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 (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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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?* 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 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
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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
Monochromatism

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
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

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

Rod (Night Vision) Disease

- Monochromatism
  - Rod monochromatism

Congenital/Stationary Retinal Disease

- VA range: 20/80–20/200
- ERG:
  - Cone response: Absent
  - Rod response: Normal

Nystagmus always present
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
--ERG:
--Cone response: Absent
--Rod response: Normal

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

Congenital/Stationary Retinal Disease

VA range: 20/80–20/200

ERG:

--Cone response: Absent

--Rod response: Normal

Nystagmus always present
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

VA range: 20/80–20/200

ERG:
--Cone response: Absent
--Rod response: Normal

Nystagmus always present
Cone (Color) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

Blue-cone monochromatism

Rod (Night Vision) Disease

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Congenital/Stationary Retinal Disease

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

Nystagmus always present
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:
Cone (Color) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

Blue-cone monochromatism

Rod (Night Vision) Disease

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
--No cones present--true color blindness
--Nystagmus always present
--VA range: 20/80–20/200
--ERG:
  --Cone response:

Blue-cone monochromatism
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

Congenital/Stationary Retinal Disease
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:
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
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

Classic presentation of rod monochromatism:

--- two words
--- and
--- and

Congenital/Stationary Retinal Disease
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:
--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:

Cone response: Absent

Rod response: Normal

Why the broad range in VA?

Congenital/Stationary Retinal Disease
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

Rod monochromatism

Blue-cone 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

ERG:

Congenital/Stationary Retinal Disease
Cone (Color) Disease

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

Rod (Night Vision) Disease

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
Cone (Color) Disease

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

Rod (Night Vision) Disease

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

Congenital/Stationary Retinal Disease
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

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

Blue-cone monochromatism
--Inheritance: X-linked
--Only blue cones present
--VA usually about 20/80

Rod (Night Vision) Disease

Congenital/Stationary Retinal Disease
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

Why is VA better than in many rod monochromats?
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

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

Congenital/Stationary Retinal Disease

Trichromatism

Dichromatism

Monochromatism

Rod 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

---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

Blue-cone monochromatism

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

What are the findings of color-ERG testing in blue-cone monochromatism?

--- VA usually about 20/80
--- Diagnose via specialized color ERG
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

Rod monochromatism

Blue-cone monochromatism

---Inheritance: X-linked

--Only blue cones present

--VA usually about 20/80

--Diagnose via specialized color ERG

**Blue-cone monochromatism**

Classic presentation of blue-cone monochromatism:

--- two words

--- and

--- and

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

(Yes, just like rod monochromatism)
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism
Dichromatism
Monochromatism

**Cone (Color) Disease**

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

**Rod (Night Vision) Disease**

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

**Congenital/Stationary Retinal Disease**

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
- ?
- ?
- ?

**Retinal Disease**

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

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

- Blue-cone monochromatism
- Rod 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

Congenital/Stationary Retinal Disease

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
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

**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
- Nystagmus
- Photophobia

If a pt has nystagmus plus good vision, 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

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?
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

- 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.

If a pt has nystagmus + good VA, it’s congenital motor nystagmus.

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

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?*

**Congenital motor nystagmus**

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 monochromatism

Blue-cone monochromatism

Rod (Night Vision) Disease

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?
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It is virtually always horizontal
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…
  - 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

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

Congenital/Stationary Retinal Disease

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

Good

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

- 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

Rod monochromatism

S-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?
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?)
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism  Dichromatism

Monochromatism

Rod monochromatism

S-cone  Blue-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?*
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

*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

Blue-cone monochromatism

Rod monochromatism

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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?

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

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

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

Congenital/Stationary Retinal Disease

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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?)

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

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Trichromatism  Dichromatism

Blue-cone monochromatism  Rod monochromatism

Congenital/Stationary Retinal Disease

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What sort of condition is it?

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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...
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.

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.

In what ways are photoreceptors affected?

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

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

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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.
In what ways are photoreceptors affected?

--Rods:

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…

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.

In what ways are photoreceptors affected?

--Rods:

--Red/green cones: Reduced in number.

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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

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 "RP" stand for?
Retinitis Pigmentosa
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism  Dichromatism

**Cone (Color) Disease**

**Rod (Night Vision) Disease**

**Congenital/Stationary Retinal Disease**

**Trichromatism**

**Dichromatism**

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

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

In what ways are photoreceptors affected?

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**Enhanced S-cone syndrome**

---

What sort of condition is it?

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Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

**Congenital/Stationary Retinal Disease**

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

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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**

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

Trichromatism
Dichromatism

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

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

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.

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 is the presentation of enhanced S-cone syndrome?

With decreased acuity as well as night blindness.

What sort of condition is it?

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

Speaking of conditions with two names…What else is it known as?

Goldmann-Favre syndrome

Enhanced S-cone syndrome
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Speaking of conditions with two names, it is also known as what?
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What are the ERG findings?
--Rod response:

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enhanced S-cone syndrome

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What are the ERG findings?
--Rod response:
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

**Speaking of conditions with two names**

*a.k.a.* Goldmann-Favre syndrome

*What sort of condition is it?*

The BCSC *Retina* book calls it a "diffuse photoreceptor dystrophy" (a la RP)

*Rods: Non-functioning*

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--Blue cones: Increased in number

*What are the ERG findings?*

--Rod response: Undetectable

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Rod (Night Vision) Disease

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Speaking of conditions with two names, also known as what?
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enhanced S-cone syndrome
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Trichromatism

Dichromatism

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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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Other than knowing that the condition goes by two names, is there another reason that an awareness of the name ...
Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Congenital/Stationary Retinal Disease

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What are the ERG findings?
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--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

enhanced S-cone 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

Congenital/Stationary Retinal Disease

Trichromatism

Dichromatism

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

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
- CSNB
- Fundus appearance abnormal
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

**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
- CSNB
- Congenital Stationary Night Blindness
- CSNB
- Congenital/Stationary Retinal Disease
- Cone (Color) Disease
- Rod (Night Vision) Disease
- Trichromatism
- Dichromatism
- Monochromatism

Pathology: Communication failure between photoreceptors & bipolar cells

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

'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…
  *two cell types*

---

**Fundus appearance normal**

**CSNB**

**Fundus appearance abnormal**

---

**Congenital/Stationary Retinal Disease**

**Cone (Color) Disease**

**Rod (Night Vision) Disease**

---

**Trichromatism**

**Dichromatism**

**Monochromatism**

---

**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
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.
• **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 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:

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

---

**CSNB**
**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 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...
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
**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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*When VA is poor in CSNB, it's usually due to the (high) myopia, not the photoreceptors*
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Trichromatism

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Rod (Night Vision) Disease

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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:
  --
  --
  --

Fundus appearance

normal

CSNB

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
**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
  - Night blindness

**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

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
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What is nyctalopia?
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Many CSNB children do **not** complain of nyctalopia. Why not?
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What is nyctalopia?
Night blindness

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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

Fundus appearance normal

Fundus appearance abnormal

**Congenital Stationary Night Blindness (CSNB)**
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  --Nystagmus
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  --Nyctalopia
--Classified according to…[Psychophysical test]
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- Classified according to... Scotopic ERG pattern

**Fundus appearance**
- normal
- abnormal

**CSNB**
Congenital Stationary Night Blindness (CSNB)

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  --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 Night Blindness (CSNB)**
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- 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 Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism
Dichromatism
Monochromatism

Fundus appearance normal
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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*
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-- Presents in childhood with:
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  -- Nyctalopia
-- Classified according to Scotopic ERG pattern
  -- Most common pattern: Negative ERG
  -- Negative ERG =
Congenital Stationary Retinal Disease

Cone (Color) Disease

- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease

- Fundus appearance normal
- Fundus appearance abnormal

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- Presents in childhood with:
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  - Negative ERG = Large a-wave, no b-wave
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

Another condition presents with c/o night blindness, and ERG reveals patterns identical to those of CSNB. Thus, this condition is high on the DDx for CSNB. What is it?

--Nystagmus
--Decreased vision
--Nyctalopia
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**Congenital/Stationary Retinal Disease**

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

- Nystagmus
- Decreased vision
- Nyctalopia

--Classified according to...Scotopic ERG pattern
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Congenital/Stationary Retinal Disease

Conic (Color) Disease

Rod (Night Vision) Disease

Trichromatism

Dichromatism

Monochromatism

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MAR

What does MAR stand for in this context?

--Nystagmus

--Decreased vision

--Nystagmus

--Scotopic ERG pattern

--Most common pattern: Negative ERG

--Negative ERG = Large a-wave, no b-wave
Congenital/Stationary Retinal Disease

Cone (Color) Disease

Rod (Night Vision) Disease

Trichromatism
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MAR

What does MAR stand for in this context?
Melanoma-associated retinopathy
--Nystagopia
--Classified according to…Scotopic ERG pattern
--Most common pattern: Negative ERG
--Negative ERG = Large a-wave, no b-wave
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--Several inheritance patterns; most common = X-linked

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What does MAR stand for in this context?

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--Classified according to...Scotopic ERG pattern

What is melanoma-associated retinopathy?
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)

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-- Pathology: Communication failure between photoreceptors & bipolar cells

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

-- Refractive error: Usually myopia

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MAR

What does MAR stand for in this context?

Melanoma-associated retinopathy

-- Classified according to...Scotopic ERG pattern

What is 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.
Congenital/Stationary Retinal Disease

Cone (Color) Disease

- Trichromatism
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Rod (Night Vision) Disease

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

Another condition presents with c/o night blindness and ERG reveals patterns identical to those of CSNB. Thus, this condition is high on the DDx for CSNB. What is it?

Martha (MAR)

*What does MAR stand for in this context?*

Melanoma-associated retinopathy

*What is melanoma-associated retinopathy?*

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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**
- Trichromatism
- Dichromatism
- Monochromatism

**Rod (Night Vision) Disease**
- Fundus appearance normal
- CSNB
- 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
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**In addition to night blindness, there is another complaint that is classic for MAR. What is it?**
**Congenital Stationary Retinal Disease**

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

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

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What is 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.

In addition to night blindness, there is another complaint that is classic for MAR. What is it? **Photopsias**, often described as shimmering. 
Congenital Stationary Night Blindness (CSNB)

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Melanoma-associated retinopathy

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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 other psychophysical test is always abnormal in CSNB?

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- Refractive error: Usually myopia
- Presents in childhood with:
  - Nystagmus
  - Decreased vision
  - Nyctalopia
- Classified according to Scotopic ERG pattern
- Most common pattern: Negative ERG
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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

Monochromatism

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
**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?

--- Fundus appearance normal

--- CSNB

--- Fundus appearance abnormal

--- Trichromatism

--- Dichromatism

--- Monochromatism

--- Cone (Color) Disease

--- Rod (Night Vision) Disease

--- Congenital/Stationary Retinal Disease
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 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).
This is what a dark adaptation curve looks like. As you can see, sensitivity increases to a maximum after about 30 minutes.
This is what a dark adaptation curve looks like. As you can see, sensitivity increases to a maximum after about 30 minutes.

But what's up this weird hiccup in the curve?
Congenital/Stationary Retinal Disease

Here’s what’s up. Early on, the response is dominated by the cones.

Less sensitive

More sensitive

Time in dark (minutes)

Dark adaptation level

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.
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).
**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)**
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  - Refractive error: Usually myopia
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    - Nyctalopia
  - Classified according to Scotopic ERG pattern
  - Most common pattern: Negative ERG
    - Negative ERG = Large \(a\)-wave, no \(b\)-wave

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Congenital/Stationary Retinal Disease

Cone (Color) Disease

- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease

- Fundus appearance normal
- Fundus appearance abnormal

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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
  - CSNB
- Fundus appearance abnormal
  - ?
  - ?

Next question
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
**Congenital/Stationary Retinal Disease**

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

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

**Fundus Albipunctatus**
--Pathology: Delayed regeneration of the photopigment…
Congenital/Stationary Retinal Disease

Cone (Color) Disease
- Trichromatism
- Dichromatism
- Monochromatism

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

Cone (Color) Disease
- Trichromatism
- Dichromatism
- Monochromatism

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

**Fundus Albipunctatus**
- Pathology: Delayed regeneration of the photopigment...rhodopsin
- Dark adaptation is abnormal:
  - Initially, patients are...[condition], with abnormal...[test]
**Congenital/Stationary Retinal Disease**

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

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

Cone (Color) Disease

- Trichromatism
- Dichromatism
- Monochromatism

Rod (Night Vision) Disease

- Fundus appearance normal
- CSNB

Fundus Albinopunctatus

-- 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

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?
Congenital/Stationary Retinal Disease

Cone (Color) Disease
- Trichromatism
- Dichromatism
- Monochromatism

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

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

Oguchi disease

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

Delayed dark adaptation in fundus albipunctatus
Congenital/Stationary Retinal Disease

Cone (Color) Disease
- Trichromatism
- Dichromatism
- Monochromatism

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

Fundus albipunctatus
Congenital/Stationary Retinal Disease

Cone (Color) Disease
- Trichromatism
- Dichromatism
- Monochromatism

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

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...
**Congenital/Stationary Retinal Disease**

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

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

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: Like other forms of RP, retinitis punctata albescens 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 albescens is a photoreceptor disease; therefore, dark adaptation does not occur and the ERG never normalizes, no matter how much time is allowed to elapse.
What is the main disease that must be differentiated from fundus albipunctatus?
Retinitis punctata albsens

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?

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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**How do fundus albipunctatus and retinitis punctata albescens differ?**
--On DFE: Like other forms of RP, retinitis punctata albescens 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 albescens is a photoreceptor disease; therefore, dark adaptation does not occur and the ERG never normalizes, no matter how much time is allowed to elapse.
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Congenital/Stationary Retinal Disease

Cone (Color) Disease
- Trichromatism
- Dichromatism
- Monochromatism

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

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

Delayed dark adaptation in Oguchi dz
Congenital/Stationary Retinal Disease

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  - Trichromatism
  - Dichromatism
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  - Fundus appearance abnormal
    - 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...[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
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--DFE:
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- 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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--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
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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
  -- 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

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- Once dark-adapted... bright flash
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