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—and 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?
What are the hereditary macular dystrophies covered in the Retina book?

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

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 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
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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
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
- North Carolina macular dystrophy
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)
How does Best dz present?
With a vitelliform lesion (or lesions) during childhood (the condition is also known as 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 → Macular

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

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
Diffuse Retinal/Choroidal Dystrophies

Macular

Hereditary

Sorsby macular dystrophy

Best dz

Stargardt

Pattern dystrophies

Central areolar choroidal dystrophy

North Carolina macular dystrophy

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Diffuse Retinal/Choroidal Dystrophies

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Diffuse

Hereditary Retinal/Choroidal Dystrophies

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Macular

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Hereditary Retinal/Choroidal Dystrophies

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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…
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…shape? Round
…contour? Domed

- 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)
**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*).

*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
Diffuse

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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?

Macular

Hereditary Retinal/Choroidal Dystrophies

- Sorsby macular dystrophy
- Best dz
- Stargardt
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Other than its name, what makes Stargardt’s the ‘star of the macular dystrophies’?

The star of the mac dystrophies

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

Hereditary Retinal/Choroidal Dystrophies

Macular

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It is the most common

The star of the mac dystrophies
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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?
(That means fish-shaped) lesions in the macula
Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular

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

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

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

Sorsby macular dystrophy
Best dz

Stargardt

Pattern dystrophies
Central areolar choroidal dystrophy
North Carolina macular dystrophy
Stargardt—*dark choroid* appearance on FA
**Diffuse**

**Hereditary Retinal/Choroidal Dystrophies**

**Macular**

- 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?*
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?
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A ring of perifoveal hypo-autofluorescence surrounding a central foveal area of hyper-autofluorescence
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A ring of perifoveal hyper-autofluorescence surrounding a central foveal area of hypo-autofluorescence
Stargardt—hyper/hypofluorescence on FAF
Briefly, what is a pattern dystrophy?
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An inherited macular dystrophy that has a characteristic appearance (ie, a particular ‘pattern’)
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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
--R
--F

With apologies, the mnemonic is…BARF
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
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’)

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---Butterfly dystrophy
---Adult-onset foveomacular vitelliform dystrophy
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**Are pattern dystrophies associated with severe vision loss?**
Briefly, what is a pattern dystrophy?
An inherited macular dystrophy that has a characteristic appearance (i.e., 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 mildly affected
Diffuse

Retinal/Choroidal Dystrophies

Macular

Hereditary

Sorsby macular dystrophy

Best dz

Stargardt

Pattern dystrophies

Central areolar choroidal dystrophy

North Carolina macular dystrophy

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Do the macular ‘patterns’ appear early in life?
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Are pattern dystrophies associated with severe vision loss?
Generally no—vision is only mildly affected

Do the macular ‘patterns’ appear early in life?
Generally no—they usually show up in middle adulthood
**Hereditary Retinal/Choroidal Dystrophies**

### Diffuse

### Macular

**Sorsby macular dystrophy**

**Best dz**

**Stargardt**

### Pattern dystrophies

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

**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

**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?
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Are the vitelliform lesions of AOFVD similar to those of Best dz?

- 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?
-- 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.
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

Pattern dystrophies
- Sorsby macular dystrophy
- Best dz
- Stargardt
- Central areolar choroidal dystrophy
- North Carolina macular dystrophy
Typical vitelliform lesion of AOFVD

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

--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’)

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

### Diffuse

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

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- Butterfly dystrophy
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<table>
<thead>
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<th>Pattern dystrophies</th>
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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
A pic of fundus pulverulentus, just because

--Butterfly dystrophy
--Adult-onset foveomacular vitelliform dystrophy
--Reticular dystrophy
--Fundus pulverulentus
How does central areolar choroidal dystrophy present?
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
Diffuse Retinal/Choroidal Dystrophies

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?
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‘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.
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?*
**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
Diffuse Retinal/Choroidal Dystrophies

 Macroneritic 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?
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?
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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?
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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
Diffuse Retinal/Choroidal 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.

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

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

Diffuse

Macular

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Hereditary Retinal/Choroidal Dystrophies

Diffuse

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

Macular

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

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Quite rare

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Diffuse

Hereditary Retinal/Choroidal Dystrophies

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 an AD fashion)
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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**Hereditary Retinal/Choroidal Dystrophies**

**Diffuse**

**Macular**

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

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**How does central areolar choroidal dystrophy present?**
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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
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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
There are three subtypes of diffuse dystrophies—what are they?
There are three subtypes of diffuse dystrophies—what are they?
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)
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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?
Diffuse

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

Nyctalopia

Type of ‘blindness’ produced

Hemeralopia

- 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

Nyctalopia
Type of ‘blindness’ produced
Hemeralopia
- Enhanced S-cone dz

What is…
--nyctalopia?
--hemeralopia?

Macular
- Sorsby macular dystrophy
- Best dz
- Stargardt
- Pattern dystrophies
- Central areolar choroidal dystrophy
- North Carolina macular dystrophy
What is…
--nyctalopia? Night blindness
--hemeralopia? Day blindness
Hereditary Retinal/Choroidal Dystrophies

Diffuse

Rod-dominant

Cone-dominant

Rod-cone (RP)

Cone/cone-rod

Retinal/Choroidal Dystrophies

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

Type of ‘blindness’ produced

Hemeralopia

Chief visual defect(s)
Hereditary Retinal/Choroidal Dystrophies

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

Hereditary Macular Dystrophies

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

Rod-dominant

Cone-dominant

Macular

Sorsby macular dystrophy

Best dz

Stargardt

Pattern dystrophies

Central areolar choroidal dystrophy

North Carolina macular dystrophy

Hereditary Retinal/Choroidal Dystrophies

Chief visual defect(s)

Acuity, and color loss

Type of ‘blindness’ produced

This makes sense, as rods dominate the retinal periphery

VF loss

Nyctalopia

Hemeralopia

Rod-cone (RP)

Cone/cone-rod
Diffuse

Rod-dominant
- Rod-cone (RP)

Cone-dominant
- Cone/cone-rod

Hereditary Retinal/Choroidal Dystrophies

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

<table>
<thead>
<tr>
<th></th>
<th>Type of ‘blindness’ produced</th>
<th>Hemeralopia</th>
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<tr>
<td>Nyctalopia</td>
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<tr>
<td>VF loss</td>
<td>Chief visual defect(s)</td>
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<td>?</td>
<td>c/o photophobia?</td>
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</table>
Hereditary Retinal/Choroidal Dystrophies

Diffuse

Rod-dominant

Cone-dominant

Rod-cone (RP)

Cone/cone-rod

Nyctalopia

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) Cone/cone-rod

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

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

No question—proceed when ready
Macular Sorsby macular dystrophy
Best dz
Stargardt
Central areolar choroidal dystrophy
North Carolina macular dystrophy
Pattern dystrophies

Diffuse

Rod-dominant
- Rod-cone (RP)

Cone-dominant
- Cone/cone-rod

Choroidal

Hereditary Retinal/Choroidal Dystrophies

Enhanced S-cone dz

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

Retinal/Choroidal Dystrophies
Hereditary

Leber's congenital amaurosis
Best dz
Stargardt
Pattern dystrophies
What does LCA stand for in this context? Leber's congenital amaurosis
Diffuse

Hereditary
Retinal/Choroidal
Dystrophies

Macular

Sorsby macular dystrophy
Best dz
Stargardt
Pattern dystrophies

Rod-dominant
Rod-cone (RP)

Cone-dominant
Cone/cone-rod

Choroidal

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

Choroidal
- Cone-dominant
  - Cone/cone-rod

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

Rod-dominant

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

What are its three hallmark findings?
-- Profoundly poor VA from birth
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What does LCA stand for in this context? Leber’s congenital amaurosis

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--Profoundly poor VA from birth
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Macular Sorsby macular dystrophy
Best dz
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Central areolar choroidal dystrophy
North Carolina macular dystrophy
Pattern dystrophies

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

Cone-dominant

LCA Leber’s congenital amaurosis

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…
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.)

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.

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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?
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.

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 focus again, we 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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One word captures the dz process common to all—what is it?
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‘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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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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Bietti crystalline dystrophy

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

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

structure
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**...
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 Bietti 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 Bietti present clinically?
With visual symptom and location scotomas

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

Pattern dystrophies
- Enhanced S-cone dz
- Gyrate
- Choroideremia

Choroidal

Bietti

Rod-dominant choroidal dystrophy
Rod-cone (RP)
Cone-dominant choroideremia
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 Bietti present clinically?
With nyctalopia and paracentral 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 Bietti present clinically?
With nyctalopia and paracentral scotomas in life period years
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?
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How does Bietti present clinically?
With nyctalopia and paracentral scotomas in teen/young adult years
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 nyctalopia and paracentral scotomas in teen/young adult years.

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

Is it treatable?
No.

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 the teen/young adult years.

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

Is Bietti present clinically?
With nyctalopia and paracentral scotomas.

How does Bietti present clinically?
Teen/young adult years.

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

Is it treatable?
No.
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

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**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 areas of RPE and choriocapillaris atrophy.

**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

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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.
Gyrate atrophy: Pavingstones
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?
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

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

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, and progressive VF loss

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, and progressive VF loss

**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, and progressive VF loss

What is the underlying pathophysiology? A defect in the gene coding for the ornithine aminotransferase enzyme leads to excess serum levels of ornithine-ase enzyme substrate.
**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, and progressive VF loss

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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 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.
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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.
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.
**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.

**Clinical presentation:**
- Severe nyctalopia commencing in childhood, and progressive VF loss.

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

**Inheritance pattern:**
- X-linked.
Choroideremia
Choroideremia

FAF reveals the expected hypofluorescent lesions
**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.
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 symptom commencing in life period

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.

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

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

**Inheritance pattern for this gene?**
X-linked.
**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.
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
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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**
- LCA
- Enhanced S-cone dz

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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.

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

For more on gyrate and choroideremia, see slide-set R42.
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.)
Hereditary Retinal/Choroidal Dystrophies

Diffuse

Rod-dominant

Choroidal

Rod-cone (RP)

Choroideremia

Gyrate Bietti

Cone-dominant

Cone/cone-rod

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?

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Diffuse

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Enhanced S-cone dz
Retinal/Choroidal Dystrophies Hereditary

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?

**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.

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

**X-linked retinoschisis**
Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular

Rod-dominant

Choroidal

Rod-cone (RP)

Choroideremia

Gyrate Bietti

Inner retinal dystrophy

X-linked retinoschisis

Cone-dominant

Cone/cone-rod

LCA

Enhanced S-cone dz

Retinal/Choroidal 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.

Sorsby macular dystrophy

Best dz

Stargardt

Central areolar choroidal dystrophy

North Carolina macular dystrophy

Pattern dystrophies

Note: There is some disagreement within the BCSC regarding how X-linked retinoschisis should be classified.
Hereditary Retinal/Choroidal Dystrophies

Diffuse

Macular

Rod-dominant

Choroidal

Rod-cone (RP)

Choroideremia

Gyrate Bietti

Inner retinal dystrophy (per the Retina book)

X-linked retinoschisis

Cone-dominant

Cone/cone-rod

LCA

Enhanced S-cone dz

Retinal/Choroidal Dystrophies

Sorsby macular dystrophy

Best dz

Stargardt central areolar choroidal dystrophy

North Carolina macular dystrophy

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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

Rod-dominant

Choroideremia

Gyrate Bietti

X-linked retinoschisis

Cone-dominant

Cone/cone-rod

LCA

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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)

X-linked retinoschisis
Consider the following diagnostic categories:

- **Diffuse Rod-dominant Choroidal Rod-cone (RP) Choroideremia Gyrate Bietti X-linked retinoschisis**
- **Cone-dominant Cone/cone-rod**

**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.

**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?**

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

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**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

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?
The vitreous—it is syneretic, and contains veils and other opacities

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

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

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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
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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

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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What do these stand for in this context?
NFL: ?
OPL: ?
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**NFL**: Nerve fiber layer
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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)
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Inner retinal dystrophy

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HVF 60-4, normal (I think)
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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?
Electroretinography (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
Inner retinal dystrophy

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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?

The vitreous—it is syneretic, and contains veils and other opacities

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

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Before we get into the weeds on this…**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

How is it performed?
The pt is dilated, and usually dark-adapted. Electrodes are attached to the pt’s cornea and/or periocular skin, and a series of standardized visual stimuli (usually brief flashes) are presented.

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

Which type is typically employed in diagnosing XLR?
Full-field
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Pt's must adhere to a specific lifestyle modification—what is it? No contact sports—XLR retinas are highly susceptible to trauma

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

How is it performed?
The pt is **dilated** vs **undilated** and **dark- vs light-adapted**

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**ERG**

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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 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.

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).

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.

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.

Before we get into the weeds on this… **What does ERG stand for?**

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

**How is it performed?**
The pt is **dilated**, and usually **dark-adapted**.

**X-linked retinoschisis**
By what other (very similar) name is X-linked retinoschisis (XLR) known?
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How does it present on DFE?
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What layer(s) of the retina are involved in the schisis?
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[![ERG](image)](image) 

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

Which type is typically employed in diagnosing XLR?
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Before we answer this, let's get familiar with the normal ffERG
Normal ffERG

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--The response commences with the stimulus flash
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--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**

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

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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
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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)

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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?
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**ERG**

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

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**Negative** or **electronegative** waveform
By what other (very similar) name is X-linked retinoschisis (XLR) known? X-linked juvenile retinoschisis

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Normal ERG

ERG in XLJR

This constitutes a ‘negative’ or (‘electronegative’) ERG

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

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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)

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

Is a negative ERG pathognomonic for XLR?
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

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

We mentioned CNSB previously. In a nutshell, what is it?
A congenital condition in which a dearth of functioning rods leads to nyctalopia, nystagmus, and variably decreased VA.

*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.

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

*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)

*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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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 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

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What is the term for any retinal condition which the b-wave is preserved but the a-wave is diminished?
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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

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

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**ERG**

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

For more on CSNB, see slide-set R9
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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?**  
The vitreous—it is syneretic, and contains veils and other opacities

**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)
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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 to severely decreased VA in life stage

What ocular structure—unmentioned previously in this set—is affected by XLR?
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With modestly decreased VA in childhood

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Defective function of the retinoschisin protein

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Inner retinal dystrophy

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

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.

What gene codes for retinoschisin?
RS1
By what other (very similar) name is X-linked retinoschisis (XLR) known?
X-linked juvenile retinoschisis

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Is testing for RS1 mutations commercially available? Yes

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

Inner retinal dystrophy

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The vitreous

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What ocular structure—unmentioned previously in this set—is affected by XLR?
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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.

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

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)

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

Inner retinal dystrophy

- X-linked retinoschisis

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

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

How does it present on DFE? 
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Inner retinal dystrophy

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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? The vitreous—it is syneretic, and contains veils and other opacities

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

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)

Inner retinal dystrophy
X-linked retinoschisis