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.
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.
*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.)
What are the hereditary macular dystrophies covered in the Retina book?
Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular

One of two with macular in its name
This one is the best
The star of the mac dys's
A group of non-lookalike conditions
Looks like GA
The other with macular in its name

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

What are the hereditary macular dystrophies covered in the Retina book?
What is the classic (and unfortunate) manifestation of Sorsby’s?
What is the classic (and unfortunate) manifestation of Sorsby’s?
Bilateral CNVMs occurring at around age 40.

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

How does Best dz present?

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

Diffuse

Macular

- Sorsby macular dystrophy
- Best dz
- Stargardt
- Pattern dystrophies
- Central areolar choroidal dystrophy
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Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular

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

- Sorsby macular dystrophy
- Best dz
- Stargardt
- Pattern dystrophies
- Central areolar choroidal dystrophy
- North Carolina macular dystrophy
How does Best dz present?
With a vitelliform lesion (or lesions) during childhood (the condition is aka Best vitelliform dystrophy)

What does vitelliform mean?
It means 'egg-yolk-like'
Because they’re egg-yolk-like, what can you infer about a vitelliform lesion’s…
…color? Yellow
…shape? Round
…contour? Domed

Hereditary Retinal/Choroidal Dystrophies

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

Macular
Diffuse Retinal/Choroidal Dystrophies

Macular

Sorsby macular dystrophy

Best dz

Stargardt

Pattern dystrophies

Central areolar choroidal dystrophy

North Carolina macular dystrophy

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How does Best dz present?
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Diffuse Retinal/Choroidal Dystrophies

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…color?

Macular

Sorsby macular dystrophy

Best dz

Stargardt

Pattern dystrophies

Central areolar choroidal dystrophy

North Carolina macular dystrophy

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

Hereditary Retinal/Choroidal Dystrophies

Macular

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

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Hereditary Retinal/Choroidal Dystrophies

Diffuse

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Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular

- Sorsby macular dystrophy
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- Stargardt
- Pattern dystrophies
- Central areolar choroidal dystrophy
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Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular

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

Retinal/Choroidal Dystrophies

Hereditary

Macular

What does vitelliform mean?
It means ‘egg-yolk-like’

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

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

- Sorsby macular dystrophy
- Best dz
- Stargardt
- Pattern dystrophies
- Central areolar choroidal dystrophy
- North Carolina macular dystrophy
Best dz: Vitelliform lesion
How does Best dz present?
With a vitelliform lesion (or lesions) during childhood (the condition is aka *Best vitelliform dystrophy*)

*What psychophysical test is abnormal in Best dz?*
How does Best dz present?
With a vitelliform lesion (or lesions) during childhood (the condition is aka Best vitelliform dystrophy)

What psychophysical test is abnormal in Best dz?
The electro-oculogram (EOG)
How does Best dz present?
With a vitelliform lesion (or lesions) during childhood (the condition is aka Best vitelliform dystrophy)

What psychophysical test is abnormal in Best dz?
The electro-oculogram (EOG)

In a nutshell, what does an EOG measure?
How does Best dz present?
With a vitelliform lesion (or lesions) during childhood (the condition is aka Best vitelliform dystrophy)

What psychophysical test is abnormal in Best dz?
The electro-oculogram (EOG)

In a nutshell, what does an EOG measure?
RPE function

Sorsby macular dystrophy
Best dz
Stargardt
Pattern dystrophies
Central areolar choroidal dystrophy
North Carolina macular dystrophy
How does Best dz present?
With a vitelliform lesion (or lesions) during childhood (the condition is aka Best vitelliform dystrophy)

What psychophysical test is abnormal in Best dz?
The electro-oculogram (EOG)

In a nutshell, what does an EOG measure?
RPE function

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

Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular

- Sorsby macular dystrophy
- Best dz
- Stargardt
- Pattern dystrophies
- Central areolar choroidal dystrophy
- North Carolina macular dystrophy
How does Best dz present?
With a vitelliform lesion (or lesions) during childhood (the condition is aka *Best vitelliform dystrophy*).

*What psychophysical test is abnormal in Best dz?*
The electro-oculogram (EOG).

*In a nutshell, what does an EOG measure?*
RPE function.

*The results of EOG testing are reported in terms of a ratio. What is its eponymous name?*
The *Arden ratio*.
Other than its name, what makes Stargardt’s the ‘star of the macular dystrophies’?

The star of the mac dys’s

- Sorsby macular dystrophy
- Best dz
- Stargardt
- Pattern dystrophies
- Central areolar choroidal dystrophy
- North Carolina macular dystrophy
Other than its name, what makes Stargardt’s the ‘star of the macular dystrophies’?
It is the most common

*The star of the mac dys’s*
Other than its name, what makes Stargardt’s the ‘star of the macular dystrophies’?
It is the most common

What is the classic finding on DFE?
Other than its name, what makes Stargardt’s the ‘star of the macular dystrophies’?
It is the most common

What is the classic finding on DFE?
(pisciform—fish-shaped) lesions in the macula
Other than its name, what makes Stargardt’s the ‘star of the macular dystrophies’?
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
Other than its name, what makes Stargardt’s the ‘star of the macular dystrophies’? It is the most common.

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

What is the classic finding on fluorescein angiography?
Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular

Other than its name, what makes Stargardt’s the ‘star of the macular dystrophies’?
It is the most common

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
Diffuse

Retinal/Choroidal
Dystrophies

Hereditary

Macular

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’

What is the classic finding on autofluorescence imaging?

Other than its name, what makes Stargardt’s the ‘star of the macular dystrophies’?
It is the most common

What is the classic finding on fluorescein angiography?
‘Dark choroid’

Sorsby macular dystrophy
Best dz
Stargardt
Pattern dystrophies
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Other than its name, what makes Stargardt’s the ‘star of the macular dystrophies’? It is the most common

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’

What is the classic finding on autofluorescence imaging? A ring of perifoveal hyperautofluorescence surrounding a central foveal area of hypoautofluorescence
Stargardt—hyper/hypopigmentation on FAF
Diffuse

Macular

Hereditary Retinal/Choroidal Dystrophies

Briefly, what is a pattern dystrophy?

- Sorsby macular dystrophy
- Best dz
- Stargardt
- **Pattern dystrophies**
  - Central areolar choroidal dystrophy
  - North Carolina macular dystrophy
Briefly, what is a pattern dystrophy?
An inherited macular dystrophy that has a characteristic appearance (i.e., a particular 'pattern')
Diffuse

Retinal/Choroidal Dystrophies

Hereditary

Macular

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

The Retina book identifies four pattern dystrophies—what are they?

--?

--?

With apologies, the mnemonic is…

--?

--?
Briefly, what is a pattern dystrophy?
An inherited macular dystrophy that has a characteristic appearance (ie, a particular ‘pattern’)

The Retina book identifies four pattern dystrophies—what are they?

--B

--A  With apologies, the mnemonic is…BARF

--R

--F

Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular

Sorsby macular dystrophy

Best dz

Stargardt

Pattern dystrophies

Central areolar choroidal dystrophy

North Carolina macular dystrophy
Diffuse

Hereditary Retinal/Choroidal Dystrophies

Macular

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

The Retina book identifies four pattern dystrophies—what are they?
--Butterfly dystrophy
--Adult-onset foveomacular vitelliform dystrophy
--Reticular dystrophy
--Fundus pulverulentus

--Sorsby macular dystrophy
--Best dz
--Stargardt

Pattern dystrophies
--Central areolar choroidal dystrophy
--North Carolina macular dystrophy
Briefly, what is a pattern dystrophy?
An inherited macular dystrophy that has a characteristic appearance (ie, a particular ‘pattern’)

*The Retina book identifies four pattern dystrophies—what are they?*
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---Fundus pulverulentus

Are pattern dystrophies associated with severe vision loss?
**Briefly, what is a pattern dystrophy?**
An inherited macular dystrophy that has a characteristic appearance (ie, a particular ‘pattern’)

*The Retina book identifies four pattern dystrophies—what are they?*

--Butterfly dystrophy
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*Are pattern dystrophies associated with severe vision loss?*
Generally no—vision is only mildly affected
Briefly, what is a pattern dystrophy?
An inherited macular dystrophy that has a characteristic appearance (ie, a particular ‘pattern’)

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Generally no—vision is only mildly affected

Do the macular ‘patterns’ appear early in life?
Briefly, what is a pattern dystrophy?
An inherited macular dystrophy that has a characteristic appearance (ie, a particular ‘pattern’)

*The Retina* book identifies four pattern dystrophies—what are they?
--Butterfly dystrophy
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Generally no—vision is only mildly affected

Do the macular ‘patterns’ appear early in life?
Generally no—they usually show up in middle adulthood
Briefly, what is a pattern dystrophy?
An inherited macular dystrophy that has a characteristic appearance (ie, a particular ‘pattern’)

The Retina book identifies four pattern dystrophies—what are they?
- Butterfly dystrophy
- Adult-onset foveomacular vitelliform dystrophy
- Reticular dystrophy
- Fundus pulverulentus

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

Do the macular ‘patterns’ appear early in life?
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.

--Butterfly dystrophy
--Adult-onset foveomacular vitelliform dystrophy
--Reticular dystrophy
--Fundus pulverulentus
FAF demonstrates a butterfly-shaped pattern in the macula OU. Hypofluorescent areas corresponded to pigment clumping seen on fundoscopy.

--Butterfly dystrophy
--Adult-onset foveomacular vitelliform dystrophy
--Reticular dystrophy
--Fundus pulverulentus
Briefly, what is a pattern dystrophy?
An inherited macular dystrophy that has a characteristic appearance (ie, a particular ‘pattern’)

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--Butterfly dystrophy
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Are the vitelliform lesions of AOFVD similar to those of Best dz?

Pattern dystrophies
--Sorsby macular dystrophy
--Best dz
--Stargardt
--Central areolar choroidal dystrophy
--North Carolina macular dystrophy
Briefly, what is a pattern dystrophy?
An inherited macular dystrophy that has a characteristic appearance (ie, a particular ‘pattern’)

The Retina book identifies four pattern dystrophies—what are they?
--Butterfly dystrophy
--Adult-onset foveomacular vitelliform dystrophy

*Are the vitelliform lesions of AOFVD similar to those of Best dz?*
Yes, although they tend to be somewhat smaller vs larger

Pattern dystrophies

- Sorsby macular dystrophy
- Best dz
- Stargardt
- Central areolar choroidal dystrophy
- North Carolina macular dystrophy
Briefly, what is a pattern dystrophy?
An inherited macular dystrophy that has a characteristic appearance (ie, a particular ‘pattern’)

The Retina book identifies four pattern dystrophies—what are they?
--Butterfly dystrophy
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Are the vitelliform lesions of AOFVD similar to those of Best dz?
Yes, although they tend to be somewhat smaller
Typical vitelliform lesion of AOFVD

Best dz lesion for comparison

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

--Butterfly dystrophy
--Adult-onset foveomacular vitelliform dystrophy
--Reticular dystrophy
--Fundus pulverulentus
Diffuse Retinal/Choroidal Dystrophies

Macular

Breitly, what is a pattern dystrophy?
An inherited macular dystrophy that has a characteristic appearance (ie, a particular ‘pattern’)

The Retina book identifies four pattern dystrophies—what are they?
---Butterfly dystrophy
---Adult-onset foveomacular vitelliform dystrophy

Are the vitelliform lesions of AOFVD similar to those of Best dz?
Yes, although they tend to be somewhat smaller

Do Best dz and AOFVD share a common genetic heritage?
Briefly, what is a pattern dystrophy?
An inherited macular dystrophy that has a characteristic appearance (ie, a particular ‘pattern’)

The Retina book identifies four pattern dystrophies—what are they?
--Butterfly dystrophy
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Are the vitelliform lesions of AOFVD similar to those of Best dz? 
Yes, although they tend to be somewhat smaller

Do Best dz and AOFVD share a common genetic heritage? 
You’d think so, but no—they’re genetically unrelated
Briefly, what is a pattern dystrophy?
An inherited macular dystrophy that has a characteristic appearance (ie, a particular ‘pattern’)

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Do Best dz and AOFVD share a common genetic heritage?
You’d think so, but no—they’re genetically unrelated

Is the EOG abnormal in AOFVD, like it is in Best dz?
Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular

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Yes, although they tend to be somewhat smaller

Do Best dz and AOFVD share a common genetic heritage?
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
Briefly, what is a pattern dystrophy?
An inherited macular dystrophy that has a characteristic appearance (ie, a particular ‘pattern’)

_The Retina book identifies four pattern dystrophies—what are they?
--Butterfly dystrophy
--Adult-onset foveomacular vitelliform dystrophy
--Reticular dystrophy
--Fundus pulverulentus

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.

--Butterfly dystrophy
--Adult-onset foveomacular vitelliform dystrophy
--Reticular dystrophy
--Fundus pulverulentus
Symmetrical and reticular pattern of accumulated pigment in the fovea that radiates from the center to the periphery like a “fishnet with knots.”

--Butterfly dystrophy
--Adult-onset foveomacular vitelliform dystrophy
--Reticular dystrophy
--Fundus pulverulentus
--Butterfly dystrophy
--Adult-onset foveomacular vitelliform dystrophy
--Reticular dystrophy
--Fundus pulverulentus

A pic of fundus pulverulentus, just because
How does central areolar choroidal dystrophy present?
**Hereditary Retinal/Choroidal Dystrophies**

- **Diffuse**
- **Macular**

*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)
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
Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular

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. Given that it looks like GA, why not call it Geographic atrophy choroidal dystrophy?
Diffuse

Hereditary Retinal/Choroidal Dystrophies

Macular

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. **Given that it looks like GA, why not call it Geographic atrophy choroidal dystrophy?**

it is, sort of. What we now call **geographic atrophy** was formerly known as **central areolar atrophy**.
How does central areolar choroidal dystrophy present? With well-circumscribed, bilateral and symmetric areas of choroidal and retinal atrophy

Is it common, or rare?
How does central areolar choroidal dystrophy present?
With well-circumscribed, bilateral and symmetric areas of choroidal and retinal atrophy

Is it common, or rare?
Quite rare
**Diffuse**

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

Is it common, or rare?
Quite rare

At what age are the changes first discernable?

**Macular**

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

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

Is it common, or rare?
Quite rare

At what age are the changes first discernable?
Between 20 and 40 or so
How does central areolar choroidal dystrophy present?
With well-circumscribed, bilateral and symmetric areas of choroidal and retinal atrophy

Is it common, or rare?
Quite rare

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Between 20 and 40 or so

Is the visual prognosis good, like in the pattern dystrophies?
How does central areolar choroidal dystrophy present?
With well-circumscribed, bilateral and symmetric areas of choroidal and retinal atrophy

Is it common, or rare?
Quite rare

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

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 common, or rare?
Quite rare

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

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

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 in pts with CACD because it is inherited in AD fashion)
- The lack of drusen in the macula of CACD pts
How does central areolar choroidal dystrophy present?
With well-circumscribed, bilateral and symmetric areas of choroidal and retinal atrophy.

Is it common, or rare?
Quite rare.

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

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.

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Diffuse Retinal/Choroidal Dystrophies

Hereditary 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 common, or rare?
Quite rare

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

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

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Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular

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

Is it common, or rare?
Quite rare

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Between 20 and 40 or so

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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)
--Family hx (will be stronger in pts with CACD because it is inherited in AD fashion)
Diffuse

Hereditary Retinal/Choroidal Dystrophies

Macular

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

Is it common, or rare?
Quite rare.

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

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.

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 in pts with CACD because it is inherited in AD fashion)

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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 common, or rare?
Quite rare.

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

Is the visual prognosis good, like in the pattern dystrophies?
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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 drusen in the macula of CACD pts.

Central areolar choroidal dystrophy

Sorsby macular dystrophy

Best dz

Stargardt

Pattern dystrophies

North Carolina macular dystrophy
Hereditary Retinal/Choroidal Dystrophies

**Diffuse**

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

**Macular**

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

- Is it common or rare?
  Quite rare

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

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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)
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- The lack of drusen in the macula of CACD pts
North Carolina macular dystrophy is called (by the Retina book) an “atypical” macular dystrophy. What makes it atypical?
Diffuse Retinal/Choroidal Dystrophies

Hereditary Macular

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

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?
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
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)

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

Do NCMD pts tend to have good vision, or poor?
Good, often surprisingly so given the appearance of their macula
There are three subtypes of diffuse dystrophies—what are they?
There are three subtypes of diffuse dystrophies—what are they?

- Rod-dominant
- Cone-dominant
- Choroidal

Hereditary Retinal/Choroidal Dystrophies

- 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.
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.
What is the catch-all name for the rod-dominant dystrophies?
What is the catch-all name for the rod-dominant dystrophies? The ‘rod-cone dystrophies’
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?
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
- North Carolina macular dystrophy

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?
Retinitis pigmentosa (RP)
What are the three cone-dominant conditions highlighted in the Retina book?
What are the three cone-dominant conditions highlighted in the Retina book?
Hereditary Retinal/Choroidal Dystrophies

Diffuse

Rod-dominant

Rod-cone (RP)

Cone-dominant

Cone/cone-rod

Type of ‘blindness’ produced

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

Macular

Nyctalopia

Type of ‘blindness’ produced

Hemeralopia

Enhanced S-cone dz

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

Nystagmus

Type of ‘blindness’ produced

Hemeralopia

What is…

--nystagmus?

--hemeralopia?
What is…
--nyctalopia? Night blindness
--hemeralopia? Day blindness
Hereditary Retinal/Choroidal Dystrophies

Diffuse

Rod-dominant

Cone-dominant

Rod-cone (RP)

Cone/cone-rod

Nyctalopia

Type of ‘blindness’ produced

Hemeralopia

? Chief visual defect(s)

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

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
Diffuse Choroidal Cone-dominant LCA Enhanced S-cone dz
Rod-dominant Cone/cone-rod Rod-cone (RP)

This makes sense, as rods dominate the retinal periphery

Hereditary Retinal/Choroidal Dystrophies

VF loss

Chief visual defect(s)

Acuity, and color loss

No question—proceed when ready
Diffuse

Hereditary Retinal/Choroidal Dystrophies

Diffuse

Rod-dominant

- Rod-cone (RP)

Cone-dominant

- Cone/cone-rod

Hereditary Retinal/Choroidal Dystrophies

Rods

- Rod-dominant
  - Rod-cone (RP)

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

Cone-dominant

- Cone-cone-rod

- Macular
  - Sorsby macular dystrophy
  - Best dz
  - Stargardt
  - 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

Retinal/Choroidal Dystrophies

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

Chief visual defect(s)
- Acuity, and color loss
- ?

Type of ‘blindness’ produced
- ?

Nyctalopia
- ?

VF loss
- c/o photophobia?

Hemeralopia
- ?
Hereditary Retinal/Choroidal Dystrophies

Diffuse

Rod-dominant

Rod-cone (RP)

Cone-dominant

Cone/cone-rod

rod-dominant

Nystagmus

Type of ‘blindness’ produced

Hemeralopia

VF loss

Chief visual defect(s)

Acuity, and color loss

No

c/o photophobia?

Yes

Macular

Sorsby macular dystrophy

Best dz

Stargardt

Pattern dystrophies

Central areolar choroidal dystrophy

North Carolina macular dystrophy
Diffuse Choroidal Cone-dominant LCA Enhanced S-cone dz

Rod-dominant Cone/cone-rod Rod-cone (RP)

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

Hereditary Retinal/Choroidal Dystrophies

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)
- Choroidal
- Cone-dominant
- Cone/cone-rod

Macular
- Sorsby macular dystrophy
- Best dz
- Stargardt
- Pattern dystrophies

What does LCA stand for in this context?

Leber’s congenital amaurosis

Profoundly poor VA from birth
Wandering nystagmus from birth
Pts engage in eye rubbing behavior
What does LCA stand for in this context?
Leber's congenital amaurosis
Macular Sorsby macular dystrophy Best dz Stargardt Central areolar choroidal dystrophy North Carolina macular dystrophy Pattern dystrophies

Diffuse Rod-dominant Rod-cone (RP) Choroidal Cone-dominant Cone/cone-rod

Hereditary Retinal/Choroidal Dystrophies

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

What are its three hallmark findings?
--?
--?
--?
Hereditary Retinal/Choroidal Dystrophies

Diffuse

- Rod-dominant
  - Rod-cone (RP)

- Cone-dominant
  - Cone/cone-rod

Macular

- Sorsby macular dystrophy
- Best dz
- Stargardt
- Pattern dystrophies

LCA

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

Diffuse

- Rod-dominant
  - Rod-cone (RP)

- Cone-dominant
  - Cone/cone-rod
    - LCA

Macular

- Sorsby macular dystrophy
- Best dz
- Stargardt
- Pattern dystrophies

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
By what eponymous name is enhanced S-cone dystrophy also known?
By what eponymous name is enhanced S-cone dystrophy also known?
Goldmann-Favre syndrome
Hereditary Retinal/Choroidal Dystrophies

Diffuse

- Rod-dominant
  - Rod-cone (RP)

- 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

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

...and night-blindness conditions?

Hol up—where’s all those classic inherited color-blindness conditions…
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.)

...and night-blindness conditions?

Hol up—where’s all those classic inherited color-blindness conditions...
To be sure, these are hereditary diseases of the retina and/or choroid. However, they are not dystrophies—rather, they are stationary conditions, i.e., they do not worsen over time. (One of them, congenital stationary night blindness, [CSNB], even has the word stationary in its name.) If we were to pull back our focus a little, we can see that the stationary conditions form a separate branch on the organizational tree of hereditary retinal/choroidal conditions. It is on this branch that these conditions belong.

...and night-blindness conditions?

Hol up—where’s all those classic inherited color-blindness conditions…
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.) If we were to pull back our focus a little, we can see that the stationary conditions form a separate branch on the organizational tree of hereditary retinal/choroidal conditions. It is on this branch that these conditions belong.

For more on these conditions, see slide-set R9

Hol up—where’s all those classic inherited color-blindness conditions…
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.) If we were to pull back our focus a little, we can see that the stationary conditions form a separate branch on the organizational tree of hereditary retinal/choroidal conditions. It is on this branch that these conditions belong.

(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)
OK, but what about the RP-lookalike conditions? They’re all inherited and progressive. Why aren’t they listed here?
OK, but what about the RP-lookalike conditions? They’re all inherited and progressive. Why aren’t they listed here?
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 our focus a little further still, we can see that the degenerations form yet another separate branch on the organizational tree of ‘hereditary retinal/choroidal conditions.’

OK, but what about the RP-lookalike conditions? They’re all inherited and progressive. Why aren’t they listed here?
For more on these conditions, see slide-set R5
The Retina book lists three choroidal dystrophies—what are they?
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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?
The Retina book lists three choroidal dystrophies—what are they?

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

‘Atrophy’
The Retina book lists three choroidal dystrophies—what are they?

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

'Atrophy'

These conditions share a common feature on one retinal-imaging modality. What is the modality, and what is the feature?
The Retina book lists three choroidal dystrophies—what are they?

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

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

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

‘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 hypoautofluorescent on fundus autofluorescence (FAF).
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 it present clinically?

With nyctalopia and paracentral scotomas in teens/young adults.
What is the full name of this condition?
Bietti crystalline dystrophy

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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 it present clinically?
With nyctalopia and paracentral scotomas in teens/young adults.
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 it present clinically? With nyctalopia and paracentral scotomas in teens/young adults.
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 and in the cornea near the limbus.

How does it present clinically?
With nyctalopia and paracentral scotomas in teens/young adults.

Pattern dystrophies
- Sorsby macular dystrophy
- Best disease
- Stargardt disease
- 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.

How does it present clinically?
With nyctalopia and paracentral scotomas in teens/young adults.
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?
**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 it present clinically?**
With **nyctalopia and paracentral scotomas in teen/young adult years**
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
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

Choroidal

Bietti

Gyrate

Choroideremia

Central areolar choroidal dystrophy
North Carolina macular dystrophy
Pattern dystrophies
Best dz
Stargardt central areolar choroidal dystrophy
Sorsby macular dystrophy
Retinal/Choroidal dystrophies
Hereditary macular dystrophy
Cone-rod (RP)
Cone-dominant choroideremia
Enhanced S-cone dz
**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.
Bietti crystalline dystrophy: Cornea
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 it present clinically?
With nyctalopia and paracentral scotomas in teen/young adult years
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 it present clinically?
With visual symptom and location scotomas
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 it present clinically?
With nyctalopia and paracentral scotomas
What is the full name of this condition?
Bietti crystalline dystrophy

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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 it present clinically?
With nyctalopia and paracentral scotomas in life period years
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 it present clinically?
With nyctalopia and paracentral scotomas in teen/young adult years
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 it present clinically?
With nyctalopia and paracentral scotomas in the teen/young adult years.

Is Bietti’s common, or rare?
Very rare.

Is it treatable?
No.
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 it present clinically? With nyctalopia and paracentral scotomas in teen/young adult years.

Is Bietti’s common, or rare? Very rare.
**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 it present clinically?**
With nyctalopia and paracentral scotomas in teen/young adult years.

**Is Bietti’s common, or rare?**
Very rare

**Is it treatable?**
No
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 it present clinically? With nyctalopia and paracentral scotomas in teenage adult years

Is Bietti's common, or rare? Very rare

Is it treatable? No

Pattern dystrophies

Central areolar choroidal dystrophy

North Carolina macular dystrophy

Retinal/Choroidal Dystrophies Hereditary Macular Sorsby macular dystrophy Best dz Stargardt 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. Over time these areas will coalesce, ultimately forming a scalloped perimeter around the posterior pole.

How does it present clinically?
Severe nyctalopia commencing in childhood

What is the underlying pathophysiology?
A defect in the gene coding for the ornithine aminotransferase enzyme leads to excess serum levels of ornithine.
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.

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What is the underlying pathophysiology?
A defect in the gene coding for the ornithine aminotransferase enzyme leads to excess serum levels of ornithine.
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?

Severe nyctalopia commencing in childhood

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A defect in the gene coding for the ornithine aminotransferase enzyme leads to excess serum levels of ornithine.
**What is the full name of this condition?**
Gyrate atrophy

**How does it present on DFE?**
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What is the full name of this condition?
Gyrate atrophy

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How does it present clinically?
Severe nyctalopia commencing in childhood

What is the underlying pathophysiology?
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Gyrate atrophy: Pavingstones
What is the full name of this condition?
Gyrate atrophy

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Severe nyctalopia commencing in childhood

What is the underlying pathophysiology?
A defect in the gene coding for the ornithine aminotransferase enzyme leads to excess serum levels of ornithine
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

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

What is the underlying pathophysiology?
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How does it present clinically?
Severe nyctalopia commencing in childhood, and progressive VF loss

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

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A defect in the gene coding for the *ornithine aminotransferase* enzyme leads to excess serum levels of ornithine

What lab test supports the dx?
Finding elevated levels of ornithine in the serum. Additionally, testing for the *OAT* gene defect is available.

Is gyrate treatable?
Yes, via restriction of dietary arginine (the metabolic precursor to ornithine). Vitamin B6 supplementation is helpful in some cases.
What is the full name of this condition?
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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.

What is the inheritance pattern for this gene?

X-linked 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.

Severe nyctalopia commencing in childhood, and progressive VF loss.

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What is the inheritance pattern for this gene?

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Choroideremia
FAF reveals the expected hypofluorescent lesions

Choroideremia
**How does choroideremia present on DFE?**
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**How does it present clinically?**

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**What is the inheritance pattern for this gene?**
X-linked.
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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 symptom commencing in childhood, and progressive VF loss.

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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**

Hol up: ‘Atrophic changes with a scalloped border and nyctalopia + VF loss’? That’s the description just given for gyrate. How are you supposed to tell these apart in the clinic?
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.

Hol up: ‘Atrophic changes with a scalloped border and nyctalopia + VF loss’? That’s the description just given for gyrate. How are you supposed to tell these apart in the clinic? 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.
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

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A defect in the gene coding for the geranylgeranyl transferase enzyme.
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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.
**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

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Severe nyctalopia commencing in childhood, and progressive VF loss

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What is the inheritance pattern for this gene?
X-linked.

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Choroidal

- Bietti
- Gyrate
- **Choroideremia**

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

Rod-dominant

- Rod-cone (RP)

Hereditary macular dystrophies
- Sorsby macular dystrophy
- Best dz
- Stargardt
- 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.

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

Pattern dystrophies
Central areolar choroidal dystrophy
North Carolina macular dystrophy
Recall the above foreshadowing from earlier in the set—it’s time to address it.

*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?*
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. **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
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.

What is the lone such condition discussed in the Retina book?
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? (We'll get to it at the end of the set.)

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**
Hereditary Retinal/Choroidal Dystrophies

- Diffuse
- Macular

Rod-dominant
- Rod-cone (RP)
- Cone-dominant
- Cone/cone-rod
- LCA

Cone-dominant
- Enhanced S-cone dz

Retinal/Choroidal Dystrophies

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

Scopier 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.

Inner retinal dystrophy
- X-linked retinoschisis
Hereditary Retinal/Choroidal Dystrophies

- Diffuse
- Macular
  - Sorsby macular dystrophy
  - Central areolar choroidal dystrophy
  - North Carolina macular dystrophy
- Cone-dominant
  - Cone/cone-rod LCA
- Enhanced S-cone dz

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

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.)
Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular

Rod-dominant

Choroidal

Cone-dominant

Rod-cone (RP)

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LCA

Enhanced S-cone dz

Retinal/Choroidal Dystrophies

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

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

*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.

(per the Peds book)
Hereditary Retinal/Choroidal Dystrophies

Diffuse

Rod-dominant

Choroidal

Cone-dominant

Cone/cone-rod

LCA

Enhanced S-cone dz

Macular

Sorsby macular dystrophy

Central areolar choroidal dystrophy

North Carolina macular dystrophy

Pattern dystrophies

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.

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

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.
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?

Inner retinal dystrophy

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

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

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

Inner retinal dystrophy

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

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?

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)

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

How does it present on DFE?

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
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
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?

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.

**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

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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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
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

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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? Diamox (PO and/or topical)

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

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Inner retinal dystrophy

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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)
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HVF 60-4, normal (I think)
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What is the classic ERG finding in XLR?
Loss of the **b-wave** with preservation of the **a-wave**

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What does ERG stand for? Electroretinogram (or electroretinography)

In one sentence, what is it? An **electrophysiologic** test that measures how **retinal** cells respond to a light stimulus

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

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

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.
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

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

Now we’re ready to answer this question.

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

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?
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)

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
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 patients manifest foveal schisis?
Almost all of them, essentially

What proportion of patients manifest peripheral schisis?
About half

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
In a normal full-field ERG, the $b$-wave is substantially larger than the $a$-wave.
However, in XLJR the $b$-wave is substantially **smaller** than the $a$-wave, which is preserved in size.
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

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**Inner retinal dystrophy**

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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.
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

Is a negative ERG pathognomonic for XLJR?
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All of them, essentially

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Is a negative ERG pathognomonic for XLJR?
No, several conditions can cause it. That said, the only other one likely to present in childhood is CSNB.
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

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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?

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

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**Is a negative ERG pathognomonic for XLJR?** No, several conditions can cause it

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

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**Inner retinal dystrophy**

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What does CSNB stand for in this context?
Congenital stationary night blindness

In a nutshell, what's CSNB?
A congenital condition in which a dearth of functioning rods leads to nyctalopia, nystagmus, and variably decreased VA.
By what other (very similar) name is X-linked retinoschisis (XLR) known?
X-linked juvenile retinoschisis

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

In a nutshell, what’s ERG?
A psychophysical test for evaluating retinal function by measuring electrical responses to light stimulation

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)

In addition to OCT and HVF, there’s a psychophysical test that’s very useful in diagnosing XLR—what is it?
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Inner retinal dystrophy
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What is the classic ERG finding in XLR?
Loss of the b-wave with preservation of the a-wave

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

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

What does CSNB stand for in this context?
Congenital stationary night blindness

In a nutshell, what’s CSNB?
A congenital condition in which a dearth of functioning rods leads to nyctalopia, nystagmus, and variably decreased rods vs cones

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Congenital stationary night blindness

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What does CSNB stand for in this context?
**Congenital stationary night blindness**

In a nutshell, what’s CSNB?
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What ocular structure—unmentioned previously in this set—is affected by XLR?
The vitreous—it is syneretic, and contains veils and other opacities

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

Pts must adhere to a specific lifestyle modification—what is it?
No contact sports—XLR retinas are highly susceptible to trauma

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?
**Diamox (PO and/or topical)**

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)

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?
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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

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

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All of them, essentially

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For more on CSNB, see slide-set R9
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

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

How does it present clinically?
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

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In a nutshell, what is retinoschisin?
An adhesion protein that plays an important role in Müller cell viability.

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

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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.

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