Retinitis Pigmentosa

A note before we begin: The Academy seems to be phasing out the term *retinitis pigmentosa*. Further, the scope of conditions covered by the term is shrinking. The point being, the facts concerning RP are in flux at the moment, and may have changed by the time you read this. Caveat emptor.
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor vision

Retinitis Pigmentosa
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor *scotopic* vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:

--poor scotopic vision
--constricted VF

Night blindness and progressive peripheral VF loss are the two hallmark symptoms of RP
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
- poor scotopic vision
- constricted VF

Night blindness and progressive peripheral VF loss are the two hallmark symptoms of RP.

Some RP pts do not c/o poor night vision. Why not?
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:

- poor scotopic vision
- constricted VF

Night blindness and progressive peripheral VF loss are the two hallmark symptoms of RP.

Some RP pts do not c/o poor night vision. Why not? Such pts have had poor scotopic vision their entire lives, and thus are unable to recognize it; ie, their poor night vision seems normal to them.
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal...
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG

Night blindness, VF loss and abnormal ERG are the defining features of RP. If it ain’t got all three, it very likely ain’t RP

Retinitis Pigmentosa
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance

Classic fundus appearance:
-- two words

Retinitis Pigmentosa
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance

Classic fundus appearance:
--Bone spicules
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance

Classic fundus appearance:
--Bone spicules

What are bone spicules?

Bone spicules: Focal accumulations of pigment released when dying RPE cells disintegrate.
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance

Classic fundus appearance:
--Bone spicules

What are bone spicules?
Focal accumulations of pigment released when dying RPE cells disintegrate
Retinitis Pigmentosa

RP: Bone spicules
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
-- poor scotopic vision
-- constricted VF
-- abnormal ERG
-- characteristic fundus appearance

Classic fundus appearance:
-- Bone spicules
-- disc pallor
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance

Classic fundus appearance:
--Bone spicules
--Waxy disc pallor
Retinitis Pigmentosa

RP: Waxy disc pallor
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance

Classic fundus appearance:
--Bone spicules
--Waxy disc pallor
--Arteriolar
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance

Classic fundus appearance:
--Bone spicules
--Waxy disc pallor
--Arteriolar narrowing
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance

Classic fundus appearance:
--Bone spicules
--Waxy disc pallor
--Arteriolar narrowing

Which of the three appears first?

Retinitis Pigmentosa
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance

Classic fundus appearance:
--Bone spicules
--Waxy disc pallor
--Arteriolar narrowing

Which of the three appears first?
Arteriolar narrowing
Retinitis Pigmentosa

RP: Arteriolar narrowing
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance

Possible fundus appearance:
--Bone spicules
--Waxy disc pallor
--Arteriolar narrowing

What infectious (and therefore treatable) disease can produce a similar fundus appearance, and must always, **always** be at least considered in a patient with an RP-like fundus?
Retinitis Pigmentosa

- Group of inherited retinal diseases characterized by:
  - poor scotopic vision
  - constricted VF
  - abnormal ERG
  - characteristic fundus appearance

Possible fundus appearance:
- Bone spicules
- Waxy disc pallor
- Arteriolar narrowing

What infectious (and therefore treatable) disease can produce a similar fundus appearance, and must always, **always** be at least considered in a patient with an RP-like fundus?

**Syphilis**
Retinitis Pigmentosa

RP-like fundus in syphilis
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
-- poor scotopic vision
-- constricted VF
-- abnormal ERG
-- characteristic fundus appearance

Classic fundus appearance:
-- Bone spicules
-- Waxy disc pallor
-- Arteriolar narrowing

Two well-recognized non-classic phenotypes:
-- Retinitis punctata albescens
-- Choroideremia
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
-- poor scotopic vision
-- constricted VF
-- abnormal ERG
-- characteristic fundus appearance

Classic fundus appearance:
-- Bone spicules
-- Waxy disc pallor
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Retinitis Pigmentosa

Two well-recognized non-classic phenotypes:
-- Retinitis punctata albescens
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Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance

Retinitis Pigmentosa

Two well-recognized non-classic phenotypes:
--Retinitis punctata albescens
--Choroideremia

Appearance-wise, what is the hallmark of retinitis punctata albescens?

Retinitis Pigmentosa

Myriad white dots/flecks in the deep retina

What is the hallmark of choroideremia?

Pronounced atrophic changes of the RPE, choriocapillaris and choroid
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
-- poor scotopic vision
-- constricted VF
-- abnormal ERG
-- characteristic fundus appearance

Retinitis Pigmentosa

Two well-recognized non-classic phenotypes:
-- Retinitis punctata albescens
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Appearance-wise, what is the hallmark of retinitis punctata albescens?
Myriad white dots/flecks in the deep retina

Appearance-wise, what is the hallmark of choroideremia?
Pronounced atrophic changes of the RPE, choriocapillaris and choroid
Retinitis Pigmentosa

Retinitis punctata albescens
Retinitis Pigmentosa

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Appearance-wise, what is the hallmark of retinitis punctata albescens?
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What is the hallmark of choroideremia?
**Retinitis Pigmentosa**

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-- poor scotopic vision
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**Retinitis Pigmentosa**

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What is the hallmark of choroideremia?
Pronounced atrophic changes of the RPE, choriocapillaris and choroid

Two well-recognized non-classic phenotypes:
-- Retinitis punctata albescens
-- Choroideremia
Retinitis Pigmentosa

Choroideremia
Classic fundus appearance:
--Bone spicules
--Waxy disc pallor
--Arteriolar narrowing

Retinitis Pigmentosa
Group of inherited retinal diseases characterized by:
--poor scotopic vision
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--characteristic fundus appearance

Retinitis Pigmentosa
Two well-recognized non-classic phenotypes:
--Retinitis punctata albescens
--Choroideremia

What is the hallmark of retinitis punctata albescens?
Myriad white dots/flecks in the deep retina

What is the hallmark of choroideremia?
Pronounced atrophic changes of the RPE, choriocapillaris and choroid

Note: Whether choroideremia is a form of RP is one of the points in flux. The most recent Retina revision states it is not, whereas the immediately preceding version--which, at the time of this writing, is still being used by residents-in-training--maintains that it is.
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance

Retinitis Pigmentosa

Appearance-wise, what is the hallmark of retinitis punctata albescens? Myriad white dots/flecks in the deep retina

In terms of both its name and appearance, with what disease is retinitis punctata albescens often confused?

Two well-recognized non-classic phenotypes:
--Retinitis punctata albescens
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Retinitis pigmentosa

Characterized by:
--poor scotopic vision
--constricted VF
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Retinitis pigmentosa

Two well-recognized non-classic phenotypes:
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Appearance-wise, what is the hallmark of retinitis punctata albescens? Myriad white dots/flecks in the deep retina

In terms of both its name and appearance, with what disease is retinitis punctata albescens often confused?

Fundus albipunctatus

No, and it's very important to know the difference

OK, what's the difference?

Whereas retinitis punctata albescens is a non-classic phenotype of RP, fundus albipunctatus is a non-classic phenotype of CSNB

What does CSNB stand for?

Congenital stationary night blindness
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance

Retinitis Pigmentosa

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Two well-recognized non-classic phenotypes:
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Congenital stationary night blindness

Whereas retinitis punctata albescens is a non-classic phenotype of RP, fundus albipunctatus is a non-classic phenotype of CSNB.
Retinitis Pigmentosa

Retinitis punctata albescens

Fundus albipunctatus
Retinitis Pigmentosa

- Bone spicules
- Waxy disc pallor
- Arteriolar narrowing

Retinitis Pigmentosa

- Group of inherited retinal diseases characterized by:
  - Poor scotopic vision
  - Constricted VF
  - Abnormal ERG
  - Characteristic fundus appearance

Retinitis Pigmentosa

- Two well-recognized non-classic phenotypes:
  - Retinitis punctata albescens
  - Choroideremia

Appearance-wise, what is the hallmark of retinitis punctata albescens?

Myriad white dots/flecks in the deep retina

In terms of both its name and appearance, with what disease is retinitis punctata albescens often confused?

Fundus albipunctatus

Is retinitis punctata albescens the same thing as fundus albipunctatus?

No, and it's very important to know the difference.

Whereas retinitis punctata albescens is a non-classic phenotype of RP, fundus albipunctatus is a non-classic phenotype of CSNB.

What does CSNB stand for?

Congenital stationary night blindness
Classic fundus appearance:
-- Bone spicules
-- Waxy disc pallor
-- Arteriolar narrowing

Retinitis Pigmentosa
Group of inherited retinal diseases characterized by:
-- Poor scotopic vision
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Retinitis Pigmentosa

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Is retinitis punctata albescens the same thing as fundus albipunctatus?
No, and it’s very important to know the difference

OK, what’s the difference?

Congenital stationary night blindness (CSNB)
Retinitis Pigmentosa

- Bone spicules
- Waxy disc pallor
- Arteriolar narrowing

Retinitis Pigmentosa

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- Poor scotopic vision
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Retinitis Pigmentosa

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--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance

Two well-recognized non-classic phenotypes:
--Retinitis punctata albscens
--Choroideremia

Appearance-wise, what is the hallmark of retinitis punctata albscens?
Myriad white dots/flecks in the deep retina

In terms of both its name and appearance, with what disease is retinitis punctata albscens often confused?
Fundus albipunctatus

Is retinitis punctata albscens the same thing as fundus albipunctatus?
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What does CSNB stand for?

Congenital stationary night blindness
Retinitis Pigmentosa

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What does CSNB stand for?
Congenital stationary night blindness

Two well-recognized non-classic phenotypes:
-- Retinitis punctata albescens
-- Choroideremia
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance

Retinitis Pigmentosa

Two well-recognized non-classic phenotypes:
--Retinitis punctata albsencens
--Choroideremia

Appearance-wise, what is the hallmark of retinitis punctata albsencens?
Myriad white dots/flecks in the deep retina

In terms of both its name and appearance, with what disease is retinitis punctata albsencens often confused?
Fundus albipunctatus

Is retinitis punctata albsencens the same thing as fundus albipunctatus?
No, and it’s very important to know the difference

OK, what’s the difference?
Whereas retinitis punctata albsencens is a non-classic phenotype of RP, fundus albipunctatus is a non-classic phenotype of CSNB

What does it mean to say the night blindness is stationary?
Congenital stationary night blindness

Congenital stationary night blindness

CSNB stands for:
Congenital stationary night blindness

What does it mean to say the night blindness is stationary?
It means it is nonprogressive, an important way in which fundus albipunctatus differs from retinitis punctata albsencens (which, like all RP, is relentlessly progressive)
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance

Two well-recognized non-classic phenotypes:
--Retinitis punctata albescens
--Choroideremia

Appearance-wise, what is the hallmark of retinitis punctata albescens?
Myriad white dots/flecks in the deep retina

In terms of both its name and appearance, with what disease is retinitis punctata albescens often confused?
Fundus albipunctatus

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OK, what’s the difference?
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What does it mean to say the night blindness is stationary?
It means it is nonprogressive, an important way in which fundus albipunctatus differs from retinitis punctata albescens (which, like all RP, is relentlessly progressive)

Congenital stationary night blindness
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance

Retinitis Pigmentosa is a group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance

Two well-recognized non-classic phenotypes:
--Retinitis punctata albsencens
--Choroideremia

Appearance-wise, what is the hallmark of retinitis punctata albsencens?
Myriad white dots/flecks in the deep retina

In terms of both its name and appearance, with what disease is retinitis punctata albsencens often confused?
Fundus albipunctatus

Is retinitis punctata albsencens the same thing as fundus albipunctatus?
No, and it’s very important to know the difference

OK, what’s the difference?
Whereas retinitis punctata albsencens is a non-classic phenotype of RP, fundus albipunctatus is a non-classic phenotype of CSNB.

What does it mean to say the night blindness is stationary?
It means it is nonprogressive, an important way in which fundus albipunctatus differs from retinitis punctata albsencens

How else do fundus albipunctatus and retinitis punctata albsencens differ?

On DFE:
Like other forms of RP, retinitis punctata albsencens demonstrates arteriolar narrowing, whereas fundus 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 albsencens is a photoreceptor disease; therefore, dark adaptation does not occur and the ERG never normalizes, no matter how much time is allowed to elapse.
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance

Two well-recognized non-classic phenotypes:
--Retinitis punctata albscens
--Choroideremia

**Appearance-wise, what is the hallmark of retinitis punctata albscens?**
Myriad white dots/flecks in the deep retina

**In terms of both its name and appearance, with what disease is retinitis punctata albscens often confused?**
Fundus albipunctatus

Is retinitis punctata albscens the same thing as fundus albipunctatus?
No, and it’s very important to know the difference

OK, what’s the difference?
Whereas retinitis punctata albscens is a non-classic phenotype of RP, fundus albipunctatus is a non-classic phenotype of CSNB

What does it mean to say the night blindness is stationary?
It means it is nonprogressive, an important way in which fundus albipunctatus differs from retinitis punctata albscens (which, like all RP, is relentlessly progressive)

**How else do fundus albipunctatus and retinitis punctata albscens differ?**
--On DFE:
Like other forms of RP, retinitis punctata albscens demonstrates arteriolar narrowing, whereas fundus 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.
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance

Retinitis Pigmentosa

Appearance-wise, what is the hallmark of retinitis punctata albescens?
Myriad **white dots/flecks** in the deep retina

In terms of both its name and appearance, with what disease is retinitis punctata albescens often confused?
**Fundus albinpunctatus**

In terms of both its name and appearance, with what disease is fundus albipunctatus often confused?

Two well-recognized non-classic phenotypes:
--Retinitis punctata albescens
--Choroideremia

Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
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Retinitis Pigmentosa

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--poor scotopic vision
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Appearance-wise, what is the hallmark of retinitis punctata albescens?
Myriad white dots/flecks in the deep retina

In terms of both its name and appearance, with what disease is retinitis punctata albescens often confused?
Fundus albipunctatus

In terms of both its name and appearance, with what disease is fundus albipunctatus often confused?
Fundus flavimaculatus
Retinitis Pigmentosa

Fundus albipunctatus

Fundus flavimaculatus
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance

Retinitis Pigmentosa

Two well-recognized non-classic phenotypes:
--Retinitis punctata albsenes
--Choroideremia

Appearance-wise, what is the hallmark of retinitis punctata albsenes?
Myriad **white dots/flecks** in the deep retina

In terms of both its name and appearance, with what disease is retinitis punctata albsenes often confused?
**Fundus albipectatus**

In terms of both its name and appearance, with what disease is fundus albipectatus often confused?
**Fundus flavimaculatus**

Is fundus albipectatus the same thing as fundus flavimaculatus?

**CSNB**

Congenital stationary night blindness

In terms of both its name and appearance, with what disease is fundus flavimaculatus often confused?
Fundus flavimaculatus

Juvenile macular dystrophy

Pts with fundus albipectatus c/o night blindness. What do pts with fundus flavimaculatus complain of?
Decreased visual acuity
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance

Retinitis Pigmentosa

Two well-recognized non-classic phenotypes:
--Retinitis punctata albscens
--Choroideremia

Appearance-wise, what is the hallmark of retinitis punctata albscens?
Myriad white dots/flecks in the deep retina

In terms of both its name and appearance, with what disease is retinitis punctata albscens often confused?
Fundus albipunctatus

In terms of both its name and appearance, with what disease is fundus albipunctatus often confused?
Fundus flavimaculatus

Is fundus albipunctatus the same thing as fundus flavimaculatus?
No, and it’s very important to know the difference

Fundus albipunctatus is a non-classic phenotype of RP, whereas fundus flavimaculatus is a classic variant of Stargardt disease, also known as Juvenile macular dystrophy.

Fundus flavimaculatus is often confused with fundus albipunctatus, whereas fundus albipunctatus is a non-classic phenotype of CSNB.

CSNB stands for Congenital stationary night blindness.

Fundus flavimaculatus is often confused with fundus albipunctatus, whereas fundus albipunctatus is a non-classic phenotype of CSNB, whereas fundus flavimaculatus is a classic variant of Stargardt disease, also known as Juvenile macular dystrophy.
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance

Classic fundus appearance:
--Bone spicules
--Waxy disc pallor
--Arteriolar narrowing

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Appearance-wise, what is the hallmark of retinitis punctata albescens?
Myriad white dots/flecks in the deep retina

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Fundus alipunctatus

In terms of both its name and appearance, with what disease is fundus alipunctatus often confused?
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No, and it's very important to know the difference

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Fundus albipunctatus

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OK, what's the difference?

Fundus albipunctatus

In terms of both its name and appearance, with what disease is fundus albipunctatus often confused?
Fundus flavimaculatus

Is fundus albipunctatus the same thing as fundus flavimaculatus?
No, and it's very important to know the difference

OK, what's the difference?
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance

Appearance-wise, what is the hallmark of retinitis punctata albscents?
Myriad white dots/flecks in the deep retina

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By what other name is fundus flavimaculatus/Stargardt disease known?
Juvenile macular dystrophy

Pts with fundus albipunctatus c/o night blindness. What do pts with fundus flavimaculatus complain of?
Decreased visual acuity

two words
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Appearance-wise, what is the hallmark of retinitis punctata albescens?
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Classic fundus appearance:
--Bone spicules
--Waxy disc pallor
--Arteriolar narrowing

Other common signs:
--ONH: not waxy pallor
(ONH = optic nerve head)

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--Fovea: abb./abb.

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Retinitis Pigmentosa
Retinitis Pigmentosa

Cystic foveal changes in RP
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CME:
CMD:

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CME is...
CMD is...

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CME is…‘wet’ (ie, leaks on FA)
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Retinitis Pigmentosa

Other common signs:
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How is CME usually treated?

With PO acetazolamide

What about CMD?
It’s not treatable

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Cystoid macular edema (CME) and Cystoid macular degeneration (CMD) are both conditions characterized by fluid accumulation in the macula, but they differ in their clinical presentation and management.
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Vitreous cell? Does this mean RP is an inflammatory condition?

Retinitis Pigmentosa

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Classic fundus appearance:

Vitreous cell? Does this mean RP is an inflammatory condition?
No. The vitreous cells seen in RP are mainly RPE cells liberated from the degeneration of that structure

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Retinitis Pigmentosa
Fundamentally, RP is a disease of photoreceptor dysfunction; pigment changes are secondary.

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What is the difference between a rod-cone dystrophy and a cone-rod dystrophy?

The difference is the order in which those two populations of photoreceptors are affected by the dystrophy.

Is RP a rod-cone dystrophy, or a cone-rod dystrophy?

It is a rod-cone dystrophy.

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Per the newest (at the time of this writing) edition of the Retina book, the term retinitis pigmentosa is “no longer preferred.” Per the book, what term is preferred instead?

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4 most common inheritance patterns:
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--
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4 most common inheritance patterns:
--Sporadic
--AD
--AR
--X-linked

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Of the 4 most common inheritance patterns, which is... most common?

Retinitis Pigmentosa

Two well-recognized non-classic phenotypes:
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Of the 4 most common inheritance patterns, which is... least common?

Of the 4 most common inheritance patterns, which... carries the worst visual prognosis?

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Classic fundus appearance:
--Bone spicules
--Waxy disc pallor
--Arteriolar narrowing

Retinitis Pigmentosa

Of the 4 most common inheritance patterns, which is…
…most common? Sporadic

4 most common inheritance patterns:
--Sporadic
--AD
--AR
--X-linked

Other common signs:
--ONH: Drusen
--Fovea: CME/AMD
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Typical pattern of VF loss: [specific pattern of VF loss found in early RP]

(Start here)
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Group of inherited retinal diseases characterized by:
--poor scotopic vision
--constricted VF
--abnormal ERG
--characteristic fundus appearance

3 classic 'variant' forms of RP:
--Sectorial
--Sine pigmento
--Central

Characteristic ERG changes in RP:
--Early:
--Late:

Two well-recognized non-classic phenotypes:
--Retinitis punctata albescens
--Choroideremia

Other common signs:
--ONH: Drusen
--Fovea: CME/CMD
--Vitreous: Cell

Classic fundus appearance:
--Bone spicules
--Waxy disc pallor
--Arteriolar narrowing

Normal ERG

Early RP

Typical pattern of VF loss: Mid-peripheral scotomata → coalesce into partial ring → coalesce into complete ring → expand rapidly outward → expand slowly inward

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--Early: Reduced $a$ and $b$ waves
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Whether LCA is a form of RP is another issue currently in flux!

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**There are three subtypes of Usher syndrome**—what are they called? How do they differ?

- **Type I** manifests in the first decade with profound hearing loss, RP and vestibular dysfunction
- **Type II** manifests in the second decade with moderate hearing loss, RP; vestibular function is intact
- **Type III** has progressive hearing loss; the RP varies in severity; vestibular function is sporadic
**Retinitis Pigmentosa**

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Where does Usher syndrome rank as a cause of deaf-blindness in the US?
It is the most common cause thereof

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