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.
Hereditary Retinal/Choroidal Dystrophies

- Diffuse
- Macular

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

?
Hereditary Retinal/Choroidal Dystrophies

- Diffuse
- Macular

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

- One of two with macular in its name
- 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
What are the hereditary macular dystrophies covered in the Retina book?

- **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 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 #
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
Diffuse

How does Best dz present?

Hereditary Retinal/Choroidal Dystrophies

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 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
Diffuse

Hereditary Retinal/Choroidal Dystrophies

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

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 does vitelliform mean?

- Yellow
- Round
- 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)
Diffuse

Hereditary Retinal/Choroidal Dystrophies

Macular

Sorsby macular dystrophy

Best dz

Stargardt

Pattern dystrophies

Central areolar choroidal dystrophy

North Carolina macular 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?

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

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

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 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?
Hereditary Retinal/Choroidal Dystrophies

Diffuse

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

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

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?
Diffuse Retinal/Choroidal Dystrophies

Hereditary Macular

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

Macular

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

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?

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.
Diffuse Retinal/Choroidal Dystrophies

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?

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*
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

- 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

What is the classic finding on DFE?
Diffuse Retinal/Choroidal Dystrophies

Hereditary Macular Dystrophies

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?
Fish-shaped lesions in the macula

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

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?
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
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?
Hereditary Retinal/Choroidal Dystrophies

**Diffuse**

**Macular**

- 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

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

**Hereditary Retinal/Choroidal Dystrophies**

- Diffuse
- Macular
  - 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?
--?
--? 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

--- 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—which are they?*
---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
--Adult-onset foveomacular vitelliform dystrophy
--Reticular dystrophy
--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
--Adult-onset foveomacular vitelliform dystrophy
--Reticular dystrophy
--Fundus pulverulentus

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’)

*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

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

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
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 slightly affected

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

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

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

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?
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*

---

**Sorsby macular dystrophy**

**Best dz**

**Stargardt**

**Pattern dystrophies**

**Central areolar choroidal dystrophy**

**North Carolina macular dystrophy**
**Hereditary Retinal/Choroidal Dystrophies**

**Diffuse**

**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

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

- Sorsby macular dystrophy
- Best dz
- Stargardt
- **Pattern dystrophies**
- 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 (i.e., 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?
Hereditary Retinal/Choroidal Dystrophies

Diffuse

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

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
Diffuse

Hereditary Retinal/Choroidal Dystrophies

Macular

- 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’)

*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?**
You’d think so, but no—they’re genetically unrelated

**Is the EOG abnormal in AOFVD, like it is in Best dz?**
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 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

Is the EOG abnormal in AOFVD, like it is in Best dz?
Again, you’d think so, but no—EOG is normal in AOFVD
Hereditary Retinal/Choroidal Dystrophies

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
Diffuse

Macular

Hereditary Retinal/Choroidal Dystrophies

How does central areolar choroidal dystrophy present?

- 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
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.
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
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?
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
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?
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

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

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)?

---

Central areolar choroidal dystrophy

Sorsby macular dystrophy

Best dz

Stargardt

Pattern dystrophies

North Carolina macular dystrophy
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.

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)
--

Central areolar choroidal dystrophy

Sorsby macular dystrophy

Best dz

Stargardt

Pattern dystrophies

North Carolina macular dystrophy
**Diffuse**

Hereditary Retinal/Choroidal Dystrophies

- 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

*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)
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

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)
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)

---
Diffuse Retinal/Choroidal Dystrophies

Macular

How does central areolar choroidal dystrophy present?
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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.

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

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---

Sorsby macular dystrophy
Best dz
Stargardt
Pattern dystrophies
Central areolar choroidal dystrophy
North Carolina macular dystrophy
North Carolina macular dystrophy is called (by the Retina book) an “atypical” macular dystrophy. What makes it atypical?
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.”
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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
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?

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

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

No question—proceed when ready
Hereditary Retinal/Choroidal Dystrophies

Diffuse

- Rod-dominant
- Cone-dominant

Choroidal

Macular

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

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

Diffuse

Rod-dominant
  └ Rod-cone (Abb.)

Cone-dominant

Choroidal

Macular

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

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

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

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)

For more on RP, see slide-set R38
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 Choroidal Cone-dominant LCA Enhanced S-cone dz

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

Cone-dominant Cone/cone-rod

Diffuse Macular

Hereditary Retinal/Choroidal Dystrophies

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

Type of ‘blindness’ produced

Enhanced S-cone dz
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
106

Diffuse

Choroidal

Cone-dominant

LCA

Enhanced S-cone dz

Rod-dominant

Cone/cone-rod

Nyctalopia

Type of ‘blindness’ produced

Hemeralopia

What is…
--nyctalopia?
--hemeralopia?

Rod-cone (RP)

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

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

- 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
  - Cone/cone-rod

<table>
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<tr>
<th>Nyctalopia</th>
<th>Type of ‘blindness’ produced</th>
<th>Hemeralopia</th>
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</thead>
<tbody>
<tr>
<td>?</td>
<td>Chief visual defect(s)</td>
<td>?</td>
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</tbody>
</table>

Macular

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

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

Hereditary Retinal/Choroidal Dystrophies

Nyctalopia Type of ‘blindness’ produced Hemeralopia

VF loss Chief visual defect(s) Acuity, and color loss
Hereditary Retinal/Choroidal Dystrophies

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

- Macular
  - 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
  - Hemeralopia

- Nyctalopia

- VF loss

This makes sense, as rods dominate the retinal periphery.
**Hereditary Retinal/Choroidal Dystrophies**

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

**Chief visual defect(s)**

- **Acuity, and color loss**
- **VF loss**
- **Type of ‘blindness’ produced**
- **Nyctalopia**
- **Hemeralopia**

- 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)

**Macular**

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

**No question—proceed when ready**

---

- **Retinal/Choroidal Dystrophies**
- **Hereditary**
- **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>
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<tr>
<th>Condition</th>
<th>Type of ‘blindness’ produced</th>
<th>Chief visual defect(s)</th>
<th>Acuity, and color loss</th>
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<tbody>
<tr>
<td>Nyctalopia</td>
<td>Hemeralopia</td>
<td>VF loss</td>
<td>?</td>
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?
### 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>Rod-cone (RP)</th>
<th>Nyctalopia</th>
<th>Type of ‘blindness’ produced</th>
<th>Hemeralopia</th>
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<tbody>
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<td></td>
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<td>No</td>
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<th>Rod-dominant</th>
<th>Chief visual defect(s)</th>
<th>Acuity, and color loss</th>
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<tr>
<td>Cone-dominant</td>
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<table>
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<tr>
<th>Macular</th>
<th>c/o photophobia?</th>
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<td>Yes</td>
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Hereditary Retinal/Choroidal Dystrophies

Diffuse

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

Macular

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

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<tr>
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<td>c/o photophobia?</td>
<td>Yes</td>
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</table>

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

No question—proceed when ready.
What does LCA stand for in this context?

Leber's congenital amaurosis
Hereditary Retinal/Choroidal Dystrophies

Diffuse

Rod-dominant

Rod-cone (RP)

Choroidal

Cone-dominant

Cone/cone-rod

LCA

Macular

Sorsby macular dystrophy

Best dz

Stargardt

Pattern dystrophies

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
-- Patients engage in eye rubbing behavior
Retinal/Choroidal Dystrophies

Hereditary

Diffuse

Rod-dominant

Rod-cone (RP)

Choroidal

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?
--?
--?
--?
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

LCA

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

What are its three hallmark findings?
-- Profoundly poor VA from birth
-- Nystagmus from birth
-- Pts engage in eye rubbing behavior
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
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.)

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For more on these conditions, see slide-set R9
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(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.

Rod-dominant
- Rod-cone (RP)
  - Usher syndrome
  - Bardet-Biedl syndrome
  - Bassen-Kornzweig dz
  - Infantile Refum
  - (a number of others)

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?
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.

For more on these conditions, see slide-set R5

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

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

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.

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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—two colors crystals are present within the 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 (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

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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
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
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 location.
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

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 life period years.

OPTIONS AND CONSIDERATION

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

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 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

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

What is the full name of this condition?
Gyrate atrophy

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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

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 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

How does it present on DFE?
Early dz is marked by the presence of large peripheral ‘pavingstone’ 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
Gyrate atrophy: Pavingstones
What is the full name of this condition?
Gyrate atrophy

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

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?**
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.

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

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

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

**How does it present clinically?**
Severe 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.
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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 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

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 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?
Gyrate atrophy

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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

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What is the full name of this condition?
Gyrate atrophy

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Severe nyctalopia commencing in childhood, and progressive VF loss

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Severe nyctalopia commencing in childhood, and progressive VF loss

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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 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.
How does choroideremia present on DFE?

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.
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.
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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**Choroidal**
- Bietti
- Gyrate
- Choroideremia

- LCA
  - Enhanced S-cone dz
  - Central areolar choroidal dystrophy
  - Pattern dystrophies
  - 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 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**

**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

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, 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

Choroidal
- Bietti
- Gyrate

Choroideremia

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.
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.

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**Choroidal**
- Bietti
- Gyrate
- **Choroideremia**

- LCA
- Enhanced S-cone dz
- Central areolar choroidal dystrophy
- Pattern dystrophies
- 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.
Hereditary Retinal/Choroidal Dystrophies

Diffuse

- Rod-dominant Choroidal Rod-cone (RP)

Macular

- Chorioretinal dystrophies
  - 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.

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

No question—proceed when ready
Diffuse vs Macular

Diffuse

Rod-dominant

Rod-cone (RP)

Choroidal

Choroideremia

Gyrate Bietti

Cone-dominant

Cone/cone-rod

LCA

Enhanced S-cone dz

Retinal/Choroidal Dystrophies

Hereditary

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?**

*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

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

Retinal/Choroidal Dystrophies

Diffuse vs Macular system

Rod-cone (RP)

Cone-dominant

Cone/cone-rod

Outer retinal dystrophies

Macular dystrophies

Chorioideremia

Bietti

Inner retinal dystrophy

X-linked retinoschisis

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?

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.

Inner retinal dystrophy

X-linked retinoschisis
Hereditary Retinal/Choroidal Dystrophies

Diffuse

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

Macular

Sorsby macular dystrophy

Best dz

Stargardt

Central areolar choroidal dystrophy

North Carolina macular dystrophy

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

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…

Inner retinal dystrophy (per the Retina book)

X-linked retinoschisis
Hereditary Retinal/Choroidal Dystrophies

Diffuse

Rod-dominant

Choroidal

Rod-cone (RP)

Choroideremia

Gyrate Bietti

X-linked retinoschisis

Cone-dominant

Cone/cone-rod

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Enhanced S-cone dz

Retinal/Choroidal Dystrophies

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Central areolar choroidal dystrophy

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

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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* (per the *Peds book*)

X-linked retinoschisis
Hereditary Retinal/Choroidal Dystrophies

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.)

Macular

Sorsby macular dystrophy

Rod-dominant

Cone-dominant

Cone/cone-rod

Choroidal

Hereditary vitreoretinopathy (per the Peds book)

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.

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?

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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**X-linked retinoschisis**

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**Inner retinal dystrophy**
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?
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 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

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

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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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X-linked juvenile retinoschisis

How does it present on DFE?
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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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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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X-linked retinoschisis

Inner retinal dystrophy

Retinal/Choroidal Dystrophies

Hereditary Macular

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

Pattern dystrophies

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HVF 60-4, normal (I think)
By what other (very similar) name is X-linked retinoschisis (XLR) known? X-linked juvenile retinoschisis.

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

Mainly the **NFL**, but the **OPL** can be involved as well.

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

All of what about what?

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

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

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

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)

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

Electroretinography (ERG)

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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?

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Electroretinography (ERG)

What is the classic ERG finding in XLR?

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

a vs. b-wave a vs. b-wave

What HVF protocol is needed?

60-4 (I know, I’ve never seen one either)

Previous slide notwithstanding, photographic documentation of peripheral schisis? What's the protocol?

What's the protocol?
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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60-4 (I know, I’ve never seen one either)

Inner retinal dystrophy

X-linked retinoschisis
X-linked retinoschisis: ERG

Normal

Patient

Absent $b$-wave

$A$-wave

$B$-wave
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

Is this ERG finding pathognomonic for XLR? No

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

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Rod-dominant Choroidal Rod-cone (RP)

Choroideremia Gyrate Bietti Inner retinal dystrophy X-linked retinoschisis
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**Inner retinal dystrophy**

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

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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?  
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With macular schisis in a radial pattern, +/- peripheral schisis

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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.

What gene codes for retinoschisin?
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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

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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.

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