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
--brightness
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

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Group of inherited retinal diseases characterized by:
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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
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Night blindness and progressive peripheral VF loss are the two hallmark symptoms of RP

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

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

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

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

Group of inherited retinal diseases characterized by:
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Group of inherited retinal diseases characterized by:
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-- 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:
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Classic fundus appearance:
--Bone spicules
--Waxy disc pallor
--Arteriolar narrowing

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Classic fundus appearance:
--Bone spicules
Retinitis Pigmentosa

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Classic fundus appearance:
-- Bone spicules

What are bone spicules?

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

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
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Retinitis Pigmentosa

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Which of the three appears first?

Retinitis Pigmentosa
Retinitis Pigmentosa

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Which of the three appears first?
Arteriolar narrowing
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Possible fundus appearance:
--Bone spicules
--Waxy disc pallor
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What infectious (and therefore treatable) disease can produce a similar fundus appearance, and must always, always be at least considered in a patient with an RP-like fundus?
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Syphilis
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Classic fundus appearance:
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Two well-recognized non-classic phenotypes:

Retinitis Pigmentosa
Retinitis Pigmentosa

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

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--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
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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.

-- abnormal ERG
-- characteristic fundus appearance

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Fundus albipunctatus

Retinitis Pigmentosa

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

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

Choroideremia

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Retinitis

Punctata

Albescens

Congenital stationary night blindness

CSNB
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OK, what’s the difference?
Retinitis Pigmentosa

Group of inherited retinal diseases characterized by:
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Retinitis Pigmentosa

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

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

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

Is retinitis punctata albscens the same thing as fundus albipunctatus?
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Congenital stationary night blindness
Retinitis Pigmentosa

- Bone spicules
- Waxy disc pallor
- Arteriolar narrowing

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

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

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

**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

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How else do fundus albipunctatus and retinitis punctata albescens differ?

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

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

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

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Retinitis Pigmentosa

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

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

In terms of both its name and appearance, with what disease is fundus albi-punctatus 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:
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-- constricted VF
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-- characteristic fundus appearance

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

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Fundus albipunctatus

Fundus albipunctatus

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

Two well-recognized non-classic phenotypes:
-- Retinitis punctata albescens
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Retinitis Pigmentosa is a group of inherited retinal diseases characterized by:
- Poor scotopic vision
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Two well-recognized non-classic phenotypes are:
- Retinitis punctata albescens
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Fundus albipunctatus

Fundus albipunctatus is often confused with:
- Fundus flavimaculatus

Fundus flavimaculatus is often confused with:
- Fundus albipunctatus

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

Fundus flavimaculatus is known by other names:
- Juvenile macular dystrophy

Patients with fundus albipunctatus complain of:
- Decreased visual acuity

Fundus albipunctatus is a non-classic phenotype of CSNB.

Fundus albipunctatus

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

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

Fundus flavimaculatus is a classic variant of Stargardt disease.

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

Patients with fundus albipunctatus complain of:
- Night blindness
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Is fundus albipunctatus the same thing as fundus flavimaculatus?

CSNB stands for Congenital Stationary Night Blindness.

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

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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

Fundus albipunctatus

Fundus flavimaculatus

CSNB

Congenital stationary night blindness

Juvenile macular dystrophy
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:
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Pts with fundus albipunctatus c/o night blindness. What do pts with fundus flavimaculatus complain of?
Retinitis Pigmentosa

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

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

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

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--characteristic fundus appearance

Classic fundus appearance:
--Bone spicules
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--Arteriolar narrowing

Other common signs:
--ONH: Drusen

Two well-recognized non-classic phenotypes:
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Other common signs:
--ONH: Drusen
--Fovea: abb./abb.
Retinitis Pigmentosa

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

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What do these stand for?
CME:
CMD:
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CME: Cystoid macular edema
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What is the difference between the two?
CME is…'wet' (ie, leaks on FA)
CMD is…'dry' (no leakage on FA)
Retinitis Pigmentosa

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CMD is...‘dry’ (no leakage on FA)

How is CME usually treated?

With PO acetazolamide

What about CMD?
It's not treatable
Retinitis Pigmentosa

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--ONH: Drusen
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CMD is…‘dry’ (no leakage on FA)

How is CME usually treated?
With PO acetazolamide

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Retinitis Pigmentosa

- Bone spicules
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Classical fundus appearance:
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How is CME usually treated?
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What about CMD?

Retinitis Pigmentosa
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CME: Cystoid macular edema
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CME is...‘wet’ (ie, leaks on FA)
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How is CME usually treated?
With PO acetazolamide

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It’s not treatable
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Retinitis Pigmentosa

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

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

Retinitis Pigmentosa

--Bone spicules
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Other common signs:
--ONH: Drusen
--Fovea: CME/CMD
--Lens: PSC

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

Retinitis Pigmentosa

Two well-recognized non-classic phenotypes:
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Vitreous: Cell
Retinitis Pigmentosa

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Classic fundus appearance:
--Bone spicules
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Other common signs:
--ONH: Drusen
--Fovea: CME/CMD
--Vitreous Cell

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

Other common signs:
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Fundamentally, RP is a disease of dysfunction; pigment changes are secondary.
Fundamentally, RP is a disease of photoreceptor dysfunction; pigment changes are secondary.

Retinitis Pigmentosa

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

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

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

It is a rod-cone dystrophy.
Retinitis Pigmentosa

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--Fovea: CME/CMD
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Retinitis Pigmentosa

Classic fundus appearance:
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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

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Rod-cone dystrophy
Retinitis Pigmentosa

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4 most common inheritance patterns:
--

Other common signs:
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Two well-recognized non-classic phenotypes:
--Retinitis punctata albescens
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Fundamentally, RP is a disease of photoreceptor dysfunction; pigment changes are secondary.

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

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Retinitis Pigmentosa

Retinitis Pigmentosa

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

4 most common inheritance patterns:
--Sporadic
--AD
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Retinitis Pigmentosa

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

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

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

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Of the 4 most common inheritance patterns, which is…
…most common? Sporadic
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Classic fundus appearance:
--Bone spicules
--Waxy disc pallor
--Arteriolar narrowing

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Other common signs:
--ONH: Drusen
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(Start here)

Typical pattern of VF loss: [specific pattern of VF loss found in early RP]
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Typical pattern of VF loss: Mid-peripheral scotomata

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Typical pattern of VF loss: **Mid-peripheral scotomata** ➔ [how the VF evolves next]
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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

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

Normal ERG

Early RP

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Characteristic ERG changes in RP:
--Early:
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- Sectorial
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**Normal ERG**
- a-wave
- b-wave

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Normal ERG

Early RP

Late RP

<table>
<thead>
<tr>
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</tr>
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<tbody>
<tr>
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Eponym word is an age-related variant of RP
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Whether LCA is a form of RP is another issue currently in flux!

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

It is the most common cause thereof.

There are three subtypes of Usher syndrome:
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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

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

Other common signs:
- ONH: Drusen
- Fovea: CME/CMD
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Two well-recognized non-classic phenotypes:
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Characteristic ERG changes in RP:
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Usher syndrome tl;dr
= RP + hearing loss +/- vestibular dysfunction
--Type I: Early, severe
--Type II: Later, less severe
--Type III: Variable

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