Congenital/Stationary Retinal Disease

Two very basic categories
Congenital/Stationary Retinal Disease

Two very basic categories

Cone (Color) Disease

Rod (Night Vision) Disease
Cone (Color) Disease

Rod (Night Vision) Disease

Three very basic categories
Congenital/Stationary Retinal Disease

Cone (Color) Disease
  Trichromatism
  Dichromatism
  Monochromatism

Rod (Night Vision) Disease

*Three very basic categories*
What does it mean to say someone is a ‘trichromat’?

Trichromatism

Dichromatism
Trichromatism

What does it mean to say someone is a ‘trichromat’?
It concerns performance on a color-matching test. In this test, the participant is asked to match a test color by mixing primary-color lights (note--not mixing paints!).
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What does it mean to say someone is an ‘anomalous’ trichromat?
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What does it mean to say someone is an ‘anomalous’ trichromat?
It means he needs all three colored lights to do the matching, but that the relative intensities among the lights differs significantly from that employed by people with normal color vision (which color is abnormally intense is a function of what sort of anomalous trichromacy he has)
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*Dude, wussup with the gendered language?*
The genetics relevant to anomalous color vision are predominantly X-linked recessive, so the vast majority of individuals with color deficiencies are males (including yours truly)
What does it mean to say someone is a dichromat?

It means that, on the color-matching test, he can match any test color using only two lights. (Which two depends upon the form of dichromacy, but the missing one is almost never blue.)

The fact that dichromats can match any color with two primaries indicates what about their cones?

It indicates that his cones possess only two photopigments, not three as do trichromats.

Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism → Dichromatism

What does it mean to say someone is a dichromat?
What does it mean to say someone is a **dichromat**?
It means that, on the color-matching test, he can match any test color using only *two* lights. (Which two depends upon the form of dichromacy, but the missing one is almost always vs never **blue**.)
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The fact that a dichromat can match any color with only two primaries indicates what about his cones? It indicates his cones possess only two photopigments, not three as do the cones in trichromats.
By what other name is monochromatism known?
By what other name is monochromatism known?
Achromatopsia
By what other name is monochromatism known? Achromatopsia

Does monochromatism/achromatopsia mean what I think it does?
By what other name is monochromatism known?
Achromatopsia

Does monochromatism/achromatopsia mean what I think it does?
Yes--it is the state in which an individual can match any test color using just one color of light
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

? 

? 

The two types of monochromatism are…
The two types of monochromatism are…
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

Blue-cone monochromatism

Rod monochromatism

--Inheritance…

Congenital/Stationary Retinal Disease

Nystagmus

-- VA range: 20/80–20/200

ERG:

-- Cone response: Absent

-- Rod response: Normal
Cone (Color) Disease

Rod (Night Vision) Disease

- Trichromatism
- Dichromatism
- Monochromatism
  - Rod monochromatism
  - Blue-cone monochromatism

Rod monochromatism
  -- Inheritance ... AR

Congenital/Stationary Retinal Disease

20/80–20/200

Nystagmus always present

Cone response: Absent
Rod response: Normal
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

--Inheritance…AR

--No cones present—true color blindness

Rod monochromatism

Blue-cone monochromatism

Congenital/Stationary Retinal Disease
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

--Inheritance…AR

--No cones present—true color blindness

--EOM issue always present

Blue-cone monochromatism

Nystagmus

VA range: 20/80–20/200

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--Cone response: Absent
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Congenital/Stationary Retinal Disease
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

-- Inheritance... AR

-- No cones present—true color blindness

-- Nystagmus always present
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

Rod monochromatism

- Inheritance…AR
- No cones present—true color blindness
- Nystagmus always present
- VA range:
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism
Dichromatism

Monochromatism

Rod monochromatism

-- Inheritance... AR
-- No cones present—true color blindness
-- Nystagmus always present
-- VA range: 20/80–20/200

Blue-cone monochromatism

Congenital/Stationary Retinal Disease
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism → Dichromatism → Monochromatism

Rod monochromatism
-- Inheritance... AR
-- No cones present—true color blindness
-- Nystagmus always present
-- VA range: 20/80–20/200

Why the broad range in VA?

Why the broad range in VA?

Because the dz manifests partial expression in some cases (ie, some pts will have a few functioning cones)
Cone (Color) Disease

- Trichromatism
- Dichromatism
- Monochromatism
  - Rod monochromatism
  - Blue-cone monochromatism

Rod (Night Vision) Disease

Monochromatism

- Rod monochromatism

Why the broad range in VA?
Because the dz manifests partial expression in some cases (ie, some pts will have a few functioning cones)

Rod monochromatism
-- Inheritance…AR
-- No cones present—true color blindness
-- Nystagmus always present

VA range: 20/80–20/200

Congenital/Stationary Retinal Disease
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism
Dichromatism

Monochromatism

Rod monochromatism

--Inheritance…AR
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--Nystagmus always present
--VA range: 20/80–20/200
--ERG: ?
Before we get into the weeds on this… *What does ERG stand for?*
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
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How is it performed?

Congenital/Stationary Retinal Disease

- Nystagmus always present
- VA range: 20/80–20/200
- Cone response: absent
- Rod response: normal
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**Full-field (ffERG, aka Ganzfeld ERG), multifocal (mfERG), and pattern (pERG)**

- **ffERG**: Demonstrates the response of the central vs entire retina to flash stimuli
- **mfERG**
- **pERG**
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What are the three main types of ERG?
**Full-field** (ffERG, aka Ganzfeld ERG), **multifocal** (mfERG), and **pattern** (pERG)

ffERG: Demonstrates the response of the entire retina to flash stimuli
mfERG: Produces a map of central cone vs rod function
pERG
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mfERG: Produces a topographic map of central cone function
pERG
Cone (Color) Disease

Rod (Night Vision) Disease

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Trichromatism
Dichromatism

Monochromatism
Rod monochromatism
Blue-cone monochromatism

Rod monochromatism
---Inheritance…
AR
---No cones present—true color blindness
---Nystagmus
---VA range: 20/80–20/200
---ERG:
---Cone response:
---Rod response: Normal

Congenital/Stationary Retinal Disease

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ffERG: Demonstrates the response of the entire retina to flash stimuli
mfERG: Produces a topographic map of central cone function
pERG: Flashes a pattern of rapidly alternating light-and-dark areas
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**Full-field (ffERG, aka Ganzfeld ERG), multifocal (mfERG), and pattern (pERG)**

ffERG: Demonstrates the response of the entire retina to flash stimuli

mfERG: Produces a topographic map of central cone function

pERG: Flashes a checkerboard pattern of rapidly alternating light and dark areas

How does a mfERG accomplish this?
Before we get into the weeds on this…What does ERG stand for?
Electroretinogram (or electroretinography)

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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)

ffERG: Demonstrates the response of the entire retina to flash stimuli.
mfERG: Produces a topographic map of central cone function.
pERG: Flashes a checkerboard pattern of rapidly alternating light and dark areas.

How does a mfERG accomplish this?
Instead of flashing the entire retina, mfERG flashes are limited to small, hexagon-shaped areas of the macula. By divvying the macula up into hexagons and systematically testing each, mfERG can map out the functional status of the macula.
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism
Dichromatism
Monochromatism

Rod monochromatism
--Inheritance... AR
--No cones present—true color blindness
--Nystagmus always present
--VA range: 20/80–20/200
--ERG:
  --Cone response: ?
  --Rod response: ?

Next questions
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

--Inheritance…AR

--No cones present—true color blindness

--Nystagmus always present

--VA range: 20/80–20/200

--ERG:

--Cone response: Absent

--Rod response: Normal
Rod monochromatism: ERG

Congenital/Stationary Retinal Disease

In rod monochromatism, the rod response is (relatively) normal.
However, the cone response is essentially nonexistent, as expected.
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

Blue-cone monochromatism

Rod monochromatism

--Inheritance…AR
--No cones present—true color blindness
--Nystagmus always present
--VA range: 20/80–20/200
--ERG:
  --Cone response: Absent
  --Rod response: Normal

Classic presentation of rod monochromatism:

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and
---
and
---

Congenital/Stationary Retinal Disease

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Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

Blue-cone monochromatism

--- Inheritance... AR
--- No cones present—true color blindness
--- Nystagmus always present
--- VA range: 20/80–20/200
--- ERG:
   -- Cone response: Absent
   -- Rod response: Normal

Classic presentation of rod monochromatism:
--- Poor acuity and
--- Nystagmus and
--- Photophobia

Congenital/Stationary Retinal Disease
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

Blue-cone monochromatism

Inheritance: ?
Cone (Color) Disease

- Trichromatism
- Dichromatism

Monochromatism

- Rod monochromatism
- Blue-cone monochromatism

Rod (Night Vision) Disease

- Blue-cone monochromatism
  -- Inheritance: X-linked

Congenital/Stationary Retinal Disease
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

Blue-cone monochromatism

--Inheritance: X-linked

--Only cones present
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism
Dichromatism

Monochromatism

Blue-cone monochromatism

--Inheritance: X-linked
--Only **blue** cones present
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

Blue-cone monochromatism

--Inheritance: X-linked
--Only blue cones present
--VA usually about…?
Cone (Color) Disease

Rod (Night Vision) Disease

- Trichromatism
- Dichromatism
- Monochromatism
  - Rod monochromatism
  - Blue-cone monochromatism

*Blue-cone monochromatism*
- Inheritance: X-linked
- Only blue cones present
- VA usually about… 20/80
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism  Dichromatism

Monochromatism

Rod monochromatism

Blue-cone monochromatism

---Inheritance: X-linked
---Only blue cones present
---VA usually about... 20/80

Why is VA better than in many rod monochromats?
Cone (Color) Disease

- Trichromatism
- Dichromatism
- Monochromatism
  - Rod monochromatism
  - Blue-cone monochromatism

Rod (Night Vision) Disease

- Inheritance: X-linked
- Only blue cones present
- VA usually about…20/80

Why is VA better than in many rod monochromats? Because all blue-cone monochromats have a set of functioning cones (specifically, the blue ones)
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Blue-cone monochromatism

--Inheritance: X-linked
--Only blue cones present
--VA usually about…20/80
--Diagnose via specialized…?
Cone (Color) Disease

- Trichromatism
- Dichromatism

Monochromatism

- Rod monochromatism
- Blue-cone monochromatism

Blue-cone monochromatism
-- Inheritance: X-linked
-- Only blue cones present
-- VA usually about... 20/80
-- Diagnose via specialized... color ERG

Rod (Night Vision) Disease
Trichromatism → Dichromatism → Monochromatism

- Rod monochromatism
- **Blue-cone monochromatism**

**Blue-cone monochromatism**
--Inheritance: X-linked
--Only blue cones present
--VA usually about 20/80
--Diagnose via specialized color ERG

What are the findings of color-ERG testing in blue-cone monochromatism?
Blue-cone monochromatism
--Inheritance: X-linked
--Only blue cones present
--Visual acuity (VA) usually about 20/80
--Diagnose via specialized color ERG

What are the findings of color-ERG testing in blue-cone monochromatism?
Perhaps unsurprisingly, findings include a normal blue-cone response along with absent or greatly attenuated green- and red-cone responses
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

Blue-cone monochromatism

--- Inheritance: X-linked
--- Only blue cones present
--- VA usually about... 20/80
--- Diagnose via specialized... color ERG

Classic presentation of blue-cone monochromatism:
--- two words
--- and
--- and
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

Blue-cone monochromatism

-- Inheritance: X-linked
-- Only blue cones present
-- VA usually about…20/80
-- Diagnose via specialized…color ERG

Classic presentation of blue-cone monochromatism:
-- Poor acuity and
-- Nystagmus and
-- Photophobia

(Yes, just like rod monochromatism)
Speaking of conditions that present very early in life with poor VA, nystagmus and photophobia...While there are many, the others that should come first to mind are what?
--Rod monochromatism
--Blue-cone monochromatism
--?
--?
--?

Classic presentation of blue-cone monochromatism:
-- Poor acuity and
-- Nystagmus and
-- Photophobia
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

-- Blue-cone monochromatism
-- Rod monochromatism
-- Blue-cone monochromatism
-- Albinism
-- Aniridia
-- Leber’s congenital amaurosis

Speaking of conditions that present very early in life with poor VA, nystagmus and photophobia…While there are many, the others that should come first to mind are what?
-- Rod monochromatism
-- Blue-cone monochromatism
-- Albinism
-- Aniridia
-- Leber’s congenital amaurosis

Classic presentation of blue-cone monochromatism:
-- Poor acuity and
-- Nystagmus and
-- Photophobia

Inheritance:
-- Only blue cones present
-- VA usually about...20/80
-- Diagnose via specialized...color ERG
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism → Dichromatism → Monochromatism

- Rod monochromatism
- Blue-cone monochromatism

**Blue-cone monochromatism**
- Inheritance: X-linked
- Only blue cones present
- VA usually about...20/80
- Diagnose via specialized...color ERG

Classic presentation of blue-cone monochromatism:
- Poor acuity and
- Nystagmus and
- Photophobia

If a pt has nystagmus plus good vision, what condition does s/he most likely have?
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism → Dichromatism → Monochromatism
- Rod monochromatism
- Blue-cone monochromatism

Classic presentation of Blue-cone monochromatism:
- Poor acuity
- Nystagmus
- Photophobia

**Blue-cone monochromatism**
- Inheritance: X-linked
- Only blue cones present
- VA usually about... 20/80
- Diagnose via specialized... color ERG

If a pt has nystagmus plus good vision, what condition does s/he most likely have?
Congenital motor nystagmus
Cone (Color) Disease

Trichromatism

Dichromatism

Monochromatism

Rod (Night Vision) Disease

-- Inheritance: X-linked
-- Only blue cones present
-- Mimics rod monochromatism
-- Diagnose via specialized color ERG

Classic presentation of blue-cone monochromatism:
-- Poor acuity and
-- Nystagmus and
-- Photophobia

^ If a pt has nystagmus plus good vision, what condition does s/he most likely have?

Congenital motor nystagmus

Briefly, what is congenital motor nystagmus?

A nystagmus arising in the first few months of life that is not secondary to either sensory or CNS pathology

It is virtually always horizontal

Rule of thumb: If a pt has nystagmus + good VA, it’s congenital motor nystagmus
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

Blue-cone monochromatism

Inheritance: X-linked

Only blue cones present

Mimics rod monochromatism

Diagnose via specialized color ERG

Classic presentation of blue-cone monochromatism:

-- Poor acuity
-- Nystagmus
-- Photophobia

If a pt has nystagmus plus good vision, what condition does s/he most likely have? Congenital motor nystagmus

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Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

Blue-cone monochromatism

Inheritance: X-linked

-- Only blue cones present

-- Mimics rod monochromatism

-- Diagnose via specialized… color ERG

Congenital/Stationary Retinal Disease

Classic presentation of blue-cone monochromatism:

-- Poor acuity

-- Nystagmus

-- Photophobia

If a pt has nystagmus plus good vision, what condition does s/he most likely have?

Congenital motor nystagmus

Briefly, what is congenital motor nystagmus?

A nystagmus arising in the first few months of life that is not secondary to either sensory or CNS pathology

Is the nystagmus vertical, horizontal or both/either?

Rule of thumb: If a pt has nystagmus + good VA, it's congenital motor nystagmus
Cone (Color) Disease

- Trichromatism
- Dichromatism
- Monochromatism
  - Rod monochromatism
  - Blue-cone monochromatism

Rod (Night Vision) Disease

- 80

**Inheritance:**
- X-linked
- Only blue cones present
- Mimics rod monochromatism
- Diagnose via specialized color ERG

**Congenital/Stationary Retinal Disease**

**Classic presentation of blue-cone monochromatism:**
- Poor acuity
- Nystagmus
- Photophobia

Good

**If a pt has nystagmus plus good vision, what condition does s/he most likely have?**

**Congenital motor nystagmus**

**Briefly, what is congenital motor nystagmus?**

A nystagmus arising in the first few months of life that is not secondary to either sensory or CNS pathology

**Is the nystagmus vertical, horizontal or both/either?**

It is virtually always horizontal.
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

Blue-cone monochromatism

-- Inheritance: X-linked

-- Only blue cones present

-- Mimics rod monochromatism

-- Diagnose via specialized... color ERG

Congenital/Stationary Retinal Disease

Classic presentation of blue-cone monochromatism:

-- Poor acuity and

-- Nystagmus and

-- Photophobia

Good

Is the nystagmus vertical, horizontal or both/either?
It is virtually always horizontal

Briefly, what is congenital motor nystagmus?
A nystagmus arising in the first few months of life that is not secondary to either sensory or CNS pathology

If a pt has nystagmus plus good vision, what condition does s/he most likely have?
Congenital motor nystagmus
**Congenital/Stationary Retinal Disease**

**Cone (Color) Disease**
- Trichromatism
- Dichromatism
  - Monochromatism
    - Blue-cone monochromatism
      - Inheritance: X-linked
      - Only blue cones present
      - Mimics rod monochromatism
      - Diagnose via specialized color ERG

**Rod (Night Vision) Disease**
- Monochromatism
  - Rod monochromatism

**Classic presentation of blue-cone monochromatism:**
- Poor acuity and nystagmus and photophobia

**Briefly, what is congenital motor nystagmus?**
A nystagmus arising in the first few months of life that is not secondary to either sensory or CNS pathology

Is the nystagmus vertical, horizontal or both/either?
It is virtually always horizontal

**Rule of thumb:** If a pt has nystagmus + good VA, it's congenital motor nystagmus

**What condition does s/he most likely have?**
Congenital motor nystagmus
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

Blue-cone monochromatism

**aka...**

Finally: Note that blue-cone monochromatism is also known as monochromatism.
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

S-cone monochromatism

Blue-cone monochromatism

Finally: Note that blue-cone monochromatism is also known as S-cone monochromatism.
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

S-cone monochromatism

Blue-cone monochromatism

Finally: Note that blue-cone monochromatism is also known as S-cone monochromatism.

Why is it aka S-cone monochromatism? What’s the ‘S’ stand for?
Trichromatism  
Dichromatismo  
Monochromatism  

Why is it aka S-cone monochromatism? What’s the ‘S’ stand for?  
As noted earlier in the slide-set, blue light is of short wavelength,  
so blue cones are aka short-wavelength cones--S-cones for short  
(see what I did there?)
Note that blue-cone monochromatism is also known as S-cone monochromatism.

Other than knowing that the condition goes by two names, is there another reason that an awareness of the name S-cone monochromatism is noteworthy?
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Blue-cone monochromatism

Rod monochromatism

S-cone monochromatism

Finally: **Note that** blue-cone monochromatism is also known as **S-cone monochromatism**

*Other than knowing that the condition goes by two names, is there another reason that an awareness of the name S-cone monochromatism is noteworthy?*  
Indeed there is, and it's this: To make certain not to confuse S-cone monochromatism with the similarly-named but completely different condition, **enhanced S-cone syndrome**
Finally:

Note that blue-cone monochromatism is also known as S-cone monochromatism. Why is it aka S-cone monochromatism? What's the 'S' stand for?

As noted earlier in the slide-set, blue light is of short wavelength, so blue cones are aka short-wavelength cones--S-cones for short (see what I did there?)

Other than knowing that the condition goes by two names, is there another reason that an awareness of the name S-cone monochromatism is noteworthy?

Indeed there is, and it’s this: To make certain not to confuse S-cone monochromatism with the similarly named but completely different condition, enhanced S-cone syndrome.

Speaking of conditions with two names…'Enhanced S-cone syndrome’ is also known as what?

Goldmann-Favre syndrome

What sort of condition is it?

The BCSC Retina book calls it a “diffuse photoreceptor dystrophy” a la RP

How does it present?

With decreased acuity as well as night blindness

What is the appearance of enhanced S-cone syndrome on DFE?

Unlike the relatively normal appearance of the posterior pole in S-cone monochromatism, the posterior pole in enhanced S-cone syndrome is decidedly abnormal--retinoschisis as well as RP-like changes are the rule.
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Blue-cone monochromatism

Rod monochromatism

Congenital/Stationary Retinal Disease

Finally:

Note that blue-cone monochromatism is also known as S-cone monochromatism.

Why is it aka S-cone monochromatism? What's the 'S' stand for?

As noted earlier in the slide-set, blue light is of short wavelength, so blue cones are aka short-wavelength cones— S-cones for short (see what I did there?)

Other than knowing that the condition goes by two names, is there another reason that an awareness of the name S-cone monochromatism is noteworthy?

Indeed there is, and it’s this: To make certain not to confuse S-cone monochromatism with the similarly named but completely different condition, enhanced S-cone syndrome is also known as Goldmann-Favre syndrome.

Speaking of conditions with two names…’Enhanced S-cone syndrome’ is also known as what?

Goldmann-Favre syndrome

The BCSC Retina book calls it a “diffuse photoreceptor dystrophy” a la RP

How does it present?

With decreased acuity as well as night blindness

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Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Congenital/Stationary Retinal Disease

Finally:

Note that blue-cone monochromatism is also known as S-cone monochromatism.

Why is it aka S-cone monochromatism? What's the 'S' stand for?

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Other than knowing that the condition goes by two names, is there another reason that an awareness of the name S-cone monochromatism is noteworthy?

Indeed there is, and it's this. To make certain not to confuse S-cone monochromatism with the similarly named but completely different condition...

enhanced S-cone syndrome

Speaking of conditions with two names…'Enhanced S-cone syndrome’ is also known as what?

Goldmann-Favre syndrome

What sort of condition is it?

The BCSC Retina book calls it a "diffuse photoreceptor dystrophy" a la RP.

How does it present?

With decreased acuity as well as night blindness

What is the appearance of enhanced S-cone syndrome on DFE?

Unlike the relatively normal appearance of the posterior pole in S-cone monochromatism, the posterior pole in enhanced S-cone syndrome is decidedly abnormal—retinoschisis as well as RP-like changes are the rule.
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Indeed there is, and it’s this: To make certain not to confuse S-cone monochromatism with the similarly named but completely different condition enhanced S-cone syndrome.

Speaking of conditions with two names…’Enhanced S-cone syndrome’ is also known as what?
Goldmann-Favre syndrome

What sort of condition is it?
The BCSC Retina book calls it a “diffuse photoreceptor dystrophy” a la RP.

What is the appearance of enhanced S-cone syndrome on DFE?

Unlike the relatively normal appearance of the posterior pole in S-cone monochromatism, the posterior pole in enhanced S-cone syndrome is decidedly abnormal—retinoschisis as well as RP-like changes are the rule.
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

**In what ways are photoreceptors affected?**

Speaking of conditions with two names… Goldmann-Favre syndrome

What sort of condition is it?

The BCSC *Retina* book calls it a “diffuse photoreceptor dystrophy” a la RP

Indeed there is, and it’s this: To make certain not to confuse S-cone monochromatism with the similarly named but completely different condition **enhanced S-cone syndrome**
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Blue-cone monochromatism

Rod monochromatism

Congenital/Stationary Retinal Disease

Finally:

Note that blue-cone monochromatism is also known as S-cone monochromatism.

Why is it aka S-cone monochromatism? What's the 'S' stand for?

As noted earlier in the slide-set, blue light is of short wavelength, so blue cones are aka short-wavelength cones--S-cones for short (see what I did there?)

Other than knowing that the condition goes by two names, is there another reason that an awareness of the name S-cone monochromatism is noteworthy?

Indeed there is, and it’s this: To make certain not to confuse S-cone monochromatism with the similarly-named but completely different condition, enhanced S-cone syndrome.

Speaking of conditions with two names…'Enhanced S-cone syndrome' is also known as what?

Goldmann-Favre syndrome

What sort of condition is it?

The BCSC Retina book calls it a “diffuse photoreceptor dystrophy” a la RP

How does it present?

With decreased acuity as well as night blindness

What is the appearance of enhanced S-cone syndrome on DFE?

Unlike the relatively normal appearance of the posterior pole in S-cone monochromatism, the posterior pole in enhanced S-cone syndrome is decidedly abnormal--retinoschisis as well as RP-like changes are the rule

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Finally:

Note that blue-cone monochromatism is also known as S-cone monochromatism.

Why is it aka S-cone monochromatism? What's the 'S' stand for?

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Speaking of conditions with two names…'Enhanced S-cone syndrome' is also known as what?

Goldmann-Favre syndrome

What sort of condition is it?

The BCSC Retina book calls it a “diffuse photoreceptor dystrophy” a la RP

How does it present?

With decreased acuity as well as night blindness

What is the appearance of enhanced S-cone syndrome on DFE?

Unlike the relatively normal appearance of the posterior pole in S-cone monochromatism, the posterior pole in enhanced S-cone syndrome is decidedly abnormal—retinoschisis as well as RP-like changes are the rule

In what ways are photoreceptors affected?

--Rods: Non-functioning

--Red/green cones: Reduced in number.

--Blue cones: Increased in number
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Speaking of conditions with two names, blue-cone monochromatism is also known as S-cone monochromatism. What does the 'S' stand for?

As noted earlier in the slide-set, blue light is of short wavelength, so blue cones are also known as S-cones for short (see what I did there?).

Other than knowing that the condition goes by two names, is there another reason that an awareness of the name S-cone monochromatism is noteworthy?

Indeed there is, and it’s this: To make certain not to confuse S-cone monochromatism with the similarly-named but completely different condition enhanced S-cone syndrome.

S-cone monochromatism

Enlarged S-cone syndrome

What sort of condition is it?

The BCSC Retina book calls it a “diffuse photoreceptor dystrophy” a la RP.

How does it present?

With decreased acuity as well as night blindness.

What is the appearance of enhanced S-cone syndrome on DFE?

Unlike the relatively normal appearance of the posterior pole in S-cone monochromatism, the posterior pole in enhanced S-cone syndrome is decidedly abnormal—retinoschisis as well as RP-like changes are the rule.

In what ways are photoreceptors affected?

--Rods: Non-functioning

--Red/green cones: Reduced in number.

--Blue cones: Increased in number.

Enhanced S-cone syndrome
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism  Dichromatism

Speaking of conditions with two names... aka what?
Goldmann-Favre syndrome

What sort of condition is it?
The BCSC Retina book calls it a “diffuse photoreceptor dystrophy” a la RP

Indeed there is, and it’s this. To make certain not to confuse S-cone monochromatism with the similarly named but completely different condition...

enhanced S-cone syndrome

In what ways are photoreceptors affected?
--Rods: Non-functioning
--Red/green cones: Reduced in number
In what ways are photoreceptors affected?
--Rods: Non-functioning
--Red/green cones: Reduced in number
--Blue cones: ?

Why is it aka S-cone monochromatism? What's the 'S' stand for?
As noted earlier in the slide-set, blue light is of short wavelength, so blue cones are aka short-wavelength cones--S-cones for short (see what I did there?)

Other than knowing that the condition goes by two names, is there another reason that an awareness of the name S-cone monochromatism is noteworthy?
Indeed there is, and it’s this: To make certain not to confuse S-cone monochromatism with the similarly named but completely different condition: enhanced S-cone syndrome.
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Speaking of conditions with two names…

In what ways are photoreceptors affected?

- Rods: Non-functioning
- Red/green cones: Reduced in number
- Blue cones: \textbf{Increased} in number

Goldmann-Favre syndrome

What sort of condition is it?

The BCSC Retina book calls it a “diffuse photoreceptor dystrophy” a la RP

Indeed there is, and it’s this: To make certain not to confuse S-cone monochromatism with the similarly named but completely different condition: \textbf{enhanced S-cone syndrome}
**In what ways are photoreceptors affected?**

- Rods: Non-functioning
- Red/green cones: Reduced in number
- Blue cones: Increased in number

**What are the ERG findings?**

- Rod response: Undetectable
- Red/green cone response: Attenuated
- Blue cones: Enhanced (hence the name of the syndrome)

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**Speaking of conditions with two names…**

- Enhanced S-cone syndrome is also known as Goldmann-Favre syndrome.

**What sort of condition is it?**

The BCSC Retina book calls it a “diffuse photoreceptor dystrophy” a la RP.

**What are the ERG findings?**

- Rod response: Undetectable
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- Blue cones: Enhanced (hence the name of the syndrome)
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Blue-cone monochromatism

Rod monochromatism

Congenital/Stationary Retinal Disease

Finally:

Note that blue-cone monochromatism is also known as S-cone monochromatism.

Why is it aka S-cone monochromatism? What's the 'S' stand for?

As noted earlier in the slide-set, blue light is of short wavelength, so blue cones are aka short-wavelength cones—S-cones for short (see what I did there?)

Other than knowing that the condition goes by two names, is there another reason that an awareness of the name S-cone monochromatism is noteworthy?

Indeed there is, and it's this: To make certain not to confuse S-cone monochromatism with the similarly-named but completely different condition, enhanced S-cone syndrome.

Speaking of conditions with two names…'Enhanced S-cone syndrome' is also known as what?

Goldmann-Favre syndrome

What sort of condition is it?
The BCSC Retina book calls it a “diffuse photoreceptor dystrophy” a la RP

What are the ERG findings?

--Rod response: ?

--Red/green cone response: Attenuated

--Blue cones: Increased in number

In what ways are photoreceptors affected?

--Rods: Non-functioning

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enhanced S-cone syndrome
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Speaking of conditions with two names, what is also known as what?
Goldmann-Favre syndrome

What sort of condition is it?
The BCSC Retina book calls it a “diffuse photoreceptor dystrophy” a la RP

Indeed there is, and it’s this: To make certain not to confuse S-cone monochromatism with the similarly named but completely different condition, "enhanced S-cone syndrome"

In what ways are photoreceptors affected?
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Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Finally:

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Speaking of conditions with two names…'Enhanced S-cone syndrome' is also known as Goldmann-Favre syndrome.

What sort of condition is it?

The BCSC Retina book calls it a “diffuse photoreceptor dystrophy” a la RP.

How does it present?

With decreased acuity as well as night blindness.

What is the appearance of enhanced S-cone syndrome on DFE?

Unlike the relatively normal appearance of the posterior pole in S-cone monochromatism, the posterior pole in enhanced S-cone syndrome is decidedly abnormal--retinoschisis as well as RP-like changes are the rule.

What are the ERG findings?

--Rod response: Undetectable
--Red/green cone response: ?

In what ways are photoreceptors affected?

--Rods: Non-functioning
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What are the ERG findings?

--Red/green cone response: ?

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--Rod response: Undetectable
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Cone (Color) Disease

Rod (Night Vision) Disease

Congenital/Stationary Retinal Disease

Trichromatism

Dichromatism

Speaking of conditions with two names…

In what ways are photoreceptors affected?

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What sort of condition is it?

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enhanced S-cone syndrome
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Blue-cone monochromatism

Rod monochromatism

Congenital/Stationary Retinal Disease

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What sort of condition is it? The BCSC Retina book calls it a “diffuse photoreceptor dystrophy” a la RP

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--Rod response: Undetectable
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--Blue cones: ?

In what ways are photoreceptors affected?

--Rods: Non-functioning
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--Blue cones: Increased in number

Enhanced S-cone syndrome

Indeed there is, and it’s this: To make certain not to confuse S-cone monochromatism with the similarly named but completely different condition... **enhanced S-cone syndrome**
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Blue-cone monochromatism

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Congenital/Stationary Retinal Disease

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In what ways are photoreceptors affected?

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What are the ERG findings?

--Rod response: Undetectable
--Red/green cone response: Attenuated
--Blue cones: Enhanced (hence the name of the syndrome)
Full-field ERG in response to color stimuli for an unaffected individual and a patient with enhanced S-cone syndrome. Note that in the patient, responses to blue stimuli are larger than that of the unaffected individual. Note further that the pt’s response to the red stimulus is essentially nonexistent.
Cone (Color) Disease

Rod (Night Vision) Disease

Finally:

Note that blue-cone monochromatism is also known as S-cone monochromatism.

Why is it aka S-cone monochromatism? What's the 'S' stand for?

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Speaking of conditions with two names…'Enhanced S-cone syndrome' is also known as what?
Goldmann-Favre syndrome

What sort of condition is it?
The BCSC Retina book calls it a “diffuse photoreceptor dystrophy” a la RP

How does it present?

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Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Congenital/Stationary Retinal Disease

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The BCSC Retina book calls it a “diffuse photoreceptor dystrophy” a la RP

How does it present?

With decreased acuity as well as night blindness
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

110

Finally:

Note that blue-cone monochromatism is also known as S-cone monochromatism.

Why is it aka S-cone monochromatism? What's the 'S' stand for?

As noted earlier in the slide-set, blue light is of short wavelength, so blue cones are aka short-wavelength cones--S-cones for short (see what I did there?)

Other than knowing that the condition goes by two names, is there another reason that an awareness of the name S-cone monochromatism is noteworthy?

Indeed there is, and it's this: To make certain not to confuse S-cone monochromatism with the similarly named but completely different condition enhanced S-cone syndrome.

Speaking of conditions with two names…'Enhanced S-cone syndrome’ is also known as what?
Goldmann-Favre syndrome

What sort of condition is it?
The BCSC Retina book calls it a “diffuse photoreceptor dystrophy” a la RP

How does it present?
With decreased acuity as well as night blindness

What is the appearance of enhanced S-cone syndrome on DFE?

Unlike the relatively normal appearance of the posterior pole in S-cone monochromatism, the posterior pole in enhanced S-cone syndrome is decidedly abnormal--retinoschisis as well as RP-like changes are the rule.
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

1. Cone (Color) Disease
2. Rod (Night Vision) Disease
3. Trichromatism
4. Dichromatism

Finally:

Note that blue-cone monochromatism is also known as S-cone monochromatism.

Why is it aka S-cone monochromatism? What's the 'S' stand for?

As noted earlier in the slide-set, blue light is of short wavelength, so blue cones are aka short-wavelength cones—S-cones for short (see what I did there?)

Other than knowing that the condition goes by two names, is there another reason that an awareness of the name S-cone monochromatism is noteworthy?

Indeed there is, and it's this: To make certain not to confuse S-cone monochromatism with the similarly-named but completely different condition, enhanced S-cone syndrome.

Speaking of conditions with two names…'Enhanced S-cone syndrome' is also known as what?

Goldmann-Favre syndrome

What sort of condition is it?
The BCSC Retina book calls it a “diffuse photoreceptor dystrophy” a la RP

How does it present?
With decreased acuity as well as night blindness

What is the appearance of enhanced S-cone syndrome on DFE?
Unlike the relatively normal appearance of the posterior pole in S-cone monochromatism, the posterior pole in enhanced S-cone syndrome is decidedly abnormal—retinoschisis as well as RP-like changes are the rule.

enhanced S-cone syndrome
Enhanced S-cone syndrome
Congenital/Stationary Retinal Disease

Cone (Color) Disease
- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease
- ?
- ?

Two general categories, not specific conditions
Congenital/Stationary Retinal Disease

Cone (Color) Disease

- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease

- Fundus appearance normal
- Fundus appearance abnormal

Fundus appearance normal

?
Congenital/Stationary Retinal Disease

Cone (Color) Disease
- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease
- Fundus appearance: normal
- Fundus appearance: abnormal
- CSNB
Congenital/Stationary Retinal Disease

Cone (Color) Disease

- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease

- Fundus appearance normal
- Fundus appearance abnormal

CSNB

What does CNSB stand for in this context?
Congenital/Stationary Retinal Disease

Cone (Color) Disease
- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease
- Fundus appearance
- Fundus appearance
  - normal
  - abnormal

What does CNSB stand for in this context?
Congenital stationary night blindness
Congenital/Stationary Retinal Disease

- Cone (Color) Disease
  - Trichromatism
  - Dichromatism
  - Monochromatism

- Rod (Night Vision) Disease
  - Fundus appearance normal
  - CSNB
  - Fundus appearance abnormal

Foreshadowing alert: We will soon see that while, strictly speaking, the fundus appears normal in CSNB, the posterior pole may not!
**Congenital/Stationary Retinal Disease**

- **Cone (Color) Disease**
- **Rod (Night Vision) Disease**

  **Trichromatism** → **Dichromatism** → **Monochromatism**

**Congenital Stationary Night Blindness (CSNB)**

-- Several inheritance patterns; most common =

- **Fundus appearance normal**
- **Fundus appearance abnormal**

**CSNB**
**Congenital/Stationary Retinal Disease**

- **Cone (Color) Disease**
  - Trichromatism
  - Dichromatism
  - Monochromatism

- **Rod (Night Vision) Disease**
  - Fundus appearance normal
  - Fundus appearance abnormal

---

**Congenital Stationary Night Blindness (CSNB)**

-- Several inheritance patterns; most common = X-linked

- VA 20/20 - 20/200
- Refractive error: Usually myopia
- Presents in childhood with:
  - Nystagmus
  - Decreased vision
  - Nyctalopia

-- 'Most common pattern: 'Negative' ERG

- 'Negative' ERG = Large \(a\)-wave, no \(b\)-wave
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Trichromatism

Monochromatism

Dichromatism

Rod (Night Vision) Disease

Fundus appearance normal

Fundus appearance abnormal

Congenital Stationary Night Blindness (CSNB)
--Several inheritance patterns; most common = X-linked
--Pathology: Communication failure between…
[two cell types]
Congenital/Stationary Retinal Disease

Cone (Color) Disease

- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease

- Fundus appearance normal
- Fundus appearance abnormal

**Congenital Stationary Night Blindness (CSNB)**

- Several inheritance patterns; most common = X-linked
- Pathology: Communication failure between… photoreceptors & bipolar cells
Retinal Layers
- Internal limiting membrane
- Nerve fiber layer
- Ganglion cell layer
- Inner plexiform layer
- Inner nuclear layer
- Outer plexiform layer (Henle’s layer)
- Outer nuclear layer
- External limiting membrane
- Rod & cone inner and outer segments

RPE

Bruch’s membrane

The photoreceptors, bipolar and ganglion cells comprise the vertical retinal pathway—vertical in the sense that it is the direct path from photic stimulation to the CNS processing centers.

It is at the PR-bipolar cell interface that the pathology of CSNB resides.
Congenital Stationary Night Blindness (CSNB)

--Several inheritance patterns; most common = X-linked
--Pathology: Communication failure between… photoreceptors & bipolar cells
--VA range: ?
Cone (Color) Disease

Rod (Night Vision) Disease

-- Several inheritance patterns; most common = X-linked
-- Pathology: Communication failure between photoreceptors & bipolar cells
-- VA range: 20/20 - 20/200

Congenital Stationary Night Blindness (CSNB)

Fundus appearance

normal

abnormal

CSNB
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Fundus appearance normal

Fundus appearance abnormal

**Congenital Stationary Night Blindness (CSNB)**

--Several inheritance patterns; most common = X-linked
--Pathology: Communication failure between photoreceptors & bipolar cells
--VA range: 20/20 - 20/200
--Refractive error: Usually…?
**Congenital Stationary Night Blindness (CSNB)**

--Several inheritance patterns; most common = X-linked
--Pathology: Communication failure between... photoreceptors & bipolar cells
--VA range: 20/20 - 20/200
--Refractive error: Usually... myopia
**Congenital/Stationary Retinal Disease**

**Cone (Color) Disease**
- Trichromatism
- Dichromatism
- Monochromatism

**Rod (Night Vision) Disease**
- Fundus appearance normal
- Fundus appearance abnormal

**Congenital Stationary Night Blindness (CSNB)**
- Several inheritance patterns; most common = X-linked
- Pathology: Communication failure between photoreceptors & bipolar cells
- VA range: 20/20 - \(20/200\)
- Refractive error: Usually... **myopia**

*When VA is poor in CSNB, it’s usually due to the (high) myopia, not the photoreceptors*
**Congenital Stationary Night Blindness (CSNB)**
--Several inheritance patterns; most common = X-linked
--Pathology: Communication failure between… photoreceptors & bipolar cells
--VA range: 20/20 - 20/200
--Refractive error: Usually…myopia
--Presents in childhood with:
  --?
  --?
  --?
**Congenital Stationary Night Blindness (CSNB)**

--Several inheritance patterns; most common = X-linked
--Pathology: Communication failure between... photoreceptors & bipolar cells
--VA range: 20/20 - 20/200
--Refractive error: Usually...myopia
--Presents in childhood with:
  --Nystagmus
  --Decreased vision
  --Nyctalopia
Congenital/Stationary Retinal Disease

- Cone (Color) Disease
- Rod (Night Vision) Disease

Trichromatism  Dichromatism  Monochromatism

Fundus appearance normal  Fundus appearance abnormal

### Congenital Stationary Night Blindness (CSNB)
- Several inheritance patterns; most common = X-linked
- Pathology: Communication failure between... photoreceptors & bipolar cells
- VA range: 20/20 - 20/200
- Refractive error: Usually...myopia
- Presents in childhood with:
  - Nystagmus
  - Decreased vision
  - Nyctalopia

**What is nyctalopia?**

As they have had extremely poor night vision their entire lives, it seems normal to them—they don't know any different.
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Congenital Stationary Night Blindness (CSNB)
--Several inheritance patterns; most common = X-linked
--Pathology: Communication failure between... photoreceptors & bipolar cells
--VA range: 20/20 - 20/200
--Refractive error: Usually...myopia
--Presents in childhood with:
  --Nystagmus
  --Decreased vision
  --Nyctalopia

What is nyctalopia?
Night blindness

Fundus appearance normal

CSNB

Fundus appearance abnormal
**Congenital/Stationary Retinal Disease**

- **Cone (Color) Disease**
- **Rod (Night Vision) Disease**

**Trichromatism**
- **Dichromatism**
- **Monochromatism**

---

**Congenital Stationary Night Blindness (CSNB)**
- Several inheritance patterns; most common = X-linked
- Pathology: Communication failure between... photoreceptors & bipolar cells
- VA range: 20/20 - 20/200
- Refractive error: Usually...myopia
- Presents in childhood with:
  - Nystagmus
  - Decreased vision
  - **Nyctalopia**

---

**Fundus appearance**
- **normal**
- **abnormal**

---

**What is nyctalopia?**
Night blindness

Many CSNB children do **not** complain of nyctalopia. Why not?
**Congenital Stationary Night Blindness (CSNB)**

--Several inheritance patterns; most common = X-linked
--Pathology: Communication failure between photoreceptors & bipolar cells
--VA range: 20/20 - 20/200
--Refractive error: Usually...myopia
--Presents in childhood with:
  --Nystagmus
  --Decreased vision
  --**Nyctalopia**

**What is nyctalopia?**
Night blindness

**Many CSNB children do not complain of nyctalopia. Why not?**
As they have had extremely poor night vision their entire lives, it seems normal to them—they don’t know any different
**Congenital Stationary Night Blindness (CSNB)**

--Several inheritance patterns; most common = X-linked
--Pathology: Communication failure between photoreceptors & bipolar cells
--VA range: 20/20 - 20/200
--Refractive error: Usually…myopia
--Presents in childhood with:
  --Nystagmus
  --Decreased vision
  --Nyctalopia
--Classified according to…[Psychophysical test]
**Congenital Stationary Night Blindness (CSNB)**

- Several inheritance patterns; most common = X-linked
- Pathology: Communication failure between... photoreceptors & bipolar cells
- VA range: 20/20 - 20/200
- Refractive error: Usually... myopia
- Presents in childhood with:
  - Nystagmus
  - Decreased vision
  - Nyctalopia
- Classified according to... Scotopic ERG pattern

---

**Congenital/Stationary Retinal Disease**

- Cone (Color) Disease
- Rod (Night Vision) Disease

**Fundus appearance**

- normal
- abnormal

---

**CSNB**
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Fundus appearance normal

Fundus appearance abnormal

Congenital Stationary Night Blindness (CSNB)

--Several inheritance patterns; most common = X-linked
--Pathology: Communication failure between... photoreceptors & bipolar cells
--VA range: 20/20 - 20/200
--Refractive error: Usually...myopia
--Presents in childhood with:
  --Nystagmus
  --Decreased vision
  --Nyctalopia
--Classified according to... Scotopic ERG pattern

In other words, the *dark-adapted* ERG. (Remember, it’s in the dark that CSNB pts have their difficulty.) Abnormalities of the photopic or light-adapted ERG also occur in CSNB, but are much more subtle.
Congenital/Stationary Retinal Disease

Cone (Color) Disease

- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease

- Fundus appearance normal
- Fundus appearance abnormal

**Congenital Stationary Night Blindness (CSNB)**

- Several inheritance patterns; most common = X-linked
- Pathology: Communication failure between… photoreceptors & bipolar cells
- VA range: 20/20 - 20/200
- Refractive error: Usually…myopia
- Presents in childhood with:
  - Nystagmus
  - Decreased vision
  - Nyctalopia
- Classified according to…Scotopic ERG pattern
- Most common pattern: ?
### Congenital Stationary Night Blindness (CSNB)

- Several inheritance patterns; most common = X-linked
- Pathology: Communication failure between… photoreceptors & bipolar cells
- VA range: 20/20 - 20/200
- Refractive error: Usually… myopia
- Presents in childhood with:
  - Nystagmus
  - Decreased vision
  - Nyctalopia
- Classified according to… Scotopic ERG pattern
- Most common pattern: **Negative ERG**
**Congenital Stationary Night Blindness (CSNB)**
--Several inheritance patterns; most common = X-linked
--Pathology: Communication failure between… photoreceptors & bipolar cells
--VA range: 20/20 - 20/200
--Refractive error: Usually…myopia
--Presents in childhood with:
  --Nystagmus
  --Decreased vision
  --Nyctalopia
--Classified according to…Scotopic ERG pattern
--Most common pattern: *Negative* ERG
  --*Negative* ERG =
**Congenital Stationary Night Blindness (CSNB)**
--Several inheritance patterns; most common = X-linked
--Pathology: Communication failure between photoreceptors & bipolar cells
--VA range: 20/20 - 20/200
--Refractive error: Usually...myopia
--Presents in childhood with:
  --Nystagmus
  --Decreased vision
  --Nyctalopia
--Classified according to...Scotopic ERG pattern
  --Most common pattern: *Negative* ERG
  --*Negative* ERG = Large *a*-wave, smaller *b*-wave
In a normal ERG, the $b$-wave is much larger than the $a$-wave.
In a normal ERG, the $b$-wave is much larger than the $a$-wave.

The $b$-wave is said to be ‘negative’ when it is smaller than the $a$-wave, as is the case in CSNB.
**Congenital/Stationary Retinal Disease**

- **Cone (Color) Disease**
- **Rod (Night Vision) Disease**

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**Cone (Color) Disease**
- **Trichromatism**
- **Dichromatism**
- **Monochromatism**

**Rod (Night Vision) Disease**
- **Fundus appearance normal**
- **Fundus appearance abnormal**

---

**Congenital Stationary Night Blindness (CSNB)**
- Several inheritance patterns; most common = X-linked
- Pathology: Communication failure between... photoreceptors & bipolar cells
- **VA range:** 20/20 - 20/200
- **Refractive error:** Usually... myopia
- Presents in childhood with:
  - Nystagmus
  - Decreased vision
  - Nyctalopia
- Classified according to... Scotopic ERG pattern
  - Most common pattern: **Negative ERG**
  - **Negative ERG = Large a-wave, smaller b-wave**

Another condition—not common, but moreso than CSNB—also presents with a negative ERG. What is it?

**XLR**
- X-linked retinoschisis
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Fundus appearance normal

Fundus appearance abnormal

Congenital Stationary Night Blindness (CSNB)
--Several inheritance patterns; most common = X-linked
--Pathology: Communication failure between... photoreceptors & bipolar cells

Another condition—not common, but moreso than CSNB—also presents with a negative ERG. What is it?
XLR

Nystagmus

--Classified according to...Scotopic ERG pattern
--Most common pattern: Negative ERG
--Negative ERG = Large a-wave, smaller b-wave
**Congenital Stationary Night Blindness (CSNB)**

- Several inheritance patterns; most common = X-linked
- Pathology: Communication failure between...photoreceptors & bipolar cells

Another condition—not common, but moreso than CSNB—also presents with a negative ERG. What is it?

XLR

*What does XLR stand for in this context?*

Nystagmus

- Classified according to...Scotopic ERG pattern
  -- Most common pattern: *Negative ERG*
  -- *Negative ERG* = Large $a$-wave, smaller $b$-wave
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

**Congenital Stationary Night Blindness (CSNB)**

--Several inheritance patterns; most common = X-linked
--Pathology: Communication failure between...photoreceptors & bipolar cells

Another condition—not common, but moreso than CSNB—also presents with a negative ERG. What is it? XLR

What does XLR stand for in this context?
X-linked retinoschisis

--Classified according to...Scotopic ERG pattern
--Most common pattern: **Negative ERG**
--**Negative ERG** = Large $a$-wave, smaller $b$-wave
XLR patient

Light flash

a-wave

Negative b-wave

X-linked retinoschisis: ERG
**Congenital/Stationary Retinal Disease**

- **Cone (Color) Disease**
- **Rod (Night Vision) Disease**

**Trichromatism**

**Dichromatism**

**Monochromatism**

**Congenital Stationary Night Blindness (CSNB)**
- Several inheritance patterns; most common = X-linked
- Pathology: Communication failure between... photoreceptors & bipolar cells
- VA range: 20/20 - 20/200
- Refractive error: Usually...myopia
- Presents in childhood with:
  - Nystagmus
  - Decreased vision
  - Nyctalopia
- Classified according to...Scotopic ERG pattern
- Most common pattern: **Negative ERG**
- **Negative ERG** = Large a-wave, smaller b-wave

Another condition—not common, but moreso than CSNB—also presents with a negative ERG. What is it?

- XLR

What does XLR stand for in this context?

X-linked retinoschisis

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

- Splitting within the layers of the neurosensory retina
Congenital/Stationary Retinal Disease

Cone (Color) Disease

- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease

- Fundus appearance: normal
- Fundus appearance: abnormal

**Congenital Stationary Night Blindness (CSNB)**

- Several inheritance patterns; most common = X-linked
- Pathology: Communication failure between... photoreceptors & bipolar cells
- VA range: 20/20 - 20/200
- Refractive error: Usually... myopia
- Presents in childhood with:
  - Nystagmus
  - Decreased vision
  - Nyctalopia
- Classified according to... Scotopic ERG pattern
  - Most common pattern: *Negative* ERG
  - *Negative* ERG = Large *a*-wave, smaller *b*-wave

Another condition—not common, but moreso than CSNB—also presents with a negative ERG. What is it?

**XLR**

What does XLR stand for in this context?

**X-linked retinoschisis**

---

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

Splitting within the layers of the neurosensory retina
Congenital/Stationary Retinal Disease

X-linked retinoschisis
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Trichromatism

Dichromatism

Monochromatism

Rod (Night Vision) Disease

Fundus appearance normal

Fundus appearance abnormal

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

X-linked retinoschisis

What does XLR stand for in this context?

X-linked retinoschisis

Classified according to...Scotopic ERG pattern

Most common pattern: Negative ERG

Negative ERG = Large $a$-wave, smaller $b$-wave
**Congenital/Stationary Retinal Disease**

- **Cone (Color) Disease**
  - Trichromatism
  - Dichromatism
  - Monochromatism

- **Rod (Night Vision) Disease**
  - Fundus appearance normal
  - Fundus appearance abnormal

---

**Congenital Stationary Night Blindness (CSNB)**
- Several inheritance patterns; most common = X-linked
- Pathology: Communication failure between photoreceptors & bipolar cells
- VA range: 20/20 - 20/200
- Refractive error: Usually...myopia
- Presents in childhood with:
  - Nystagmus
  - Decreased vision
  - Nyctalopia
- Classified according to...Scotopic ERG pattern
  - Most common pattern: Negative ERG
  - Negative ERG = Large a-wave, smaller b-wave

Another condition—not common, but moreso than CSNB—also presents with a negative ERG. What is it?

**XLR**

What does XLR stand for in this context?

X-linked retinoschisis (XLR) known as

- X-linked juvenile retinoschisis (XLJR)

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

X-linked juvenile retinoschisis (XLJR)

---

What does XLR stand for in this context?

**X-linked retinoschisis**

Nystagopia

- Classified according to...Scotopic ERG pattern
  - Most common pattern: Negative ERG
  - Negative ERG = Large a-wave, smaller b-wave
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Trichromatism

Dichromatism

Monochromatism

Rod (Night Vision) Disease

Fundus appearance normal

Fundus appearance abnormal

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

X-linked juvenile retinoschisis (XLJR)

What does XLR stand for in this context?

X-linked retinoschisis

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

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

---

What is the most common pattern of Scotopic ERG for CSNB?

Negative ERG

---

Negative ERG = Large a-wave, smaller b-wave

---

Another condition—not common, but moreso than CSNB—also presents with a negative ERG. What is it?

XLR

What does XLR stand for in this context?

X-linked retinoschisis

---

X-linked juvenile retinoschisis (XLJR)

---

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 implied—correctly—by the word juvenile above?

That the condition manifests early in life (in fact, it is congenital)
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Trichromatism

Dichromatism

Monochromatism

Rod (Night Vision) Disease

Fundus appearance normal

Fundus appearance abnormal

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

What is implied—correctly—by the word juvenile above?  
That the condition manifests early in life (in fact, it is congenital)

What does XLR stand for in this context?  
X-linked retinoschisis

--Classified according to...Scotopic ERG pattern

--Most common pattern: Negative ERG

--Negative ERG = Large $a$-wave, smaller $b$-wave
Congenital/Stationary Retinal Disease

Cone (Color) Disease

- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease

- Fundus appearance normal
- Fundus appearance abnormal

Congenital Stationary Night Blindness (CSNB)

- Several inheritance patterns; most common = X-linked
- Pathology: Communication failure between photoreceptors & bipolar cells
- VA range: 20/20 - 20/200
- Refractive error: Usually...myopia
- Presents in childhood with:
  - Nystagmus
  - Decreased vision
  - Nyctalopia
- Classified according to...Scotopic ERG pattern
- Most common pattern: Negative ERG
- Negative ERG = Large a-wave, smaller b-wave

Another condition—not common, but moreso than CSNB—also presents with a negative ERG. What is it? XLR

What does XLR stand for in this context?

X-linked retinoschisis

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

X-linked juvenile retinoschisis (XLJR)

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

That the condition manifests early in life (in fact, it is congenital)
Congenital/Stationary Retinal Disease

Cone (Color) Disease
- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease
- Fundus appearance normal
- Fundus appearance abnormal

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

How does it present on DFE?

What does XLR stand for in this context?
- X-linked retinoschisis

Classified according to...Scotopic ERG pattern
- Most common pattern: Negative ERG
  - Negative ERG = Large a-wave, smaller b-wave
By what other (very similar) name is X-linked retinoschisis (XLR) known?
X-linked juvenile retinoschisis (XLJR)

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

Cone (Color) Disease

- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease

- Fundus appearance normal
- Fundus appearance abnormal

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

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

What does XLR stand for in this context?
X-linked retinoschisis

- classified according to...Scotopic ERG pattern
- most common pattern: Negative ERG
- Negative ERG = Large a-wave, smaller b-wave
**Congenital/Stationary Retinal Disease**

**Cone (Color) Disease**
- Trichromatism
- Dichromatism
- Monochromatism

**Rod (Night Vision) Disease**
- Fundus appearance normal
- Fundus appearance abnormal

---

**Congenital Stationary Night Blindness (CSNB)**
- Several inheritance patterns; most common = X-linked
- Pathology: Communication failure between photoreceptors & bipolar cells
- VA range: 20/20 - 20/200
- Refractive error: Usually…myopia
- Presents in childhood with:
  - Nystagmus
  - Decreased vision
  - Nyctalopia
- Classified according to...Scotopic ERG pattern
  - Most common pattern: Negative ERG
  - Negative ERG = Large $a$-wave, smaller $b$-wave

---

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

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 does XLR stand for in this context?
X-linked retinoschisis

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Another condition—not common, but moreso than CSNB—also presents with a negative ERG. What is it?
XLR

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Congenital/Stationary Retinal Disease

Cone (Color) Disease

- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease

- Fundus appearance normal
- Fundus appearance abnormal

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

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

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

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--Classified according to...Scotopic ERG pattern
--Most common pattern: Negative ERG
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Congenital Stationary Night Blindness (CSNB)

--Several inheritance patterns; most common = X-linked
--Pathology: Communication failure between...photoreceptors & bipolar cells
--VA range: 20/20 - 20/200
--Refractive error: Usually...myopia
--Presents in childhood with:
  --Nystagmus
  --Decreased vision
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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.

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What does X-linked retinoschisis (XLR) mean in context?

X-linked retinoschisis (XLR) is a genetic condition that affects the macula of the retina. It is characterized by the presence of a macular schisis, which can be radially oriented and may involve peripheral schisis as well. The condition usually presents in early childhood with reduced visual acuity and may progress over time.

On DFE (Digital Fundus Examination), XLR typically manifests as macular schisis, often with a radial pattern. Peripheral schisis may also be present.

The condition is primarily associated with a negative ERG (electroretinogram), where the a-wave is larger than the b-wave. This is in contrast to CSNB, which typically shows a positive ERG pattern.

XLR is also known as X-linked juvenile retinoschisis (XLJR) and shares some similarities with CSNB in terms of its presentation and clinical features. However, XLR is less common than CSNB but is more prevalent than CSNB in the context of negative ERG patterns.
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism
Dichromatism
Monochromatism

Fundus appearance normal
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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
Congenital/Stationary Retinal Disease

Cone (Color) Disease  
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CSNB  
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Fundus appearance normal

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Congenital Stationary Night Blindness (CSNB)
--Several inheritance patterns; most common = X-linked

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--Nystagmus

--Decreased vision

--Nyctalopia

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Trichromatism

Dichromatism

Monochromatism

MAR

What does MAR stand for in this context?

Melanoma-associated retinopathy
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

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Monochromatism

Fundus appearance normal

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Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

Fundus appearance abnormal

Cone (Color) Disease

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Trichromatism

Dichromatism

Monochromatism

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Melanoma-associated retinopathy

Melanoma-associated retinopathy
--A paraneoplastic process in which retinal cells display antigens that are identical to, or cross-react with, melanoma cells within the body. Subsequent to sensitization to these antigens on the melanoma cells, the immune system attacks the same/similar antigens in the retina.

What is melanoma-associated retinopathy?
Congenital/Stationary Retinal Disease

Cone (Color) Disease

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Cone (Color) Disease

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- Dichromatism
- Monochromatism

Rod (Night Vision) Disease

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- CSNB

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Trichromatism  Dichromatism  Monochromatism

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In what very important way does the night blindness of MAR differ from that of CSNB? The night blindness in MAR is acquired, not congenital.
**Congenital/Stationary Retinal Disease**

**Cone (Color) Disease**

**Rod (Night Vision) Disease**

**Trichromatism**

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**Congenital Stationary Night Blindness (CSNB)**

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Photopsias, often described as **shimmering**
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Congenital Stationary Night Blindness (CSNB)

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What other psychophysical test is always abnormal in CSNB?

Fundus appearance normal

Rod (Night Vision) Disease

Fundus appearance abnormal

Cone (Color) Disease

Trichromatism

Dichromatism

Monochromatism

Congenital/Stationary Retinal Disease

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-- Presents in childhood with:

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-- Decreased vision

-- Nyctalopia

-- Classified according to...Scotopic ERG pattern

-- Most common pattern:

Negative ERG = Large a-wave, no b-wave

What other psychophysical test is always abnormal in CSNB?

Dark adaptometry

What does dark adaptometry assess?
The increase in sensitivity that occurs when the background illumination is low. That is to say, the longer an eye is in the dark, the dimmer the light it can perceive (up to a point).

In what way is dark adaptometry abnormal in CSNB?

Due to the lack of functioning rods, the cone-rod break never kicks in—adaptation remains at the cone maximum, with the result being poor vision under very dim conditions.
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism
Dichromatism
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- Fundus appearance: normal
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**Congenital Stationary Night Blindness (CSNB)**
- Several inheritance patterns; most common = X-linked
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- **What does dark adaptometry assess?**
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This is what a *dark adaptation curve* looks like. As you can see, sensitivity increases to a maximum after about 30 minutes.
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But what's up with this weird hiccup in the curve?
Congenital/Stationary Retinal Disease

Here's what's up. Early on, the response is dominated by the cones.
Here’s what’s up. Early on, the response is dominated by the cones. However, note that the cones’ ability to dark-adapt is limited, and plateaus after ~10 minutes.
Congenital/Stationary Retinal Disease

Meanwhile, the rods have been quietly dark-adapting along with the cones.
Meanwhile, the rods have been quietly dark-adapting along with the cones. Note that their dark-adaptation capacity is much greater than that of the cones.
If you overlay the rod curve atop the cone curve, you end up with the classic dark-adaptation curve.
If you overlay the rod curve atop the cone curve, you end up with the classic dark-adaptation curve. It turns out the ‘hiccup’ is the point where rod sensitivity overtakes that of the cones (and hence is called the *cone-rod break*).
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Dark adaptometry

What does dark adaptometry assess?

The increase in sensitivity that occurs when the background illumination is low. That is to say, the longer an eye is in the dark, the dimmer the light it can perceive (up to a point).

In what way is dark adaptometry abnormal in CSNB?
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

Fundus appearance normal

Fundus appearance abnormal

**Congenital Stationary Night Blindness (CSNB)**

-- Several inheritance patterns; most common = X-linked

-- What other psychophysical test is always abnormal in CSNB? Dark adaptometry

-- What does dark adaptometry assess?
  The increase in sensitivity that occurs when the background illumination is low. That is to say, the longer an eye is in the dark, the dimmer the light it can perceive (up to a point).

-- In what way is dark adaptometry abnormal in CSNB?
  Due to the lack of functioning rods, the cone-rod break never kicks in—adaptation remains at the cone maximum, with the result being poor vision under very dim conditions.
Dark-adaptometry curve in CSNB (filled circles). Note the lack of rod adaptation (ie, it looks just like the *cones only* graph a few slides back).
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Trichromatism

Dichromatism

Monochromatism

Rod (Night Vision) Disease

Fundus appearance normal

Fundus appearance abnormal

**Congenital Stationary Night Blindness (CSNB)**

--Several inheritance patterns; most common = X-linked
--Pathology: Communication failure between... photoreceptors & bipolar cells
--VA range: 20/20 - 20/200
--Refractive error: Usually...myopia
--Presents in childhood with:
  - Nystagmus
  - Decreased vision
  - Nyctalopia
--Classified according to...Scotopic ERG pattern
--Most common pattern: Negative ERG = Large a-wave, no b-wave

Recall we said earlier that the posterior pole exam may not be normal.

(No question yet—keep going)
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Congenital Stationary Night Blindness (CSNB)

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  - Nystagmus
  - Decreased vision
  - Nyctalopia
-- Classified according to... Scotopic ERG pattern
-- Most common pattern: Negative ERG; large a-wave, no b-wave

Recall we said earlier that the posterior pole exam may not be normal.

What structure in particular may be abnormal in appearance?
Congenital/Stationary Retinal Disease

Cone (Color) Disease

- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease

- Fundus appearance normal
- Fundus appearance abnormal

**Congenital Stationary Night Blindness (CSNB)**

- Several inheritance patterns; most common = X-linked
- Pathology: Communication failure between... photoreceptors & bipolar cells
- VA range: 20/20 - 20/200
- Refractive error: Usually... myopia
  - Presents in childhood with:
    - Nystagmus
    - Decreased vision
    - Nyctalopia

Recall we said earlier that the posterior pole exam may not be normal. **What structure in particular may be abnormal in appearance?**

The optic nerve head
Congenital Stationary Night Blindness (CSNB)
--Several inheritance patterns; most common = X-linked
--Pathology: Communication failure between... photoreceptors & bipolar cells
--VA range: 20/20 - 20/200
--Refractive error: Usually...myopia
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  --Nystagmus
  --Decreased vision
  --Nyctalopia
--Classified according to...Scotopic ERG pattern
  --Most common pattern: Negative ERG = Large a-wave, no b-wave

Recall we said earlier that the posterior pole exam may not be normal. What structure in particular may be abnormal in appearance? The optic nerve head

In what manner is the ONH likely to be abnormal in CSNB?
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Congenital Stationary Night Blindness (CSNB)
--Several inheritance patterns; most common = X-linked
--Pathology: Communication failure between… photoreceptors & bipolar cells
--VA range: 20/20 - 20/200
--Refractive error: Usually…myopia
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  - Decreased vision
  - Nyctalopia
--Classified according to…Scotopic ERG pattern
--Most common pattern:
  - Negative ERG = Large a-wave, no b-wave

Recall we said earlier that the posterior pole exam may not be normal.
What structure in particular may be abnormal in appearance?
The optic nerve head

In what manner is the ONH likely to be abnormal in CSNB?
It may manifest a... two words
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Fundus appearance normal

Fundus appearance abnormal

Congenital Stationary Night Blindness (CSNB)

--Several inheritance patterns; most common = X-linked
--Pathology: Communication failure between…photoreceptors & bipolar cells
--VA range: 20/20 - 20/200
--Refractive error: Usually…myopia
--Presents in childhood with:
  - Nystagmus
  - Decreased vision
  - Nyctalopia
--Classified according to…Scotopic ERG pattern
--Most common pattern: Negative ERG
  - Negative ERG = Large a-wave, no b-wave

Recall we said earlier that the posterior pole exam may not be normal. What structure in particular may be abnormal in appearance?
The optic nerve head

In what manner is the ONH likely to be abnormal in CSNB?
It may manifest a myopic tilt
**Congenital/Stationary Retinal Disease**

- **Cone (Color) Disease**
- **Rod (Night Vision) Disease**

**Cone (Color) Disease**
- Trichromatism
- Dichromatism
- Monochromatism

**Rod (Night Vision) Disease**
- Fundus appearance normal
- Fundus appearance abnormal

---

**Congenital Stationary Night Blindness (CSNB)**

- Several inheritance patterns; most common = X-linked
- Pathology: Communication failure between... photoreceptors & bipolar cells
- VA range: 20/20 - 20/200
- Refractive error: Usually... **myopia**
- Presents in childhood with:
  - Nystagmus
  - Decreased vision
  - Nyctalopia

---

Recall we said earlier that the posterior pole exam may not be normal. What structure in particular may be abnormal in appearance?

The optic nerve head

*If you can remember one of these facts, it should help you remember the other*

In what manner is the ONH likely to be abnormal in CSNB?

It may manifest a **myopic tilt**
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Fundus appearance normal

Fundus appearance abnormal

**Congenital Stationary Night Blindness (CSNB)**

-- Several inheritance patterns; most common = X-linked
-- Pathology: Communication failure between photoreceptors & bipolar cells
-- VA range: 20/20 - 20/200
-- Refractive error: Usually...myopia
-- Presents in childhood with:
  - Nystagmus
  - Decreased vision
  - Nyctalopia
-- Classified according to...Scotopic ERG pattern
-- Most common pattern: Negative ERG = Large a-wave, no b-wave

Recall we said earlier that the posterior pole exam may not be normal. What structure in particular may be abnormal in appearance?
The optic nerve head

*In what manner is the ONH likely to be abnormal in CSNB?*
It may manifest a myopic tilt, and its nasal vs temporal aspect may be pallorous
**Congenital Stationary Night Blindness (CSNB)**
--Several inheritance patterns; most common = X-linked
--Pathology: Communication failure between…photoreceptors & bipolar cells
--VA range: 20/20 - 20/200
--Refractive error: Usually…myopia
--Presents in childhood with:
  --Nystagmus
  --Decreased vision
  --Nyctalopia
--Classified according to…Scotopic ERG pattern
--Most common pattern:
  --Negative ERG = Large a-wave, no b-wave

Recall we said earlier that the posterior pole exam may not be normal. **What structure in particular may be abnormal in appearance?**
The optic nerve head

**In what manner is the ONH likely to be abnormal in CSNB?**
It may manifest a **myopic tilt**, and its temporal aspect may be pallorous
Congenital Stationary Night Blindness (CSNB)
--Several inheritance patterns; most common = X-linked
--Pathology: Communication failure between... photoreceptors & bipolar cells
--VA range: 20/20 - 20/200
--Refractive error: Usually...myopia
--Presents in childhood with:
  --Nystagmus
  --Decreased vision
  --Nyctalopia
--Classified according to...Scotopic ERG pattern
--Most common pattern: Negative ERG = Large a-wave, no b-wave

Recall we said earlier that the posterior pole exam may not be normal. What structure in particular may be abnormal in appearance?
The optic nerve head

In what manner is the ONH likely to be abnormal in CSNB?
It may manifest a myopic tilt, and its temporal aspect may be pallorous

What is the eponymous name for a tilted disc of this sort?
Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)
Congenital Stationary Night Blindness (CSNB)
--Several inheritance patterns; most common = X-linked
--Pathology: Communication failure between… photoreceptors & bipolar cells
--VA range: 20/20 - 20/200
--Refractive error: Usually…myopia
--Presents in childhood with:
  --Nystagmus
  --Decreased vision
  --Nyctalopia
--Classified according to…Scotopic ERG pattern
  --Most common pattern: Negative ERG
    --Negative ERG = Large $a$-wave, no $b$-wave

Recall we said earlier that the posterior pole exam may not be normal.
*What structure in particular may be abnormal in appearance?*
The optic nerve head

*In what manner is the ONH likely to be abnormal in CSNB?*
It may manifest a myopic tilt, and its temporal aspect may be pallorous

*What is the eponymous name for a tilted disc of this sort?*
Fuchs coloboma
**Congenital/Stationary Retinal Disease**

- **Cone (Color) Disease**
- **Rod (Night Vision) Disease**

**Trichromatism** → **Dichromatism** → **Monochromatism**

---

**Congenital Stationary Night Blindness (CSNB)**

--Several inheritance patterns; most common = X-linked
--Pathology: Communication failure between... photoreceptors & bipolar cells
--VA range: 20/20 - 20/200
--Refractive error: Usually...myopia
--Presents in childhood with:
  - Nystagmus
  - Decreased vision
  - Nyctalopia
--Classified according to...Scotopic ERG pattern

--Most common pattern: Negative ERG = Large a-wave, no b-wave

Recall we said earlier that the posterior pole exam may not be normal.

*What structure in particular may be abnormal in appearance?*

The optic nerve head

*In what manner is the ONH likely to be abnormal in CSNB?*

It may manifest a **myopic tilt**, and its **temporal** aspect may be pallorous

*What is the eponymous name for a tilted disc of this sort?*

**Fuchs coloboma** (Dr Fuchs strikes again—I stan an eye king!)

---

Ernst Fuchs
1851-1930
**Congenital/Stationary Retinal Disease**

- **Cone (Color) Disease**
- **Rod (Night Vision) Disease**

**Trichromatism** → **Dichromatism** → **Monochromatism**

**Congenital Stationary Night Blindness (CSNB)**
--Several inheritance patterns; most common = X-linked

- **CSNB**
  - Fundus appearance: normal
  - Fundus appearance: abnormal

---

**What does it mean to say the ONH is ‘tilted’?**

**In what manner is the ONH likely to be abnormal in CSNB?**
It may manifest a myopic tilt, and its temporal aspect may be pallorous.

**What is the eponymous name for a tilted disc of this sort?**
Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Fundus appearance normal

Fundus appearance abnormal

Congenital Stationary Night Blindness (CSNB)
--Several inheritance patterns; most common = X-linked

What does it mean to say the ONH is ‘tilted’?
It means its inferior vs superior pole is elevated and its inferior vs superior pole is posteriorly displaced (ie, staphyloma-like)

In what manner is the ONH likely to be abnormal in CSNB?
It may manifest a myopic tilt and its temporal aspect may be pallorous

What is the eponymous name for a tilted disc of this sort?
Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

Fundus appearance
normal

Fundus appearance
abnormal

Trichromatism

Dichromatism

Monochromatism

Congenital Stationary Night Blindness (CSNB)
--Several inheritance patterns; most common = X-linked

What does it mean to say the ONH is ‘tilted’?
It means its superior pole is elevated and its inferior pole is posteriorly displaced (ie, staphyloma-like)

In what manner is the ONH likely to be abnormal in CSNB?
It may manifest a myopic tilt and its temporal aspect may be pallorous

What is the eponymous name for a tilted disc of this sort?
Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)
Congenital/Stationary Retinal Disease

Tilted disc
Congenital/Stationary Retinal Disease

Cone (Color) Disease

- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease

- Fundus appearance normal
- Fundus appearance abnormal

Congenital Stationary Night Blindness (CSNB)
--Several inheritance patterns; most common = X-linked

**What does it mean to say the ONH is ‘tilted’?**
It means its superior pole is elevated and its inferior pole is posteriorly displaced (ie, staphyloma-like)

*The vessels on a tilted disc may run in an unusual pattern. What is the two-word name for this pattern?*

**In what manner is the ONH likely to be abnormal in CSNB?**
It may manifest a myopic tilt and its temporal aspect may be pallorous

*What is the eponymous name for a tilted disc of this sort? Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)*
**Congenital Stationary Night Blindness (CSNB)**

--Several inheritance patterns; most common = X-linked

---

**What does it mean to say the ONH is 'tilted'?**

It means its superior pole is elevated and its inferior pole is posteriorly displaced (ie, staphyloma-like)

---

**The vessels on a tilted disc may run in an unusual pattern. What is the two-word name for this pattern?**

'Situs inversus'

---

**In what manner is the ONH likely to be abnormal in CSNB?**

It may manifest a myopic tilt and its temporal aspect may be pallorous

---

**What is the eponymous name for a tilted disc of this sort?**

Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)
### Congenital Stationary Night Blindness (CSNB)

--Several inheritance patterns; most common = X-linked

What does it mean to say the ONH is ‘tilted’?
It means its superior pole is elevated and its inferior pole is posteriorly displaced (ie, staphyloma-like)

The vessels on a tilted disc may run in an unusual pattern. What is the two-word name for this pattern? ‘Situs inversus’

Huh? I thought situs inversus meant all the organs were on the wrong side of the body, or something. What does it mean in this context?

It may manifest a myopic tilt and its temporal aspect may be pallorous

What is the eponymous name for a tilted disc of this sort? Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)
Congenital Stationary Night Blindness (CSNB)
--Several inheritance patterns; most common = X-linked

What does it mean to say the ONH is ‘tilted’?
It means its superior pole is elevated and its inferior pole is posteriorly displaced (ie, staphyloma-like)

The vessels on a tilted disc may run in an unusual pattern. What is the two-word name for this pattern?
‘Situs inversus’

Huh? I thought situs inversus meant all the organs were on the wrong side of the body, or something. What does it mean in this context?
It means the vessels run nasally for a short interval before heading off in the right direction

It may manifest a myopic tilt and its temporal aspect may be pallorous

What is the eponymous name for a tilted disc of this sort?
Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)
**Congenital Stationary Night Blindness (CSNB)**

--Several inheritance patterns; most common = X-linked

What does it mean to say the ONH is ‘tilted’?
It means its superior pole is elevated and its inferior pole is posteriorly displaced (ie, staphyloma-like)

The vessels on a tilted disc may run in an unusual pattern. What is the two-word name for this pattern? 'Situs inversus'

Huh? I thought situs inversus meant all the organs were on the wrong side of the body, or something. What does it mean in this context? It means the temporal vessels run nasally for a short interval before heading off in the right direction

It may manifest a myopic tilt and its temporal aspect may be pallorous

What is the eponymous name for a tilted disc of this sort? Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)
Congenital/Stationary Retinal Disease

Tilted disc: Situs inversus
Congenital Stationary Night Blindness (CSNB)
--Several inheritance patterns; most common = X-linked

What does it mean to say the ONH is ‘tilted’?
It means its superior pole is elevated and its inferior pole is posteriorly displaced (ie, staphyloma-like)

The vessels on a tilted disc may run in an unusual pattern. What is the two-word name for this pattern?
‘Situs inversus’

Does any of this impact vision?

In what manner is the ONH likely to be abnormal in CSNB?
It may manifest a myopic tilt and its temporal aspect may be pallorous

What is the eponymous name for a tilted disc of this sort?
Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)
**Congenital Stationary Night Blindness (CSNB)**

--Several inheritance patterns; most common = X-linked

**What does it mean to say the ONH is ‘tilted’?**
It means its superior pole is elevated and its inferior pole is posteriorly displaced (ie, staphyloma-like)

**The vessels on a tilted disc may run in an unusual pattern. What is the two-word name for this pattern?**
‘Situs inversus’

**Does any of this impact vision?**
It does indeed—the tilt of the ONH may produce a visual field defect (classically, a bitemporal defect).

**In what manner is the ONH likely to be abnormal in CSNB?**
It may manifest a myopic tilt, and its temporal aspect may be pallorous

**What is the eponymous name for a tilted disc of this sort?**
Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)
**Congenital/Stationary Retinal Disease**

- **Cone (Color) Disease**
- **Rod (Night Vision) Disease**

**Trichromatism** → **Dichromatism** → **Monochromatism**

---

**Congenital Stationary Night Blindness (CSNB)**

-- Several inheritance patterns; most common = X-linked

---

- **Fundus appearance normal**
- **Fundus appearance abnormal**

---

**Congenital Stationary Night Blindness (CSNB)**

What does it mean to say the ONH is ‘tilted’?
It means its superior pole is elevated and its inferior pole is posteriorly displaced (ie, staphyloma-like)

The vessels on a tilted disc may run in an unusual pattern. What is the two-word name for this pattern?
‘Situs inversus’

Does any of this impact vision?
It does indeed—the tilt of the ONH may produce a visual field defect

In what manner is the ONH likely to be abnormal in CSNB?
It may manifest a myopic tilt, and its temporal aspect may be pallorous

What is the eponymous name for a tilted disc of this sort?
Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)
**Congenital Stationary Night Blindness (CSNB)**
--Several inheritance patterns; most common = X-linked

*What does it mean to say the ONH is ‘tilted’?*
It means its superior pole is elevated and its inferior pole is posteriorly displaced (i.e., staphyloma-like).

*The vessels on a tilted disc may run in an unusual pattern. What is the two-word name for this pattern?*
‘Situs inversus’

*Does any of this impact vision?*
It does indeed—the tilt of the ONH may produce a visual field defect (classically, a bitemporal defect).

*In what manner is the ONH likely to be abnormal in CSNB?*
It may manifest a **myopic tilt** and its temporal aspect may be pallorous.

*What is the eponymous name for a tilted disc of this sort?*
Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)
**Congenital Stationary Night Blindness (CSNB)**

--Several inheritance patterns; most common = X-linked

- **Fundus appearance**
  - normal
  - **CSNB**

**What does it mean to say the ONH is ‘tilted’?**
It means its superior pole is elevated and its inferior pole is posteriorly displaced (ie, staphyloma-like)

**The vessels on a tilted disc may run in an unusual pattern. What is the two-word name for this pattern?**
‘Situs inversus’

**Does any of this impact vision?**
It does indeed—the tilt of the ONH may produce a visual field defect *(classically, a bitemporal defect)*

**In what manner is the ONH likely to be abnormal in CSNB?**
It may manifest a myopic *tilt* and its temporal aspect may be pallorous

**What is the eponymous name for a tilted disc of this sort?**
Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Fundus appearance normal

Fundus appearance abnormal

How on earth does a tilted disc produce a bitemporal VF defect?

Does any of this impact vision?

It does indeed—the tilt of the ONH may produce a visual field defect (classically, a bitemporal defect).

In what manner is the ONH likely to be abnormal in CSNB?

It may manifest a myopic tilt, and its temporal aspect may be pallorous.

What is the eponymous name for a tilted disc of this sort?

Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Fundus appearance normal

Fundus appearance abnormal

Congenital Stationary Night Blindness (CSNB)

-- Several inheritance patterns; most common = X-linked

-- Pathology: Communication failure between photoreceptors & bipolar cells

-- VA range: 20/20 - 20/200

-- Refractive error: Usually...myopia

-- Presents in childhood with:

-- Nystagmus

-- Decreased vision

-- Nyctalopia

-- Classified according to...Scotopic ERG pattern

-- Most common pattern:

  - **Negative** ERG = Large a-wave, no b-wave

Recall we said earlier that the posterior pole exam may not be normal. What structure in particular may be abnormal in appearance?

The optic nerve head

In what manner is the ONH likely to be abnormal in CSNB?

It may manifest a myopic tilt, and its temporal aspect may be pallorous

What is the eponymous name for a tilted disc of this sort?

Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)

How on earth does a tilted disc produce a bitemporal VF defect?

It's actually pretty straightforward. Recall that the area including and adjacent to the inferior pole of the ONH is staphylomatous. This means the ‘axial length’ of the photoreceptors within this region is greater than that of the rest of the posterior pole. Because of this extra axial length, the correction used during VF testing (which is based on the refraction of the non-staphylomatous fovea) is not myopic enough for the inferior peripapillary region.

Does any of this impact vision?

It does indeed—the tilt of the ONH may produce a visual field defect (classically, a bitemporal defect).

In what manner is the ONH likely to be abnormal in CSNB?

It may manifest a myopic tilt, and its temporal aspect may be pallorous.

What is the eponymous name for a tilted disc of this sort?

Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Fundus appearance normal

Fundus appearance abnormal

How on earth does a tilted disc produce a bitemporal VF defect?
It's actually pretty straightforward.
Recall that the area including and adjacent to the inferior pole of the ONH is staphylomatous.
This means the ‘axial length’ of the photoreceptors within this region is greater than that of the rest of the posterior pole. Because of this extra axial length, the correction used during VF testing (which is based on the refraction of the non-staphylomatous fovea) is not myopic enough for the inferior peripapillary region. Because this region is out of focus, it will manifest a refractive scotoma on the test. And because the retina involved in this scotoma is inferonasal to the fovea, it follows that the resulting VF defect will be superotemporal to fixation.

In what manner is the ONH likely to be abnormal in CSNB?
It may manifest a myopic tilt and its temporal aspect may be pallorous.

What is the eponymous name for a tilted disc of this sort?
Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)
How on earth does a tilted disc produce a bitemporal VF defect?
It's actually pretty straightforward.
Recall that the area including and adjacent to the inferior pole of the ONH is staphylomatous. This means the ‘axial length’ of the photoreceptors within this region is greater than that of the rest of the posterior pole. Because of this extra axial length, the correction used during VF testing (which is based on the refraction of the non-staphylomatous fovea) is not myopic enough for the inferior peripapillary region. Because this region is out of focus, it will manifest a refractive scotoma on the test. And because the retina involved in this scotoma is inferonasal to the fovea, it follows that the resulting VF defect will be superotemporal to fixation. And, as Fuchs coloboma is virtually always bilateral, these superotemporal VF defects are present bilaterally.

Does any of this impact vision?
It does indeed—the tilt of the ONH may produce a visual field defect (classically, a bitemporal defect).

In what manner is the ONH likely to be abnormal in CSNB?
It may manifest a myopic tilt and its temporal aspect may be pallorous.

What is the eponymous name for a tilted disc of this sort?
Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)
Tilted disc: Superior bitemporal VF defects
**How on earth does a tilted disc produce a bitemporal VF defect?**

It's actually pretty straightforward. Recall that the area including and adjacent to the inferior pole of the ONH is staphylomatous. This means the ‘axial length’ of the photoreceptors within this region is greater than that of the rest of the posterior pole. Because of this extra axial length, the correction used during VF testing (which is based on the refraction of the non-staphylomatous fovea) is not myopic enough for the inferior peripapillary region. Because this region is out of focus, it will manifest a refractive scotoma on the test. And because Fuchs coloboma is virtually always bilateral, these superotemporal VF defects are present bilaterally.

Does any of this impact vision? It does indeed—the tilt of the ONH may produce a visual field defect (classically, a bitemporal defect).

In what manner is the ONH likely to be abnormal in CSNB? It may manifest a myopic tilt and its temporal aspect may be pallorous.

What is the eponymous name for a tilted disc of this sort? Fuchs coloboma (Dr. Fuch's strikes again—I stan for an eye king!)

This implies the VF defects resolve if the proper refractive correction is employed. Do they?
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Fundus appearance normal

Fundus appearance abnormal

Congenital Stationary Night Blindness (CSNB)

--Several inheritance patterns; most common = X-linked
--Pathology: Communication failure between photoreceptors & bipolar cells
--VA range: 20/20 - 20/200
--Refractive error: Usually...myopia
--Presents in childhood with:
  --Nystagmus
  --Decreased vision
  --Nyctalopia
--Classified according to...Scotopic ERG pattern
--Most common pattern:
  Negative ERG = Large a-wave, no b-wave

Recall we said earlier that the posterior pole exam may not be normal.
What structure in particular may be abnormal in appearance?
The optic nerve head

In what manner is the ONH likely to be abnormal in CSNB?
It may manifest a myopic tilt, and its temporal aspect may be pallorous

What is the eponymous name for a tilted disc of this sort?
Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)

How on earth does a tilted disc produce a bitemporal VF defect?
It's actually pretty straightforward.
Recall that the area including and adjacent to the inferior pole of the ONH is staphylomatous.
This means the ‘axial length’ of the photoreceptors within this region is greater than that of the rest of the posterior pole. Because of this extra axial length, the correction used during VF testing (which is based on the refraction of the non-staphylomatous fovea) is not myopic enough for the inferior peripapillary region. Because this region is out of focus, it will manifest a refractive scotoma on the test. And because Fuchs coloboma is virtually always bilateral, these refractive scotomas are expected to be bilateral as well.

Does any of this impact vision?
It does indeed—the tilt of the ONH may produce a visual field defect (classically, a bitemporal defect)

In what manner is the ONH likely to be abnormal in CSNB?
This implies the VF defects resolve if the proper refractive correction is employed. Do they?
Indeed they do, and this cinches the diagnosis

What is the eponymous name for a tilted disc of this sort?
Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

When I hear ‘superior bitemporal VF defect’ two words come to mind, and they ain’t Fuchs coloboma—they are **pituitary tumor**.

How on earth does a tilted disc produce a bitemporal VF defect?

It’s actually pretty straightforward.

Recall that the area including and adjacent to the inferior pole of the ONH is staphylomatous. This means the ‘axial length’ of the photoreceptors within this region is greater than that of the rest of the posterior pole. Because of this extra axial length, the correction used during VF testing (which is based on the refraction of the non-staphylomatous fovea) is not myopic enough for the inferior peripapillary region. Because this region is out of focus, it will manifest a refractive scotoma on the test. And because the retina involved in this scotoma is inferonasal to the fovea, it follows that the resulting VF defect will be superotemporal to fixation. And, as Fuchs coloboma is virtually always bilateral, these superotemporal VF defects are present bilaterally.

In what manner is the ONH likely to be abnormal in CSNB?

It may manifest a myopic **tilt** and its temporal aspect may be pallorous.

What is the eponymous name for a tilted disc of this sort?

Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

When I hear ‘superior bitemporal VF defect’ two words come to mind, and they ain’t Fuchs coloboma—they are pituitary tumor.

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In what manner is the ONH likely to be abnormal in CSNB? It may manifest a myopic tilt, and its temporal aspect may be pallorous.

What is the eponymous name for a tilted disc of this sort? Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

**When I hear ‘superior bitemporal VF defect’ two words come to mind, and they ain’t Fuchs coloboma—they are pituitary tumor. How am I supposed to know whether a bitemporal VF cut results from a pituitary tumor as opposed to Fuchs coloboma?**

How do I know whether a bitemporal VF defect comes from a pituitary tumor or not?

It's actually pretty straightforward.

Recall that the area including and adjacent to the inferior pole of the ONH is staphylomatous. This means the ‘axial length’ of the photoreceptors within this region is greater than that of the rest of the posterior pole. Because of this extra axial length, the correction used during VF testing (which is based on the refraction of the non-staphylomatous fovea) is not myopic enough for the inferior peripapillary region. Because this region is out of focus, it will manifest a refractive scotoma on the test. And because the retina involved in this scotoma is inferonasal to the fovea, it follows that the resulting VF defect will be superotemporal to fixation. And, as Fuchs coloboma is virtually always bilateral, these superotemporal VF defects are present bilaterally.

Does any of this impact vision?

It does indeed—the tilt of the ONH may produce a visual field defect (classically, a bitemporal defect).

In what manner is the ONH likely to be abnormal in CSNB?

It may manifest a myopic tilt and its temporal aspect may be pallorous.

What is the eponymous name for a tilted disc of this sort?

Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)
When I hear ‘superior bitemporal VF defect’ two words come to mind, and they ain’t Fuchs coloboma—they are pituitary tumor. How am I supposed to know whether a bitemporal VF cut results from a pituitary tumor as opposed to Fuchs coloboma? You mean, other than the fact that the ONHs in a Fuchs pt will be highly tilted and manifest situs inversus, whereas the ONHs of a pituitary-tumor pt will be edematous? (Remember: When all else fails, examine the pt.)

Recall we said earlier that the posterior pole exam may not be normal. What structure in particular may be abnormal in appearance? The optic nerve head

In what manner is the ONH likely to be abnormal in CSNB? It may manifest a myopic tilt and its temporal aspect may be pallorous

What is the eponymous name for a tilted disc of this sort? Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

When I hear ‘superior bitemporal VF defect’ two words come to mind, and they ain’t Fuchs coloboma—they are pituitary tumor. How am I supposed to know whether a bitemporal VF cut results from a pituitary tumor as opposed to Fuchs coloboma? You mean, other than the fact that the ONHs in a Fuchs pt will be highly tilted and manifest situs inversus, whereas the ONHs of a pituitary-tumor pt will be edematous? (Remember: When all else fails, examine the pt.) OK, fair. (Unnecessarily rude, but fair.) But is there a way to tell from the VF itself? Indeed there is—a bitemporal VF defect secondary to a pituitary tumor will always respect the vertical midline, whereas one secondary to Fuchs coloboma will not. Put another way: A pituitary tumor, but not a Fuchs coloboma, is expected to produce bitemporal hemianopic VF loss.

In what manner is the ONH likely to be abnormal in CSNB? It may manifest a myopic tilt and its temporal aspect may be pallorous. What is the eponymous name for a tilted disc of this sort? Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!).
When I hear ‘superior bitemporal VF defect’ two words come to mind, and they ain’t Fuchs coloboma—they are pituitary tumor. How am I supposed to know whether a bitemporal VF cut results from a pituitary tumor as opposed to Fuchs coloboma?

You mean, other than the fact that the ONHs in a Fuchs pt will be highly tilted and manifest situs inversus, whereas the ONHs of a pituitary-tumor pt will be edematous? (Remember: When all else fails, examine the pt.)

OK, fair. (Unnecessarily rude, but fair.) But is there a way to tell from the VF itself? Indeed there is—a bitemporal VF defect 2ndry to a pituitary tumor will always respect the vertical vs horizontal midline, whereas one 2ndry to Fuchs coloboma will not.

In what manner is the ONH likely to be abnormal in CSNB? It may manifest a myopic tilt and its temporal aspect may be pallorous.

What is the eponymous name for a tilted disc of this sort? Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

When I hear ‘superior bitemporal VF defect’ two words come to mind, and they ain’t Fuchs coloboma— they are **pituitary tumor**.  **How am I supposed to know whether a bitemporal VF cut results from a pituitary tumor as opposed to Fuchs coloboma?**  You mean, other than the fact that the ONHs in a Fuchs pt will be highly tilted and manifest situs inversus, whereas the ONHs of a pituitary-tumor pt will be edematous?  (Remember: **When all else fails, examine the pt.**)

*OK, fair. (Unnecessarily rude, but fair.) But is there a way to tell from the VF itself?** Indeed there is—a bitemporal VF defect 2ndry to a pituitary tumor will always respect the vertical midline, whereas one 2ndry to Fuchs coloboma will not.

**In what manner is the ONH likely to be abnormal in CSNB?**  It may manifest a **myopic tilt** and its temporal aspect may be pallorous

**What is the eponymous name for a tilted disc of this sort?**  Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

When I hear ‘superior bitemporal VF defect’ two words come to mind, and they ain’t Fuchs coloboma—they are **pituitary tumor**. How am I supposed to know whether a bitemporal VF cut results from a pituitary tumor as opposed to Fuchs coloboma?

You mean, other than the fact that the ONHs in a Fuchs pt will be highly tilted and manifest situs inversus, whereas the ONHs of a pituitary-tumor pt will be edematous? (Remember: When all else fails, examine the pt.)

**OK, fair. (Unnecessarily rude, but fair.) But is there a way to tell from the VF itself?** Indeed there is—a bitemporal VF defect 2ndry to a pituitary tumor will always respect the vertical midline, whereas one 2ndry to Fuchs coloboma will not. **Put another way:** A pituitary tumor, but not a Fuchs coloboma, is expected to produce bitemporal hemianopic VF loss.

Does any of this impact vision? It does indeed—the tilt of the ONH may produce a visual field defect (classically, a superotemporal defect). These superotemporal VF defects are present bilaterally.

**In what manner is the ONH likely to be abnormal in CSNB?** It may manifest a myopic tilt and its temporal aspect may be pallorous.

**What is the eponymous name for a tilted disc of this sort?** Fuchs coloboma (Dr Fuchs strikes again—I stan for an eye king!)
Congenital/Stationary Retinal Disease

Cone (Color) Disease
- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease
- Fundus appearance normal
- Fundus appearance abnormal
  - CSNB
  - ?
  - ?

Next question
Congenital/Stationary Retinal Disease

Cone (Color) Disease
- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease
- Fundus appearance normal
  - CSNB
- Fundus appearance abnormal
  - Fundus albipunctatus
  - Oguchi disease
Cone (Color) Disease

- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease

- Fundus appearance normal
- Fundus appearance abnormal
  - CSNB
  - Fundus albipunctatus
  - Oguchi disease

**Fundus Albipunctatus**
--Pathology: Delayed regeneration of the photopigment...
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Fundus appearance normal

Fundus appearance abnormal

CSNB

Fundus albipunctatus

Oguchi disease

Fundus Albipunctatus

--Pathology: Delayed regeneration of the photopigment…rhodopsin
Fundus Albipunctatus
--Pathology: Delayed regeneration of the photopigment…rhodopsin
--Dark adaptation is abnormal:
   --Initially, patients are…[condition], with abnormal… [test]
**Fundus Albipunctatus**

--Pathology: Delayed regeneration of the photopigment…rhodopsin
--Dark adaptation is abnormal:
   --Initially, patients are…night-blind, with abnormal…rod ERG
Fundus Albipunctatus
--Pathology: Delayed regeneration of the photopigment…rhodopsin
--Dark adaptation is abnormal:
  --Initially, patients are…night-blind, with abnormal…rod ERG
  --With enough time, will dark-adapt, and ERG normalizes
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Trichromatism

Dichromatism

Monochromatism

Rod (Night Vision) Disease

Fundus Albipunctatus

--Pathology: Delayed regeneration of the photopigment...rhodopsin
--Dark adaptation is abnormal:
  --Initially, patients are...night-blind, with abnormal...rod ERG
  --With enough time, will

How much time are we talking about?
Congenital/Stationary Retinal Disease

Cone (Color) Disease

- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease

- Fundus appearance normal
- Fundus appearance abnormal
- CSNB
- Fundus albipunctatus
- Oguchi disease

Fundus Albipunctatus

--- Pathology: Delayed regeneration of the photopigment...rhodopsin
--- Dark adaptation is abnormal:
--- Initially, patients are...night-blind, with abnormal...rod ERG
--- With enough time...

How much time are we talking about?
Several hours at least
Congenital/Stationary Retinal Disease

Delayed dark adaptation in fundus albipunctatus
**Fundus Albipunctatus**

--Pathology: Delayed regeneration of the photopigment...rhodopsin
--Dark adaptation is abnormal:
  --Initially, patients are...night-blind, with abnormal...rod ERG
  --With enough time, will dark-adapt, and ERG normalizes
--DFE: Striking array of...?
**Fundus Albipunctatus**

--Pathology: Delayed regeneration of the photopigment…rhodopsin
--Dark adaptation is abnormal:
  --Initially, patients are…night-blind, with abnormal…rod ERG
  --With enough time, will dark-adapt, and ERG normalizes
--DFE: Striking array of…**yellow - white dots**
Fundus albipunctatus
Fundus Albipunctatus
--Pathology: Delayed regeneration of the photopigment…rhodopsin
--Dark adaptation is abnormal:
  --Initially, patients are…night-blind, with abnormal…rod ERG
  --With enough time, will dark-adapt, and ERG normalizes
--DFE: Striking array of…**yellow - white dots**
--Dots found in entire posterior pole except…?
**Fundus Albipunctatus**

--Pathology: Delayed regeneration of the photopigment...rhodopsin
--Dark adaptation is abnormal:
  --Initially, patients are...night-blind, with abnormal...rod ERG
  --With enough time, will dark-adapt, and ERG normalizes
--DFE: Striking array of...**yellow** - **white dots**
--Dots found in entire posterior pole except...fovea
What is the main disease that must be differentiated from fundus albipunctatus?

Fundus Albipunctatus
--Pathology: Delayed regeneration of the photopigment...rhodopsin
--Dark adaptation is abnormal:
  --Initially, patients are...night-blind, with abnormal...rod ERG
  --With enough time, will dark-adapt, and ERG normalizes
--DFE: Striking array of...yellow - white dots
--Dots found in entire posterior pole except...fovea

Fundus Albipunctatus
--Pathology: Delayed regeneration of the photopigment...rhodopsin
--Dark adaptation is abnormal:
  --Initially, patients are...night-blind, with abnormal...rod ERG
  --With enough time, will dark-adapt, and ERG normalizes
--DFE: Striking array of...yellow - white dots
--Dots found in entire posterior pole except...fovea
What is the main disease that must be differentiated from fundus albipunctatus?
Retinitis punctata albscens

**Fundus Albipunctatus**
--Pathology: Delayed regeneration of the photopigment...rhodopsin
--Dark adaptation is abnormal:
  --Initially, patients are...night-blind, with abnormal...rod ERG
  --With enough time, will dark-adapt, and ERG normalizes
--DFE: Striking array of...yellow - white dots
--Dots found in entire posterior pole except...fovea
**Fundus Albinotatus**

--Pathology: Delayed regeneration of the photopigment…rhodopsin
--Dark adaptation is abnormal:
  --Initially, patients are…night-blind, with abnormal…rod ERG
  --With enough time, will dark-adapt, and ERG normalizes
--DFE: Striking array of…*yellow - white dots*
--Dots found in entire posterior pole except…fovea

What is the main disease that must be differentiated from *fundus albinotatus*?
Retinitis punctata albscens

What is *retinitis punctata albscens*?

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Trichromatism
Dichromatism
Monochromatism

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Fundus appearance normal

Fundus appearance abnormal

CSNB

Fundus albinotatus

Oguchi disease

Fundus Albipunctatus

--Pathology: Delayed regeneration of the photopigment…rhodopsin
--Dark adaptation is abnormal:
  --Initially, patients are…night-blind, with abnormal…rod ERG
  --With enough time, will dark-adapt, and ERG normalizes
--DFE: Striking array of…*yellow - white dots*
--Dots found in entire posterior pole except…fovea

What is the main disease that must be differentiated from *fundus albinotatus*?
Retinitis punctata albscens

What is *retinitis punctata albscens*?
What is the main disease that must be differentiated from fundus albipunctatus? Retinitis punctata albescens

What is retinitis punctata albescens?
An variant characterized by white - yellow dots similar to those of albipunctatus

Fundus Albipunctatus
--Pathology: Delayed regeneration of the photopigment...rhodopsin
--Dark adaptation is abnormal:
  --Initially, patients are...night-blind, with abnormal...rod ERG
  --With enough time, will dark-adapt, and ERG normalizes
--DFE: Striking array of...yellow - white dots
--Dots found in entire posterior pole except...fovea
**Fundus Albipunctatus**

--Pathology: Delayed regeneration of the photopigment...rhodopsin
--Dark adaptation is abnormal:
  --Initially, patients are...night-blind, with abnormal...rod ERG
  --With enough time, will dark-adapt, and ERG normalizes
--DFE: Striking array of...**yellow - white dots**
--Dots found in entire posterior pole except...fovea

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**What is the main disease that must be differentiated from fundus albipunctatus?**
Retinitis punctata albscens

**What is retinitis punctata albscens?**
An RP variant characterized by white - yellow dots similar to those of albipunctatus

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**Fundus Albipunctatus**

--Pathology: Delayed regeneration of the photopigment...rhodopsin
--Dark adaptation is abnormal:
  --Initially, patients are...night-blind, with abnormal...rod ERG
  --With enough time, will dark-adapt, and ERG normalizes
--DFE: Striking array of...**yellow - white dots**
--Dots found in entire posterior pole except...fovea
Retinitis punctata albescens
What is the main disease that must be differentiated from fundus albinpunctatus?  
Retinitis punctata albecens

What is retinitis punctata albecens? 
An RP variant characterized by white - yellow dots similar to those of albinpunctatus

How do fundus albinpunctatus and retinitis punctata albecens differ?

Fundus Albinpunctatus
--Pathology: Delayed regeneration of the photopigment...rhodopsin 
--Dark adaptation is abnormal:
  --Initially, patients are...night-blind, with abnormal...rod ERG 
  --With enough time, will dark-adapt, and ERG normalizes
--DFE: Striking array of...yellow - white dots
--Dots found in entire posterior pole except...fovea
**Fundus Albipunctatus**

--Pathology: Delayed regeneration of the photopigment...rhodopsin
--Dark adaptation is abnormal:
  --Initially, patients are...night-blind, with abnormal...rod ERG
  --With enough time, will dark-adapt, and ERG normalizes
--DFE: Striking array of...**yellow - white dots**
  --Dots found in entire posterior pole except...fovea

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**What is the main disease that must be differentiated from fundus albipunctatus?**
Retinitis punctata albescens

**What is retinitis punctata albescens?**
An RP variant characterized by white - yellow dots similar to those of albipunctatus

**How do fundus albipunctatus and retinitis punctata albescens differ?**
--On DFE: ?
--On ERG

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**Fundus Albipunctatus**

--Pathology: Delayed regeneration of the photopigment...rhodopsin
--Dark adaptation is abnormal:
  --Initially, patients are...night-blind, with abnormal...rod ERG
  --With enough time, will dark-adapt, and ERG normalizes
--DFE: Striking array of...**yellow - white dots**
  --Dots found in entire posterior pole except...fovea
What is the main disease that must be differentiated from fundus albipunctatus?
Retinitis punctata albscens

What is retinitis punctata albscens?
An RP variant characterized by white - yellow dots similar to those of albipunctatus

How do fundus albipunctatus and retinitis punctata albscens differ?
--On DFE: Like other forms of RP, retinitis punctata albscens demonstrates arteriolar narrowing, whereas albipunctatus does not
--On ERG

Fundus Albipunctatus
--Pathology: Delayed regeneration of the photopigment...rhodopsin
--Dark adaptation is abnormal:
  --Initially, patients are...night-blind, with abnormal...rod ERG
  --With enough time, will dark-adapt, and ERG normalizes
--DFE: Striking array of...yellow - white dots
--Dots found in entire posterior pole except...fovea
What is the main disease that must be differentiated from fundus albipunctatus?
Retinitis punctata albscens

What is retinitis punctata albscens?
An RP variant characterized by white - yellow dots similar to those of albipunctatus

How do fundus albipunctatus and retinitis punctata albscens differ?
--On DFE: Like other forms of RP, retinitis punctata albscens demonstrates arteriolar narrowing, whereas albipunctatus does not
--On ERG: ?

Fundus Albipunctatus
--Pathology: Delayed regeneration of the photopigment...rhodopsin
--Dark adaptation is abnormal:
  --Initially, patients are...night-blind, with abnormal...rod ERG
  --With enough time, will dark-adapt, and ERG normalizes
--DFE: Striking array of...yellow - white dots
  --Dots found in entire posterior pole except...fovea
What is the main disease that must be differentiated from fundus albipunctatus?
Retinitis punctata albscens

What is retinitis punctata albscens?
An RP variant characterized by white - yellow dots similar to those of albipunctatus

How do fundus albipunctatus and retinitis punctata albscens differ?
--On DFE: Like other forms of RP, retinitis punctata albscens demonstrates arteriolar narrowing, whereas albipunctatus does not
--On ERG: Fundus albipunctatus is a disease of abnormal rhodopsin regeneration, which manifests as slow but ultimately successful dark adaptation.

Fundus Albipunctatus
--Pathology: Delayed regeneration of the photopigment...rhodopsin
--Dark adaptation is abnormal:
  --Initially, patients are...night-blind, with abnormal...rod ERG
  --With enough time, will dark-adapt, and ERG normalizes
--DFE: Striking array of...yellow - white dots
--Dots found in entire posterior pole except...fovea
Congenital/Stationary Retinal Disease

What is the main disease that must be differentiated from fundus albipunctatus?
Retinitis punctata albscens

What is retinitis punctata albscens?
An RP variant characterized by white - yellow dots similar to those of albipunctatus

How do fundus albipunctatus and retinitis punctata albscens differ?
--On DFE: Like other forms of RP, retinitis punctata albscens demonstrates arteriolar narrowing, whereas albipunctatus does not
--On ERG: Fundus albipunctatus is a disease of abnormal rhodopsin regeneration, which manifests as slow but ultimately successful dark adaptation. In contrast, retinitis punctata albscens is a photoreceptor disease; therefore, dark adaptation does not occur and the ERG never normalizes, no matter how much time is allowed to elapse.

Fundus Albipunctatus
--Pathology: Delayed regeneration of the photopigment...rhodopsin
--Dark adaptation is abnormal:
  --Initially, patients are...night-blind, with abnormal...rod ERG
  --With enough time, will dark-adapt, and ERG normalizes
--DFE: Striking array of...yellow - white dots
--Dots found in entire posterior pole except...fovea
**Oguchi Disease**

--Also have slow dark adaptation (*not* a pigment regeneration issue, though)
Congenital/Stationary Retinal Disease

Delayed dark adaptation in Oguchi dz
Oguchi Disease
--Also have slow dark adaptation (not a pigment regeneration issue, though)
--Once dark-adapted, dark sensitivity lost with a single...[event]
Oguchi Disease
--Also have slow dark adaptation (not a pigment regeneration issue, though)
--Once dark-adapted, dark sensitivity lost with a single…bright flash
Cone (Color) Disease

- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease

- Fundus appearance normal
- Fundus appearance abnormal
- CSNB
- Fundus albin punctatus
- Oguchi disease

**Oguchi Disease**

-- Also have slow dark adaptation (*not* a pigment regeneration issue, though)
-- Once dark-adapted, dark sensitivity lost with a single... bright flash
-- DFE:
  -- Normal appearance when... *[state of adaptation]*
Congenital/Stationary Retinal Disease

Cone (Color) Disease
- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease
- Fundus appearance normal
- Fundus appearance abnormal
  - CSNB
  - Fundus albipunctatus
  - Oguchi disease

Oguchi Disease
-- Also have slow dark adaptation (not a pigment regeneration issue, though)
-- Once dark-adapted, dark sensitivity lost with a single... bright flash
-- DFE:
  -- Normal appearance when... dark-adapted
**Congenital/Stationary Retinal Disease**

- **Cone (Color) Disease**
  - Trichromatism
  - Dichromatism
  - Monochromatism

- **Rod (Night Vision) Disease**
  - Fundus appearance normal
  - CSNB
  - Fundus appearance abnormal
  - Fundus albipunctatus
  - Oguchi disease

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**Oguchi Disease**
- Also have slow dark adaptation (not a pigment regeneration issue, though)
- Once dark-adapted, dark sensitivity lost with a single…bright flash
- DFE:
  - Normal appearance when…**dark-adapted**
  - After light exposure, posterior pole takes on a…*appearance*
**Congenital/Stationary Retinal Disease**

- **Cone (Color) Disease**
  - Trichromatism
  - Dichromatism
  - Monochromatism
- **Rod (Night Vision) Disease**
  - Fundus appearance normal
  - Fundus appearance abnormal
    - CSNB
    - **Fundus albinpectatus**
    - **Oguchi disease**

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**Oguchi Disease**

--Also have slow dark adaptation (*not* a pigment regeneration issue, though)
--Once dark-adapted, dark sensitivity lost with a single… bright flash
--DFE:
  --Normal appearance when… **dark-adapted**
  --After light exposure, posterior pole takes on a… **yellow iridescent sheen**
Oguchi Disease
--Also have slow dark adaptation (not a pigment regeneration issue, though)
--Once dark-adapted, dark sensitivity lost with a single...bright flash
--DFE:
  --Normal appearance when...dark-adapted
  --After light exposure, posterior pole takes on a...yellow iridescent sheen
  --This color change is known as the...[eponym-eponym]
**Oguchi Disease**
--Also have slow dark adaptation (*not* a pigment regeneration issue, though)
--Once dark-adapted, dark sensitivity lost with a single…bright flash
--DFE:
  --Normal appearance when…**dark-adapted**
  --After light exposure, posterior pole takes on a…**yellow iridescent sheen**
  --This color change is known as the…**Mizuo-Nakamura phenomenon**
Mizuo-Nakamura phenomenon in Oguchi dz
Congenital/Stationary Retinal Disease

**Cone (Color) Disease**
- Trichromatism
- Dichromatism
- Monochromatism

**Rod (Night Vision) Disease**
- Fundus appearance normal
- CSNB
- Fundus appearance abnormal

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**Oguchi Disease**
- Also have slow dark adaptation (not a pigment regeneration issue, though)
- Once dark-adapted, dark sensitivity lost with a single... bright flash
- DFE:
  - Normal appearance when...
  - After light exposure, posterior pole takes on a... yellow iridescent sheen
- This color change is known as the Mizuo-Nakamura phenomenon

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**Is Oguchi dz common, or rare?**
- It is very rare
- With what ethnicity is it closely associated?
  - Japanese
Congenital/Stationary Retinal Disease

Cone (Color) Disease
- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease
- Fundus appearance normal
- Fundus appearance abnormal
- CSNB
- Fundus albipunctatus
- Oguchi disease

Oguchi Disease
- Also have slow dark adaptation (not a pigment regeneration issue, though)
- Once dark-adapted, dark sensitivity lost with a single...bright flash
- DFE:
  - Normal appearance when...dark-adapted
  - After light exposure, posterior pole takes on a...yellow iridescent sheen
  - This color change is known as the Mizuo-Nakamura phenomenon

Is Oguchi dz common, or rare?
It is very rare

With what ethnicity is it closely associated?
Japanese
**Congenital/Stationary Retinal Disease**

- Cone (Color) Disease
  - Trichromatism
  - Dichromatism
  - Monochromatism

- Rod (Night Vision) Disease
  - Fundus appearance normal
  - Fundus appearance abnormal

- CSNB

**Oguchi Disease**
- Also have slow dark adaptation (not a pigment regeneration issue, though)
- Once dark-adapted, dark sensitivity lost with a single... bright flash
- DFE:
  - Normal appearance when...
  - After light exposure, posterior pole takes on a... yellow iridescent sheen
- This color change is known as the Mizuo-Nakamura phenomenon

*Is Oguchi dz common, or rare?*

It is very rare

*With what ethnicity is it closely associated?*

Japanese
Oguchi Disease

--Also have slow dark adaptation (not a pigment regeneration issue, though)
--Once dark-adapted... bright flash
--DFE:
  --Normal appearance when dark-adapted
  --After light exposure, posterior pole takes on a yellow iridescent sheen
  --This color change is known as the Mizuo-Nakamura phenomenon

Is Oguchi dz common, or rare?
It is very rare

With what ethnicity is it closely associated?
Japanese

Oguchi disease