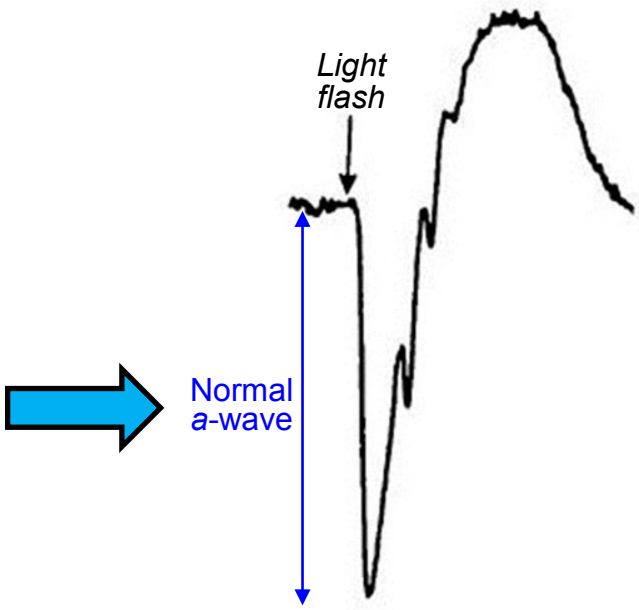
The normal ffERG. Note:

--The response commences with the stimulus flash

--The first deflection—the ***a*-wave**—is sharply downward.

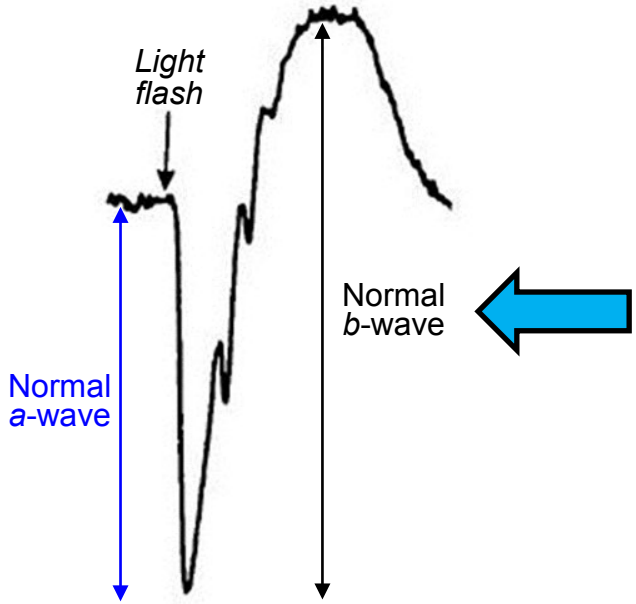
The *a*-wave represents [redacted] function

Normal ffERG



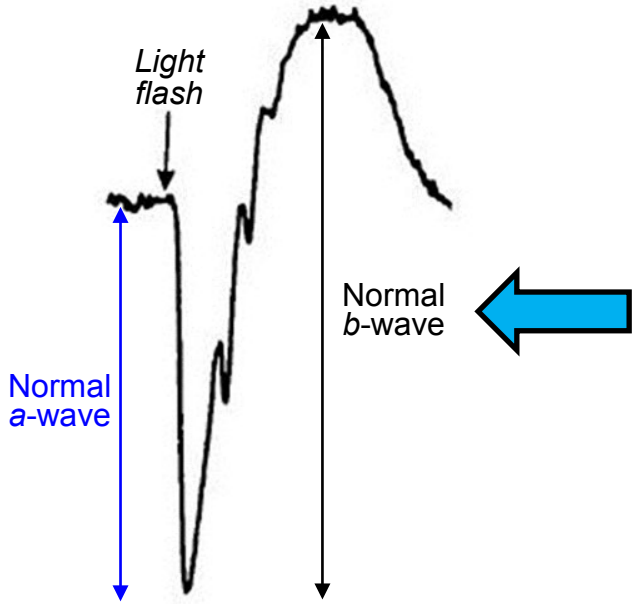
The normal ffERG. Note:
--The response commences with the stimulus flash
--The first deflection—the ***a*-wave**—is sharply downward.
The *a*-wave represents photoreceptor function

Normal ffERG



The normal ffERG. Note:
--The response commences with the stimulus flash
--The first deflection—the ***a*-wave**—is sharply downward.
The *a*-wave represents photoreceptor function
--The second deflection—the ***b*-wave**—is upward. While not quite as steep as the *a*-wave, it is of substantially greater amplitude.

Normal ffERG



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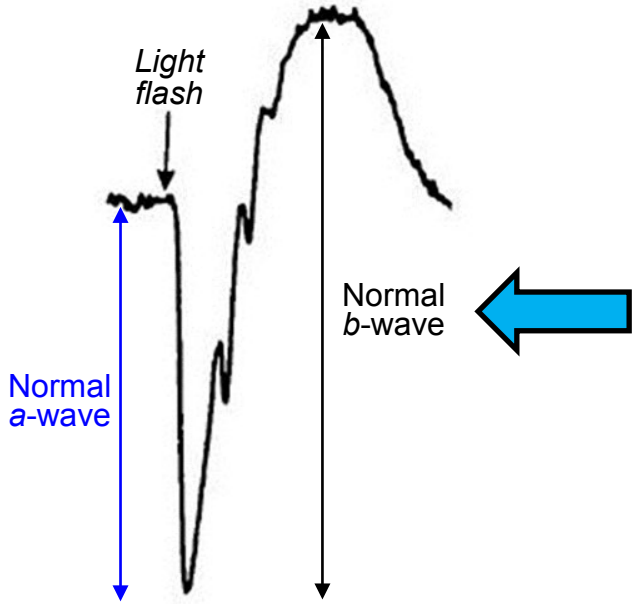
The *b*-wave represents function of the two words

Normal ffERG



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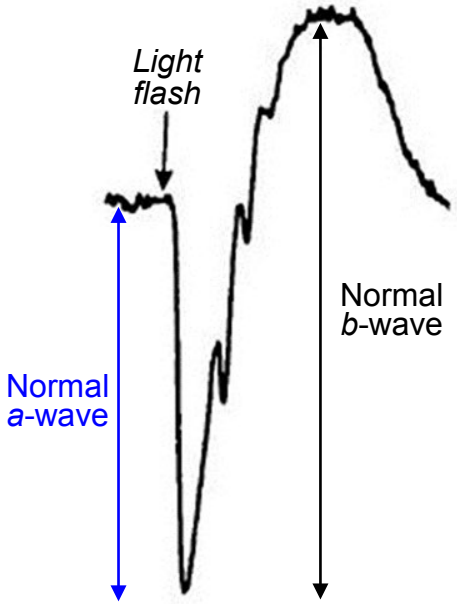
The *b*-wave represents function of the inner retina

Normal ffERG



Note: This is a significant oversimplification of ERG interpretation, That said, I *think* it's enough to get you through the OKAP and Boards. But caveat emptor, bro.

The normal ffERG. Note:

--The response commences with the stimulus flash

--The first deflection—the ***a*-wave**—is sharply downward.

The *a*-wave represents photoreceptor function

--The second deflection—the ***b*-wave**—is upward. While not quite as steep as the *a*-wave, it is of substantially greater amplitude.

The *b*-wave represents function of the inner retina

By what other (very similar) name is X-linked retinoschisis (XLR) known?
X-linked juvenile retinoschisis

How does it present on DFE?
With **macular schisis** in a radial pattern, +/- **peripheral schisis**

What layer(s) of the
Mainl

What
All of

What
About

Previous slide notwithstanding, photographic documentation of
ut there.) What's the
peripheral schisis?

In addition to OCT and HVF, there's a psychophysical test that's very useful in diagnosing XLR—what is it?
ERG

What is the classic ERG finding in XLR?

***Now** we're ready to answer this question*

Rod-
└ R

ny

Inner retinal dystrophy
└ X-linked retinoschisis

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Loss of the *a-* vs *b-*wave with preservation of the *a-* vs *b-*wave

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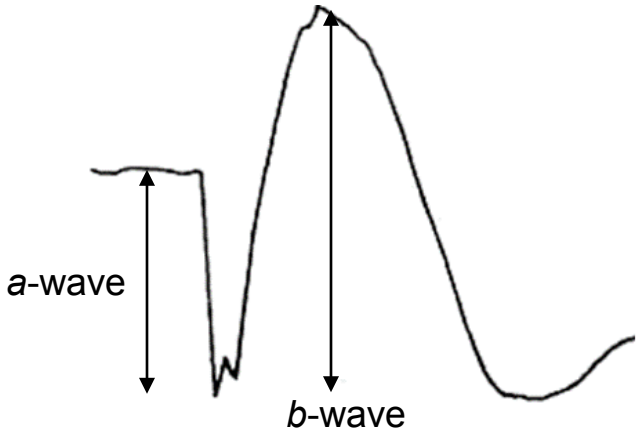
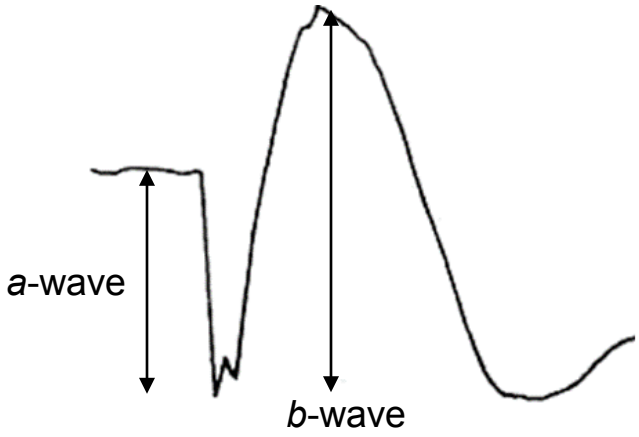
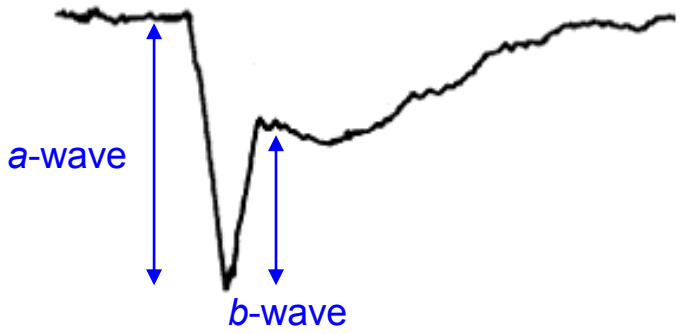


In a normal full-field ERG, the *b*-wave is substantially larger than the *a*-wave

Normal ERG



ERG in XLJR



However, in XLJR the *b*-wave is substantially **smaller** than the *a*-wave, which is preserved in size

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Loss of the *b*-wave with preservation of the *a*-wave

What is the term for an ERG in which the a*-wave is preserved but the* b*-wave is diminished?*

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How does it present on DFE?
With **macular schisis** in a radial pattern, +/- **peripheral schisis**

What layer(s) of the
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What
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Previous slide notwithstanding, photographic documentation of
ut there.) What's the
peripheral schisis?

cotoma?

In addition to OCT and HVF, there's a psychophysical test that's very useful in diagnosing XLR—what is it?
ERG

What is the classic ERG finding in XLR?
Loss of the *b*-wave with preservation of the *a*-wave

What is the term for an ERG in which the a*-wave is preserved but the* b*-wave is diminished?*
Such an ERG is said to demonstrate a 'negative' or 'electronegative' waveform

Rod-
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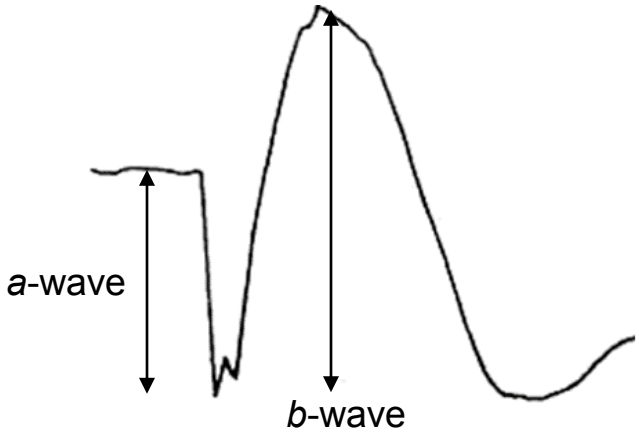
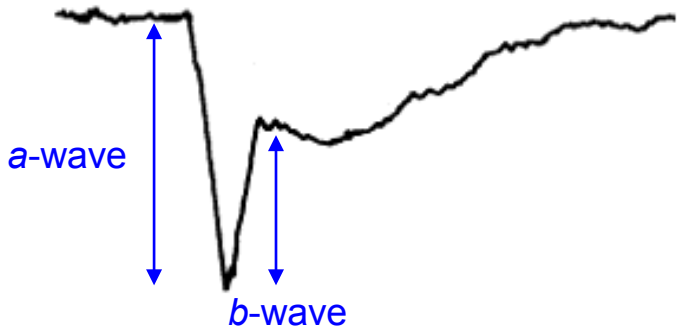
Inner retinal dystrophy
X-linked retinoschisis

Normal ERG



ERG in XLJR



the *b*-wave is substantially **smaller** than the *a*-wave, which is preserved in size

This constitutes a 'negative' or ('electronegative') ERG

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No, several conditions can cause it

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We mentioned CNSB previously. In a nutshell, what is it?

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cotoma?

We mentioned CNSB previously. In a nutshell, what is it?
A congenital condition in which a dearth of functioning [rods vs cones] leads to [symptom], [sign], and variably decreased [abb.]

a-wave

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A congenital condition in which a dearth of functioning rods leads to nyctalopia , nystagmus , and variably decreased VA

Inner retinal dystrophy

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For more on CSNB, see slide-set R9

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phy

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Rod-
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In short, retinoschisin is mission-critical to the structural integrity of the retina.

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RS1

Inner retinal dystrophy
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What ocular structure—unmentioned previously in this set—is affected by XLR?

Inner retinal dystrophy
└ X-linked retinoschisis

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What ocular structure—unmentioned previously in this set—is affected by XLR?
The vitreous—it is syneretic , and contains [redacted] and other opacities

Inner retinal dystrophy
└ X-linked retinoschisis

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Inner retinal dystrophy
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How does it present on DFE?
With macular schisis in a radial pattern, +/- peripheral schisis

How does it present clinically?

Recall that we mentioned previously that the *Peds* book classifies XL(J)R as a hereditary vitreoretinopathy, and that we would see why later.

Defective function of the *retinoschisin* protein

What ocular structure—unmentioned previously in this set—is affected by XLR?
The vitreous—it is syneretic , and contains veils and other opacities

Note: There is some disagreement within the *BCSC* regarding how X-linked retinoschisis should be classified. Whereas the *Retina* book calls it an *inner retinal dystrophy*…**the *Peds* book classifies it as a *hereditary vitreoretinopathy****. Caveat emptor.

Inner retinal dystrophy
└ X-linked retinoschisis

*As to *why* they do this, we will see shortly

By what other (very similar) name is X-linked retinoschisis (XLR) known?
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How does it present on DFE?
With macular schisis in a radial pattern, +/- peripheral schisis

How does it present clinically?

Recall that we mentioned previously that the *Peds* book classifies XL(J)R as a hereditary vitreoretinopathy, and that we would see why later. ***This is the reason why!***

Defective function of the *retinoschisin* protein

What ocular structure—as mentioned previously in this set—is affected by XLR?
The vitreous—it is syneretic , and contains veils and other opacities

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With modestly decreased VA in childhood. Over time, VA will drop to 20/200 or so.

What is the underlying pathophysiology?
Defective function of the *retinoschisin* protein

What ocular structure—unmentioned previously in this set—is affected by XLR?
The vitreous—it is syneretic , and contains ?^ veils and other opacities

Another, more ominous vitreous finding is common as well—what is it?

Inner retinal dystrophy

└ X-linked retinoschisis

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Inner retinal dystrophy
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What is the mechanism by which vitreous hemorrhage occurs?

Inner retinal dystrophy

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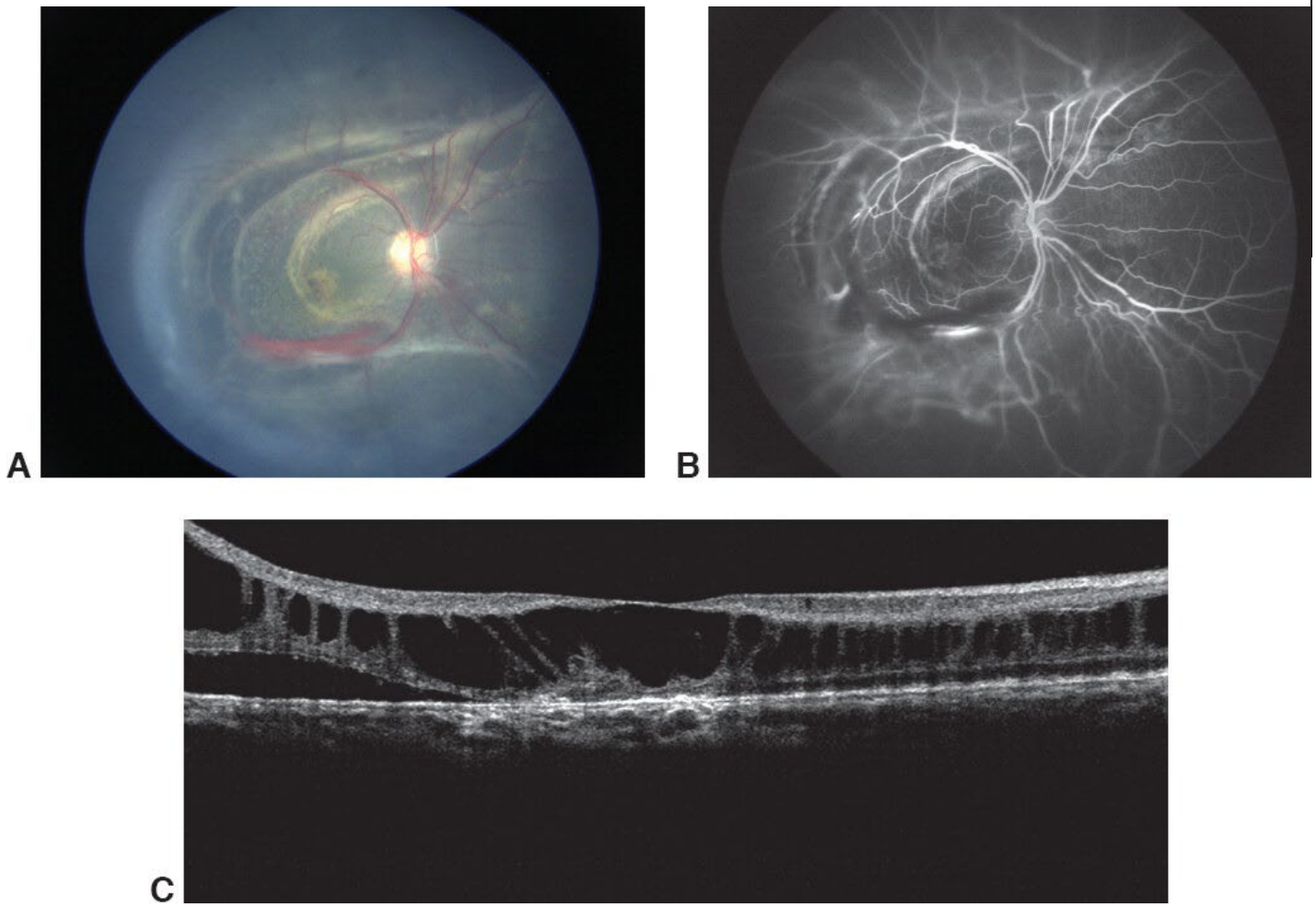
What is the mechanism by which vitreous hemorrhage occurs?
Schisis within the NFL can leave retinal vessels essentially unsupported, making them highly vulnerable to rupturing

Inner retinal dystrophy

- X-linked retinoschisis



Three-year-old with XLR. A) Central and extensive peripheral retinoschisis with mild vitreous and preretinal hemorrhage. There is a large circumferential TRD around the arcades, encroaching on the central macula. A retinal hole is also present. B) Fluorescein angiography reveals an extensive tractional detachment. C) Macular OCT image demonstrates retinoschisis with a TRD and SRF encroaching on the fovea.

X-linked retinoschisis

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Pts must adhere to a specific lifestyle modification—what is it?

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No contact sports—XLR retinas are highly susceptible to trauma-induced RD

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The chronic presence of fluid within macular schisis cavities is felt to contribute to long-term visual morbidity in XLR. What simple intervention may ameliorate this?

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Diamox (PO and/or topical)

Inner retinal dystrophy

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