Congenital/Stationary Retinal Disease

Two very basic categories
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

Two very basic categories
Congenital/Stationary Retinal 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’?

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!). A trichromat requires three lights—
hence the ‘tri’—lights—
one of short wavelength (aka blue), one of medium wavelength (= green), and one of long (= red).
(This is the normal state of color vision in humans.)
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!). A trichromat requires three (hence the ‘tri-’) lights—one of short wavelength (aka blue), one of medium wavelength (= green), and one of long (= red). (This is the normal state of color vision in humans.)
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What does it mean to say someone is an ‘anomalous’ trichromat?
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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!). A trichromat requires three (hence the ‘tri-’) lights—one of short wavelength (aka blue), one of medium wavelength (= green), and one of long (= red). (This is the normal state of color vision in humans.)

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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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).

Dude, wussup with the gendered language?
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!). A trichromat requires three (hence the ‘tri’-) lights—one of short wavelength (aka blue), one of medium wavelength (= green), and one of long (= red). (This is the normal state of color vision in humans.)

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).

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.
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.
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.)
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 a dichromat can match any color with only two primaries indicates what about his cones?*
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 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
Congenital/Stationary Retinal Disease

Cone (Color) Disease

- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease

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

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

Blue-cone monochromatism

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

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

Blue-cone monochromatism

Rod monochromatism

--Inheritance…
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

Blue-cone monochromatism

Rod (Night Vision) Disease

--Inheritance…AR

Congenital/Stationary Retinal Disease
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism
Trichromatism
Monochromatism
Rod monochromatism
Blue-cone monochromatism

Rod monochromatism
--Inheritance... AR
--No cones present--true color blindness

Congenital/Stationary Retinal Disease

--Inheritance... AR
--No cones present--true color blindness
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
-- always present

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
-- No cones present--true color blindness
-- Nystagmus always present
**Cone (Color) Disease**

**Rod (Night Vision) Disease**

---

**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

Blue-cone monochromatism

Rod monochromatism

--Inheritance...AR
--No cones present--true color blindness
--Nystagmus always present
--VA range: 20/80–20/200
Cone (Color) Disease

Rod (Night Vision) Disease

Monochromatism

Trichromatism

Dichromatism

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:
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod 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
Cone (Color) Disease

Rod (Night Vision) Disease

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:
Congenital/Stationary Retinal Disease

Cone (Color) Disease
- Trichromatism
- Dichromatism
  - Monochromatism
    - Rod monochromatism
    - Blue-cone monochromatism

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

Rod (Night Vision) Disease

Congenital/Stationary Retinal 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:

-- two words and
-- and

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**Cone (Color) Disease**

**Rod (Night Vision) Disease**

**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:**
- Poor acuity *and*
- Nystagmus *and*
- Photophobia
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:

Why the broad range in VA?
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:

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

- Blue-cone monochromatism
  -- Inheritance:
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

Blue-cone monochromatism

--Inheritance: X-linked
Congenital/Stationary Retinal Disease

Cone (Color) Disease

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

Rod (Night Vision) Disease

**Blue-cone monochromatism**
- Inheritance: X-linked
- Only [duh] cones present
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod 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

Blue-cone monochromatism

--Inheritance: X-linked

--Only blue cones present

--VA usually about...
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
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

Why is VA better than in many rod monochromats?
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

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

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
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

Blue-cone monochromatism

--- Inheritance: X-linked
--- Only blue cones present

What are the findings of color-ERG testing in blue-cone monochromatism?
Blue-cone monochromatism
--Inheritance: X-linked
--Only blue cones present

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

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

**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
Cone (Color) Disease

- Rod (Night Vision) Disease

Monochromatism

Dichromatism

Trichromatism

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

--- Only blue cones present
--- VA usually about 20/80
--- Diagnose via specialized color ERG

Congenital/Stationary Retinal Disease

- Conditions presenting very early in life with poor VA, nystagmus and photophobia (list obviously incomplete):
  - Rod monochromatism
  - Blue-cone monochromatism
  - ?
  - ?
  - ?

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Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism
Dichromatism

Monochromatism

-- Blue-cone monochromatism
-- Rod monochromatism

Monochromatism

Inheritance: X-linked

--- Only blue cones present
--- VA usually about 20/80
--- Diagnose via specialized color ERG

Conditions presenting very early in life with poor VA, nystagmus and photophobia (list obviously incomplete):

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

Classic presentation of blue-cone monochromatism:

-- Poor acuity and
-- Nystagmus and
-- Photophobia
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

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

Rod (Night Vision) Disease

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

Congenital/Stationary Retinal Disease

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
**Congenital/Stationary Retinal Disease**

- **Cone (Color) Disease**
  - Trichromatism
  - Dichromatism
  - Monochromatism
    - Rod monochromatism
    - Blue-cone monochromatism

- **Rod (Night Vision) Disease**

  **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?

- It is virtually always horizontal

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

- 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

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?
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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

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**Inheritance:**

- X-linked
- Only blue cones present
- Mimics rod monochromatism
- Diagnose via specialized…

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Congenital/Stationary Retinal Disease

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- 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

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**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?*
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

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

Congenital motor nystagmus

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

--- What condition does s/he most likely have?

--- 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

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Blue-cone monochromatism

Rod 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?

It is virtually always horizontal
Cone (Color) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

Blue-cone monochromatism

Rod (Night Vision) Disease

Congenital/Stationary Retinal Disease

Good vision and
Nystagmus
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
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism  Dichromatism

Monochromatism

Rod monochromatism

Blue-cone monochromatism

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

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

S-cone monochromatism

Rod 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
  - S-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?
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism
Dichromatism

Monochromatism

Rod monochromatism

Blue-cone monochromatism

S-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?
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?)
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?
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Blue-cone monochromatism

S-cone monochromatism

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

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_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

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?

'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.
Congenital/Stationary Retinal Disease

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?

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enhanced S-cone syndrome
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Congenital/Stationary Retinal Disease

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

What sort of condition is it?

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. (see what I did there?)
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?

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 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.
Speaking of conditions with two names, it's 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.

In what ways are photoreceptors affected?

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, they are 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

In what ways are photoreceptors affected?

--Rods:
Non-functioning

--Red/green cones:
Reduced in number.

--Blue cones:
Increased in number

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, 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” à la RP

**In what ways are photoreceptors affected?**
--Rods: Non-functioning

---

Enhanced S-cone syndrome

**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, they are 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

In what ways are photoreceptors affected?

--Rods: Non-functioning

--Red/green cones:

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

enhanced S-cone syndrome

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

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

In what ways are photoreceptors affected?

--Rods: Non-functioning

--Red/green cones:

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

enhanced S-cone syndrome

Speaking of conditions with two names, they are 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

In what ways are photoreceptors affected?

--Rods: Non-functioning

--Red/green cones:

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

enhanced S-cone syndrome
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

**Congenital/Stationary Retinal Disease**

**Speaking of conditions with two names**, blue-cone monochromatism 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

In what ways are photoreceptors affected?

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

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**

What sort of condition is it?

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

What does it present?

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, they are 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?

--Rods: Non-functioning
--Red/green cones: Reduced in number
--Blue cones:
**Cone (Color) Disease**

**Rod (Night Vision) Disease**

**Congenital/Stationary Retinal Disease**

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

---

**Speaking of conditions with two names, what are they 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

---

**In what ways are photoreceptors affected?**

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

---

**Indeed there is, and it’s this:** To make certain not to confuse S-cone monochromatism with the similarly-named but completely different condition, the list of photoreceptors affected reads as follows:

---

**enhanced S-cone syndrome**

Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Speaking of conditions with two names, what are they 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

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

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

Enhanced S-cone 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

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

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

Speaking of conditions with two names, 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

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:

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, what are they also known as?
Goldmann-Favre syndrome

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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

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

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

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--Blue cones: Increased in number

What are the ERG findings?
--Rod response: Undetectable
--Red/green cone response:

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...
**Cone (Color) Disease**

**Rod (Night Vision) Disease**

**Trichromatism**

**Dichromatism**

Speaking of conditions with two names, what are they also known as?

- Goldmann-Favre syndrome

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In what ways are photoreceptors affected?

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What are the ERG findings?

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What's the 'S' stand for?

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--Blue cones:

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enhanced S-cone syndrome
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

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

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

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)

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

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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How does it present?

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Rod (Night Vision) Disease

Trichromatism

Dichromatism

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

How does it present?

With decreased acuity as well as night blindness

enhanced S-cone syndrome
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

**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?

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

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enhanced S-cone syndrome
Congenital/Stationary Retinal Disease

Cone (Color) Disease
  - Trichromatism
  - Dichromatism
  - Monochromatism

Rod (Night Vision) Disease
Congenital/Stationary Retinal Disease

Cone (Color) Disease
- Trichromatism
- Dichromatism
- Monochromatism

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

Cone (Color) Disease
- Trichromatism
- Dichromatism
- Monochromatism

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

Fundus appearance?

90
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
  - CSNB
- Fundus appearance
  - abnormal
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 normal
- Fundus appearance abnormal
- CSNB

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
  - Fundus appearance abnormal

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**Congenital Stationary Night Blindness (CSNB)**

---Several inheritance patterns; most common =
**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*
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 *two cell types*
**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

**Fundus appearance**
- normal
- abnormal

**Fundus**
- CSNB

- VA 20/20 - 20/200
- Refractive error: Usually myopia
- Presents in childhood with:
  - Nystagmus
  - Decreased vision
  - Nyctalopia
  - Classified according to… ERG pattern
  - ‘Most common pattern: ‘Negative’ ERG
    - Large a-wave, no b-wave
**Congenital Stationary Night Blindness (CSNB)**

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

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

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

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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
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 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
**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:

---
---
---

--- 'Most common pattern: 'Negative' ERG

--- 'Negative' ERG = Large \(a\)-wave, no \(b\)-wave
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

Trichromatism
Dichromatism
Monochromatism

Rod (Night Vision) Disease

Fundus appearance
- normal
- 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?

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 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**

*What is nyctalopia?*
- Night blindness
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?
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
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- 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

- 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**

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

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:
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
  --Nystalopia

--Classified according to... Scotopic ERG pattern

--Most common pattern: Negative ERG
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 =
Congenital Stationary Night Blindness (CSNB)
--Several inheritance patterns; most common = X-linked
--Pathology: Communication failure between… photoreceptors & bipolar cells
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  --Nystagmus
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--Classified according to…Scotopic ERG pattern
--Most common pattern: Negative ERG
  --Negative ERG = Large a-wave, no b-wave
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
  - abnormal
  - CSNB
  - Fundus albipunctatus
  - Oguchi disease
Congenital/Stationary Retinal Disease

Cone (Color) Disease
  - Trichromatism
  - Dichromatism
  - Monochromatism

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

Fundus Albipunctatus
  -- Pathology: Delayed regeneration of the photopigment…

Fundus Albipunctatus
  -- Oguchi disease
Congenital/Stationary Retinal Disease

Cone (Color) Disease
- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease
- Fundus appearance normal
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- CSNB
- Fundus Albipunctatus
  -- Pathology: Delayed regeneration of the photopigment...rhodopsin

Oguchi disease
Congenital/Stationary Retinal Disease

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- Trichromatism
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- Monochromatism

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

**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

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- Fundus appearance normal
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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…
Congenital/Stationary Retinal Disease

Cone (Color) Disease

- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease

- Fundus appearance normal
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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**
Congenital/Stationary Retinal Disease

Cone (Color) Disease
- Trichromatism
- Dichromatism
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Rod (Night Vision) Disease
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    - 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...
Congenital/Stationary Retinal Disease

Cone (Color) Disease
- Trichromatism
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Rod (Night Vision) Disease
- Fundus appearance normal
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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?

**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

-Trichromatism

-Rod (Night Vision) Disease

-Cone (Color) Disease

-Congenital/Stationary Retinal Disease
What is the main disease that must be differentiated from fundus albipunctatus?
Retinitis punctata albe scens

**Fundus Albipunctatus**

--Pathology: Delayed regeneration of the photopigment...rhodopsin
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Retinitis punctata albe scens
What is the main disease that must be differentiated from fundus albipunctatus?
Retinitis punctata albescens

Retinitis punctata albescens:

- 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

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

**Fundus Albipunctatus**
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

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 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?**

**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**
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**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:

--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
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What is the main disease that must be differentiated from fundus albipunctatus?
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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
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--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
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**Fundus Albipunctatus**

--Pathology: Delayed regeneration of the photopigment…rhodopsin
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--DFE: Striking array of…**yellow - white dots**
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---

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

**What is retinitis punctata albscens?**
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**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.
Oguchi Disease
--Also have slow dark adaptation (\textit{not} a pigment regeneration issue, though)
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
**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]*
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
**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
  - Fundus albinopunctatus
  - 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*
**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]*
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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**
  - This color change is known as the...**Mizuo-Nakamura phenomenon**
Congenital/Stationary Retinal 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...
  -- 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
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Rod (Night Vision) Disease
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Oguchi Disease
- Also have slow dark adaptation (not a pigment regeneration issue, though)
- Once dark-adapted...bright flash
- DFE:
  --Normal appearance
  --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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- Fundus appearance normal
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- 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
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