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

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 ERG
--characteristic fundus appearance
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

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

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?

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

What are 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
--disc pallor

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
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
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

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

What infectious (and therefore treatable) disease can produce a similar fundus appearance, and must always, always be ruled out 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

Classic 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 ruled out in a patient with an RP-like fundus? 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:
--one word
--three words
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

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

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

Retinitis Pigmentosa

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

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

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

Retinitis Pigmentosa
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

What is the hallmark of choroideremia?
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

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

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

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.

What is the hallmark of retinitis punctata albescens?
--abnormal ERG
--characteristic fundus appearance

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

--Bone spicules
--Waxy disc pallor
--Arteriolar narrowing

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?

Retinitis punctata albescens

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

CSNB

Congenital stationary night blindness
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

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

Congenital stationary night blindness (CSNB)
**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

**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.**

**CSNB** stands for Congenital Stationary Night Blindness.
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 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

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

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

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

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

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

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

OK, what's the difference?
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

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.
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

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

CSNB

Fundus albipunctatus

Retinitis Punctata Albescens

Choroideremia

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctatus

Fundus Albipunctate
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

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 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

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?
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

CSNB Congenital stationary night blindness

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

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

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

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 it mean to say the night blindness is stationary?
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

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

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 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

Appearance-wise, 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

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

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 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

How else do fundus albipunctatus and retinitis punctata albescens differ?

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

How else do **fundus albipunctatus** and **retinitis punctata albescens** differ?
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

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

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

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 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

How else do fundus albipunctatus and retinitis punctata albescens differ?
--On DFE:
Like other forms of RP, retinitis punctata albescens 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 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.
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 alipunctatus

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

Fundus flavimaculatus

What does CSNB stand for?
Congenital stationary night blindness

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

By what other name is fundus flavimaculatus/Stargardt dz known?
Juvenile macular dystrophy
Retinitis Pigmentosa

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

---Bone spicules---Waxy disc pallor---Arteriolar narrowing

Retinitis Pigmentosa

--Retinitis punctata albescens
--Choroideremia

Retinitis Pigmentosa

Two well-recognized non-classic phenotypes:

Appearance-wise, 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

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

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

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?
Whereas fundus albipunctatus is a non-classic phenotype of CSNB, fundus flavimaculatus is a classic variant of Stargardt disease.

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

Pts with fundus albipunctatus 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

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

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

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

Pronounced atrophic changes of the RPE, choriocapillaris and choroid

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?

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

CSNB stands for Congenital Stationary Night Blindness

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

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
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 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

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

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

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

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

What disease is retinitis punctata albescens often confused with?
Fundus albipunctatus

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

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

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

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? Whereas fundus albipunctatus is a non-classic phenotype of CSNB, fundus flavimaculatus is a classic variant of...
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

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

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

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

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?
Whereas fundus albipunctatus is a non-classic phenotype of CSNB, fundus flavimaculatus is a classic variant of Stargardt disease

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

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

OK, what’s the difference?
Whereas fundus albipunctatus is a non-classic phenotype of CSNB, fundus flavimaculatus is a classic variant of Stargardt disease

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

OK, what’s the difference? Whereas fundus albipunctatus is a non-classic phenotype of CSNB, fundus flavimaculatus is a classic variant of Stargardt disease

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

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

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

OK, what’s the difference? Whereas fundus albipunctatus is a non-classic phenotype of CSNB, fundus flavimaculatus is a classic variant of Stargardt disease

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

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

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

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

In terms of both 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?
Whereas fundus albipunctatus is a non-classic phenotype of CSNB, fundus flavimaculatus is a classic variant of Stargardt disease

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

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 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?
Whereas fundus albipunctatus is a non-classic phenotype of CSNB, fundus flavimaculatus is a classic variant of Stargardt disease

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

OK, what’s the difference?
Whereas fundus albipunctatus is a non-classic phenotype of CSNB, fundus flavimaculatus is a classic variant of Stargardt disease

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

Pts with fundus albipunctatus 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

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

Other common signs:
--ONH: not waxy pallor

(ONH = optic nerve head)

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

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

Other common signs:
--ONH: Drusen

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
--Arteriolar narrowing

Other common signs:
--ONH: Drusen
--Fovea: Corrected

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

Retinitis Pigmentosa

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

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

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
--Arteriolar narrowing

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

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

What do these stand for?
CME: Cystoid macular degeneration
CMD:

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

What do these stand for?
CME: Cystoid macular edema
CMD: Cystoid macular degeneration

Retinitis Pigmentosa

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

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

Retinitis Pigmentosa

What is the difference between the two?
CME is…'wet' (ie, leaks on FA)
CMD is…'dry' (no leakage on FA)
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

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

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

What do these stand for?
CME: Cystoid macular edema
CMD: Cystoid macular degeneration

What is the difference between the two?
CME is...
CMD is...

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

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

Retinitis Pigmentosa

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

What do these stand for?
CME: Cystoid macular edema
CMD: Cystoid macular degeneration

What is the difference between the two?
CME is…’wet’ (ie, leaks on FA)
CMD is…’dry’ (no leakage on FA)
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

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

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

Retinitis Pigmentosa

What do these stand for?
CME: Cystoid macular edema
CMD: Cystoid macular degeneration

What is the difference between the two?
CME is…‘wet’ (ie, leaks on FA)
CMD is…‘dry’ (no leakage on FA)

How is CME usually treated?
With PO acetazolamide

It's not treatable
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

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

Retinitis Pigmentosa

What do these stand for?
CME: Cystoid macular edema
CMD: Cystoid macular degeneration

What is the difference between the two?
CME is…’wet’ (ie, leaks on FA)
CMD is…’dry’ (no leakage on FA)

How is CME usually treated?
With PO acetazolamide
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

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

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

Retinitis Pigmentosa

What do these stand for?
CME: Cystoid macular edema
CMD: Cystoid macular degeneration

What is the difference between the two?
CME is…‘wet’ (ie, leaks on FA)
CMD is…‘dry’ (no leakage on FA)

How is CME usually treated?
With PO acetazolamide

What about CMD?
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

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

Retinitis Pigmentosa

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

What do these stand for?
CME: Cystoid macular edema
CMD: Cystoid macular degeneration

What is the difference between the two?
CME is…‘wet’ (ie, leaks on FA)
CMD is…‘dry’ (no leakage on FA)

How is CME usually treated?
With PO acetazolamide

What about CMD?
It’s not treatable
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

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

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

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

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

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

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

Vitreous cell? Does this mean RP is an inflammatory condition?

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

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

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

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

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

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

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

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

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

(posterior subcapsular cataract)

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

Retinitis Pigmentosa

Retinitis Pigmentosa

Retinitis Pigmentosa

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

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

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

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

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

Retinitis Pigmentosa

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

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

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

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

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

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

Retinitis Pigmentosa

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

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

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

Retinitis Pigmentosa

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

Other common signs:
--ONH: Drusen
--Fovea: CME/CMD
--Vitreous: Cells
--Lens: PSC

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

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.

It is a rod-cone dystrophy.
Fundamentally, RP is a disease of photoreceptor dysfunction; pigment changes are secondary.

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

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

Retinitis Pigmentosa

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

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

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

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

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?
**Fundamentally,** RP is a disease of photoreceptor dysfunction; pigment changes are secondary.

**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

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

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

---

**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.
Retinitis Pigmentosa

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

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

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

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

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

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

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

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

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

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

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

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?
Rod-cone 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

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

4 most common inheritance patterns:
--
--
--

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

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
--Arteriolar narrowing

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

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

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

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

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

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

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

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

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

Retinitis Pigmentosa is a 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

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

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

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

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

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

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

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

Of the 4 most common inheritance patterns, which is...
- ...most common? Sporadic
- ...least common? X-linked

Of the 4 most common inheritance patterns, which carries the worst visual prognosis?
- X-linked

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

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
-- Arteriolar narrowing

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

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

Retinitis Pigmentosa

Of the 4 most common inheritance patterns, which is...
... most common? Sporadic
... least common? X-linked

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

Other common signs:
--ONH: Drusen
--Fovea: CME/CMD
--Vitreous: Cells
--Lens: PSC

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

Retinitis

Of the 4 most common inheritance patterns, which is...
...most common? Sporadic
...least common? X-linked
...carries the worst visual prognosis?

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

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

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

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

Of the 4 most common inheritance patterns, which is...
...most common? Sporadic
...least common? X-linked
...carries the worst visual prognosis? X-linked
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

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

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

Typical pattern of VF loss:
[specific pattern of VF loss found in early RP]

(Start here)
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

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

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

Typical pattern of VF loss: Mid-peripheral scotomata
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

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

- Typical pattern of VF loss: Mid-peripheral scotomata ➔ [how the VF evolves next]
Two well-recognized non-classic phenotypes:
--Retinitis punctata albescens
--Choroideremia

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

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

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

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

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
Typical pattern of VF loss: Mid-peripheral scotomata $\rightarrow$ coalesce into partial ring $\rightarrow$ expand rapidly outward $\rightarrow$ expand slowly inward.

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

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

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

Other common signs:
--Bone spicules
--Waxy disc pallor
--Arteriolar narrowing

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

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

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

Retinitis Pigmentosa
Retinitis Pigmentosa

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

Typical pattern of VF loss: Mid-peripheral scotomata → coalesce into partial ring → coalesce into complete ring

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

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

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

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

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

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

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

Typical pattern of VF loss: Mid-peripheral scotomata → coalesce into partial ring → coalesce into complete ring
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

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

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

Retinitis Pigmentosa?

Typical pattern of VF loss: Mid-peripheral scotomata $\rightarrow$ coalesce into partial ring $\rightarrow$ coalesce into complete ring $\rightarrow$ [how the VF evolves next]
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

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

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

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

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

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

Typical pattern of VF loss: Mid-peripheral scotomata → coalesce into partial ring → coalesce into complete ring → expand rapidly outward → [how the VF evolves next]
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

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

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

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

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

3 classic ‘variant’ forms of RP:
--
--
--

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

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

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

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

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

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

Retinitis Pigmentosa
Retinitis Pigmentosa

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

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

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

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

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

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

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

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

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

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

- What does it mean to say RP is ‘sectorial’?
  - Simply that it is limited to one or two sectors of the fundus
  - Is it symmetric between the two eyes?
    - Yes, which is an important clue that it’s RP (as opposed to an acquired insult in one eye)
  - What does *sine pigmento* mean?
    - It’s Latin for ‘without pigment.’ It refers to a variant of RP in which the spicules are absent
  - What is central RP?
    - In essence, a reversed form of RP. Whereas in ‘normal’ RP visual acuity and the central visual field are spared until late in the disease process, in central RP acuity is profoundly affected early on, and VF loss progresses outward from fixation.
Retinitis Pigmentosa

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

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

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

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

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

Retinitis

What does it mean to say RP is ‘sectorial’?
Simply that it is limited to one or two sectors of the fundus
Retinitis Pigmentosa

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

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

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

Other common signs:
--ONH: Drusen
--Fovea: CME/CMD
--Vitreous: Cells
--Lens: PSC

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

Retinitis Pigmentosa

What does it mean to say RP is 'sectorial'?
Simply that it is limited to one or two sectors of the fundus

Is it symmetric between the two eyes?

What does sine pigmento mean?
It's Latin for 'without pigment.' It refers to a variant of RP in which the spicules are absent

Central RP
In essence, a reversed form of RP. Whereas in 'normal' RP visual acuity and the central visual field are spared until late in the disease process, in central RP acuity is profoundly affected early on, and VF loss progresses outward from fixation.
Retinitis Pigmentosa

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

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

Retinitis Pigmentosa

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

Other common signs:
--ONH: Drusen
--Fovea: CME/CMD
--Vitreous: Cells
--Lens: PSC

Retinitis Pigmentosa

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

What does it mean to say RP is ‘sectorial’?
Simply that it is limited to one or two sectors of the fundus

Is it symmetric between the two eyes?
Yes, which is an important clue that it’s RP (as opposed to an acquired insult in one eye)

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

Other common signs:
--ONH: Drusen
--Fovea: CME/CMD
--Vitreous: Cells
--Lens: PSC

What does it mean to say RP is ‘sectorial’?
Simply that it is limited to one or two sectors of the fundus

Is it symmetric between the two eyes?
Yes, which is an important clue that it’s RP (as opposed to an acquired insult in one eye)

What does sine pigmento mean?

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

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

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

- Other common signs:
  - ONH: Drusen
  - Fovea: CME/CMD
  - Vitreous: Cells
  - Lens: PSC

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

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

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

Retinitis

- What does it mean to say RP is ‘sectorial’?
  - Simply that it is limited to one or two sectors of the fundus

- Is it symmetric between the two eyes?
  - Yes, which is an important clue that it’s RP (as opposed to an acquired insult in one eye)

- What does sino pigmento mean?
  - It’s Latin for ‘without pigment.’ It refers to a variant of RP in which the spicules are absent

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

- Other common signs:
  - ONH: Drusen
  - Fovea: CME/CMD
  - Vitreous: Cells
  - Lens: PSC

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

- Other common signs:
  - ONH: Drusen
  - Fovea: CME/CMD
  - Vitreous: Cells
  - Lens: PSC

Retinitis

- What does it mean to say RP is ‘sectorial’?
  - Simply that it is limited to one or two sectors of the fundus

- Is it symmetric between the two eyes?
  - Yes, which is an important clue that it’s RP (as opposed to an acquired insult in one eye)

- What does sine pigmento mean?
  - It’s Latin for ‘without pigment.’ It refers to a variant of RP in which the spicules are absent

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

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

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

What does it mean to say RP is ‘sectorial’?
Simply that it is limited to one or two sectors of the fundus

Is it symmetric between the two eyes?
Yes, which is an important clue that it’s RP (as opposed to an acquired insult in one eye)

What does sine pigmento mean?
It’s Latin for ‘without pigment.’ It refers to a variant of RP in which the spicules are absent

What is central RP?
In essence, a reversed form of RP. Whereas in ‘normal’ RP visual acuity and the central visual field are spared until late in the disease process, in central RP acuity is profoundly affected early on, and VF loss progresses outward from fixation.

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

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

Retinitis Pigmentosa

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

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

Retinitis Pigmentosa

What does it mean to say RP is ‘sectorial’?
Simply that it is limited to one or two sectors of the fundus

Is it symmetric between the two eyes?
Yes, which is an important clue that it’s RP (as opposed to an acquired insult in one eye)

What does sine pigmento mean?
It’s Latin for ‘without pigment.’ It refers to a variant of RP in which the spicules are absent

What is central RP?
In essence, a reversed form of RP. Whereas in ‘normal’ RP visual acuity and the central visual field are spared until late in the disease process, in central RP acuity is profoundly affected early on, and VF loss progresses outward from fixation.
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

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

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

What does it mean to say RP is ‘sectorial’?
Simply that it is limited to one or two sectors of the fundus

Is it symmetric between the two eyes?
Yes, which is an important clue that it’s RP (as opposed to an acquired insult in one eye)

What does sine pigmento mean?
It’s Latin for ‘without pigment.’ It refers to a variant of RP in which the spicules are absent

What is central RP?
In essence, a reversed form of RP. Whereas in ‘normal’ RP visual acuity and the central visual field are spared until late in the disease process, in central RP acuity is profoundly affected early on, and VF loss progresses outward from fixation.

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

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

Characteristic ERG changes in RP:
--Early: Reduced a and b waves
--Late: Undetectable b-wave

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

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

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

Other common signs:

Other common signs:

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

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

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

Characteristic ERG changes in RP:
--Early: Reduced a and b waves
--Late: Undetectable b-wave

Normal ERG

Early RP
Retinitis Pigmentosa

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

Characteristic ERG changes in RP:
--Early: Reduced a and b waves
--Late:

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

Retinitis Pigmentosa

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

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

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

Normal ERG

Early RP

a-wave

b-wave

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

Characteristic ERG changes in RP:
--Early: Reduced a and b waves
--Late:

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

Retinitis Pigmentosa

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

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

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

Normal ERG

Early RP

a-wave

b-wave

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

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

Characteristic ERG changes in RP:
--Early: Reduced $a$ and $b$ waves
--Late:

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

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

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

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

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

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

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

Characteristic ERG changes in RP:
--Early: Reduced $a$ and $b$ waves
--Late: Undetectable $b$-wave

Normal ERG

Early RP

Late RP

$\rightarrow$ $\rightarrow$ $\rightarrow$
Retinitis Pigmentosa

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

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

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

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

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

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

- Characteristic ERG changes in RP:
  - Early: Reduced $a$ and $b$ waves
  - Late: Undetectable $b$-wave

- Normal ERG:
  - $a$-wave
  - $b$-wave

- Early RP:
  - $a$-wave
  - Undetectable $b$-wave

- Late RP:
  - ‘$b$-wave’
  - ‘$a$-wave’

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

- Fundamentally, RP is a disease of photoreceptor dysfunction; pigment changes are secondary
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

- Characteristic ERG changes in RP:
  - Early: Reduced a and b waves
  - Late: Undetectable

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

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

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

- Fundamentally, RP is a disease of photoreceptor dysfunction; pigment changes are secondary

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

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

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

- Leber’s congenital amaurosis is an age-related variant of RP

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

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

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

- Characteristic ERG changes in RP:
  - Early: Reduced a and b waves
  - Late: Undetectable

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

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

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

- Fundamentally, RP is a disease of photoreceptor dysfunction; pigment changes are secondary

- Retinitis Pigmentosa

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

- Leber’s congenital amaurosis is an age-related variant of RP

- Characteristic ERG changes in RP:
  - Early: Reduced a and b waves
  - Late: Undetectable

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

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

Retinitis Pigmentosa is a group of inherited retinal diseases characterized by:
- Poor scotopic vision
- Constricted VF
- Abnormal ERG
- Characteristic fundus appearance

Leber's congenital amaurosis is an age-related variant of RP.

Whether LCA is a form of RP is another issue currently in flux!

4 classic 'variant' forms of RP:
- Sectorial
- Sin pigmento
- Pigmented paravenous atrophy
- Centro-peripheral

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

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

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

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

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

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

4 classic 'variant' forms of RP:
- Sectorial
- Sin pigmento
- Pigmented paravenous atrophy
- Centro-peripheral

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

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

**Usher syndrome** = *Retinitis pigmentosa* + two words

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

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

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

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

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

4 classic ‘variant’ forms of RP:
--Sectorial
--sin pigimento
--Pigmented paravenous atrophy
--centro-peripheral

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

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

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

Other common signs:

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

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

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

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

Retinitis Pigmentosa

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

4 classic ‘variant’ forms of RP:
--Sectorial
--sin pigimento
--Pigmented paravenous atrophy
--centro-peripheral

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

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

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

Other common signs:

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

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

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

Typical pattern of VF loss: Mid-peripheral scotomata coalesce into partial ring coalesce into complete ring expand rapidly outward expand slowly inward
Leber's congenital amaurosis is an age-related variant of RP. Characteristic ERG changes in RP:
- Early: Reduced a and b waves
- Late: Undetectable

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

4 classic 'variant' forms of RP:
- Sectorial
- Sin pigmento
- Pigmented paravenous atrophy
- Centro-peripheral

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

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

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

Where does Usher syndrome rank as a cause of deaf-blindness in the US?

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

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

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

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

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

Where does Usher syndrome rank as a cause of deaf-blindness in the US?
Retinitis Pigmentosa

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

Characteristic ERG changes in RP:
-- Early: Reduced a and b waves
-- Late: Undetectable

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

4 classic ‘variant’ forms of RP:
-- Sectorial
-- sin pigmen
t-- Pigmented paravenous atrophy
-- centro-peripheral

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

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

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

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

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

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

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

There are three types of Usher syndrome--what are they called?

4 classic 'variant' forms of RP:
--Sectorial
--sin pigmento
--Pigmented paravenous atrophy
--centro-peripheral

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

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

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

Characteristic ERG changes in RP:
--Early: Reduced a and b waves
--Late: Undetectable

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

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

**Leber's congenital amaurosis** is an age-related variant of RP

Characteristic ERG changes in RP:
- Early: Reduced a and b waves
- Late: Undetectable

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

4 classic 'variant' forms of RP:
- Sectorial
- Sin pigmento
- Pigmented paravenous atrophy
- Centro-peripheral

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

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

**Usher syndrome** = Retinitis pigmentosa + sensorineural deafness

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

*There are three types of Usher syndrome--what are they called?*
- **Type I**
- **Type II**
- **Type III**

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

**Other 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

**Usher syndrome** = Retinitis pigmentosa + sensorineural deafness

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

*There are three types of Usher syndrome--what are they called?*
- **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

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

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

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

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

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

There are three types of Usher syndrome--what are they called? How do they manifest?
--Type I manifests…
--Type II
--Type III

4 classic ‘variant’ forms of RP:
--Sectorial
--sin pigmento
--Pigmented paravenous atrophy
--centro-peripheral

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

Typical pattern of VF loss: Mid-peripheral scotomata ➔ coalesce into partial ring ➔ coalesce into complete ring ➔ expand rapidly outward ➔ expand slowly inward
Two well-recognized non-classic phenotypes:
--Retinitis punctata albescens
--Choroideremia

Leber’s congenital amaurosis is an age-related variant of RP.

Characteristic ERG changes in RP:
--Early: Reduced a and b waves
--Late: Undetectable

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

4 classic ‘variant’ forms of RP:
--Sectorial
--sin pigmento
--Pigmented paravenous atrophy
--centro-peripheral

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

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

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

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

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

There are three types of Usher syndrome--what are they called? How do they manifest?
--Type I manifests...in the first decade with profound hearing loss, RP and vestibular dysfunction
--Type II
--Type III

Typical pattern of VF loss: Mid-peripheral scotomata $\Rightarrow$ coalesce into partial ring $\Rightarrow$ coalesce into complete ring $\Rightarrow$ expand rapidly outward $\Rightarrow$ expand slowly inward
Leber’s congenital amaurosis is an age-related variant of RP.

Characteristic ERG changes in RP:
- Early: Reduced a and b waves
- Late: Undetectable

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

4 classic 'variant' forms of RP:
- Sectorial
- Sin pigmeno
- Pigmented paravenous atrophy
- Centro-peripheral

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

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

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

There are three types of Usher syndrome—what are they called? How do they manifest?
- **Type I** manifests...in the first decade with profound hearing loss, RP and vestibular dysfunction
- **Type II**
- **Type III** has progressive hearing loss; the RP varies in severity; vestibular function is sporadic

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

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
- Arteriolar narrowing

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

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

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

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

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

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

There are three types of Usher syndrome—what are they called? How do they manifest?
- **Type I** manifests...in the first decade with profound hearing loss, RP and vestibular dysfunction
- **Type II**
- **Type III** has...progressive hearing loss; the RP varies in severity; vestibular function is sporadic

4 classic 'variant' forms of RP:
- Sectorial
- Sin pigmeno
- Pigmented paravenous atrophy
- Centro-peripheral

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

Typical pattern of VF loss: Mid-peripheral scotomata → coalesce into partial ring → coalesce into complete ring → expand rapidly outward → expand slowly inward
Leber’s congenital amaurosis is an age-related variant of RP. Characteristic ERG changes in RP:
--Early: Reduced a and b waves
--Late: Undetectable

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

4 classic ‘variant’ forms of RP:
--Sectorial
--sin pigmento
--Pigmented paravenous atrophy
--centro-peripheral

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

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

Retinitis Pigmentosa

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

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

Other common signs:

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

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

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

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

There are three types of Usher syndrome--what are they called? How do they manifest?
--Type I manifests…in the first decade with profound hearing loss, RP and vestibular dysfunction
--Type II manifests…
--Type III

Retinitis Pigmentosa

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

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

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

Retinitis Pigmentosa

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

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

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

Retinitis Pigmentosa

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

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

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

Retinitis Pigmentosa

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

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

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

Retinitis Pigmentosa

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

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

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

Retinitis Pigmentosa

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

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

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

Retinitis Pigmentosa

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

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

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

Retinitis Pigmentosa

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

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

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

Retinitis Pigmentosa

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

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

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

Retinitis Pigmentosa

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

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

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

Retinitis Pigmentosa

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

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

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

Retinitis Pigmentosa

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

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

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

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

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

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

There are three types of Usher syndrome--what are they called? How do they manifest?
--Type I manifests...in the first decade with profound hearing loss, RP and vestibular dysfunction
--Type II manifests...in the decade with hearing loss, RP; vestibular function is
--Type III

4 classic 'variant' forms of RP:
--Sectorial
--sin pigmento
--Pigmented paravenous atrophy
--centro-peripheral

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

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

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

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

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

There are three types of Usher syndrome--what are they called? How do they manifest?
--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

4 classic 'variant' forms of RP:
--Sectorial
--sin pigmento
--Pigmented paravenous atrophy
--centro-peripheral

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

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

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

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

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

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

There are three types of Usher syndrome--what are they called? How do they manifest?
--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

4 classic 'variant' forms of RP:
--Sectorial
--sin pigmento
--Pigmented paravenous atrophy
--centro-peripheral

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

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

Two well-recognized non-classic phenotypes:
--Retinitis punctata albescens
--Choroideremia
Leber's congenital amaurosis is an age-related variant of RP.

Characteristic ERG changes in RP:
- Early: Reduced a and b waves
- Late: Undetectable

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

4 classic 'variant' forms of RP:
- Sectorial
- Sin pigmento
- Pigmented paravenous atrophy
- Centro-peripheral

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

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

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

There are three types of Usher syndrome--what are they called? How do they manifest?
- 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...

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

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
- Arteriolar narrowing

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

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

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

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

**Usher syndrome** = Retinitis pigmentosa + sensorineural deafness

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

There are three types of Usher syndrome--what are they called? How do they manifest?

- **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 hearing loss; the RP varies in severity; vestibular function is sporadic

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

**Typical pattern of VF loss:** Mid-peripheral scotomata → coalesce into partial ring → coalesce into complete ring → expand rapidly outward → expand slowly inward
Leber's congenital amaurosis is an age-related variant of RP.

Characteristic ERG changes in RP:

- Early: Reduced a and b waves
- Late: Undetectable

Typical pattern of VF loss:

Mid-peripheral scotomata → coalesce into partial ring → coalesce into complete ring → expand rapidly outward → expand slowly inward

4 classic 'variant' forms of RP:
- Sectorial
- Sin pigmento
- Pigmented paravenous atrophy
- Centro-peripheral

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

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

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

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

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

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

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

There are three types of Usher syndrome—what are they called? How do they manifest?
- **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

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

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

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

There are three types of Usher syndrome—what are they called? How do they manifest?
- **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

4 classic 'variant' forms of RP:
- Sectorial
- Sin pigmento
- Pigmented paravenous atrophy
- Centro-peripheral

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

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

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

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

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

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

There are three types of Usher syndrome:
--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

Usher syndrome tl;dr
= RP + hearing loss +/- vestibular dysfunction
--Type I: Early, severe
--Type II: Later, less severe
--Type III: Variable

4 classic ‘variant’ forms of RP:
--Sectorial
--sin pigmento
--Pigmented paravenous atrophy
--centro-peripheral

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

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