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 300).

Now, let’s get this bread!
DDx for an
RP-like Fundus

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?
DDx for an 
RP-like Fundus

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)
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?
--Nyctalopia
--Visual field loss

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

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

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

*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' eponym-eponym)

- Ciliopathies

- Abetalipoproteinemia
  (aka 'Bassen-Kornzweig dz' 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

- 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* +

- 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 + sensorineural deafness

- 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?

- 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’

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

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  --aka ‘Bassen-Kornzweig dz’
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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?

- 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’

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’
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(aka typical RP)

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- 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…
--Type II
--Type III

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

Primary RP

(aka typical RP)

Secondary RP

(aka Complex RP; Syndromic RP)

- Usher syndrome
- Peroxisomal disorders

‘Pseudo-RP’

- 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**
- **Type III**
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 + sensorineural deafness**

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

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‘Pseudo-RP’

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It is the most common cause thereof

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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... 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)

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- **Type III** has... hearing loss; the RP severity? in severity; vestibular function is

- 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* + sensorineural deafness

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It is the most common cause thereof

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---**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’**

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

---

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

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*There are three types of Usher syndrome—what are they called? How do they manifest?*
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- **Type III** has...progressive hearing loss; the RP varies in severity; vestibular function is sporadic

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

Peroxisomal disorders are a heterogeneous group of disorders of peroxisome function. They are characterized by intracellular organelles that play key roles in many aspects of cell metabolism. The hallmark lab abnormality of peroxisomal disorders that present with LCA is abnormally high serum levels of very long chain fatty acids (VLCFA). Specific peroxisomal disorders that can manifest an LCA-type presentation include Zellweger syndrome, Neonatal adrenoleukodystrophy (NALD), Infantile Refsum disease, Usher syndrome, and Neuronal ceroid lipofuscinoses.
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)
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 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?
--
--
--
**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?**

--Zellweger syndrome
--Neonatal adrenoleukodystrophy (NALD)
--Infantile Refsum 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

‘Pseudo-RP’

What is the nonponymous name for Zellweger 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

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’

What is the non-pseudonym for Zellweger syndrome?
Cerebrohepatorenal 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
- Neuronal ceroid lipofuscinoses
- Ciliopathies
- Abetalipoproteinemia

‘Pseudo-RP’

What is the non-eponymous name for Zellweger syndrome? Cerebrohepatorenal syndrome

What is its inheritance pattern?

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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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‘Pseudo-RP’

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‘Pseudo-RP’

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In the neonatal period
DDx for an RP-like Fundus

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‘Pseudo-RP’

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:
--?
--?
--?
--?
--?
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)
DDx for an RP-like Fundus

Zellweger syndrome facies: High forehead; hypertelorism
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:
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‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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How do Zellweger syndrome pts present? In the neonatal period with:
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--Deafness
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--Seizures
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Secondary RP (aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy

‘Pseudo-RP’

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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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‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
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How do Zellweger syndrome pts present?
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-- Deafness
-- Hypotonia
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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)
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’

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

How is Zellweger managed?

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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‘Pseudo-RP’

What is the non-eponymous 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

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

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

- **Primary RP**
  - (aka typical RP)
  - --Zellweger syndrome
  - --Neonatal adrenoleukodystrophy
  - --Infantile Refsum dz

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

- **'Pseudo-RP'**

---

**What is the nonepithelial form of 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 age **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
- Neuronal ceroid lipofuscinoses
- Ciliopathies
- Abetalipoproteinemia

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
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'age'
Secondary RP

(aka Complex RP; Syndromic RP)

- Usher syndrome
- Peroxisomal disorders
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
- Neuronal ceroid lipofuscinoses
- Ciliopathies
- Abetalipoproteinemia

Primary RP

(aka typical RP)

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

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Note: NALD’s onset occurs a little later than does Zellweger’s
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Note that NALD has the same S/S as Zellweger, except it’s missing the last one on the list
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And albeit dreadful, the prognosis for NALD is better than that for Zellweger
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**What is the non-epithelial Cerebrohepatorenal syndrome?**
- Neonatal adrenoleukodystrophy

**What is its inheritance pattern?**
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**How do Zellweger syndrome patients present?**
- In the neonatal period with:
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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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  - In NALD, the VLCFAs impair the function of adrenal glands by compromising the integrity of adrenal-cell membranes
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- Infantile Refsum dz = adult-onset Refsum?

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

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‘Pseudo-RP’
- Infantile Refsum dz = adult-onset Refsum? **No!**

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

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Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

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

How do Zellweger syndrome pts present?
In the neonatal period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures
-- Abnormal facies
What is its inheritance pattern?
AR
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 its inheritance pattern?
AR
What is the prognosis?
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-- Deafness
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What is the prognosis?
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Primary RP
(aka typical RP)

Secondary RP
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- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum disease
  aka...Infantile phytanic acid storage disease

‘Pseudo-RP’

**What is the noneponymous 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

**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 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)
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’

### Primary 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

### Secondary 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**

### Infantile Refsum dz

#### 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’

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? It is uniformly fatal by age 1 year.

What is the noneponymous name for Zellweger syndrome? Cerebrohepatorenal syndrome

What is its inheritance pattern? AR

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)
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
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- Abnormal facies (high forehead; hypertelorism)

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

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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 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)

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
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-- Deafness
-- Hypotonia
-- Seizures

What is the prognosis?
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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
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In the late infancy period with:
- RP-like fundus
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- Hypotonia
- Seizures

What is the prognosis?
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- Peroxisomal disorders
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Infantile Refsum dz (aka... Infantile phytanic acid storage dz)

'Pseudo-RP'

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Cerebrohepatorenal syndrome

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In the late infancy period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures

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

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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

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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 dz pts present?
In the early childhood period with:
- ?
- ?
- ?

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
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‘Pseudo-RP’

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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 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.
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 (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
- Seizures

What is the prognosis?

It is uniformly fatal by *early adulthood* (if treatment is unsuccessful)

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**)
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

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

‘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

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

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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’

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 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 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’**

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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* (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 *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’

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

Is Zellweger syndrome the same condition as infantile Refsum disease?
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.

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

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

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

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal 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’

- 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 (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

And just as the prognosis for NALD was better than for Zellweger, so too in turn is the prognosis for infantile Refsum disease 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
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-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures
-- Abnormal facies

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
-- 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
Pseudo-RP

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 non-epithelial 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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- RP-like fundus
- Deafness
- Hypotonia
- Seizures

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

By what non-epithelial 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)

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)
'Pseudo-RP'
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

---

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

**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 late childhood

---

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

**Refsum disease known?**
Yes

**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 early adulthood (if treatment is unsuccessful)

---

**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
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 Infantile Refsum disease known?
Via elevated serum phytanic acid levels (and VLCFAs)

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

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 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??!!

How is infantile Refsum treated?
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

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

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

How is infantile Refsum disease known?

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

‘Pseudo-RP’

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

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
- Infantile Refsum NALD
- 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
  --Batten dz
  --Bassen-Kornzweig dz
  --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’

- Abetalipoproteinemia

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

Infantile Refsum
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

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

Infantile Refsum
Zellweger syndrome
Worse prognosis

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

‘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

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
  --Batten dz
  --Bassen-Kornzweig dz
  --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’

- Usher syndrome
- Peroxisomal disorders
- Only infantile Refsum is treatable

RP-like fundus

Deafness

Hypotonia

Seizures

Abnormal facies

Peroxisomal disorders tl;dr

(Review slides--no questions)
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

‘Pseudo-RP’
- Abetalipoproteinemia
 -- aka ‘Bassen-Kornzweig dz’
- -- Zellweger syndrome
- -- Neonatal adrenoleukodystrophy
- -- Infantile Refsum dz

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

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

What is 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 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 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 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 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
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:
- 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
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 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?

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
  -- 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

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

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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
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
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What is the prognosis?
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It is a mélange of imaging, genetic and other tests. If asked, punt to a geneticist.
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?
Abetalipoproteinemia (Batten dz; ‘Bassen-Kornzweig dz’; 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)

‘Pseudo-RP’

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

What is the prognosis?

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

How is Batten dz managed?

Supportively

How is Batten dz diagnosed?

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

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

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)

Pseudo-RP

Neuronal ceroid lipofuscinoses

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

Neuronal ceroid lipofuscinoses (NCLs; collectively referred to as Batten dz):

Neurodegenerative conditions stemming from the buildup of ceroid & lipofuscin in cells
What is a ciliopathy?
What is a ciliopathy?
An inherited condition marked by abnormal structure and/or function of cilia
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
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?
An inherited condition marked by abnormal structure and/or function of cilia

Cilia are organelles that protrude from the cell surface and are found in the
The eyes

The eyes??!! Which part of the eye contains cilia wiggling about?
What is a ciliopathy?
An inherited condition marked by abnormal structure and/or function of cilia.

Cilia are organelles found in many organs, including the eye, brain, and kidney.

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 **non**motile 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

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, liver, and kidney. How do you know? 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)
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  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka ‘Batten dz’
- Ciliopathies

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 one word disorder.

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**

---

**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.

**Cilia are organized in the eye, brain, skin, etc.**

**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
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  --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 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).

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
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  -- 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
  --Neonatal adrenoleukodystrophy
  --Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  --aka ‘Batten dz’
- **Ciliopathies**
  --Bardet-Biedl syndrome
  --Alström syndrome
  --Joubert syndrome
  --Senior-Løken syndrome

‘Pseudo-RP’

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
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka 'Batten dz'
- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome

‘Pseudo-RP’

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

Note that all of the ciliopathies are marked by relentlessly progressive renal failure resulting in ESRD early in life!
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Got a mnemonic for remembering the ciliopathies?
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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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- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

What findings define the Bardet-Biedl complex?
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---
---

The mnemonic is:...
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‘Pseudo-RP’

What findings define the Bardet-Biedl complex?
- H
- O
- M
- E
- R

(as in Simpson)
Not surprisingly, the ‘R’ stands for RP-like fundus.

As for the rest…
Pseudo-RP

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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…
DDx for an RP-like Fundus

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What findings define the Bardet-Biedl complex?
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- Extra fingers (polydactyly)
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And yeah, I know, Homer only has four digits per hand--paucidactyly, not polydactyly. But the rest fits him pretty well.
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- Hypogonadism?
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- Extra fingers (polydactyly)?
- RP-like fundus

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

‘Pseudo-RP’
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- Mental retardation
- Extra fingers (polydactyly)
- RP-like fundus

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

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

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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?
  - Cardiomyopathy
  - Early-onset type 2 DM

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

Both occur in childhood to teens
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‘Pseudo-RP’

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

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‘Pseudo-RP’

How are BBS and AS managed? Supportively
Pseudo-RP

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‘Pseudo-RP’

DDx for an RP-like Fundus

What are the main nonocular structures affected in JS?

What classic MRI finding is the hallmark of JS?

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
DDx for an RP-like Fundus

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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?
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‘Pseudo-RP’

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Joubert syndrome: Molar-tooth sign (look at the brainstem)
What are the main nonocular structures affected in JS?
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--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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It is highly variable

What is the management of JS?
Supportive

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The brainstem and cerebellum

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How do JS pts present?
How is JS managed? Supportively

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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.
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‘Pseudo-RP’

For completeness’ sake:
In three words (including syndromes), what sort of condition are these ciliopathies?
They are Ciliopathies
- Bardet-Biedl syndrome
- Alström syndrome
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Abetalipoproteinemia
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DDx for an RP-like Fundus

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

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How many familial oculorenal syndromes are there?
Including the four ciliopathies–six
What are the other two?
Alport syndrome and Lowe syndrome
DDx for an RP-like Fundus

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Are Alport and Lowe syndromes ciliopathies?
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Are Alport and Lowe syndromes ciliopathies? No
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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

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

What are the other two?
Alport syndrome and Lowe 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

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
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- Usher syndrome
- Peroxisomal disorders
- Neuronal ceroid lipofuscinoses
- Ciliopathies
- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’
- Bardet-Biedl syndrome
- Alström syndrome
- Joubert syndrome
- Senior-Løken syndrome

For completeness’ sake:
In three words (including syndromes), what sort of condition 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

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

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic)

- Usher syndrome
- Peroxisomal disorders

‘Pseudo-RP’

- Zellweger syndrome
- Neonatal adrenoleukodystrophy
- Infantile Refsum dz
- aka ‘Batten dz’

Secondary RP

- Usher syndrome
- Peroxisomal disorders
- Neuronal ceroid lipofuscinoses
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Ciliopathies
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Abnormalities of what intraocular structure are associated with Alport and Lowe syndromes?
DDx for an RP-like Fundus

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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
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
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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

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

What lens pathologies occur with Alport and Lowe syndromes?
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What are the other two?
Alport syndrome and Lowe syndrome
DDx for an RP-like Fundus

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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

What lens pathologies occur with Alport and Lowe syndromes?
-- Lenticonus
-- 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
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

For completeness’ sake:
In three words (including syndromes), what sort of condition are these ciliopathies?
They are familial oculoventral 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?
- Lenticonus?
- Cataracts?
- Microspherophakia?

Are these associated with the ciliopathies?
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’

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

For completeness’ sake:

In three words (including syndromes), what sort of condition are these ciliopathies?

They are familial oculo renal syndromes

What are the other two?

Alport syndrome and Lowe syndrome

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

No

Are these associated with the ciliopathies?

- Lenticonus
- Cataracts
- Microspherophakia

Are these associated with the ciliopathies?

- Lenticonus
- Cataracts
- Microspherophakia

What lens pathologies occur with Alport and Lowe syndromes?
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 Oculo-renal Syndromes *tl;dr*

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

- Not Ciliopathies
  - Alport syndrome
  - Lowe syndrome
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
- 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

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 *tl;dr*

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

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

**Key Features**

<table>
<thead>
<tr>
<th>Renal failure</th>
<th>Classic eye finding:</th>
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<tr>
<td><em>without</em> hematuria</td>
<td><em>Pigmentary retinopathy</em></td>
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Familial Oculorenal Syndromes

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

Not Ciliopathies

Renal failure
- without hematuria

Classic eye finding:
- Pigmentary retinopathy

Inheritance:

Renal failure
- with hematuria

Classic eye finding:
- Lenticonus

Inheritance:
Familial Oculorenal Syndromes *tl;dr*

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:
*AR*

Renal failure
*with* hematuria

Classic eye finding:
*Lenticonus*

Inheritance:
*X-linked*
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: AR

For more info on Alport and Lowe syndromes, see slide-set L4

Renal failure
- with hematuria
- Classic eye finding: Lenticulus
- Inheritance: X-linked
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
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  -- aka ‘Batten dz’
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  -- 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’
What is the underlying problem in abetalipoproteinemia?
One of the lipoproteins (ApoB) is not synthesized by the body

---Senior-Løken syndrome

- **Abetalipoproteinemia**
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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

**How is abetalipoproteinemia inherited?**
AR

**How does the absence of ApoB lead to secondary RP?**

- 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).

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How is it diagnosed?

How is it treated?
With supplementary vitamins A & E

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Checking vitamin A levels is a good start

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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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Acanthocytosis of the RBCs

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DDx for an RP-like Fundus

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

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- 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 hypoabetalipoproteinemia a thing?
In abetalipoproteinemia, \( \beta \)-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

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

*Is a-*alpha*-lipoproteinemia a thing?*
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
- A\text{beta} lipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

Is a \text{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)

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  -- Joubert syndrome
  -- Senior-Løken syndrome
- A-beta lipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

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

Does the absence of α-lipoprotein affect the retina?
DDx for an RP-like Fundus

Primary RP (aka typical RP)

Secondary RP (aka Complex RP; Syndromic RP)
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  -- Zellweger syndrome
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- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Alpha-lipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

Is 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
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- Usher syndrome
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  -- Zellweger syndrome
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- Alpha-lipoproteinemia
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‘Pseudo-RP’

*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

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(aka typical 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’

‘Pseudo-RP’

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
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Low levels of α-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'

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.
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
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

Infection
DDx for an RP-like Fundus

Primary RP (aka typical 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’

Secondary RP (aka Complex RP; Syndromic RP)
- Congenital syphilis
- Congenital rubella

‘Pseudo-RP’
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

- 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
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- Neuronal ceroid lipofuscinoses
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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
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

---

Secondary RP:

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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
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  --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
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.
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’
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  --Bardet-Biedl syndrome
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  --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

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- Usher syndrome
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  -- Zellweger syndrome
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  -- aka ‘Batten dz’
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  -- Joubert syndrome
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‘Pseudo-RP’
- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  -- Toxoplasmosis
  -- HSV
  -- Others
- Cancer-associated retinopathy
- Kearns-Sayre syndrome

-- Zellweger syndrome
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DDx for an RP-like Fundus

Primary RP
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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?

- Cardiac conduction abnormalities
- Ophthalmoplegia (usually the ptosis)
- Others

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)

--aka ‘Bassen-Kornzweig dz’

--aka ‘Bassen-Kornzweig dz’
What is the classic triad of Kearns-Sayre syndrome?
--Pigmentary retinopathy
--CPEO
--Cardiac conduction abnormalities

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Kears-Sayre syndrome
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.
DDx for an RP-like Fundus

Primary RP (aka typical RP)

Secondary RP

‘Pseudo-RP’

Congenital syphilis
Congenital rubella
Infectious retinitis
Cancer-associated retinopathy

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

What does CPEO stand for?
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--aka ‘Bassen-Kornzweig dz’

--aka ‘Senior-Løken syndrome’

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Toxoplasmosis
HSV
Others

Kearns-Sayre syndrome
DDx for an RP-like Fundus

Primary RP (aka typical RP)

Secondary RP

‘Pseudo-RP’

- Congenital syphilis
- Congenital rubella
- Infectious retinitis
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
- Usher syndrome
- Peroxisomal disorders
- Neuronal ceroid lipofuscinoses
- Ciliopathies
- Abetalipoproteinemia

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

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Which EOMs are typically affected first?

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

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

Secondary RP
- Usher syndrome
- Peroxisomal disorders
- Neuronal ceroid lipofuscinoses
- Ciliopathies

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

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DDx for an RP-like Fundus

CPEO
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’

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  

In very general terms, what sort of disease is K-SS?  
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The ophthalmoplegia (usually the ptosis)

Are the cardiac-conduction problems serious?  
Yes, they can be life-threatening
What is the classic triad of Kearns-Sayre syndrome?

--Pigmentary retinopathy
--CPEO
--Cardiac conduction abnormalities

Are the cardiac-conduction problems serious?
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DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP

‘Pseudo-RP’

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

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--CPEO
--Cardiac conduction abnormalities

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Kearns-Sayre syndrome
--aka ‘Bassen-Kornzweig dz’
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

Kears-Sayre syndrome

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

Primary RP
(aka typical RP)

Secondary RP

‘Pseudo-RP’

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

Kearns-Sayre syndrome

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-- CPEO
-- Cardiac conduction abnormalities

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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
--Toxoplasmosis
--HSV
--Others
Cancer-associated retinopathy

Kearns-Sayre syndrome

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--CPEO
--Cardiac conduction abnormalities

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A mitochondrial disease

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

--aka ‘Bassen-Kornzweig dz’

--Zellweger syndrome
--Neonatal adrenoleukodystrophy
--Infantile Refsum dz
--Bardet-Biedl syndrome
--Alström syndrome
--Joubert syndrome
--Senior-Løken syndrome

--aka ‘Batten dz’

--aka ‘Bassen-Kornzweig dz’

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

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

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--CPEO
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A mitochondrial disease

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

--aka ‘Bassen-Kornzweig dz’
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--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?

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

**Primary RP**
(aka typical RP)

**Secondary RP**

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

**‘Pseudo-RP’**

- Kearns-Sayre syndrome
- Usher syndrome
- Peroxisomal disorders
- Neuronal ceroid lipofuscinoses
- Ciliopathies
- Abetalipoproteinemia

**Primary RP**
(aka typical RP)

**Secondary RP**

- Zellweger syndrome
- Neonatal adrenoleukodystrophy
- Infantile Refsum dz
- Bardet-Biedl syndrome
- Alström syndrome
- Joubert syndrome
- Senior-Løken syndrome

--aka 'Bassen-Kornzweig dz'

**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

**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?
‘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?
--aka ‘Bassen-Kornzweig dz’
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)

--aka ‘Bassen-Kornzweig dz’
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

---

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

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A mitochondrial disease

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

At what age do symptoms begin occurring?
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Of the classic triad, which is the first to occur?
The ophthalmoplegia (usually the ptosis)

Why is the order important?

Because in making the proper diagnosis, the astute ophthalmologist can refer the pt to a cardiologist before s/he has a fatal dysrhythmia.

---

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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?
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Kearns-Sayre 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
- 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

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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
- Kearns-Sayre syndrome
- Severe uveitis

four-letter abb. for infectious cause
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
- Kearns-Sayre syndrome
- Severe uveitis
- DUSN
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
  
  **What does DUSN stand for?**
- DUSN

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

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

What does DUSN stand for?
Diffuse unilateral subacute neuroretinitis
**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
  "What does DUSN stand for?"
  "Diffuse unilateral subacute neuroretinitis"

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

---

**What is the cause?**
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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- Ciliopathies
  -- Bardet-Biedl syndrome

What does DUSN stand for?
- Diffuse unilateral subacute neuroretinitis

What is the cause?
Infestation by a bug (most commonly, specific bug)
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
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  -- What does DUSN stand for?
    -- Diffuse unilateral subacute neuroretinitis

‘Pseudo-RP’
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  -- Others
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- 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

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

Primary RP

(aka typical RP)

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(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

What does DUSN stand for?
Diffuse unilateral subacute neuroretinitis

When should you consider that a case of ‘RP’ might be DUSN?

‘Pseudo-RP’

- Congenital syphillis
- Congenital rubella
- Infectious retinitis
  --Toxoplasmosis
  --HSV
  --Others
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
- Severe uveitis
- DUSN

Toxoplasmosis

HSV
DDx for an RP-like Fundus

Primary RP (aka typical RP)
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  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
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- Ciliopathies
  -- Bardet-Biedl syndrome

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’

‘Pseudo-RP’
- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  -- Toxoplasmosis
  -- HSV
  -- Others
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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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- CRAO
- Drug toxicity
  --?
  --?
  --Others
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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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    --Thioridazine
    --Others

‘Pseudo-RP’
- Hydroxychloroquine (Plaquenil) toxicity is addressed in detail in its own slide-set

Note: Hydroxychloroquine (Plaquenil) toxicity is addressed in detail in its own slide-set
DDx for an RP-like Fundus

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Drug toxicity
--Hydroxychloroquine
--Thioridazine
--Others

What class of medicine is thioridazine?

It is a phenothiazine.

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.
Primary RP (aka typical RP)

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

**Drug toxicity**
--Hydroxychloroquine
--Thioridazine
--Others
What class of medicine is thioridazine?
It is a phenothiazine

What are the phenothiazines used to treat?

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

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What are the phenothiazines used to treat?
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Drug toxicity
-- Hydroxychloroquine
-- Thioridazine
-- Others
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Drug toxicity
-- Hydroxychloroquine
  -- Others
  -- Thioridazine
  -- Others
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  -- Bardet-Biedl syndrome
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- CRAO
- Drug toxicity
  -- Hydroxychloroquine
  -- Thioridazine
  -- Others

What tests should be run to determine whether a pt has RP?
--
--
--
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

Primary RP
(aka typical RP)

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- CRAO
- Drug toxicity
  --Hydroxychloroquine
  --Thioridazine
  --Others

How do you rule-in pseudo-RP?

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

Primary RP
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Secondary RP
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- Usher syndrome
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‘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
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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…
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‘Pseudo-RP’
- Congenital syphilis

How do you rule-in the causes of secondary RP?

-- Usher syndrome:

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**
- Peroxisomal disorders
  --Zellweger syndrome
  --Neonatal adrenoleukodystrophy
  --Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  --aka ‘Batten dz’
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  --Bardet-Biedl syndrome
  --Alström syndrome
  --Joubert 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: Molar-tooth sign on brainstem MRI
  - Senior-Løken: Check renal

---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

How do you rule-in the causes of secondary RP?

--Usher syndrome: Check hearing

--The peroxisomal disorders:
DDx for an RP-like Fundus

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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

-- Toxoplasmosis
-- HSV
-- Others

-- Hydroxychloroquine
-- Thioridazine
-- Others

-- Usher syndrome: Check hearing

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Usher syndrome

Neuronal ceroid lipofuscinoses

Ciliopathies

Abetalipoproteinemia

Primary RP

Secondary RP

‘Pseudo-RP’

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How do you rule-in the causes of secondary RP?

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-- The peroxisomal disorders: Check serum levels of ‘very long chain fatty acids’ +/- phytanic acid levels

-- Batten disease: Punt to a geneticist
How do you rule-in the causes of secondary RP?

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-- 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?

- **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
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-- 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*...)

**DDx for an RP-like Fundus**

? ? ?
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

‘Pseudo-RP’

(When you hear RP-like fundus...these three categories should instantly spring to mind—make sure they do!)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- ?
- ?
- ?
- ?
- ?

‘Pseudo-RP’

Next, make sure you can name the five categories of secondary/syndromic 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)
- Ciliopathies
- Abetalipoproteinemia
  (aka Bassen-Korzyweig dz)

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!
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)

‘Pseudo-RP’

Then, make sure you can name the three *peroxisomal disorders* and the four *ciliopathies*. 
DDx for an RP-like Fundus

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(aka typical RP)

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‘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.
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(aka typical RP)

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  (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)

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‘Pseudo-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--
- Neuronal ceroid lipofuscinoses (aka Batten dz)

Cilia JABS you in the eye--
Relentless renal failure--
‘Molar tooth sign’ in Joubert--

Acanthocytosis of RBCs--
- Ciliopathies
  - Bardet-Biedl syndrome
  - Alström syndrome
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Abetalipoproteinemia
(aka Bassen-Korzweig dz)

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  - --Senior-Løken syndrome
- Acanthocytosis of RBCs--
- Abetalipoproteinemia
  (aka Bassen-Korzwieg 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

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Speaking of guesses…These are the causes of pseudo-RP I would pay particular attention to.
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DUSN
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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.