In my teaching experience, this is one of the more challenging topics in ophthalmology. (The corneal dystrophies are up there too.) While I won’t say this set makes learning the material easy—there’s simply too much esoterica for that—I do think it makes it easier. My point here is not to toot my own horn, but rather to give hope and encouragement—you can master this topic!

My advice: In prepping for the OKAP, try to run through this slide-set once or twice a month. When you reach the final, frantic few weeks of cramming, don’t wade through the whole thing—just do the tl;dr at the end (it starts around slide 315).

Now, let’s get this bread!
First things first. An RP-like fundus has three characteristic attributes. What are they?
First things first. An RP-like fundus has three characteristic attributes. What are they? --Bone spicules --Waxy disc pallor --Arteriolar narrowing
DDx for an RP-like Fundus

RP
First things first. An RP-like fundus has three characteristic attributes. What are they?
--Bone spicules
--Waxy disc pallor
--Arteriolar narrowing

What two vision issues are the defining characteristics of RP?
--
--
First things first. An RP-like fundus has three characteristic attributes. What are they?
--Bone spicules
--Waxy disc pallor
--Arteriolar narrowing

What two vision issues are the defining characteristics of RP?
--Nyctalopia
--Visual field loss
Typical pattern of VF loss in RP: Mid-peripheral scotomata → coalesce into *partial* rings → coalesce into *complete* ring → expand rapidly *outward* → expand slowly *inward*
First things first. An RP-like fundus has three characteristic attributes. What are they?
--Bone spicules
--Waxy disc pallor
--Arteriolar narrowing

What two vision issues are the defining characteristics of RP?
--Nyctalopia
--Visual field loss

Results of what specialized testing modality are always abnormal in RP?
First things first. An RP-like fundus has three characteristic attributes. 
What are they?
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--Waxy disc pallor
--Arteriolar narrowing

What two vision issues are the defining characteristics of RP?
--Nyctalopia
--Visual field loss

Results of what specialized testing modality are always abnormal in RP?
Electroretinogram (ERG)
DDx for an RP-like Fundus

Characteristic ERG changes in RP:
--**Early**: Reduced $a$ and $b$ waves
--**Late**: Undetectable
First things first. An RP-like fundus has three characteristic attributes. What are they?
--Bone spicules
--Waxy disc pallor
--Arteriolar narrowing

What two vision issues are the defining characteristics of RP?
--Nyctalopia
--Visual field loss

Results of what specialized testing modality are always abnormal in RP?
Electroretinogram (ERG)

These are the defining features of RP
DDx for an RP-like Fundus

These next few slides lay out the overall way you should think about an RP-like fundus (especially on the OKAP/Boards)
When a pt’s fundi have an RP-like appearance, one of three things is going on:
1)
2)
3)
When a pt's fundi have an RP-like appearance, one of three things is going on:
1) The pt has RP (duh)
2) 
3)
When a pt’s fundi have an RP-like appearance, one of three things is going on:
1) The pt has RP; or
2) s/he has a systemic condition in which retinal manifestations c/w RP occur
3)
When a pt’s fundi have an RP-like appearance, one of three things is going on:  
1) The pt has RP; or  
2) s/he has a systemic condition in which retinal manifestations c/w RP occur; or  
3) s/he has a condition that has nothing to do with RP--it just happens to have a similar appearance
For more on RP itself, see slide-set R38.
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- (the most common syndromic association)
- (one of the ‘inborn errors of metabolism’ families)

- (a family of neurodegenerative diseases)

- (a family of diseases related to dysfunction of a ubiquitous organelle)

- (a disorder of fat metabolism)

‘Pseudo-RP’
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
- Neuronal ceroid lipofuscinoses
- Ciliopathies
- Abetalipoproteinemia

‘Pseudo-RP’
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders

- Neuronal ceroid lipofuscinoses
  --aka 'Batten dz'
  --aka 'Bassen-Kornzweig dz'
- Ciliopathies

- Abetalipoproteinemia
  --aka 'eponym-eponym'
DDx for an RP-like Fundus

Primary RP (aka typical RP)

Secondary RP (aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
- Neuronal ceroid lipofuscinoses
  --aka ‘Batten dz’
- Ciliopathies

‘Pseudo-RP’
- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders

Usher syndrome = Retinitis pigmentosa + two words

- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’
DDx for an RP-like Fundus

Primary RP (aka typical RP)

Secondary RP (aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
- Abetalipoproteinemia  
  --aka ‘Bassen-Kornzweig dz’

Usher syndrome = Retinitis pigmentosa + sensorineural deafness
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders

‘Pseudo-RP’

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

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

- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’
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

Abetalipoproteinemia
--aka ‘Bassen-Kornzweig dz’
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?*

- Abetalipoproteinemia
  -- *aka* ‘Bassen-Kornzweig dz’
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders

‘Pseudo-RP’

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

Abetalipoproteinemia
--aka ‘Bassen-Kornzweig dz’
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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(aka Complex RP; Syndromic RP)
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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

- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
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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 types of Usher syndrome--what are they called? How do they manifest?
--- Type I manifests…in the[ ] decade with[ ] hearing loss, RP and vestibular dysfunction
--- Type II
--- Type III

- Abetalipoproteinemia
  --- aka ‘Bassen-Kornzweig dz’
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It is the most common cause thereof

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-- Type III

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(aka typical RP)

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‘Pseudo-RP’

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**
**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 hearing loss, RP; vestibular function is intact

--**Type III**

- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’
**DDx for an RP-like Fundus**

**Primary RP**

(aka typical RP)

**Secondary RP**

(aka Complex RP; Syndromic RP)

- **Usher syndrome**
- Peroxisomal disorders

**‘Pseudo-RP’**

**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**

- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
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‘Pseudo-RP’

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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 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...

- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’
Pseudo-RP

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders

Primary RP
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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 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 in severity; vestibular function is

Abetalipoproteinemia
--aka ‘Bassen-Kornzweig dz’
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

Abetalipoproteinemia
--aka 'Bassen-Kornzweig dz'
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders

‘Pseudo-RP’

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 early, severe
- Type II later, less severe
- Type III variable

Usher syndrome tl;dr
= RP + hearing loss +/- vestibular dysfunction
--Type I early, severe
--Type II later, less severe
--Type III variable

Abetalipoproteinemia
--aka ‘Bassen-Kornzweig dz’
What are peroxisomal disorders?

- Usher syndrome
- Neuronal ceroid
- Peroxisomal disorders

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

‘Pseudo-RP’
What are peroxisomal disorders?
A heterogeneous group of disorders of peroxisome function
What are peroxisomal disorders?
A heterogeneous group of disorders of peroxisome function

What is/are peroxisomes?
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- **Peroxisomal disorders**
- Neuronal ceroid

**What are peroxisomal disorders?**
A heterogeneous group of disorders of peroxisome function

**What is/are peroxisomes?**
Intracellular organelles that play key roles in many aspects of cell metabolism
What are peroxisomal disorders?
A heterogeneous group of disorders of peroxisome function

What is/are peroxisomes?
Intracellular organelles that play key roles in many aspects of cell metabolism

What is the hallmark lab abnormality of the peroxisomal disorders that present with LCA?

(LCA = Leber’s congenital amaurosis, an age-related form of RP)
What are peroxisomal disorders?
A heterogeneous group of disorders of peroxisome function

What is/are peroxisomes?
Intracellular organelles that play key roles in many aspects of cell metabolism

What is the hallmark lab abnormality of the peroxisomal disorders that present with LCA?
Abnormally high serum levels of very long chain fatty acids (VLCFA)
What are peroxisomal disorders?
A heterogeneous group of disorders of peroxisome function

What is/are peroxisomes?
Intracellular organelles that play key roles in many aspects of cell metabolism

What is the hallmark lab abnormality of the peroxisomal disorders that present with LCA?
Abnormally high serum levels of very long chain fatty acids (VLCFA)

What specific peroxisomal disorders can manifest an LCA-type presentation?
--
--
--
**DDx for an RP-like Fundus**

- **Primary RP** (aka typical RP)
- **Secondary RP** (aka Complex RP; Syndromic RP)
  - Usher syndrome
  - **Peroxisomal disorders**
    - Zellweger syndrome
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    - Infantile Refsum dz
  - Neuronal ceroid

What are peroxisomal disorders?
A heterogeneous group of disorders of peroxisome function

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Abnormally high serum levels of **very long chain fatty acids** (VLCFA)

What specific peroxisomal disorders can manifest an LCA-type presentation?
-- Zellweger syndrome
-- Neonatal adrenoleukodystrophy (NALD)
-- Infantile Refsum dz
DDx for an RP-like Fundus

Primary RP
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  -- Infantile Refsum dz
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‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?

Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
-- LCA
-- Deafness
-- Hypotonia
-- Seizures
-- Abnormal facies (high forehead; hypertelorism)

What is the prognosis?
It is uniformly fatal by age 1 year
DDx for an RP-like Fundus

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(aka typical RP)

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  - Neonatal adrenoleukodystrophy
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DDx for an RP-like Fundus

Zellweger syndrome facies: High forehead; hypertelorism
DDx for an RP-like Fundus

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What is the prognosis?
It is uniformly fatal by age 1 year

How is Zellweger syndrome diagnosed?
By the constellation of findings (along with elevated levels of VLCFA in the blood)

How is Zellweger syndrome managed?
Supportively
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How is Zellweger syndrome diagnosed?
By the constellation of findings (along with elevated levels of VLCFA in the blood)

How is Zellweger managed?
Supportively

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the **neonatal** period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures
-- Abnormal facies (high forehead; hypertelorism)

What is the prognosis?
It is uniformly fatal by age **1 year**
DDx for an RP-like Fundus

- **Primary RP** (aka typical RP)
- **Secondary RP** (aka Complex RP; Syndromic RP)
  - Usher syndrome
  - **Peroxisomal disorders**
    - Zellweger syndrome
    - Neonatal adrenoleukodystrophy (NALD)
- **‘Pseudo-RP’**

*What is the noneponymous name for Zellweger syndrome?*
Cerebrohepatorenal syndrome

*What is its inheritance pattern?*
AR

*How do Zellweger syndrome pts present?*
In the neonatal period with:
  - RP-like fundus
  - Deafness
  - Hypotonia
  - Seizures
  - Abnormal facies (high forehead; hypertelorism)

*What is the prognosis?*
It is uniformly fatal by age 1 year

*Is NALD the same condition as adrenoleukodystrophy?*
No, that is an X-linked condition that presents later in childhood

*What is its inheritance pattern?*
AR

*How do NALD pts present?*
In the late infancy period with:
  - RP-like fundus
  - Deafness
  - Hypotonia
  - Seizures

*What is the prognosis?*
It is uniformly fatal by late childhood
What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that presents later in childhood

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures
- Abnormal facies (high forehead; hypertelorism)

What is the prognosis?
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DDx for an RP-like Fundus
**DDx for an RP-like Fundus**

- **Primary RP**
  - (aka typical RP)
- **Secondary RP**
  - (aka Complex RP; Syndromic RP)
    - Usher syndrome
    - **Peroxisomal disorders**
      - Zellweger syndrome
    - Neuronal ceroid lipofuscinoses
    - Ciliopathies
    - Abetalipoproteinemia
- **‘Pseudo-RP’**

**What is the non-epithelial form of Zellweger syndrome?**
- Cerebrohepatorenal syndrome

**What is its inheritance pattern?**
- AR

**How do Zellweger syndrome patients present?**
- In the neonatal period with:
  - RP-like fundus
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(aka typical RP)

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  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
- Infantile Refsum dz

‘Pseudo-RP’

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  -- Neonatal adrenoleukodystrophy
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‘Pseudo-RP’

What is the non-penetrant Cerebrohepatorenal syndrome?

Is NALD the same condition as adrenoleukodystrophy?
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What is its inheritance pattern?
AR

Note: Both are inherited AR

What is the neonatal presentation?
- RP-like fundus
- Deafness
- Hypotonia
- Seizures
- Abnormal facies

What is the prognosis?
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Primary RP
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What is the prognosis?
It is uniformly fatal by age **late childhood**
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
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- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
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  -- Infantile Refsum dz

‘Pseudo-RP’

- Abetalipoproteinemia

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
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How do Zellweger syndrome pts present?
In the neonatal period with:
-- RP-like fundus
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What is the prognosis?
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What is its inheritance pattern?
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In the late infancy period
-- Seizures
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- **Peroxisomal disorders**
  - Zellweger syndrome
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  - Infantile Refsum dz

‘Pseudo-RP’

**What is the non-pseudonym for Zellweger syndrome?**
**Cerebrohepatorenal syndrome**

**What is its inheritance pattern?**
AR

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In the **neonatal** period with:
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No, that is an X-linked condition that presents later in childhood

**What is its inheritance pattern?**
AR

**How do NALD pts present?**
In the **late infancy** period with:
- Seizures

**Note:** NALD’s onset occurs a little later than does Zellweger’s
DDx for an RP-like Fundus

Primary RP

(aka typical RP)

Secondary RP

(aka Complex RP; Syndromic RP)

- Usher syndrome
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  -- Zellweger syndrome
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No, that is an X-linked condition that presents later in childhood

What is its inheritance pattern?
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How do NALD pts present?
In the late infancy period with:
--?
--?
--?
--?

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‘Pseudo-RP’

---

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Pseudo-RP

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In the neonatal period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures
-- Abnormal facies (high forehead; hypertelorism)

What is the prognosis?
It is uniformly fatal by age 1 year

Note that NALD has the same S/S as Zellweger, except it’s missing the last one on the list

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that presents later in childhood

What is its inheritance pattern?
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-- RP-like fundus
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What is the prognosis?
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- RP-like fundus
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What is the prognosis?
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And albeit dreadful, the prognosis for NALD is better than that for Zellweger
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So NALD and Zellweger are quite similar, except that NALD is a somewhat milder condition:
-- Later onset
-- One fewer S/S (= no abnormal facies)
-- Pts live a little longer
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Why does neonatal ADRENOleukodystrophy have adreno- in the name?
In NALD, the VLCFAs impair the function of adrenal glands by compromising the integrity of adrenal-cell membranes

How is NALD diagnosed?
Via its clinical presentation, coupled with MRI of the brain revealing gross white-matter abnormalities, and the elevated serum VLCFAs

How is NALD managed?
Supportively (just like Zellweger pts)

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‘Pseudo-RP’

Why does neonatal ADRENOleukodystrophy have adreno- in the name? In NALD, the VLCFAs impair the function of adrenal glands by compromising the integrity of adrenal-cell membranes.

How is NALD diagnosed? Via its clinical presentation, coupled with MRI of the brain revealing gross white-matter abnormalities, and the elevated serum VLCFAs.

How do Zellweger syndrome pts present? In the neonatal period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures
- Abnormal facies (high forehead; hypertelorism)

What is the prognosis? It is uniformly fatal by age 1 year.

How do Neonatal adrenoleukodystrophy pts present? In the neonatal period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures
- Abnormal facies (high forehead; hypertelorism)

What is its inheritance pattern? AR

How do Neonatal adrenoleukodystrophy pts present? In the late infancy period with:
- Hypotonia
- Seizures

What is its inheritance pattern? AR

How is NALD managed? Supportively (just like Zellweger pts)

What is the prognosis? It is uniformly fatal by late childhood
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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- Usher syndrome
- Peroxisomal disorders
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‘Pseudo-RP’

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Cerebrohepatorenal syndrome

What is its inheritance pattern?
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-- Hypotonia
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‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
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AR

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In the **neonatal** period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures
- Abnormal facies

What is the prognosis?
It is uniformly fatal by age **1 year**

Before we get into it: Is **infantile** Refsum dz the same as **adult-onset** Refsum dz?

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that presents later in childhood

What is its inheritance pattern?
AR

How do NALD pts present?
In the **late infancy** period with:
- RP-like fundus
- Deafness
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- Seizures

What is the prognosis?
It is uniformly fatal by **late childhood**
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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- Usher syndrome
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‘Pseudo-RP’

Is NALD the same condition as adrenoleukodystrophy?
No

Before we get into it: Is infantile Refsum dz the same as adult-onset Refsum dz?
No

What is its inheritance pattern?
AR

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Primary RP
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- Usher syndrome
- **Peroxisomal disorders**
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz *aka...*

‘Pseudo-RP’

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What is its inheritance pattern?
AR

How do NALD pts present?
In the late infancy period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures

What is the prognosis?
It is uniformly fatal by age **late childhood**

By what non-eponymous name is infantile Refsum disease known?
Infantile phytanic acid storage disease

What is its inheritance pattern?
AR

How do infantile Refsum dz pts present?
In the early childhood period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures

What is the prognosis?
It is uniformly fatal by age **1 year**

Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Infantile Refsum dz aka...Infantile phytanic acid storage dz

‘Pseudo-RP’

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that presents later in childhood

What is its inheritance pattern?
AR

How do NALD pts present?
In the late infancy period with:
--RP-like fundus
--Deafness
--Hypotonia
--Seizures

What is the prognosis?
It is uniformly fatal by late childhood

By what noneponymous name is infantile Refsum disease known?
Infantile phytanic acid storage disease

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
--RP-like fundus
--Deafness
--Hypotonia
--Abnormal facies

What is the prognosis?
It is uniformly fatal by age 1 year

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
--RP-like fundus
--Deafness
--Hypotonia
--Abnormal facies

What is the prognosis?
It is uniformly fatal by age 1 year
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz aka... Infantile phytanic acid storage dz

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures
-- Abnormal facies (high forehead; hypertelorism)

What is the prognosis?
It is uniformly fatal by age 1 year

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No, that is an X-linked condition that presents later in childhood

What is its inheritance pattern?
AR

How do NALD pts present?
In the late infancy period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures

What is the prognosis?
It is uniformly fatal by late childhood

By what noneponymous name is infantile Refsum disease known?
Infantile phytanic acid storage disease

What is its inheritance pattern?
AR

How do infantile Refsum disease pts present?
In the early childhood period with:
-- RP-like fundus
-- Deafness
-- Hypotonia

What is the prognosis?
It is uniformly fatal by early adulthood (if treatment is unsuccessful)
DDx for an RP-like Fundus

Primary RP (aka typical RP)
- Usher syndrome
- Peroxisomal disorders
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Infantile Refsum dz (aka...Infantile phytanic acid storage dz)

Secondary RP (aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Infantile Refsum dz (aka...Infantile phytanic acid storage dz)

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
- In the neonatal period with:
  - RP-like fundus
  - Deafness
  - Hypotonia
  - Seizures
  - Abnormal facies (high forehead; hypertelorism)

What is the prognosis?
It is uniformly fatal by age 1 year

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What is its inheritance pattern?
AR

How do NALD pts present?
- In the late infancy period with:
  - RP-like fundus
  - Deafness
  - Hypotonia
  - Seizures

What is the prognosis?
It is uniformly fatal by late childhood

By what noneponymous name is infantile Refsum disease known?
Infantile phytanic acid storage disease

What is its inheritance pattern?
AR

How do infantile Refsum disease pts present?
- In the early childhood period with:
  - RP-like fundus
  - Deafness
  - Hypotonia

What is the prognosis?
It is uniformly fatal by early adulthood (if treatment is unsuccessful)

What is the noneponymous name for infantile phytanic acid storage disease?
Infantile Refsum disease

What is its inheritance pattern?
AR

How do infantile phytanic acid storage disease pts present?
- In the late infancy period with:
  - RP-like fundus
  - Deafness
  - Hypotonia
  - Seizures

What is the prognosis?
It is uniformly fatal by late childhood
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- **Peroxisomal disorders**
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz  *aka...Infantile phytanic acid storage dz*

‘Pseudo-RP’

---

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures
- Abnormal facies (high forehead; hypertelorism)

What is the prognosis?
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that presents later in childhood

What is its inheritance pattern?
AR

How do NALD pts present?
In the late infancy period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures

What is the prognosis?
It is uniformly fatal by **late childhood**

By what noneponymous name is infantile Refsum disease known?
Infantile phytanic acid storage disease

What is its inheritance pattern?
AR

How do infantile Refsum dz pts present?
In the early childhood period with:
- RP-like fundus
- Deafness
- Hypotonia

What is the prognosis?
It is uniformly fatal by **early adulthood** (if treatment is unsuccessful)

Like Zellweger’s and NALD, infantile Refsum’s is inherited AR
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz aka... Infantile phytanic acid storage dz

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures
-- Abnormal facies (high forehead; hypertelorism)

What is the prognosis?
It is uniformly fatal by age 1 year

Is NALD the same condition as Zellweger syndrome?
No, that is an X-linked condition that presents later in childhood

What is its inheritance pattern?
AR

How do NALD pts present?
In the late infancy period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures

What is the noneponymous name for NALD?
Infantile phytanic acid storage disease

What is its inheritance pattern?
AR

How do infantile Refsum dz pts present?
In the early childhood period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures

What is the prognosis?
It is uniformly fatal by age 1 year

By what noneponymous name is infantile Refsum disease known?
Infantile phytanic acid storage disease

How do infantile Refsum dz pts present?
In the late infancy period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures

What is the prognosis?
It is uniformly fatal by age late childhood

If treatment is unsuccessful
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Infantile Refsum dz aka...Infantile phytanic acid storage dz

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures
- Abnormal facies (high forehead; hypertelorism)

What is the prognosis?
It is uniformly fatal by age 1 year

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that presents later in childhood

What is its inheritance pattern?
AR

How do NALD pts present?
In the late infancy period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures

What is the prognosis?
It is uniformly fatal by late childhood

By what noneponymous name is infantile Refsum disease known?
Infantile phytanic acid storage disease

What is its inheritance pattern?
AR

How do infantile Refsum dz pts present?
In the early childhood period with:
- RP-like fundus
- Deafness
- Hypotonia

What is the prognosis?
It is uniformly fatal by age 1 year (if treatment is unsuccessful)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Infantile Refsum dz aka...Infantile phytanic acid storage dz

‘Pseudo-RP’

---

Is NALD the same condition as Zellweger syndrome?
No, that is an X-linked condition that presents later in childhood.

What is its inheritance pattern?
AR

How do NALD pts present?
In the late infancy period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures

What is the prognosis?
It is uniformly fatal by age 1 year.

By what noneponymous name is infantile Refsum disease known?
Infantile phytanic acid storage disease

What is its inheritance pattern?
AR

How do infantile Refsum dz pts present?
In the early childhood period
- RP-like fundus
- Deafness
- Hypotonia
- Seizures

What is the prognosis?
It is uniformly fatal by late childhood
DDx for an RP-like Fundus

Secondary RP

(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Ciliopathies
- Abetalipoproteinemia

'Pseudo-RP'

Primary RP

(aka typical RP)

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures
- Abnormal facies (high forehead; hypertelorism)

What is the prognosis?
It is uniformly fatal by age 1 year

Is NALD the same condition as Zellweger syndrome?
No, that is an X-linked condition that presents later in childhood

What is its inheritance pattern?
AR

How do NALD pts present?
In the late infancy period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures

What is the prognosis?
It is uniformly fatal by late childhood

By what noneponymous name is infantile Refsum disease known?
Infantile phytanic acid storage disease

What is its inheritance pattern?
AR

How do infantile Refsum dz pts present?
In the early childhood period with:
- RP-like fundus
- Deafness
- Hypotonia

What is the prognosis?
It is uniformly fatal by early adulthood (if treatment is unsuccessful)

Just as NALD’s onset is a little later than Zellweger’s, infantile Refum’s is a little later than that of NALD
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Infantile Refsum dz aka... Infantile phytanic acid storage dz
- "Pseudo-RP"

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures
- Abnormal facies (high forehead; hypertelorism)

What is the prognosis?
It is uniformly fatal by age 1 year

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that presents later in childhood

What is its inheritance pattern?
AR

How do NALD pts present?
In the late infancy period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures

What is the prognosis?
It is uniformly fatal by late childhood

By what noneponymous name is infantile Refsum disease known?
Infantile phytanic acid storage disease

What is its inheritance pattern?
AR

How do infantile Refsum dz pts present?
In the early childhood period with:
- ?
- ?
- ?

What is the prognosis?
It is uniformly fatal by age 1 year
DDx for an RP-like Fundus

Primary RP (aka typical RP)

Secondary RP (aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Infantile Refsum dz aka... Infantile phytanic acid storage dz

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
  - RP-like fundus
  - Deafness
  - Hypotonia
  - Seizures
  - Abnormal facies (high forehead; hypertelorism)

What is the prognosis?
It is uniformly fatal by age 1 year

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that presents later in childhood

What is its inheritance pattern?
AR

How do NALD pts present?
In the late infancy period with:
  - RP-like fundus
  - Deafness
  - Hypotonia
  - Seizures

What is the prognosis?
It is uniformly fatal by late childhood

By what noneponymous name is infantile Refsum disease known?
Infantile phytanic acid storage disease

What is its inheritance pattern?
AR

How do infantile Refsum disease pts present?
In the early childhood period with:
  - RP-like fundus
  - Deafness
  - Hypotonia

What is the prognosis?
It is uniformly fatal by early adulthood (if treatment is unsuccessful)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- **Peroxisomal disorders**
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Infantile Refsum dz (aka Infantine phytanic acid storage dz)

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures
- Abnormal facies (high forehead; hypertelorism)

What is the prognosis?
It is uniformly fatal by age 1 year

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that presents later in childhood

What is its inheritance pattern?
AR

How do NALD pts present?
In the late infancy period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures

What is the prognosis?
It is uniformly fatal by late childhood

And just as NALD has the same S/S as Zellweger save one, so too with infantile Refsum--it has the same as NALD, except, again, for the last one on the list (note that this means it has the same S/S as Zellweger, save **two**)

By what noneponymous name is infantile Refsum disease known?
Infantine phytanic acid storage dz

In the neonatal period:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures
- Abnormal facies

What is the prognosis?
It is uniformly fatal by age 1 year

What is the noneponymous name for Infantile Refsum disease?
Infantine phytanic acid storage dz

What is its inheritance pattern?
AR

How do Infantile Refsum dz pts present?
In the early childhood period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures

What is the prognosis?
It is uniformly fatal by early adulthood (if treatment is unsuccessful)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

‘Pseudo-RP’

Usher syndrome

Peroxisomal disorders
- Zellweger syndrome
- Neonatal adrenoleukodystrophy
- Infantile Refsum dz aka...Infantile phytanic acid storage dz

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures
- Abnormal facies

What is the prognosis?
It is uniformly fatal by age 1 year

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that presents later in childhood

What is its inheritance pattern?
AR

How do NALD pts present?
In the late infancy period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures

What is the prognosis?
It is uniformly fatal by late childhood

By what noneponymous name is infantile Refsum disease known?
Infantile phytanic acid storage disease

What is its inheritance pattern?
AR

How do infantile Refsum dz pts present?
In the early childhood period with:
- RP-like fundus
- Deafness
- Hypotonia

What is the prognosis?
It is uniformly fatal by early adulthood (if treatment is unsuccessful)

Note that all three peroxisomal disorders involve deafness. Thus, if faced with a deaf-blind pt in clinic or on the OKAP/Boards, don’t automatically assume it’s Usher syndrome!
**DDx for an RP-like Fundus**

**Primary RP**
*(aka typical RP)*

**Secondary RP**
*(aka Complex RP; Syndromic RP)*
- Usher syndrome
- **Peroxisomal disorders**
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz *(aka...Infantile phytanic acid storage dz)*

**‘Pseudo-RP’**

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**What is the noneponymous name for Zellweger syndrome?**
Cerebrohepatorenal syndrome

**What is its inheritance pattern?**
AR

**How do Zellweger syndrome pts present?**
In the *neonatal* period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures
-- Abnormal facies (high forehead; hypertelorism)

**What is the prognosis?**
It is uniformly fatal by age 1 year

---

**Is NALD the same condition as adrenoleukodystrophy?**
No, that is an X-linked condition that presents later in childhood

**What is its inheritance pattern?**
AR

**How do NALD pts present?**
In the *late infancy* period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures

**What is the prognosis?**
It is uniformly fatal by late childhood

---

**By what noneponymous name is infantile Refsum disease known?**
Infantile phytanic acid storage disease

**What is its inheritance pattern?**
AR

**How do infantile Refsum dz pts present?**
In the *early childhood* period with:
-- RP-like fundus
-- Deafness
-- Hypotonia

**What is the prognosis?**
It is uniformly fatal by early adulthood (if treatment is unsuccessful)
DDx for an RP-like Fundus

Primary RP

(aka typical RP)

Secondary RP

(aka Complex RP; Syndromic RP)
- Usher syndrome
- **Peroxisomal disorders**
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Infantile Refsum dz aka...Infantile phytanic acid storage dz

‘Pseudo-RP’

- Abetalipoproteinemia

**What is the noneponymous name for Zellweger syndrome?**
Cerebrohepatorenal syndrome

**What is its inheritance pattern?**
AR

**How do Zellweger syndrome pts present?**
In the neonatal period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures
-- Abnormal facies

**What is the prognosis?**
It is uniformly fatal by age 1 year

**Is NALD the same condition as adrenoleukodystrophy?**
No, that is an X-linked condition that presents later in childhood

**How do NALD pts present?**
In the late infancy period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures

**What is the prognosis?**
It is uniformly fatal by late childhood

---

**What is the noneponymous name for infantile Refsum disease**
Infantile phytanic acid storage disease

**What is its inheritance pattern?**
AR

**How do infantile Refsum dz pts present?**
In the early childhood period with:
-- RP-like fundus
-- Deafness
-- Hypotonia

**What is the prognosis?**
It is uniformly fatal by early adulthood (if treatment is unsuccessful)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- **Peroxisomal disorders**
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz aka...Infantile phytanic acid storage dz

‘Pseudo-RP’

Is NALD the same condition as Zellweger syndrome?
No, that is an X-linked condition that presents later in childhood.

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Abnormal facies (high forehead; hypertelorism)

What is the prognosis?
It is uniformly fatal by age 1 year.

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the late infancy period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures

What is the prognosis?
It is uniformly fatal by late childhood.

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that presents later in childhood.

What is its inheritance pattern?
AR

How do NALD pts present?
In the late infancy period with:
-- RP-like fundus
-- Deafness
-- Hypotonia

What is the prognosis?
It is uniformly fatal by early adulthood (if treatment is unsuccessful).

By what noneponymous name is infantile Refsum disease known?
Infantile phytanic acid storage disease

What is its inheritance pattern?
AR

How do Infantile Refsum dz pts present?
In the early childhood period with:
-- RP-like fundus
-- Deafness
-- Hypotonia

What is the prognosis?
It is uniformly fatal by early adulthood (if treatment is unsuccessful).

Is infantile Refsum disease the same as Zellweger syndrome?
No, that is an X-linked condition that presents later in childhood.

What is its inheritance pattern?
AR

How do infantile Refsum dz pts present?
In the early childhood period with:
-- RP-like fundus
-- Deafness
-- Hypotonia

What is the prognosis?
It is uniformly fatal by early adulthood (if treatment is unsuccessful).

It is uniformly fatal by late childhood.

What is the noneponymous name for infantile Refsum disease?
Infantile phytanic acid storage disease
Pseudo-RP

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz (aka...Infantile phytanic acid storage dz)

Primary RP
(aka typical RP)
- Zellweger syndrome
- Neonatal adrenoleukodystrophy
- Infantile Refsum dz (aka...Infantile phytanic acid storage dz)

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures
- Abnormal facies (high forehead; hypertelorism)

What is the prognosis?
It is uniformly fatal by age 1 year

And just as the prognosis for NALD was better than for Zellweger, so too in turn is the prognosis for infantile Refsum better than that for NALD
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- **Peroxisomal disorders**
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Infantile Refsum dz aka...Infantile phytanic acid storage dz

‘Pseudo-RP’

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that presents later in childhood

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures
- Abnormal facies

What is the prognosis?
It is uniformly fatal by age 1 year

By what noneponymous name is infantile Refsum disease known?
Infantile phytanic acid storage disease

What is its inheritance pattern?
AR

How do infantile Refsum dz pts present?
In the early childhood period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures

What is the prognosis?
It is uniformly fatal by early adulthood (if treatment is unsuccessful)

So infantile Refsum and NALD are quite similar, except that infantile Refsum is a somewhat milder condition:
- Later onset
- One fewer S/S (= no seizures)
- Pts live longer

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do NALD pts present?
In the late infancy period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures

What is the prognosis?
It is uniformly fatal by late childhood
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Infantile Refsum dz aka...Infantile phytanic acid storage dz

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures
- Abnormal facies (high forehead; hypertelorism)

What is the prognosis?
It is uniformly fatal by age 1 year

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that presents later in childhood

What is its inheritance pattern?
AR

How do NALD pts present?
In the late infancy period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures

What is the prognosis?
It is uniformly fatal by late childhood

How is infantile Refsum diagnosed?
Via elevated serum phytanic acid levels (and VLCFAs)

By what noneponymous name is infantile Refsum disease known?
Infantile phytanic acid storage dz

What is its inheritance pattern?
AR

How do infantile Refsum dz pts present?
In the early childhood period with:
- RP-like fundus
- Deafness
- Hypotonia

What is the prognosis?
It is uniformly fatal by early adulthood (if treatment is unsuccessful)
late childhood
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Ciliopathies
- Abetalipoproteinemia

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger pts present?
In the neonatal period with:
  - RP-like fundus
  - Deafness
  - Hypotonia
  - Seizures
  - Abnormal facies (high forehead; hypertelorism)

What is the prognosis?
It is uniformly fatal by age 1 year

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that presents later in childhood

What is its inheritance pattern?
AR

How do NALD pts present?
In the late infancy period with:
  - RP-like fundus
  - Deafness
  - Hypotonia

What is the prognosis?
It is uniformly fatal by late childhood

How is infantile Refsum disease known?
Infantile phytanic acid storage disease

What is its inheritance pattern?
AR

How do infantile Refsum dz pts present?
In the early childhood period with:
  - RP-like fundus
  - Deafness
  - Hypotonia

What is the prognosis?
It is uniformly fatal by early adulthood (if treatment is unsuccessful)

How is infantile Refsum diagnosed?
Via elevated serum phytanic acid levels (and VLCFAs)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- **Peroxisomal disorders**
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz **aka**...Infantile phytanic acid storage dz

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome
What is its inheritance pattern?
AR
How do Zellweger syndrome pts present?
In the neonatal period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures
-- Abnormal facies (high forehead; hypertelorism)
What is the prognosis?
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that presents later in childhood
What is its inheritance pattern?
AR
How do NALD pts present?
In the late infancy period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures
What is the prognosis?
It is uniformly fatal by late childhood

By what noneponymous name is infantile Refsum disease known?
Infantile phytanic acid storage disease
What is its inheritance pattern?
AR
How do infantile Refsum dz pts present?
In the early childhood period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
What is the prognosis?
It is uniformly fatal by early adulthood (if treatment is unsuccessful)

How is infantile Refsum diagnosed?
Via elevated serum phytanic acid levels (and VLCFAs)

How do infantile Refsum dz pts present?
Hol up—you can **treat** this one??!! How is infantile Refsum treated?
Dietary restriction of phytanic acid and phytol (a phytanic acid precursor); plasmapheresis may be employed acutely
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz (aka...Infantile phytanic acid storage dz)

‘Pseudo-RP’

What is the non-epithelial name for Zellweger syndrome?
Cerebrohepatorenal syndrome
What is its inheritance pattern?
AR
How do Zellweger syndrome patients present?
In the neonatal period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures
-- Abnormal facies (high forehead; hypertelorism)
What is the prognosis?
It is uniformly fatal by age 1 year

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that presents later in childhood
What is its inheritance pattern?
AR
How do NALD patients present?
In the late infancy period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures
What is the prognosis?
It is uniformly fatal by late childhood

How do infantile Refsum disease patients present?
In the early childhood period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
What is the prognosis?
It is uniformly fatal by early adulthood (if treatment is unsuccessful)

How is infantile Refsum diagnosed?
Via elevated serum phytanic acid levels (and VLCFAs)
How is infantile Refsum treated?
Dietary restriction of phytanic acid and phytol (a phytanic acid precursor); plasmapheresis may be employed acutely

Hol up—you can treat this one??!!
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Infantile Refsum

Peroxisomal disorders tl;dr
(Review slides--no questions)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz

‘Pseudo-RP’

RP-like fundus
- Deafness
- Hypotonia
- Seizures
- Abnormal facies

Peroxisomal disorders tl;dr

(Review slides--no questions)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  --Zellweger syndrome
  --Neonatal adrenoleukodystrophy
  --Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
- Ciliopathies
- Abetalipoproteinemia

‘Pseudo-RP’

RP-like fundus
Deafness
Hypotonia
Seizures
Abnormal facies

Peroxisomal disorders tl;dr
(Review slides--no questions)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz

‘Pseudo-RP’

RP-like fundus
Deafness
Hypotonia
Seizures
Abnormal facies

Peroxisomal disorders tl;dr
(Review slides--no questions)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz

‘Pseudo-RP’

- Bardet-Biedl syndrome
- Alström syndrome
- Joubert syndrome
- Senior-Løken syndrome

RP-like fundus

Deafness

Hypotonia

Seizures

Abnormal facies

Peroxisomal disorders tl;dr

(Review slides--no questions)
DDx for an RP-like Fundus

Primary RP (aka typical RP)

Secondary RP (aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  --Zellweger syndrome
  --Neonatal adrenoleukodystrophy
  --Infantile Refsum dz

‘Pseudo-RP’

RP-like fundus
Deafness
Hypotonia
Seizures
Abnormal facies

Peroxisomal disorders tl;dr
(Review slides--no questions)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  --Zellweger syndrome
  --Neonatal adrenoleukodystrophy
  --Infantile Refsum dz

‘Pseudo-RP’

RP-like fundus
Deafness
Hypotonia
Seizures
Abnormal facies

Infantile Refsum
NALD
Zellweger syndrome

Best prognosis

Peroxisomal disorders tl;dr

(Review slides--no questions)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz

‘Pseudo-RP’

RP-like fundus
- Deafness
- Hypotonia
- Seizures
- Abnormal facies

Infantile Refsum
Zellweger syndrome
NALD
Worse prognosis

Peroxisomal disorders tl;dr

(Review slides--no questions)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz

‘Pseudo-RP’

Peroxisomal disorders tl;dr
(Review slides--no questions)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz

‘Pseudo-RP’

RP-like fundus
Deafness
Hypotonia
Seizures
Abnormal facies

Infantile Refsum
NALD
Zellweger syndrome

All three are inherited

Peroxisomal disorders tl;dr

(Review slides--no questions)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz

‘Pseudo-RP’

RP-like fundus
Deafness
Hypotonia
Seizures
Abnormal facies

Infantile Refsum
NALD
Zellweger syndrome

All three are inherited AR

Peroxisomal disorders tl;dr
(Review slides--no questions)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz

‘Pseudo-RP’

RP-like fundus
- Deafness
- Hypotonia
- Seizures
- Abnormal facies

Peroxisomal disorders tl;dr
(Review slides--no questions)

Only infantile Refsum is treatable
What are the neuronal ceroid lipofuscinoses (NCLs; collectively referred to as Batten dz)?

DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka ‘Batten dz’
- Ciliopathies
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’
What are the neuronal ceroid lipofuscinoses (NCLs; collectively referred to as Batten dz)?

Neurodegenerative conditions stemming from the buildup of two substances in cells

What is the inheritance pattern?

AR

How do Batten dz pts present?

In infancy to early childhood with:

-- LCA
-- Seizures
-- Myoclonus
-- Microcephaly (in the infantile forms)
-- Relentlessly progressive neurologic and cognitive decline

What is the prognosis?

Depending on the form, it is fatal by late childhood to early adulthood

Primary RP (aka typical RP)

Secondary RP (aka Complex RP; Syndromic RP)

- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka ‘Batten dz’
- Ciliopathies
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’
What are the neuronal ceroid lipofuscinoses (NCLs; collectively referred to as Batten dz)? Neurodegenerative conditions stemming from the buildup of ceroid & lipofuscin in cells.
**DDx for an RP-like Fundus**

- **Primary RP** *(aka typical RP)*
- **Secondary RP** *(aka Complex RP; Syndromic RP)*
  - Usher syndrome
  - Peroxisomal disorders
    - Zellweger syndrome
    - Neonatal adrenoleukodystrophy
    - Infantile Refsum dz
  - Neuronal ceroid lipofuscinoses *(NCLs; collectively referred to as Batten dz)*
    - Neurodegenerative conditions stemming from the buildup of ceroid & lipofuscin in cells
  - Ciliopathies
  - Abetalipoproteinemia *(aka ‘Bassen-Kornzweig dz’)*

**What are the neuronal ceroid lipofuscinoses (NCLs; collectively referred to as Batten dz)?**
Neurodegenerative conditions stemming from the buildup of ceroid & lipofuscin in cells.

**What is the inheritance pattern?**
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka ‘Batten dz’
- Ciliopathies
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

What are the neuronal ceroid lipofuscinoses (NCLs; collectively referred to as Batten dz)?
Neurodegenerative conditions stemming from the buildup of ceroid & lipofuscin in cells

What is the inheritance pattern?
AR

What is the prognosis?
Depending on the form, it is fatal by late childhood to early adulthood.
What are the neuronal ceroid lipofuscinoses (NCLs; collectively referred to as Batten dz)? Neurodegenerative conditions stemming from the buildup of ceroid & lipofuscin in cells.

What is the inheritance pattern?
AR

How do Batten dz pts present?
In infancy to early childhood with:
- LCA
- Seizures
- Myoclonus
- Microcephaly (in the infantile forms)
- Relentlessly progressive neurologic and cognitive decline

What is the prognosis?
Depending on the form, it is fatal by late childhood to early adulthood.
What are the neuronal ceroid lipofuscinoses (NCLs; collectively referred to as Batten dz)? Neurodegenerative conditions stemming from the buildup of ceroid & lipofuscin in cells.

What is the inheritance pattern? AR

How do Batten dz pts present? In infancy to early childhood with:

DDx for an RP-like Fundus

Primary RP (aka typical RP)

Secondary RP (aka Complex RP; Syndromic RP)

- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka ‘Batten dz’
- Ciliopathies
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’
What are the neuronal ceroid lipofuscinoses (NCLs; collectively referred to as Batten dz)?
Neurodegenerative conditions stemming from the buildup of ceroid & lipofuscin in cells

What is the inheritance pattern?
AR

How do Batten dz pts present?
In infancy to early childhood with:
--RP-like fundus
--Seizures
--Myoclonus
--Microcephaly (in the infantile forms)
--Relentlessly progressive neurologic and cognitive decline
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka ‘Batten dz’
- Ciliopathies
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

What are the neuronal ceroid lipofuscinoses (NCLs; collectively referred to as Batten dz)?
Neurodegenerative conditions stemming from the buildup of ceroid & lipofuscin in cells

What is the inheritance pattern?
AR

How do Batten dz pts present?
In infancy to early childhood with:
-- RP-like fundus
-- Seizures
-- Myoclonus
-- Microcephaly (in the infantile forms)
-- Relentlessly progressive neurologic and cognitive decline

What is the prognosis?
Depending on the form, it is fatal by late childhood to early adulthood.
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka ‘Batten dz’
- Ciliopathies
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

What are the neuronal ceroid lipofuscinoses (NCLs; collectively referred to as Batten dz)?
Neurodegenerative conditions stemming from the buildup of ceroid & lipofuscin in cells

What is the inheritance pattern?
AR

How do Batten dz pts present?
In infancy to early childhood with:
- RP-like fundus
- Seizures
- Myoclonus
- Microcephaly (in the infantile forms)
- Relentlessly progressive neurologic and cognitive decline

What is the prognosis?
Depending on the form, it is fatal by late childhood to early adulthood
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  --Zellweger syndrome
  --Neonatal adrenoleukodystrophy
  --Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  --aka ‘Batten dz’
- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’
  --Zellweger syndrome
  --Neonatal adrenoleukodystrophy
  --Infantile Refsum dz

What are the neuronal ceroid lipofuscinoses (NCLs; collectively referred to as Batten dz)?
Neurodegenerative conditions stemming from the buildup of ceroid & lipofuscin in cells

What is the inheritance pattern?
AR

How do Batten dz pts present?
In infancy to early childhood with:
- RP-like fundus
- Seizures
- Myoclonus
- Microcephaly (in the infantile forms)
- Relentlessly progressive neurologic and cognitive decline

How is Batten dz diagnosed?
It is a mélange of imaging, genetic and other tests. If asked, punt to a geneticist.

How is Batten dz managed?
Supportively

How is Batten dz diagnosed?
According to presentation with:
- RP-like fundus
- Seizures
- Myoclonus
- Microcephaly
- Relentlessly progressive neurologic and cognitive decline

What is the prognosis?
Depending on the form, it is fatal by late childhood to early adulthood.
What are the neuronal ceroid lipofuscinoses (NCLs; collectively referred to as Batten dz)?

Neurodegenerative conditions stemming from the buildup of ceroid & lipofuscin in cells

What is the inheritance pattern?

AR

How do Batten dz pts present?

In infancy to early childhood with:
- RP-like fundus
- Seizures
- Myoclonus
- Microcephaly (in the infantile forms)
- Relentlessly progressive neurologic and cognitive decline

How is Batten dz diagnosed?

It is a mélange of imaging, genetic and other tests. If asked, punt to a geneticist.

What is the prognosis?

Depending on the form, it is fatal by late childhood to early adulthood
What are the neuronal ceroid lipofuscinoses (NCLs; collectively referred to as Batten dz)?
Neurodegenerative conditions stemming from the buildup of ceroid & lipofuscin in cells

What is the inheritance pattern?
AR

How do Batten dz pts present?
In infancy to early childhood with:
--RP-like fundus
--Seizures
--Myoclonus
--Microcephaly (in the infantile forms)
--Relentlessly progressive neurologic and cognitive decline

How is Batten dz diagnosed?
It is a mélange of imaging, genetic and other tests. If asked, punt to a geneticist.

How is Batten dz managed?

What is the prognosis?
Depending on the form, it is fatal by late childhood to early adulthood.
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka ‘Batten dz’

What are the neuronal ceroid lipofuscinoses (NCLs; collectively referred to as Batten dz)?
Neurodegenerative conditions stemming from the buildup of ceroid & lipofuscin in cells

What is the inheritance pattern?
AR

How do Batten dz pts present?
In infancy to early childhood with:
- RP-like fundus
- Seizures
- Myoclonus
- Microcephaly (in the infantile forms)
- Relentlessly progressive neurologic and cognitive decline

What is the prognosis?
Depending on the form, it is fatal by late childhood to early adulthood

How is Batten dz diagnosed?
It is a mélange of imaging, genetic and other tests. If asked, punt to a geneticist.

How is Batten dz managed?
Supportively
- Abetalipoproteinemia (aka ‘Bassen-Kornzweig dz’; infantile forms)
- Relentlessly progressive neurologic and cognitive decline
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  --Zellweger syndrome
  --Neonatal adrenoleukodystrophy
  --Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  --aka ‘Batten dz’
- Ciliopathies

‘Pseudo-RP’

What is a ciliopathy?
**Pseudo-RP**

Primary RP (aka typical RP)

Secondary RP (aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  - aka ‘Batten dz’
- **Ciliopathies**

‘Pseudo-RP’

**What is a ciliopathy?**
An inherited condition marked by abnormal structure and/or function of cilia
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka 'Batten dz'
- Ciliopathies

What is a ciliopathy?
An inherited condition marked by abnormal structure and/or function of cilia

Cilia are ubiquitous. That said, ciliopathies primarily affect three organs--what are they?
What is a ciliopathy?
An inherited condition marked by abnormal structure and/or function of cilia

Cilia are ubiquitous. That said, ciliopathies primarily affect three organs—what are they?
The eyes, brain and kidneys
What is a ciliopathy?
An inherited condition marked by abnormal structure and/or function of cilia

Cilia are organs...
What is a ciliopathy?
An inherited condition marked by abnormal structure and/or function of cilia.

Cilia are organelles found in the eye, brain, kidney, and testes.

The eyes??!! Which part of the eye contains cilia wiggling about?
None. Remember, cilia come in two basic flavors: Motile, and nonmotile. It is the nonmotile type which is ubiquitous in the eye.
Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka ‘Batten dz’
- Ciliopathies

‘Pseudo-RP’

DDx for an RP-like Fundus

OK then, where in the eye are the nonmotile cilia, dysfunction of which leads to an RP-like fundus?

Cilia are organelles in the eye, brain, and other tissues. The eyes??!! Which part of the eye contains cilia wiggling about? None. Remember, cilia come in two basic flavors: Motile, and nonmotile. It is the nonmotile type which is ubiquitous in the eye.
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

- Usher syndrome
- Peroxisomal disorders
  --Zellweger syndrome
  --Neonatal adrenoleukodystrophy
  --Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  --aka 'Batten dz'
- Ciliopathies

‘Pseudo-RP’

---

OK then, where in the eye are the nonmotile cilia, dysfunction of which leads to an RP-like fundus? Recall that, fundamentally, RP is a disorder.

Cilia are organelles that move things. The eyes??!! Which part of the eye contains cilia wiggling about? None. Remember, cilia come in two basic flavors: Motile, and nonmotile. It is the nonmotile type which is ubiquitous in the eye.
Primary RP
*(aka typical RP)*

Secondary RP
*(aka Complex RP; Syndromic RP)*
- Usher syndrome
- Peroxisomal disorders
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  - *aka* ‘Batten dz’
- **Ciliopathies**

‘Pseudo-RP’

---

**What is a ciliopathy?**
An inherited condition marked by abnormal structure and/or function of cilia.

Cilia are organelles. That said, ciliopathies primarily affect three organs—what are they?
The eyes, brain, and kidneys.

The eyes??!! Which part of the eye contains cilia wiggling about?
None. Remember, cilia come in two basic flavors: Motile, and nonmotile.
It is the nonmotile type which is ubiquitous in the eye.

**OK then, where in the eye are the nonmotile cilia, dysfunction of which leads to an RP-like fundus?**
Recall that, fundamentally, RP is a photoreceptor disorder.

**DDx for an RP-like Fundus**
What is a ciliopathy?
An inherited condition marked by abnormal structure and/or function of cilia. Cilia are organelles.

What are ciliopathies?
The eyes, brain, kidneys

OK then, where in the eye are the nonmotile cilia, dysfunction of which leads to an RP-like fundus?
Recall that, fundamentally, RP is a photoreceptor disorder. Given this, it should come as no surprise that nonmotile cilia comprise a portion of the photoreceptors themselves (specifically, they contribute to the connection between the inner and outer segments).

Where in the eye are the nonmotile cilia, dysfunction of which leads to an RP-like fundus?
Recall that, fundamentally, RP is a photoreceptor disorder. Given this, it should come as no surprise that nonmotile cilia comprise a portion of the photoreceptors themselves (specifically, they contribute to the connection between the inner and outer segments).
Pseudo-RP
Primary RP
(aka typical RP)
Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  --Zellweger syndrome
  --Neonatal adrenoleukodystrophy
  --Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  --aka ‘Batten dz’
- Ciliopathies
  --?
  --?
  --?
  --?

Which ciliopathies present with an RP-like fundus?

What is a ciliopathy?
An inherited condition marked by abnormal structure and/or function of cilia

Cilia are ubiquitous. That said, ciliopathies primarily affect three organs--what are they?
The eyes, brain and kidneys
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
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  -- aka ‘Batten dz’

- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome

‘Pseudo-RP’

What is a ciliopathy?
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  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome

Note that all of the ciliopathies are marked by relentlessly progressive renal failure resulting in ESRD early in life!

What is a ciliopathy?
An inherited condition marked by abnormal structure and/or function of cilia

Cilia are ubiquitous. That said, ciliopathies primarily affect three organs--what are they?
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DDx for an RP-like Fundus

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Got a mnemonic for remembering the ciliopathies?

‘Pseudo-RP’
DDx for an RP-like Fundus

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(aka typical RP)

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  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
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Got a mnemonic for remembering the ciliopathies? JABS. Imagine a cilia as it jabs someone in the eye
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  --Joubert syndrome
  --Senior-Løken syndrome
- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’

What findings define the Bardet-Biedl complex?
--
--
The mnemonic is…

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‘Pseudo-RP’

What findings define the Bardet-Biedl complex?

- H
- O
- M
- E
- R

(as in Simpson)
DDx for an RP-like Fundus

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(aka typical RP)

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  --Senior-Løken syndrome
- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’

What findings define the Bardet-Biedl complex?
--H
--O
--M
--E
--RP-like fundus

Not surprisingly, the ‘R’ stands for RP-like fundus.
As for the rest…
DDx for an RP-like Fundus

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(aka typical RP)

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  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

What findings define the Bardet-Biedl complex?
-- Hypogonadism
-- Obesity
-- Mental retardation
-- Extra fingers (polydactyly)
-- RP-like fundus

Not surprisingly, the ‘R’ stands for RP-like fundus.
As for the rest…
Pseudo-RP

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‘Pseudo-RP’

What findings define the Bardet-Biedl complex?
- Hypogonadism
- Obesity
- Mental retardation
- Extra fingers (polydactyly)
- RP-like fundus

And yeah, I know, Homer only has four digits per hand—paucidactyly, not polydactyly. But the rest fits him pretty well.
DDx for an RP-like Fundus

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Secondary RP (aka Complex RP; Syndromic RP)
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  -- aka ‘Batten dz’
- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome

Alström syndrome includes an RP-like fundus (duh), but shares only one other finding with the B-B complex— which one?

What findings define the Bardet-Biedl complex?
- Hypogonadism?
- Obesity?
- Mental retardation?
- Extra fingers (polydactyly)?
- RP-like fundus

‘Pseudo-RP’
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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(aka Complex RP; Syndromic RP)
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  --Joubert syndrome

Alström syndrome includes an RP-like fundus (duh), but shares only one other finding with the B-B complex— which one?

What findings define the Bardet-Biedl complex?
--Hypogonadism
--Obesity
--Mental retardation
--Extra fingers (polydactyly)
--RP-like fundus

‘Pseudo-RP’
DDx for an RP-like Fundus

Primary RP
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  -- aka ‘Batten dz’
- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome

Alström syndrome includes an RP-like fundus (duh), but shares only one other finding with the B-B complex— which one?

It has two features not found in B-B: What are they? 

Both occur in childhood to teens

What findings define the Bardet-Biedl complex?
- Hypogonadism
- Obesity
- Mental retardation
- Extra fingers (polydactyly)
- RP-like fundus

--?
--?

What are they?
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
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  --Alström syndrome
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Alström syndrome includes an RP-like fundus (duh), but shares only one other finding with the B-B complex—which one?

It has two features not found in B-B: What are they?

What findings define the Bardet-Biedl complex?
- Hypogonadism
- Obesity
- Mental retardation
- Extra fingers (polydactyly)
- RP-like fundus
- Cardiomyopathy
- Early-onset type 2 DM

Both occur in childhood to teens
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
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‘Pseudo-RP’

- Ciliopathies
  --Bardet-Biedl syndrome
  --Alström syndrome
  --Joubert syndrome
  --Senior-Løken syndrome

- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’

How are BBS and AS managed?
DDx for an RP-like Fundus

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(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
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  -- Zellweger syndrome
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  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

How are BBS and AS managed?
Supportively

‘Pseudo-RP’
Primary RP
(aka typical RP)

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  - aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

What are the main nonocular structures affected in JS?

The brainstem and cerebellum

What classic MRI finding is the hallmark of JS?
‘Molar tooth sign’

How do JS pts present?
In the late infancy period with:
- LCA
- Hypotonia
- Abnormal breathing (hyperpnea or apnea)
- Intellectual and motor deficits
- Seizures
- Abnormal facies

What is the prognosis?
It is highly variable
What are the main nonocular structures affected in JS?
The brainstem and cerebellum
DDx for an RP-like Fundus

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What are the main nonocular structures affected in JS?
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‘Pseudo-RP’

What are the main nonocular structures affected in JS?
The brainstem and cerebellum

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DDx for an RP-like Fundus

Joubert syndrome: Molar-tooth sign (look at the brainstem)
What are the main nonocular structures affected in JS?
The brainstem and cerebellum

What classic MRI finding is the hallmark of JS?
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‘Pseudo-RP’

What are the main nonocular structures affected in JS?
The brainstem and cerebellum

What classic MRI finding is the hallmark of JS?
‘Molar tooth sign’

How do JS pts present?
In the late infancy period with:
- RP-like fundus
- Hypotonia
- Disordered breathing (hyerpnea or apnea)
- Intellectual and motor deficits
- Seizures
- Abnormal facies
DDx for an 
RP-like Fundus

Joubert syndrome: Facies. Note the large head, broad forehead
DDx for an RP-like Fundus

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(aka typical RP)

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  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
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What are the main nonocular structures affected in JS?
The brainstem and cerebellum

What classic MRI finding is the hallmark of JS?
‘Molar tooth sign’

How do JS pts present?
In the late infancy period with:
-- RP-like fundus
-- Hypotonia
-- Disordered breathing (hyperpnea or apnea)
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What are the main nonocular structures affected in JS?
The brainstem and cerebellum

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--RP-like fundus
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What is the prognosis?
It is highly variable
What are the main nonocular structures affected in JS? The brainstem and cerebellum

What classic MRI finding is the hallmark of JS? ‘Molar tooth sign’

How do JS pts present? --RP-like fundus
--Hypotonia
--Disordered breathing (hyperpnea or apnea)
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--Seizures
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What is the prognosis? It is highly variable
DDx for an RP-like Fundus

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What are the main nonocular structures affected in JS? The brainstem and cerebellum

What classic MRI finding is the hallmark of JS? ‘Molar tooth sign’

How do JS pts present? Supportively
- RP-like fundus
- Hypotonia
- Disordered breathing (hyperpnea or apnea)
- Intellectual and motor deficits
- Seizures
- Abnormal facies

What is the prognosis? It is highly variable

How is JS managed? Supportively
DDx for an RP-like Fundus

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Secondary RP (aka Complex RP; Syndromic RP)
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- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome

Senior-Løken syndrome is not listed with the other ciliopathies in the Retina book; rather, it is discussed in the Peds book, and only briefly. All you need to know about it is that, like all the ciliopathies mentioned, it involves retinal degeneration (with an LCA or RP-like fundus appearance) and relentlessly progressive renal failure.
DDx for an RP-like Fundus

Primary RP (aka typical RP)

Secondary RP (aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy

‘Pseudo-RP’

For completeness’ sake:
In three words (including syndromes), what sort of condition are these ciliopathies?
They are two words syndromes

- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome

- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’
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- Usher syndrome
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‘Pseudo-RP’

For completeness’ sake:
In three words (including syndromes), what sort of condition are these ciliopathies?
They are familial oculorenal syndromes

- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome

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‘Pseudo-RP’

For completeness’ sake:
In three words (including syndromes), what are these ciliopathies? They are familial oculorenal syndromes

Ciliopathies
- Bardet-Biedl syndrome
- Alström syndrome
- Joubert syndrome
- Senior-Løken syndrome

Abetalipoproteinemia
--aka ‘Bassen-Kornzweig dz’

How many familial oculorenal syndromes are there?

Including the four ciliopathies—six

Other two:
Alport syndrome and Lowe syndrome
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
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  -- Neonatal adrenoleukodystrophy

‘Pseudo-RP’

For completeness’ sake:
In three words (including syndromes), what are these ciliopathies?
They are familial oculorenal syndromes

How many familial oculorenal syndromes are there?
Including the four ciliopathies--six

- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome

- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’
For completeness’ sake:
*In three words (including syndromes), what are these ciliopathies?*
They are **familial oculorenal syndromes**

- **Ciliopathies**
  - Bardet-Biedl syndrome
  - Alström syndrome
  - Joubert syndrome
  - Senior-Løken syndrome

- Abetalipoproteinemia
  - *aka* ‘Bassen-Kornzweig dz’

**How many familial oculorenal syndromes are there?**
Including the four ciliopathies--six

**What are the other two?**
DDx for an RP-like Fundus

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‘Pseudo-RP’

For completeness’ sake:
In three words (including syndromes), what are these ciliopathies?
They are familial oculorenal syndromes

How many familial oculorenal syndromes are there?
Including the four ciliopathies--six

What are the other two?
Alport syndrome and Lowe syndrome

- Ciliopathies
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‘Pseudo-RP’

For completeness’ sake:
In three words (including syndromes), what are these ciliopathies?
They are familial oculorenal syndromes

How many familial oculorenal syndromes are there?
Including the four ciliopathies--six

What are the other two?
Alport syndrome and Lowe syndrome

Are Alport and Lowe syndromes ciliopathies?
DDx for an RP-like Fundus

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‘Pseudo-RP’

For completeness’ sake:
In three words (including syndromes), what are these ciliopathies?
They are familial oculorenal syndromes

How many familial oculorenal syndromes are there?
Including the four ciliopathies—six

What are the other two?
Alport syndrome and Lowe syndrome

Are Alport and Lowe syndromes ciliopathies? No
DDx for an RP-like Fundus

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- Ciliopathies
- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

For completeness’ sake:
In three words (including syndromes), what sort of condition are these ciliopathies?
They are familial oculorenal syndromes

What are the other two?
Alport syndrome and Lowe syndrome

Are Alport and Lowe syndrome in the DDx for an RP-like fundus?

Lowe syndrome does not manifest retinal changes, so it is not. Alport syndrome is associated with what the Peds book describes as a “fleck retinopathy” and the Retina book as a “pigmentary retinopathy;” for this reason the Retina book considers it to be in the DDx.
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
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  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
- Neuronal ceroid lipofuscinoses
- Ciliopathies
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

For completeness’ sake:
In three words (including syndromes), what are these ciliopathies?
They are familial oculo-renal syndromes

Are Alport and Lowe syndrome in the DDx for an RP-like fundus?
Lowe syndrome does not manifest retinal changes, so it is not. Alport syndrome is associated with what the Peds book describes as a “fleck retinopathy” and the Retina book as a “pigmentary retinopathy;” for this reason the Retina book considers it to be in the DDx.

What are the other two?
Alport syndrome and Lowe syndrome
### DDx for an RP-like Fundus

#### Primary RP
*(aka typical RP)*

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*(aka Complex RP; Syndromic RP)*
- Usher syndrome
- Peroxisomal disorders
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- Ciliopathies
  - Bardet-Biedl syndrome
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  - Senior-Løken syndrome
- Abetalipoproteinemia
  *aka ‘Bassen-Kornzweig dz’*

**‘Pseudo-RP’**

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**Are Alport and Lowe syndrome in the DDx for an RP-like fundus?**

Lowe syndrome does not manifest retinal changes, so it is not. Alport syndrome is associated with what the *Peds* book describes as a “fleck retinopathy” and the *Retina* book as a “pigmentary retinopathy;” for this reason the *Retina* book considers it to be in the DDx.

**What are the other two?**

*Alport syndrome* and *Lowe syndrome*

---

**In three words (including syndromes), what are these ciliopathies?**

**They are familial oculoarenal syndromes**
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders

‘Pseudo-RP’

For completeness’ sake: In three words (including syndromes), what sort of condition are these ciliopathies? They are familial oculorenal syndromes.

Are Alport and Lowe syndrome in the DDx for an RP-like fundus? Lowe syndrome does not manifest retinal changes, so it is not. Alport syndrome is associated with what the Peds book describes as a “fleck retinopathy” and the Retina book as a “pigmentary retinopathy;” for this reason the Retina book considers it to be in the DDx.

What are the other two? Alport syndrome and Lowe syndrome.

(Note also that Alport syndrome is associated with hearing loss, so it is in the DDx for a pt with suspected Usher syndrome.)

Ciliopathies
--Bardet-Biedl syndrome
--Alström syndrome
--Joubert syndrome
--Senior-Løken syndrome

Abetalipoproteinemia
--aka ‘Bassen-Kornzweig dz’
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
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Alport syndrome and Lowe syndrome

Abnormalities of what intraocular structure are associated with Alport and Lowe syndromes?
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What are the other two?
*Alport syndrome and Lowe syndrome*

Abnormalities of what intraocular structure are associated with Alport and Lowe syndromes?
The lens

For completeness’ sake:
*In three words (including syndromes), what are these ciliopathies?* They are **familial oculorenal syndromes**
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
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  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
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Alport syndrome and Lowe syndrome

Abnormalities of what intraocular structure are associated with Alport and Lowe syndromes?
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What lens pathologies occur with Alport and Lowe syndromes?
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### DDx for an RP-like Fundus

**Primary RP**  
*(aka typical RP)*

**Secondary RP**  
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  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
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  - Bardet-Biedl syndrome
  - Alström syndrome
  - Joubert syndrome
  - Senior-Løken syndrome
- Abetalipoproteinemia
  - *aka* ‘Bassen-Kornzweig dz’

**‘Pseudo-RP’**

---

**For completeness’ sake:**
*In three words (including syndromes), what are these ciliopathies?*
They are **familial oculorenal syndromes**

---

**Are Alport and Lowe syndrome in the DDx for an RP-like fundus?**
Lowe syndrome does not manifest retinal changes, so it is not. Alport syndrome is associated with what the *Peds* book describes as a “fleck retinopathy” and the *Retina* book as a “pigmentary retinopathy;” for this reason the *Retina* book considers it to be in the DDx.

---

**What are the other two?**
**Alport syndrome and Lowe syndrome**

---

**Abnormalities of what intraocular structure are associated with Alport and Lowe syndromes?**
**The lens**

**What lens pathologies occur with Alport and Lowe syndromes?**
- Lenticular
- Cataracts
- Microspherophakia
DDx for an RP-like Fundus

Primary RP

(aka typical RP)

Secondary RP

(aka Complex RP; Syndromic RP)

- Usher syndrome
- Peroxisomal disorders
  --Zellweger syndrome
  --Neonatal adrenoleukodystrophy
- Neuronal ceroid lipofuscinoses
- Ciliopathies
  --Bardet-Biedl syndrome
  --Alström syndrome
  --Joubert syndrome
  --Senior-Løken syndrome
- Abetalipoproteinemia
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‘Pseudo-RP’

For completeness’ sake:
In three words (including syndromes), what are these ciliopathies? They are familial oculoarenal syndromes

Are Alport and Lowe syndrome in the DDx for an RP-like fundus?
Lowe syndrome does not manifest retinal changes, so it is not. Alport syndrome is associated with what the Ped’s book describes as a “fleck retinopathy” and the Retina book as a “pigmentary retinopathy;” for this reason the Retina book considers it to be in the DDx.

What are the other two?
Alport syndrome and Lowe syndrome

Abnormalities of what intraocular structure are associated with Alport and Lowe syndromes?
The lens

What lens pathologies occur with Alport and Lowe syndromes?
--Lenticonus?
--Cataracts?
--Microspherophakia?

Which is the classic association with Alport and Lowe syndromes? Which should come first to mind?

Lenticonus
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
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- Ciliopathies
  -- Bardet-Biedl syndrome
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  -- Senior-Løken syndrome
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  -- aka ‘Bassen-Kornzweig dz’

For completeness’ sake:
In three words (including syndromes), what are these ciliopathies?
They are familial oculorenal syndromes

What are the other two?
Alport syndrome and Lowe syndrome

Abnormalities of what intraocular structure are associated with Alport and Lowe syndromes?
The lens

What lens pathologies occur with Alport and Lowe syndromes?
-- Lenticonus!
-- Cataracts
-- Microspherophakia

Which is the classic association with Alport and Lowe syndromes?
Which should come first to mind?
Lenticonus

Are Alport and Lowe syndrome in the DDx for an RP-like fundus?
Lowe syndrome does not manifest retinal changes, so it is not. Alport syndrome is associated with what the Peds book describes as a “fleck retinopathy” and the Retina book as a “pigmentary retinopathy;” for this reason the Retina book considers it to be in the DDx.
Anterior lenticonus

Posterior lenticonus
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
- Neuronal ceroid lipofuscinoses
- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

For completeness’ sake:
In three words (including syndromes), what are these ciliopathies?
They are familial oculo-renal syndromes

What are the other two?
Alport syndrome and Lowe syndrome

Abnormalities of what intraocular structure are associated with Alport and Lowe syndromes?
The lens

What lens pathologies occur with Alport and Lowe syndromes?
- Lenticonus?
- Cataracts?
- Microspherophakia?

Are Alport and Lowe syndrome in the DDx for an RP-like fundus?
Lowe syndrome does not manifest retinal changes, so it is not. Alport syndrome is associated with what the Peds book describes as a “fleck retinopathy” and the Retina book as a “pigmentary retinopathy;” for this reason the Retina book considers it to be in the DDx.

Are these associated with the ciliopathies?

Are Alport and Lowe syndrome in the DDx for an RP-like fundus?
Primary RP
(aka typical RP)

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- Usher syndrome
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For completeness’ sake:
In three words (including syndromes), what are these ciliopathies?
They are familial oculorenal syndromes

What are the other two?
Alport syndrome and Lowe syndrome

Abnormalities of what intraocular structure are associated with Alport and Lowe syndromes?
The lens

What lens pathologies occur with Alport and Lowe syndromes?
- Lenticonus
- Cataracts
- Microspherophakia

Are these associated with the ciliopathies?
No

Are Alport and Lowe syndrome in the DDx for an RP-like fundus?
Lowe syndrome does not manifest retinal changes, so it is not. Alport syndrome is associated with what the Peds book describes as a “fleck retinopathy” and the Retina book as a “pigmentary retinopathy;” for this reason the Retina book considers it to be in the DDx.
Familial Oculorenal Syndromes *tl;dr*

- One sort
- The other sort
Familial Oculorenal Syndromes *tl;dr*

- Ciliopathies
- Not Ciliopathies
Familial Oculorenal Syndromes *tl;dr*

- Ciliopathies
  - ?
  - ?
  - ?
  - ?
- Not Ciliopathies
  - ?
  - ?
Familial Oculorenal Syndromes *tl;dr*

**Ciliopathies**
- Joubert syndrome
- Alström syndrome
- Bardet-Biedl syndrome
- Senior-Løken syndrome

**Not Ciliopathies**
- Alport syndrome
- Lowe syndrome
Familial Oculorenal Syndromes

- **Ciliopathies**
  - Joubert syndrome
  - Alström syndrome
  - Bardet-Biedl syndrome
  - Senior-Løken syndrome

- **Not Ciliopathies**
  - Alport syndrome
  - Lowe syndrome

**Key Features**

- Renal failure
- Hematuria
  - w/ vs w/o
Familial Oculorenal Syndromes *tl;dr*

- Ciliopathies
  - Joubert syndrome
  - Alström syndrome
  - Bardet-Biedl syndrome
  - Senior-Løken syndrome
- Not Ciliopathies
  - Alport syndrome
  - Lowe syndrome

**Key Features**

- Renal failure *without* hematuria
- Renal failure *with* hematuria
Familial Oculorenal Syndromes *tl;dr*

- **Ciliopathies**
  - Joubert syndrome
  - Alström syndrome
  - Bardet-Biedl syndrome
  - Senior-Løken syndrome

- **Not Ciliopathies**
  - Alport syndrome
  - Lowe syndrome

**Key Features**

- **Renal failure**
  - *without* hematuria

- **Classic eye finding:**

- **Renal failure**
  - *with* hematuria

- **Classic eye finding:**
Familial Oculorenal Syndromes

Ciliopathies
- Joubert syndrome
- Alström syndrome
- Bardet-Biedl syndrome
- Senior-Løken syndrome

Not Ciliopathies
- Alport syndrome
- Lowe syndrome

Renal failure
- without hematuria

Classic eye finding:
- Pigmentary retinopathy

Renal failure
- with hematuria

Classic eye finding:
- Lenticonus
Familial Oculorenal Syndromes

Ciliopathies
- Joubert syndrome
- Alström syndrome
- Bardet-Biedl syndrome
- Senior-Løken syndrome

Not Ciliopathies
- Alport syndrome
- Lowe syndrome

Renal failure without hematuria

Classic eye finding:
- Pigmentary retinopathy

Inheritance:

Renal failure with hematuria

Classic eye finding:
- Lenticous

Inheritance:
Familial Oculorenal Syndromes \textit{tl;dr}

Ciliopathies
- Joubert syndrome
- Alström syndrome
- Bardet-Biedl syndrome
- Senior-Løken syndrome
- Alport syndrome
- Lowe syndrome

Renal failure \textit{without} hematuria

Classic eye finding: \textit{Pigmentary retinopathy}

Inheritance: \textit{AR}

Not Ciliopathies

Renal failure \textit{with} hematuria

Classic eye finding: \textit{Lenticonus}

Inheritance: \textit{X-linked}

\textbf{Key Features}
Familial Oculorenal Syndromes *tl;dr*

**Ciliopathies**
- Joubert syndrome-
- Alström syndrome-
- Bardet-Biedl syndrome-
- Senior-Løken syndrome-
- Alport syndrome
- Lowe syndrome

**Key Features**

- **Renal failure**
  - *without* hematuria
- **Classic eye finding:** *Pigmentary retinopathy*
- **Inheritance:** *AR*

- **Renal failure**
  - *with* hematuria
- **Classic eye finding:** *Lenticonus*
- **Inheritance:** *X-linked*

*For more info on Alport and Lowe syndromes, see slide-set L4*
Primary RP
(aka typical RP)

Secondary RP
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- Usher syndrome
- Peroxisomal disorders
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  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka 'Bassen-Kornzweig dz'

‘Pseudo-RP’

Worth stating explicitly that, in addition to being in the DDx for an RP-like fundus, these conditions are in the DDx for LCA. Once you get that connection locked down, learning this portion of the slide-set will constitute a twofer.
What is the underlying problem in abetalipoproteinemia?

- Abetalipoproteinemia
  --aka 'Bassen-Kornzweig dz'

--Senior-Løken syndrome
What is the underlying problem in abetalipoproteinemia?
One of the lipoproteins (ApoB) is not synthesized by the body

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- **Abetalipoproteinemia**
  --aka ‘Bassen-Kornzweig dz’
What is the underlying problem in abetalipoproteinemia?
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What is the underlying problem in abetalipoproteinemia?
One of the lipoproteins (ApoB) is not synthesized by the body

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How does the absence of ApoB lead to secondary RP?

Abetalipoproteinemia
--aka ‘Bassen-Kornzweig dz’
What is the underlying problem in abetalipoproteinemia?
One of the lipoproteins (ApoB) is not synthesized by the body

How is abetalipoproteinemia inherited?
AR

How does the absence of ApoB lead to secondary RP?
Without ApoB, fat cannot be properly absorbed. Without proper fat absorption, adequate vitamin A (a fat-soluble vitamin) levels cannot be maintained. And absent adequate vitamin A levels, the retina will cease functioning (as will other aspects of the CNS).

--Senior-Løken syndrome

- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’
### DDx for an RP-like Fundus

<table>
<thead>
<tr>
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- **Abetalipoproteinemia**
  --aka ‘Bassen-Kornzweig dz’
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In addition to low vitamin A levels, there is a classic finding on peripheral blood smear--what is it?

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Checking vitamin A levels is a good start

How is it treated?
With supplementary vitamins A & E

In addition to low vitamin A levels, there is a classic finding on peripheral blood smear--what is it?
Acanthocytosis of the RBCs

--Senior-Løken syndrome

- Abetalipoproteinemia
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In addition to low vitamin A levels, there is a classic finding on peripheral blood smear--what is it?
Acanthocytosis of the RBCs

What does acanthocytosis mean?
It means the RBCs have a 'thorny' appearance

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What does acanthocytosis mean?
It means the RBCs have a ‘thorny’ appearance.

--Senior-Løken syndrome

• Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’
DDx for an
RP-like Fundus

Acanthocytosis
What is the underlying problem in abetalipoproteinemia?
One of the lipoproteins (ApoB) is not synthesized by the body

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AR

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How is it diagnosed?
Checking vitamin A levels is a good start

How is it treated?
With supplementary vitamins A & E

Abetalipoproteinemia
--aka 'Bassen-Kornzweig dz'
**DDx for an RP-like Fundus**

- What is the underlying problem in abetalipoproteinemia?  
  One of the lipoproteins (ApoB) is not synthesized by the body

- How is abetalipoproteinemia inherited?  
  AR

- How does the absence of ApoB lead to secondary RP?  
  Without ApoB, fat cannot be properly absorbed. Without proper fat absorption, adequate vitamin A (a fat-soluble vitamin) levels cannot be maintained. And absent adequate vitamin A levels, the retina will cease functioning (as will other aspects of the CNS).

- How is it diagnosed?  
  Checking vitamin A levels is a good start

- How is it treated?  
  With supplementary vitamins A & E

---

**What is the most common cause of hypovitaminosis A? (It’s not abetalipoproteinemia)**

---

- Senior-Løken syndrome
  - Abetalipoproteinemia  
    --aka ‘Bassen-Kornzweig dz’
What is the underlying problem in abetalipoproteinemia?  
One of the lipoproteins (ApoB) is not synthesized by the body

How is abetalipoproteinemia inherited?  
AR

How does the absence of ApoB lead to secondary RP?  
Without ApoB, fat cannot be properly absorbed. Without proper fat absorption, adequate vitamin A (a fat-soluble vitamin) levels cannot be maintained. And absent adequate vitamin A levels, the retina will cease functioning (as will other aspects of the CNS).

How is it diagnosed?  
Checking vitamin A levels is a good start

How is it treated?  
With supplementary vitamins A & E

What is the most common cause of hypovitaminosis A? (It’s not abetalipoproteinemia)  
Malabsorption secondary to GI surgery (eg, gastric bypass; small-bowel resection)

DDx for an RP-like Fundus

- Abetalipoproteinemia  
  --aka ‘Bassen-Kornzweig dz’
**DDx for an RP-like Fundus**

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  - (aka typical RP)

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  - (aka Complex RP; Syndromic RP)
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    - Peroxisomal disorders
      - Zellweger syndrome
      - Neonatal adrenoleukodystrophy
      - Infantile Refsum dz
    - Neuronal ceroid lipofuscinoses
      - aka ‘Batten dz’
    - Ciliopathies
      - Bardet-Biedl syndrome
      - Alström syndrome
      - Joubert syndrome
      - Senior-Løken syndrome
    - **Abetalipoproteinemia**
      - aka ‘Bassen-Kornzweig dz’

---

*In abetalipoproteinemia, β-lipoprotein is absent (that’s what the prefix -a- indicates). Is hypobetalipoproteinemia a thing?*
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’

In abetalipoproteinemia, β-lipoprotein is absent (that’s what the prefix -a- indicates).
Is hypobetalipoproteinemia a thing?
Indeed it is, via a condition called ‘familial hypobetalipoproteinemia,’ and it can affect the retina (it’s mentioned in the BCSC books, but not addressed in detail)
DDx for an RP-like Fundus

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\textit{(aka typical RP)}

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  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
  \textcolor{red}{\textbf{alpha}}
- A\textit{beta}-lipoproteinemia
  -- \textit{aka} ‘Bassen-Kornzweig dz’

\textit{Is a-a\textbf{pha}-lipoproteinemia a thing?}
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  --Zellweger syndrome
  --Neonatal adrenoleukodystrophy
  --Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  --aka ‘Batten dz’
- Ciliopathies
  --Bardet-Biedl syndrome
  --Alström syndrome
  --Joubert syndrome
  --Senior-Løken syndrome
  --alpha
- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

Is a-alpha-lipoproteinemia a thing?
Indeed it is, but it’s not called that, for obvious reasons
DDx for an RP-like Fundus

Primary RP (aka typical RP)

Secondary RP (aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka ‘Batten dz’
- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome

‘Pseudo-RP’

Is a-\textbf{alpha}-lipoproteinemia a thing? Indeed it is, but it’s not called that, for obvious reasons

Does the absence of \textbf{\alpha}-lipoprotein affect the retina?
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka ‘Batten dz’
- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Beta-lipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

Is a-beta-lipoproteinemia a thing?
Indeed it is, but it’s not called that, for obvious reasons

Does the absence of alpha-lipoprotein affect the retina?
No, but it does affect the cornea
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka ‘Batten dz’
- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- A-beta-lipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

Is a-$\alpha$-lipoproteinemia a thing?
Indeed it is, but it’s not called that, for obvious reasons

Does the absence of $\alpha$-lipoprotein affect the retina?
No, but it does affect the cornea
DDx for an RP-like Fundus

- **Primary RP** (aka typical RP)
- **Secondary RP** (aka Complex RP; Syndromic RP)
  - Usher syndrome
  - Peroxisomal disorders
    -- Zellweger syndrome
    -- Neonatal adrenoleukodystrophy
    -- Infantile Refsum dz
  - Neuronal ceroid lipofuscinoses
    -- aka ‘Batten dz’
  - Ciliopathies
    -- Bardet-Biedl syndrome
    -- Alström syndrome
    -- Joubert syndrome
    -- Senior-Løken syndrome
  - Abetalipoproteinemia
    -- aka ‘Bassen-Kornzweig dz’

**Is α-lipoprotein a thing?**
Indeed it is, but it’s not called that, for obvious reasons.

Does the absence of α-lipoprotein affect the retina?
No, but it does affect the cornea.

Low levels of α-lipoprotein are implicated in three corneal conditions. What are they?
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  - aka ‘Batten dz’
- Ciliopathies
  - Bardet-Biedl syndrome
  - Alström syndrome
  - Joubert syndrome
  - Senior-Løken syndrome
- Abetalipoproteinemia
  - aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

Is a-\textit{alpha}-lipoprotein a thing?
Indeed it is, but it’s not called that, for obvious reasons.

Does the absence of a-\textit{alpha}-lipoprotein affect the retina?
No, but it does affect the cornea.

Low levels of a-\textit{alpha}-lipoprotein are implicated in three corneal conditions. What are they?
- LCAT deficiency
- Fish eye disease
- Tangier disease
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka ‘Batten dz’
- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

So, the hypolipoproteinemias can be divided into those involving \( \alpha \)-lipoproteins, which lead to pathology of the cornea...

Low levels of \( \alpha \)-lipoprotein are implicated in three corneal conditions. What are they?

- LCAT deficiency
- Fish eye disease
- Tangier disease

Does the absence of \( \alpha \)-lipoprotein affect the retina?

No, but it does affect the cornea
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  --Zellweger syndrome
  --Neonatal adrenoleukodystrophy
  --Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  --aka ‘Batten dz’
- Ciliopathies
  --Bardet-Biedl syndrome
  --Alström syndrome
  --Joubert syndrome
  --Senior-Løken syndrome
- Abetalipoproteinemia
  --aka ‘Bassen-Korzwieg dz’

So, the hypolipoproteinemias can be divided into those involving α-lipoproteins, which lead to pathology of the cornea...

...and β-lipoproteins, which lead to pathology of the retina

Low levels of α-lipoprotein are implicated in three corneal conditions. What are they?
- LCAT deficiency
- Fish eye disease
- Tangier disease
- Abetalipoproteinemia (Bassen-Korzwieg)
- Familial hypobetalipoproteinemia
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka ‘Batten dz’
- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’
- Congenital infection
- Congenital infection
DDx for an RP-like Fundus

Primary RP  (aka typical RP)

Secondary RP  (aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  --Zellweger syndrome
  --Neonatal adrenoleukodystrophy
  --Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  --aka ‘Batten dz’
- Ciliopathies
  --Bardet-Biedl syndrome
  --Alström syndrome
  --Joubert syndrome
  --Senior-Løken syndrome
- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’
- Congenital syphilis
- Congenital rubella
DDx for an RP-like Fundus

Congenital rubella

Congenital syphilis
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka 'Batten dz'
- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka 'Bassen-Kornzweig dz'

‘Pseudo-RP’
- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  --?
  --?
  -- Others
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka ‘Batten dz’
- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  -- Toxoplasmosis
  -- HSV
  -- Others

DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka ‘Batten dz’
- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  -- Toxoplasmosis
  -- HSV
  -- Others
DDx for an RP-like Fundus

Toxoplasmosis

HSV
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka 'Batten dz'
- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka 'Bassen-Kornzweig dz'

‘Pseudo-RP’
- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  -- Toxoplasmosis
  -- HSV
  -- Others
- Associated retinopathy
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka ‘Batten dz’
- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’
- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  -- Toxoplasmosis
  -- HSV
  -- Others
- Cancer-associated retinopathy
DDx for an RP-like Fundus

CAR
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses

‘Pseudo-RP’
- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  -- Toxoplasmosis
  -- HSV
  -- Others
- Cancer-associated retinopathy

What is cancer-associated retinopathy (CAR)?
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’
What is cancer-associated retinopathy (CAR)?
A paraneoplastic process in which retinal cells display antigens that are identical to, or cross-react with, cancer cells within the body. Subsequent to sensitization to these antigens on the cancer cells, the immune system attacks the same/similar antigens in the retina. This can produce profound retinal degeneration, resulting in loss of both visual function and the ERG response.

- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’
**DDx for an RP-like Fundus**

**Primary RP** *(aka typical RP)*

- Congenital syphilis
- Congenital rubella
- Infectious retinitis
- Cancer-associated retinopathy
- Kearns-Sayre syndrome

**Secondary RP** *(aka Complex RP; Syndromic RP)*

- Usher syndrome
- Peroxisomal disorders
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  - aka ‘Batten dz’
- Ciliopathies
  - Bardet-Biedl syndrome
  - Alström syndrome
  - Joubert syndrome
  - Senior-Løken syndrome
- Abetalipoproteinemia
  - aka ‘Bassen-Kornzweig dz’

**‘Pseudo-RP’**

- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  - Toxoplasmosis
  - HSV
  - Others
- Cancer-associated retinopathy
  - eponym-eponym syndrome
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
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- Neuronal ceroid lipofuscinoses
  -- aka ‘Batten dz’
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  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’
- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  -- Toxoplasmosis
  -- HSV
  -- Others
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
What is the classic triad of Kearns-Sayre syndrome?

- Cardiac conduction abnormalities
- Ophthalmoplegia (usually the ptosis)
- Ragged red fibers

In very general terms, what sort of disease is K-SS?

A mitochondrial disease

What is the classic finding on muscle biopsy?

Ragged red fibers

At what age do symptoms begin occurring?

Usually shortly before age 10 years

Of the classic triad, which is the first to occur?

The ophthalmoplegia (usually the ptosis)
What is the classic triad of Kearns-Sayre syndrome?
--Pigmentary retinopathy
--CPEO
--Cardiac conduction abnormalities

What is the classic finding on muscle biopsy?
Ragged red fibers

At what age do symptoms begin occurring?
Usually shortly before age 10 years

Of the classic triad, which is the first to occur?
The ophthalmoplegia (usually the ptosis)

DDx for an RP-like Fundus

Primary RP (aka typical RP)

Secondary RP

‘Pseudo-RP’

Congenital syphilis
Congenital rubella
Infectious retinitis
--Toxoplasmosis
--HSV
--Others
Cancer-associated retinopathy

Kearns-Sayre syndrome

--aka ‘Bassen-Kornzweig dz’
DDx for an RP-like Fundus

Kearns-Sayre syndrome: Pigmentary retinopathy
What is the classic triad of Kearns-Sayre syndrome?
- Pigmentary retinopathy
- CPEO
- Cardiac conduction abnormalities

What does CPEO stand for?
- Chronic progressive external ophthalmoplegia

Which EOMs are typically affected first?
The levators; ie, ptosis is the first manifestation. However, the disease is relentlessly progressive, and eventually all of the EOMs are paralyzed.

What is the classic triad of Kearns-Sayre syndrome?
- Pigmentary retinopathy
- CPEO
- Cardiac conduction abnormalities

In very general terms, what sort of disease is K-SS?
A mitochondrial disease

What is the classic finding on muscle biopsy?
Ragged red fibers

At what age do symptoms begin occurring?
Usually shortly before age 10 years

Of the classic triad, which is the first to occur?
The ophthalmoplegia (usually the ptosis)

What does CPEO stand for?
Chronic progressive external ophthalmoplegia

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DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP

What is the classic triad of Kearns-Sayre syndrome?
- Pigmentary retinopathy
- CPEO
- Cardiac conduction abnormalities

What does CPEO stand for?
Chronic progressive external ophthalmoplegia

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- Pigmentary retinopathy
- CPEO
- Cardiac conduction abnormalities

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- CPEO
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- CPEO
- Cardiac conduction abnormalities

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- CPEO
- Cardiac conduction abnormalities

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- Pigmentary retinopathy
- CPEO
- Cardiac conduction abnormalities

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What is the classic triad of Kearns-Sayre syndrome?
- Pigmentary retinopathy
- CPEO
- Cardiac conduction abnormalities

What does CPEO stand for?
Chronic progressive external ophthalmoplegia

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What is the classic triad of Kearns-Sayre syndrome?
- Pigmentary retinopathy
- CPEO
- Cardiac conduction abnormalities

What does CPEO stand for?
Chronic progressive external ophthalmoplegia

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What is the classic triad of Kearns-Sayre syndrome?
- Pigmentary retinopathy
- CPEO
- Cardiac conduction abnormalities

What does CPEO stand for?
Chronic progressive external ophthalmoplegia

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What is the classic triad of Kearns-Sayre syndrome?
- Pigmentary retinopathy
- CPEO
- Cardiac conduction abnormalities

What does CPEO stand for?
Chronic progressive external ophthalmoplegia

Which EOMs are typically affected first?
The levators; ie, ptosis is the first manifestation. However, the disease is relentlessly progressive, and eventually all of the EOMs are paralyzed.
DDx for an RP-like Fundus

Primary RP (aka typical RP)

Secondary RP

‘Pseudo-RP’

Congenital syphilis
Congenital rubella
Infectious retinitis
Cancer-associated retinopathy
--Toxoplasmosis
--HSV
--Others
Ciliopathies
Abetalipoproteinemia

What is the classic triad of Kearns-Sayre syndrome?
--Pigmentary retinopathy
--CPEO
--Cardiac conduction abnormalities

What does CPEO stand for?
Chronic progressive external ophthalmoplegia

--aka ‘Bassen-Kornzweig dz’

What is the classic finding on muscle biopsy?
Ragged red fibers

At what age do symptoms begin occurring?
Usually shortly before age 10 years

Of the classic triad, which is the first to occur?
The ophthalmoplegia (usually the ptosis)

What does CPEO stand for?
Chronic progressive external ophthalmoplegia
What is the classic triad of Kearns-Sayre syndrome?
- Pigmentary retinopathy
- CPEO (Chronic progressive external ophthalmoplegia)
- Cardiac conduction abnormalities

What does CPEO stand for?
Chronic progressive external ophthalmoplegia

Which EOMs are typically affected first?
-- aka 'Bassen-Kornzweig dz'

DDx for an RP-like Fundus

Primary RP (aka typical RP)
- Zellweger syndrome
- Neonatal adrenoleukodystrophy
- Infantile Refsum dz
- Bardet-Biedl syndrome
- Alström syndrome
- Joubert syndrome
- Senior-Løken syndrome
-- aka 'Batten dz'
-- aka 'Bassen-Kornzweig dz'

Secondary RP
- Toxoplasmosis
- HSV
- Others

‘Pseudo-RP’
- Congenital syphilis
- Congenital rubella
- Infectious retinitis
- Toxoplasmosis
- HSV
- Others
- Cancer-associated retinopathy

Kearns-Sayre syndrome
What is the classic triad of Kearns-Sayre syndrome?
- Pigmentary retinopathy
- CPEO
- Cardiac conduction abnormalities

What does CPEO stand for?  
Chronic progressive external ophthalmoplegia

Which EOMs are typically affected first?  
The levators; i.e., ptosis is the first manifestation. However, the disease is relentlessly progressive, and eventually all of the EOMs are paralyzed.

--aka 'Bassen-Kornzweig dz'
DDx for an RP-like Fundus

CPEO
DDx for an RP-like Fundus

Primary RP (aka typical RP)

Secondary RP

‘Pseudo-RP’

Congenital syphilis
Congenital rubella
Infectious retinitis
--Toxoplasmosis
--HSV
--Others
Cancer-associated retinopathy
Kearns-Sayre syndrome

Primary RP

(aka typical RP)

Secondary RP

What is the classic triad of Kearns-Sayre syndrome?
--Pigmentary retinopathy
--CPEO
--Cardiac conduction abnormalities

Are the cardiac-conduction problems serious?

--aka ‘Bassen-Kornzweig dz’

In very general terms, what sort of disease is K-SS?
A mitochondrial disease

What is the classic finding on muscle biopsy?
Ragged red fibers’

At what age do symptoms begin occurring?
Usually shortly before age 10 years

Of the classic triad, which is the first to occur?
The ophthalmoplegia (usually the ptosis)

Are the cardiac-conduction problems serious?
Yes, they can be life-threatening
DDx for an RP-like Fundus

Primary RP (aka typical RP)
- Congenital syphilis
- Congenital rubella
- Infectious retinitis
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
- Peroxisomal disorders
- Neuronal ceroid lipofuscinoses
- Ciliopathies
- Abetalipoproteinemia

Secondary RP
- Usher syndrome

‘Pseudo-RP’
- Zellweger syndrome
- Neonatal adrenoleukodystrophy
- Infantile Refsum dz
- Bardet-Biedl syndrome
- Alström syndrome
- Joubert syndrome
- Senior-Løken syndrome
- ‘Batten dz’
- ‘Bassen-Kornzweig dz’
- Toxoplasmosis
- HSV
- Others
- Cancer-associated retinopathy

Kearns-Sayre syndrome

What is the classic triad of Kearns-Sayre syndrome?
- Pigmentary retinopathy
- CPEO
- Cardiac conduction abnormalities

Are the cardiac-conduction problems serious?
Yes, they can be life-threatening

In very general terms, what sort of disease is K-SS?
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DDx for an RP-like Fundus

Primary RP (aka typical RP)

Secondary RP

‘Pseudo-RP’

- Congenital syphilis
- Congenital rubella
- Infectious retinitis
- Cancer-associated retinopathy
- Toxoplasmosis
- HSV
- Others

What is the classic triad of Kearns-Sayre syndrome?
- Pigmentary retinopathy
- CPEO
- Cardiac conduction abnormalities

In very general terms, what sort of disease is K-SS?

‘Kearns-Sayre syndrome’

--aka ‘Bassen-Kornzweig dz’

What is the classic finding on muscle biopsy?

Ragged red fibers

At what age do symptoms begin occurring?

Usually shortly before age 10 years

Of the classic triad, which is the first to occur?

The ophthalmoplegia (usually the ptosis)
**DDx for an RP-like Fundus**

- **Primary RP** (aka typical RP)
- **Secondary RP**
- ‘Pseudo-RP’

**Congenital syphilis**
- Congenital rubella
- Infectious retinitis
- Cancer-associated retinopathy

**Secondary RP** (aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
- Neuronal ceroid lipofuscinoses
- Ciliopathies
- Abetalipoproteinemia

**Primary RP** (aka typical RP)
- Zellweger syndrome
- Neonatal adrenoleukodystrophy
- Infantile Refsum dz
- Bardet-Biedl syndrome
- Alström syndrome
- Joubert syndrome
- Senior-Løken syndrome

- **What is the classic triad of Kearns-Sayre syndrome?**
  - Pigmentary retinopathy
  - CPEO
  - Cardiac conduction abnormalities

- **In very general terms, what sort of disease is K-SS?**
  - A mitochondrial disease

- **What is the classic finding on muscle biopsy?**
  - Ragged red fibers

- **At what age do symptoms begin occurring?**
  - Usually shortly before age 10 years

- **Of the classic triad, which is the first to occur?**
  - The ophthalmoplegia (usually the ptosis)

**DDx for an RP-like Fundus**

- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  - Toxoplasmosis
  - HSV
  - Others
- Cancer-associated retinopathy

**Kearns-Sayre syndrome**

- **aka ‘Bassen-Kornzweig dz’**
Primary RP (aka typical RP)

Secondary RP

‘Pseudo-RP’

Congenital syphilis
Congenital rubella
Infectious retinitis
--Toxoplasmosis
--HSV
--Others
Cancer-associated retinopathy

Kearns-Sayre syndrome

What is the classic triad of Kearns-Sayre syndrome?
--Pigmentary retinopathy
--CPEO
--Cardiac conduction abnormalities

In very general terms, what sort of disease is K-SS?
A mitochondrial disease

What is the classic finding on muscle biopsy?

--aka ‘Bassen-Kornzweig dz’
What is the classic triad of Kearns-Sayre syndrome?
--Pigmentary retinopathy
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In very general terms, what sort of disease is K-SS?
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(aka typical RP)

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Congenital rubella
Infectious retinitis
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Kearns-Sayre syndrome

DDx for an RP-like Fundus

Usually shortly before age 10 years

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DDx for an RP-like Fundus

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‘Pseudo-RP’

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--aka ‘Bassen-Kornzweig dz’
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP

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Kearns-Sayre syndrome
DDx for an RP-like Fundus

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Because in making the proper diagnosis, the astute ophthalmologist can refer the pt to a cardiologist before s/he has a fatal dysrhythmia.
DDx for an RP-like Fundus

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- Kearns-Sayre syndrome

Secondary RP
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- Peroxisomal disorders
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- Ciliopathies
- Abetalipoproteinemia

‘Pseudo-RP’
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(aka Complex RP; Syndromic RP)
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  -- aka ‘Batten dz’
- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
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four-letter abb. for infectious cause
DDx for an RP-like Fundus

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- DUSN
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What does DUSN stand for?

Diffuse unilateral subacute neuroretinitis
DDx for an RP-like Fundus

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(aka typical RP)

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(aka Complex RP; Syndromic RP)
- Usher syndrome
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  - Zellweger syndrome
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  - Others
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- Severe uveitis
- **DUSN**

**What does DUSN stand for?**
**Diffuse unilateral subacute neuroretinitis**
DDx for an RP-like Fundus

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(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

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- ‘Pseudo-RP’

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  -- Toxoplasmosis
  -- HSV
  -- Others
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
- Severe uveitis
- DUSN

What does DUSN stand for?
- Diffuse unilateral subacute neuroretinitis

What is the cause?

Infestation by a worm (most commonly, Baylisacaris)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
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- **What does DUSN stand for?**
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‘Pseudo-RP’
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  --HSV
  --Others
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
- Severe uveitis
- **DUSN**

What is the cause?
Infestation by a [type of bug] (most commonly, [specific bug])
DDx for an RP-like Fundus

Primary RP (aka typical RP)

Secondary RP (aka Complex RP; Syndromic RP)
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- **What does DUSN stand for?**
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‘Pseudo-RP’
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- **DUSN**

What is the cause?
Infestation by a worm (most commonly, *Baylisacaris* )
DDx for an RP-like Fundus

DUSN (circle indicates the worm’s location)
DDx for an RP-like Fundus

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(aka typical RP)

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- **DUSN**

**What does DUSN stand for?**

Diffuse unilateral subacute neuroretinitis

**When should you consider that a case of ‘RP’ might be DUSN?**
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
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What does DUSN stand for?
Diffuse unilateral subacute neuroretinitis

When should you consider that a case of ‘RP’ might be DUSN?
When it is so-called ‘unilateral RP’
DDx for an RP-like Fundus

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(aka Complex RP; Syndromic RP)
- Usher syndrome
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What does DUSN stand for?
- Diffuse unilateral subacute neuroretinitis

When should you consider that a case of ‘RP’ might be DUSN?
- When it is so-called ‘unilateral RP’

For more on DUSN, see slide-set R15. If it’s not available, an abbreviated coverage of DUSN can be found in the White Dot Syndrome set (R16). Or, email me and I’ll send you a copy of R15.
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four-letter abb. for a vascular cause
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- Drug toxicity
  -- ?
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Secondary RP
(aka Complex RP; Syndromic RP)
DDx for an RP-like Fundus

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- CRAO
- Drug toxicity
  -- Hydroxychloroquine
  -- Thioridazine
  -- Others
DDx for an RP-like Fundus

Thioridazine: Pigmentary retinopathy

Hydroxychloroquine retinopathy
DDx for an RP-like Fundus

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- Drug toxicity
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    -- Others

Note: Hydroxychloroquine (Plaquenil) toxicity is addressed in detail in its own slide-set
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What class of medicine is thioridazine?

It is a phenothiazine

What are the phenothiazines used to treat?
Their main use is as antipsychotics

Is thioridazine retinal toxicity dose-related?
Very much so. At doses of 800 mg/day or less, retinopathy is very rare. However, at higher doses, it can develop within a matter of weeks

How does thioridazine retinopathy present clinically?
The pt with c/o blurry vision; DFE will reveal pigment stippling in the macula

Does thioridazine use require periodic ophthalmic evaluations like hydroxychloroquine does?
Not so long as the dose is at or below 800 mg/day

Drug toxicity
-- Hydroxychloroquine
-- Thioridazine
-- Others
**DDx for an RP-like Fundus**

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DDx for an RP-like Fundus

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Drug toxicity
-- Hydroxychloroquine
  -- Thioridazine
  -- Others
DDx for an RP-like Fundus

Primary RP (aka typical RP)
- Congenital syphilis
- Congenital rubella
- Infectious retinitis
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
- Severe uveitis

Secondary RP (aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy

‘Pseudo-RP’
- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  -- Toxoplasmosis
  -- HSV
  -- Others
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
- Severe uveitis
- DUSN
- CRAO

**What class of medicine is thioridazine?**
It is a phenothiazine

**What are the phenothiazines used to treat?**
Their main use is as antipsychotics

**Is thioridazine retinal toxicity dose-related?**
Very much so. At doses of 800 mg/day or less, retinopathy is very rare. However, at higher doses, it can develop within a matter of weeks

**How does thioridazine retinopathy present clinically?**
The pt with c/o blurry vision; DFE will reveal pigment stippling in the macula

**Drug toxicity**
-- Hydroxychloroquine
  -- Thioridazine
  -- Others
DDx for an RP-like Fundus

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(aka typical RP)

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(aka Complex RP; Syndromic RP)
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The pt with c/o blurry vision; DFE will reveal pigment stippling in the macula

Does thioridazine use require periodic ophthalmic evaluations like hydroxychloroquine does?

Drug toxicity
--Hydroxychloroquine
  --Thioridazine
  --Others
DDx for an RP-like Fundus

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(aka typical RP)

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Very much so. At doses of 800 mg/day or less, retinopathy is very rare. However, at higher doses, it can develop within a matter of weeks

How does thioridazine retinopathy present clinically?
The pt with c/o blurry vision; DFE will reveal pigment stippling in the macula

Does thioridazine use require periodic ophthalmic evaluations like hydroxychloroquine does?
Not so long as the dose is at or below 800 mg/day

Drug toxicity
--Hydroxychloroquine
  --Thioridazine
  --Others
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
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  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka 'Batten dz'
- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka 'Bassen-Kornzweig dz'

‘Pseudo-RP’
- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  -- Toxoplasmosis
  -- HSV
  -- Others
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
- Severe uveitis
- DUSN
- CRAO
- Drug toxicity
  -- Hydroxychloroquine
  -- Thioridazine
  -- Others

What tests should be run to determine whether a pt has RP?
--
--
--
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
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- Kearns-Sayre syndrome
- Severe uveitis
- DUSN
- CRAO
- Drug toxicity
  -- Hydroxychloroquine
  -- Thioridazine
  -- Others

What tests should be run to determine whether a pt has RP?
-- An ERG (if it’s not markedly abnormal, it’s not RP)
-- Kinetic (ie, Goldmann, not Humphrey) VF testing. Again--if it’s not abnormal, it’s not RP.
-- Dark adaptometry (ditto)
DDx for an RP-like Fundus

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- Usher syndrome
- Peroxisomal disorders
- Zellweger syndrome

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- Kearns-Sayre syndrome
- Severe uveitis
- DUSN
- CRAO
- Drug toxicity
  --Hydroxychloroquine
  --Thioridazine
  --Others

How do you rule-in pseudo-RP?
- Congenital syphilis
- Congenital rubella
- Infectious retinitis
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
- Severe uveitis
- DUSN
- CRAO
- Drug toxicity
  --Hydroxychloroquine
  --Thioridazine
  --Others
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- Severe uveitis
- DUSN
- CRAO
- Drug toxicity
  --Hydroxychloroquine
  --Thioridazine
  --Others

How do you rule-in pseudo-RP?
Some causes can be identified by the presence (or absence) of certain signs/symptoms…

- Ciliopathies
  --Bardet-Biedl syndrome
  --Alström syndrome
  --Joubert syndrome
  --Senior-Løken syndrome
- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’
DDx for an RP-like Fundus

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  --HSV
  --Others
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
- Severe uveitis
- DUSN
- CRAO
- Drug toxicity
  --Hydroxychloroquine
  --Thioridazine
  --Others

How do you rule-in pseudo-RP?
Some causes can be identified by the presence (or absence) of certain signs/symptoms…
Some via pertinent lab results…

- Ciliopathies
  --Bardet-Biedl syndrome
  --Alström syndrome
  --Joubert syndrome
  --Senior-Løken syndrome
- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’
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(aka typical RP)

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(aka Complex RP; Syndromic RP)
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- Peroxisomal disorders
- Neuronal ceroid lipofuscinoses
- Ciliopathies
- Abetalipoproteinemia
- Zellweger syndrome

‘Pseudo-RP’
- Congenital syphilis
- Congenital rubella
- (+/- Infectious retinitis)
  --Toxoplasmosis
  --HSV
  --Others
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
- (+/- Severe uveitis)
- DUSN
- CRAO
- Drug toxicity
  --Hydroxychloroquine
  --Thioridazine
  --Others

How do you rule-in pseudo-RP?
Some causes can be identified by the presence (or absence) of certain signs/symptoms...
Some via pertinent lab results...
Some by their unilaterality...
- Ciliopathies
  --Bardet-Biedl syndrome
  --Alström syndrome
  --Joubert syndrome
  --Senior-Løken syndrome
- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’
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- Kearns-Sayre syndrome
- Severe uveitis
- DUSN
- CRAO
- Drug toxicity
  - Hydroxychloroquine
  - Thioridazine
  - Others

How do you rule-in pseudo-RP?
Some causes can be identified by the presence (or absence) of certain signs/symptoms…
Some via pertinent lab results…
Some by their unilaterality…
Some by history…

- Ciliopathies
  - Bardet-Biedl syndrome
  - Alström syndrome
  - Joubert syndrome
  - Senior-Løken syndrome

- Abetalipoproteinemia
  - aka ‘Bassen-Kornzweig dz’
DDx for an RP-like Fundus

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  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

- Congenital syphilis

How do you rule in the causes of secondary RP?

- Usher syndrome:

--- Check hearing

--- The peroxisomal disorders:
  - Check serum levels of ‘very long chain fatty acids’ +/- phytanic acid levels

--- Batten disease:
  - Punt to a geneticist

--- Ciliopathies:
  - Primarily clinical (confirmatory genetics by a geneticist), but remember the following:
    - Joubert syndrome:
      - Molar-tooth sign on brainstem MRI
    - Abetalipoproteinemia:
      - As discussed
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

- **Usher syndrome**
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- *aka* 'Batten dz'
- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- *aka* 'Bassen-Kornzweig dz'

‘Pseudo-RP’

- Congenital syphilis

--- **Usher syndrome**: Check hearing

--- *The peroxisomal disorders*: Check serum levels of 'very long chain fatty acids' +/- phytanic acid levels

--- **Batten disease**: Punt to a geneticist

--- **Ciliopathies**: Primarily clinical (confirmatory genetics by a geneticist), but remember the following:
  - **Joubert syndrome**: Molar-tooth sign on brainstem MRI
  - **Senior-Løken**: Check renal

--- **Abetalipoproteinemia**: As discussed

--- How do you rule-in the causes of secondary RP?
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
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  --Alström syndrome
  --Joubert syndrome
  --Senior-Løken syndrome
- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’
- Congenital syphilis

How do you rule-in the causes of secondary RP?

- Usher syndrome: Check hearing

The peroxisomal disorders:
- Check serum levels of ‘very long chain fatty acids’ +/- phytanic acid levels

Batten disease: Punt to a geneticist

Ciliopathies: Primarily clinical (confirmatory genetics by a geneticist), but remember the following:
- Joubert syndrome: Molar-tooth sign on brainstem MRI

Abetalipoproteinemia: As discussed

How do you rule-in the causes of secondary RP?
How do you rule-in the causes of secondary RP?

DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

- Usher syndrome
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  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

- Congenital syphilis

- Hydroxychloroquine
- Thioridazine

-- Usher syndrome: Check hearing

-- The peroxisomal disorders: Check serum levels of ‘very long chain fatty acids’ +/- phytanic acid levels

How do you rule-in the causes of secondary RP?)

- Toxoplasmosis
- HSV

-- Usher syndrome: Check hearing

-- The peroxisomal disorders: Check serum levels of ‘very long chain fatty acids’ +/- phytanic acid levels

- Neuronal ceroid lipofuscinoses
  -- aka ‘Batten dz’

- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome

- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’
DDx for an RP-like Fundus

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(aka typical RP)

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  --aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’
- Congenital syphilis

How do you rule-in the causes of secondary RP?

--Usher syndrome: Check hearing

--The peroxisomal disorders: Check serum levels of ‘very long chain fatty acids’ +/- phytanic acid levels

--Batten disease:
DDx for an RP-like Fundus

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(aka typical RP)

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(aka Complex RP; Syndromic RP)

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  --Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  --aka ‘Batten dz’
- Ciliopathies
  --Bardet-Biedl syndrome
  --Alström syndrome
  --Joubert syndrome
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- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

- Congenital syphilis

How do you rule-in the causes of secondary RP?

--Usher syndrome: Check hearing

--The peroxisomal disorders: Check serum levels of ‘very long chain fatty acids’ +/- phytanic acid levels

--Batten disease: Punt to a geneticist
DDx for an RP-like Fundus

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- Infectious retinitis
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
- Severe uveitis

Secondary RP (aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka ‘Batten dz’
- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’
- Congenital syphilis

How do you rule-in the causes of secondary RP?

-- Usher syndrome: Check hearing

-- The peroxisomal disorders: Check serum levels of ‘very long chain fatty acids’ +/- phytanic acid levels

-- Batten disease: Punt to a geneticist

-- Ciliopathies:
DDx for an RP-like Fundus

Primary RP
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  --Zellweger syndrome
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  --aka ‘Batten dz’
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  --Alström syndrome
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‘Pseudo-RP’
- Congenital syphilis

How do you rule-in the causes of secondary RP?

--Usher syndrome: Check hearing

--The peroxisomal disorders: Check serum levels of ‘very long chain fatty acids’ +/- phytanic acid levels

--Batten disease: Punt to a geneticist

--Ciliopathies: Primarily clinical (confirmatory genetics by a geneticist), but remember the following:
  ----Joubert syndrome:
How do you rule-in the causes of secondary RP?

**Primary RP** (aka typical RP)

**Secondary RP** (aka Complex RP; Syndromic RP)

- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka ‘Batten dz’
- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

**‘Pseudo-RP’**

- Congenital syphilis

--- *Usher syndrome*: Check hearing

--- *The peroxisomal disorders*: Check serum levels of ‘very long chain fatty acids’ +/- phytanic acid levels

--- *Batten disease*: Punt to a geneticist

--- *Ciliopathies*: Primarily clinical (confirmatory genetics by a geneticist), but remember the following:

--- *Joubert syndrome*: Molar-tooth sign on brainstem MRI

--- *Joubert syndrome*: Molar-tooth sign on brainstem MRI

--- *Senior-Løken*: Check renal function
### DDx for an RP-like Fundus

#### Primary RP
*(aka typical RP)*

#### Secondary RP
*(aka Complex RP; Syndromic RP)*
- **Usher syndrome**
- **Peroxisomal disorders**
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Infantile Refsum dz
- **Neuronal ceroid lipofuscinoses**
  - *aka* ‘Batten dz’
- **Ciliopathies**
  - Bardet-Biedl syndrome
  - Alström syndrome
  - Joubert syndrome
  - Senior-Løken syndrome
- **Abetalipoproteinemia**
  - *aka* ‘Bassen-Kornzweig dz’

#### ‘Pseudo-RP’
- Congenital syphilis

---

**How do you rule-in the causes of secondary RP?**

**- Usher syndrome**: Check hearing

**- The peroxisomal disorders**: Check serum levels of ‘very long chain fatty acids’ +/- phytanic acid levels

**- Batten disease**: Punt to a geneticist

**- Ciliopathies**: Primarily clinical (confirmatory genetics by a geneticist), but remember the following:
  ---- **Joubert syndrome**: Molar-tooth sign on brainstem MRI

**- Abetalipoproteinemia**: As discussed
tl;dr starts on the next slide
(When you hear *RP-like fundus*...
When you hear RP-like fundus...these three categories should instantly spring to mind—make sure they do!
Next, make sure you can name the *five categories of secondary/syndromic RP*. 
Next, make sure you can name the five categories of secondary/syndromic RP. Toggle back and forth between this slide and the previous one until you can name all five with ease!

**Primary RP** *(aka typical RP)*

**Secondary RP** *(aka Complex RP; Syndromic RP)*
- Usher syndrome
- Peroxisomal disorders
- Neuronal ceroid lipofuscinoses (aka Batten dz)
- Ciliopathies
- Abetalipoproteinemia (aka Bassen-Korzeig dz)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  --?
  --?
  --?
- Neuronal ceroid lipofuscinoses (aka Batten dz)
- Ciliopathies
  --?
  --?
  --?
- Abetalipoproteinemia
  (aka Bassen-Korzeig dz)

Then, make sure you can name the three *peroxisomal disorders* and the four *ciliopathies*. 
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Infantile Refsum dz
- Neuronal ceroid lipofuscinoses (aka Batten dz)
- Ciliopathies
  - Bardet-Biedl syndrome
  - Alström syndrome
  - Joubert syndrome
  - Senior-Løken syndrome
- Abetalipoproteinemia
  (aka Bassen-Korzweig dz)

‘Pseudo-RP’

Then, make sure you can name the three peroxisomal disorders and the four ciliopathies. Again, toggle back and forth between this slide and the previous one until you’ve mastered them all.
DDx for an RP-like Fundus

Primary RP (aka typical RP)

Secondary RP (aka Complex RP; Syndromic RP)

- Usher syndrome
- Peroxisomal disorders
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Infantile Refsum dz
- Neuronal ceroid lipofuscinoses (aka Batten dz)

- Ciliopathies
  - Bardet-Biedl syndrome
  - Alström syndrome
  - Joubert syndrome
  - Senior-Løken syndrome
- Abetalipoproteinemia (aka Bassen-Korzweig dz)

These are my best guesses (emphasis on guesses) regarding factoids that should be kept in mind for each.
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

‘Pseudo-RP’

Deaf/blind--
Usher syndrome

Peroxisomal disorders

Zellweger syndrome
Neonatal adrenoleukodystrophy
Infantile Refsum dz

Elevated VLCFAs--
Dreadful prognoses--
Only Refsum treatable--

Neuronal ceroid lipofuscinoses (aka Batten dz)

Progressive neuro decline--
Death teens/early adult--

Ciliopathies

--Bardet-Biedl syndrome
--Alström syndrome
--Joubert syndrome
--Senior-Løken syndrome

Cilia JABS you in the eye--
Relentless renal failure--
‘Molar tooth sign’ in Joubert--

Acanthocytosis of RBCs--
Abetalipoproteinemia
(aka Bassen-Korzweig dz)

These are my best guesses (emphasis on guesses) regarding factoids that should be kept in mind for each. Toggle!
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

Deaf/blind--
- Usher syndrome
- Peroxisomal disorders
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Infantile Refsum dz

Elevated VLCFAs--
Dreadful prognoses--
Only Refsum treatable--

Progressive neuro decline--
Death teens/early adult--

Cilia JABS you in the eye--
Relentless renal failure--
‘Molar tooth sign’ in Joubert--

Acanthocytosis of RBCs--

Neuronal ceroid lipofuscinoses (aka Batten dz)

Ciliopathies
- Bardet-Biedl syndrome
- Alström syndrome
- Joubert syndrome
- Senior-Løken syndrome

Abetalipoproteinemia
(aka Bassen-Korwelzeg dz)

‘Pseudo-RP’

- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  -- Toxoplasmosis
  -- HSV
  -- Others
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
- Severe uveitis
- DUSN
- CRAO
- Drug toxicity
  -- Hydroxychloroquine
  -- Thioridazine
  -- Others

Speaking of guesses…
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

Usher syndrome

Peroxisomal disorders
- Zellweger syndrome
- Neonatal adrenoleukodystrophy
- Infantile Refsum dz

Neuronal ceroid lipofuscinoses (aka Batten dz)

Ciliopathies
- Bardet-Biedl syndrome
- Alström syndrome
- Joubert syndrome
- Senior-Løken syndrome

Abetalipoproteinemia
(aka Bassen-Korzyweig dz)

Elevated VLCFAs--
Dreadful prognoses--
Only Refsum treatable--

Progressive neuro decline--
Death teens/early adult--

Congenital syphilis

Congenital rubella

Infectious retinitis
- Toxoplasmosis
- HSV
- Others

Cancer-associated retinopathy

Kearns-Sayre syndrome

Severe uveitis

DUSN

CRAO

Drug toxicity
- Hydroxychloroquine
- Thioridazine
- Others

Speaking of guesses…These are the causes of pseudo-RP I would pay particular attention to.
DDx for an RP-like Fundus

Primary RP (aka typical RP)
- Deaf/blind--
- Elevated VLCFAs--
- Dreadful prognoses--
- Only Refsum treatable--
- Progressive neuro decline--
- Death teens/early adult--

Secondary RP (aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Infantile Refsum dz
- Neuronal ceroid lipofuscinoses (aka Batten dz)
- Ciliopathies
  - Bardet-Biedl syndrome
  - Alström syndrome
  - Joubert syndrome
  - Senior-Løken syndrome
- Abetalipoproteinemia
  (aka Bassen-Korzweig dz)

‘Pseudo-RP’
- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  --Toxoplasmosis
  --HSV
  --Others
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
- Severe uveitis
- DUSN
- CRAO
- Drug toxicity
  --Hydroxychloroquine
  --Thioridazine
  --Others

Note: I’m not suggesting the other conditions are low-yield topics for the OKAP. Rather, I think they are unlikely to be the correct answer to a question intended to test your knowledge of RP imitators.

Speaking of guesses…These are the causes of pseudo-RP I would pay particular attention to.