In my experience, this is one of the most challenging topics in ophthalmology. (The corneal dystrophies are a close second.) While I won’t say I’ve made it easy (there's simply too much esoteric detail for that), I think the organization of this slide-set makes the material manageable. 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 a few times a month. Once you hit the frantic, final few weeks of cramming, stop doing the entire set—just do the *tl;dr* at the end (it starts around slide 300).

Now, let’s get this bread!
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)
RP
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)
- (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 Neuronopathies
- Ciliopathies

- Abetalipoproteinemia
  --aka Abetalipoproteinemia
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)
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‘Pseudo-RP’

Usher syndrome = Retinitis pigmentosa +

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

Primary RP
(aka typical RP)

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(aka Complex RP; Syndromic RP)
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Usher syndrome = Retinitis pigmentosa + sensorineural deafness

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

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

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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?

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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?
- Type I
- Type II
- Type III

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

Secondary RP

(aka Complex RP; Syndromic RP)

- Usher syndrome
- Peroxisomal disorders

Primary RP

(aka typical RP)

‘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**

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  --aka ‘Bassen-Kornzweig dz’
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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**
-- **Type III**

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--Type I manifests...in the first decade with profound hearing loss, RP and vestibular dysfunction
--Type II manifests...in the decade with hearing loss, RP; vestibular function is
--Type III

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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

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--Type II manifests...in the second decade with moderate hearing loss, RP; vestibular function is intact
--Type III has...

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It is the most common cause thereof

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

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  --aka ‘Bassen-Kornzweig dz’
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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

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

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‘Pseudo-RP’

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

There are three types of Usher syndrome—what are they called? How do they manifest?
-- Type I early, severe
-- Type II later, less severe
-- Type III variable

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

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

Primary RP
(aka typical RP)

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- **Peroxisomal disorders**
- Neuronal ceroid

What are peroxisomal disorders?
DDx for an RP-like Fundus

Primary RP (aka typical RP)

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- Usher syndrome
- **Peroxisomal disorders**
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy (NALD)
  - Infantile Refsum dz

‘Pseudo-RP’

What are peroxisomal disorders?
A heterogeneous group of disorders of peroxisome function
DDx for an RP-like Fundus

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

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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
DDx for an RP-like Fundus

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

What is the hallmark lab abnormality of the peroxisomal disorders that present with LCA?
(LCA = Leber’s congenital amaurosis, an age-related form of RP)
What are peroxisomal disorders?
A heterogeneous group of disorders of peroxisome function

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

What is the hallmark lab abnormality of the peroxisomal disorders that present with LCA?
Abnormally high serum levels of very long chain fatty acids (VLCFA)
DDx for an RP-like Fundus

Primary RP (aka typical RP)

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

‘Pseudo-RP’

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

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

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

**What specific peroxisomal disorders can manifest an LCA-type presentation?**
--
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‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?

Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

What is the prognosis?
It is uniformly fatal by age 1 year
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(aka typical RP)

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

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It is uniformly fatal by age 1 year

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

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

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‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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:
- LCA
- Deafness
- Hypotonia
- Seizures

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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What is its inheritance pattern?
AR

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-- 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:
-- LCA
-- Deafness
-- Hypotonia
-- Seizures

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
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- **Peroxisomal disorders**
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  - Neonatal adrenoleukodystrophy  
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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:
- LCA
- Deafness
- Hypotonia
- Seizures
- Abnormal facies (high forehead; hypertelorism)

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

**Note:** Both are inherited AR

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:
- LCA
- Deafness
- Hypotonia
- Seizures

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

Note: Both are inherited AR
DDx for an RP-like Fundus

- 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
        - aka 'Batten dz'
        - aka 'Bassen-Kornzweig dz'
      - Bardet-Biedl syndrome
      - Alström syndrome
      - Joubert syndrome
      - Senior-Løken syndrome

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
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- Hypotonia
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- Seizures

What is the prognosis?
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- Deafness
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What is its inheritance pattern?
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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

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In the late infancy period with:
- LCA
- Deafness
- Hypotonia
- Seizures

What is the prognosis?
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Secondary RP

(aka Complex RP; Syndromic RP)

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- Peroxisomal disorders
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Infantile Refsum dz

Primary RP

(aka typical RP)

- Zellweger syndrome
- Neonatal adrenoleukodystrophy
- Infantile Refsum dz

What is the noneponymous name for Zellweger syndrome?

Cerebrohepatorenal syndrome

What is its inheritance pattern?

AR

How do Zellweger syndrome pts present?

In the neonatal period with:
- LCA
- Deafness
- Hypotonia
- Seizures
- Abnormal facies

What is the prognosis?

It is uniformly fatal by age 1 year

Is NALD the same condition as adrenoleukodystrophy?

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

What is its inheritance pattern?

AR

How do NALD pts present?

In the late infancy period

What is the prognosis?

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  -- Zellweger syndrome
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- Abetalipoproteinemia

‘Pseudo-RP’

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Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger 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

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
- Seizures

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

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

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- 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:
-?
-?
-?
-?

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

- Abetalipoproteinemia

‘Pseudo-RP’

- Ciliopathies
- Neuronal ceroid lipofuscinoses

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:
  - LCA
  - Deafness
  - Hypotonia
  - Seizures
  - Abnormal facies

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

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What is its inheritance pattern?
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How do NALD pts present?
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‘Pseudo-RP’

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Cerebrohepatorenal syndrome

What is its inheritance pattern?
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In the neonatal period with:
-- LCA
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-- Seizures
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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:
-- LCA
-- Deafness
-- Hypotonia
-- Seizures

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

Secondary 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:
-- LCA
-- Deafness
-- Hypotonia
-- Seizures
-- Abnormal facies

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

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

What is its inheritance pattern?
AR

How do NALD pts present?
In the late infancy period with:
-- LCA
-- Deafness
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‘Pseudo-RP’

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

What is its inheritance pattern?
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- LCA
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- 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:
- LCA
- Deafness
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And albeit dreadful, the prognosis for NALD is better than that for Zellweger

It is uniformly fatal by late childhood
DDx for an RP-like Fundus

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

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
DDx for an RP-like Fundus

Primary RP (aka typical RP)

Secondary RP (aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  - Zellweger syndrome
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- Ciliopathies
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Why does neonatal ADRENOleukodystrophy have adreno- in the name?
In NALD, the VLCFAs impair the function of adrenal glands by compromising the integrity of adrenal-cell membranes

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

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

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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.

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

What is its inheritance pattern? AR.

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

How are NALD pts diagnosed? Supportively (just like Zellweger pts).
DDx for an RP-like Fundus

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(aka typical RP)

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What is its inheritance pattern?
AR

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In the neonatal period with:
- LCA
- Deafness
- Hypotonia
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What is the prognosis?
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Cerebrohepatorenal syndrome

What is its inheritance pattern?
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- LCA
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- LCA
- Deafness
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What is the prognosis?
It is uniformly fatal by late childhood
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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 dz = adult-onset Refsum?

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

What is its inheritance pattern?
AR

How do Zellweger pts present?
In the neonatal period with:
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

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

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

What is its inheritance pattern?
AR

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

What is the prognosis?
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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 = adult-onset Refsum? No!

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger pts present?
In the neonatal period with:
-- LCA
-- 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

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

What is its inheritance pattern?
AR

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
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-- LCA
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What is its inheritance pattern?
AR

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In the late infancy period with:
-- LCA
-- 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:
-- LCA
-- 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 phytic acid storage dz

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

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

What is its inheritance pattern?
AR

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

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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:
-- LCA
-- 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:
-- LCA
-- 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:
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies (high forehead; hypertelorism)

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

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

What is its inheritance pattern?
AR

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

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

By what noneponymous name is infantile Refsum disease known?
Infantile phytanic acid storage 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 (aka...Infantile phytanic acid storage dz)

‘Pseudo-RP’

---

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

**What is its inheritance pattern?**
AR

**How do Zellweger pts present?**
- LCA
- Deafness
- Hypotonia
- Seizures
- Abnormal facies

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

---

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

**What is its inheritance pattern?**
AR

**How do NALD pts present?**
- LCA
- Deafness
- Hypotonia
- Seizures

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

---

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger’s 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

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:
-- LCA
-- 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’s pts present?
In the early childhood period with:
-- LCA
-- Deafness
-- Hypotonia

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

- 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:
-- LCA
-- 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:
-- LCA
-- 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:
-- LCA
-- Deafness
-- Hypotonia

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

What is the noneponymous name of Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

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

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

What is its inheritance pattern?
AR

How do Infantile Refsum dz pts present?
In the late infancy period with:
-- LCA
-- 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 'age' period
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**

**Secondary RP**

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

**Primary RP**

- Zellweger syndrome
- Neonatal adrenoleukodystrophy
- Infantile Refsum disease

---

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
- LCA
- 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:
- LCA
- Deafness
- Hypotonia
- Seizures

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

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

What is its inheritance pattern?
AR

How do infantile Refsum disease pts present?
In the early childhood period with:
- LCA
- 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’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

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:
--LCA
--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:
--LCA
--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:
- LCA
- 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:
- LCA
- 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
  --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 the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

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:
--LCA
--Deafness
--Hypotonia

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

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

What is its inheritance pattern?
AR

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

What is the prognosis?
It is uniformly fatal by the 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’***

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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:
  - LCA
  - 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:
  - LCA
  - 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:
  - LCA
  - 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’

Usher syndrome

Peroxisomal disorders

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

What is the noneponymous name for Zellweger syndrome?

Cerebrohepatorenal syndrome

What is its inheritance pattern?

AR

How do Zellweger syndrome pts present?

In the neonatal period with:
- LCA
- Deafness
- Hypotonia
- Seizures
- Abnormal facies

What is the prognosis?

It is uniformly fatal by age 1 year

Is NALD the same condition as adrenoleukodystrophy?

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

What is its inheritance pattern?

AR

How do NALD pts present?

In the late infancy period with:
- LCA
- Deafness
- Hypotonia
- Seizures

What is the prognosis?

It is uniformly fatal by late childhood

By what noneponymous names is infantile Refsum disease known?

- Infantile phytanic acid storage 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:
- LCA
- 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:
- LCA
- 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:
- LCA
- 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:
- LCA
- 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’

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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:
-- LCA
-- 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:
-- LCA
-- 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:
-- LCA
-- 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’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

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:
-- LCA
-- Deafness
-- Hypotonia
-- Seizures

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

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

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

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

What is 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:
-- LCA
-- 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

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

And just as the prognosis for NALD was better than for Zellweger, so too in turn is the prognosis for infantile Refsum better than that for NALD

Is NALD the same condition as Zellweger?
No, that is an X-linked condition that presents later in 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:
-- LCA
-- Deafness
-- Hypotonia
-- Seizures

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

It is uniformly fatal by late childhood

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’

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**Is NALD the same condition as Zellweger syndrome?**

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

---

**By what non-eponymous name is infantile Refsum disease known?**

Infantile phytanic acid storage disease

---

**What is its inheritance pattern?**

AR

---

**What is its inheritance pattern?**

AR

---

**What is the prognosis?**

It is uniformly fatal by age 1 year

---

**So infantile Refsum and NALD are quite similar, except that infantile Refsum is a somewhat milder condition:**

- Later onset
- One fewer S/S (= no seizures)
- Pts live longer

---

**What is the prognosis?**

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

---

**What is the prognosis?**

It is uniformly fatal by late childhood
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger 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

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:
-- LCA
-- 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:
-- LCA
-- Deafness
-- Hypotonia

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

Is infantile Refsum disease known?
Yes

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

What is its inheritance pattern?
AR

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

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

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

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:
--LCA
--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’

---

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

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

What is its inheritance pattern?
AR

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

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

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

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

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

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

---

Hold the phone--you can treat this one??!! How is infantile Refsum treated?
Dietary restriction of phytanic acid and phytol (a phytanic acid precursor), +/- plasmapheresis acutely
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

'Pseudo-RP'

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
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- Deafness
- Hypotonia
- Seizures
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- LCA
- Deafness
- Hypotonia
- Seizures

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It is uniformly fatal by late childhood

Is infantile Refsum disease known?
Via elevated serum phytanic acid levels (and VLCFAs)

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:
- LCA
- Deafness
- Hypotonia
- Seizures

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

Hold the phone--you can treat this one??!! How is infantile Refsum treated?
Dietary restriction of phytanic acid and phytol (a phytanic acid precursor), +/- plasmapheresis acutely
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

- Infantile Refsum
- LCA
- 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
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  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz

‘Pseudo-RP’

Peroxisomal disorders tl;dr

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

Primary RP (aka typical RP)

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

‘Pseudo-RP’

Peroxisomal disorders tl;dr

- LCA
- Deafness
- Hypotonia
- Seizures
- Abnormal facies

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

Peroxisomal disorders tl;dr

LCA
Deafness
Hypotonia
Seizures
Abnormal facies

Presents earliest

(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

Presents later

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

Presents latest Peroxisomal disorders tl;dr

Infantile Refsum
NALD
Zellweger syndrome

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
  --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`

LCA
- 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

Peroxisomal disorders tl;dr

- LCA
- Deafness
- Hypotonia
- Seizures
- Abnormal facies

Worst 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
  --'Batten dz'
  --'Bassen-Kornzweig dz'
  --Bardet-Biedl syndrome
  --Alström syndrome
  --Joubert syndrome
  --Senior-Løken syndrome

LCA
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
- LCA
- Deafness
- Hypotonia
- Seizures
- Abnormal facies

‘Pseudo-RP’

Peroxisomal disorders tl;dr

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

LCA
Deafness
Hypotonia
Seizures
Abnormal facies

Only infantile Refsum is treatable

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

What are the neuronal ceroid lipofuscinoses (NCLs; collectively referred to as Batten dz)?
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

'Pseudo-RP'

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

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(aka typical RP)

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(aka Complex RP; Syndromic RP)
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  - aka ‘Batten dz’
- Ciliopathies
- Abetalipoproteinemia
  - aka ‘Bassen-Kornzweig dz’
- Zellweger syndrome
- Neonatal adrenoleukodystrophy
- Infantile Refsum dz

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

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

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(aka typical RP)

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- Usher syndrome
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‘Pseudo-RP’

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

What is the inheritance pattern?
AR

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

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

What is the prognosis?
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‘Pseudo-RP’

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

What is the inheritance pattern?
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--
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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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- Abetalipoproteinemia
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- Neonatal adrenoleukodystrophy
- Infantile Refsum dz

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

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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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- Relentlessly progressive neurologic and cognitive decline

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

How is Batten dz managed?
Supportively

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

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(aka typical RP)

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What is the inheritance pattern?
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- Relentlessly progressive neurologic and cognitive decline

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

How is Batten dz managed?
- Supportively
- Nutritional support
- Counseling
- Genetic counseling
- Physical & occupational therapy
- Relentlessly progressive neurologic and cognitive decline

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

Primary RP
(aka typical RP)

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(aka Complex RP; Syndromic RP)
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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.
DDx for an RP-like Fundus

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  -- aka ‘Batten dz’
- Ciliopathies

‘Pseudo-RP’

What is a ciliopathy?

What is a ciliopathy?

What is a ciliopathy?
DDx for an RP-like Fundus

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- Neuronal ceroid lipofuscinoses
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‘Pseudo-RP’

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

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- Ciliopathies

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

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

Primary RP
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  -- aka ‘Batten dz’
- Ciliopathies

‘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

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(aka typical RP)

Secondary RP
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  -- 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 found in the eye, brain, and...**The eyes**

The eyes??!! Which part of the eye contains cilia wiggling about?
DDx for an RP-like Fundus

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‘Pseudo-RP’

What is a ciliopathy?
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Cilia are organelles in the eyes, brain and...
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‘Pseudo-RP’

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

Cilia come in two basic flavors: Motile and nonmotile. It is the nonmotile type which is ubiquitous in the eye.

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

Cilia are organelles in the eyes and brain.

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.
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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 organelles in the eye, brain, and skin. 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.
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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).

Cilia are organized in the eye, brain, and... **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.
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‘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

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‘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

Note that all of the ciliopathies are marked by relentlessly progressive renal failure resulting in ESRD early in life!
DDx for an RP-like Fundus

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Got a mnemonic for remembering the ciliopathies?

'Pseudo-RP'
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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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What findings define the Bardet-Biedl complex?

--H
--O
--M
--E
--R

(as in Simpson)
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What findings define the Bardet-Biedl complex?
- H
- O
- M
- E
-- RP-like fundus

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

What findings define the Bardet-Biedl complex?
-- Hypogonadism
-- Obesity
-- Mental retardation
-- Extra fingers (polydactyly)
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**‘Pseudo-RP’**

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

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

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

Both occur in childhood to teens
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How are BBS and AS managed?

‘Pseudo-RP’
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How are BBS and AS managed? Supportively
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**‘Pseudo-RP’**

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**What are the main nonocular structures affected in JS?**

- The brainstem and cerebellum

**Classic MRI finding is the hallmark of JS?**  
'Molar tooth sign'

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

**What is the prognosis?**
- It is highly variable
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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’
Joubert syndrome: Molar-tooth sign
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What is the prognosis?
It is highly variable
Joubert syndrome: Facies. Note the large head, broad forehead
What are the main nonocular structures affected in JS?
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What classic MRI finding is the hallmark of JS?
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How is JS managed?
- Supportively

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How do JS pts present? How is JS managed? Supportively
- Late infancy period
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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.
**DDx for an RP-like Fundus**

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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
- Alstrøm syndrome
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For completeness’ sake:
_in three words (including syndromes), what sort of condition are these ciliopathies?_
They are familial oculorenal syndromes

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

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

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

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

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In three words (including syndromes), what are these ciliopathies?
They are familial oculorenal syndromes

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

What are the other two?

Ciliopathies
- Bardet-Biedl syndrome
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‘Pseudo-RP’

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

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

What are the other two?
Alport syndrome and Lowe syndrome

- **Ciliopathies**
  -- Bardet-Biedl syndrome
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For completeness’ sake: In three words (including syndromes), what are these ciliopathies? They are familial oculoarenal syndromes

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

What are the other two? Alport syndrome and Lowe syndrome

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

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‘Pseudo-RP’

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

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

What are the other two?
Alport syndrome and Lowe syndrome

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

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‘Pseudo-RP’

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

Alport syndrome and Lowe syndrome

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

What are the other two?
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‘Pseudo-RP’

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

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

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

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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.)
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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?
DDx for an RP-like Fundus

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‘Pseudo-RP’

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

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  - Senior-Løken syndrome
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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

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- Usher syndrome
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‘Pseudo-RP’

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

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  -- Bardet-Biedl syndrome
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  -- Joubert syndrome
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- Abetalipoproteinemia
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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?
--
--
--
DDx for an RP-like Fundus

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(aka typical RP)

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(aka Complex RP; Syndromic RP)
- Usher syndrome
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  -- Joubert syndrome
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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?
-- Lenticular
-- Cataracts
-- Microspherophakia

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

Ciliopathies
-- Bardet-Biedl syndrome
-- Alström syndrome
-- Joubert syndrome
-- Senior-Løken syndrome
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  --Neonatal adrenoleukodystrophy
- Neuronal ceroid lipofuscinoses
- Ciliopathies
  --Bardet-Biedl syndrome
  --Alström syndrome
  --Joubert syndrome
  --Senior-Løken syndrome
- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

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

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

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

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

Are these associated with the ciliopathies?
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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- Usher syndrome
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- Ciliopathies
  --Bardet-Biedl syndrome
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  --Joubert syndrome
  --Senior-Løken syndrome
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‘Pseudo-RP’

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

Are Alport and Lowe syndrome in the 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

Are these associated with the ciliopathies?
No
Familial Oculorenal Syndromes *tl;dr*

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

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

- Ciliopathies
  - ?
  - ?
  - ?
  - ?

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

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

- Not Ciliopathies
  - Alport syndrome
  - Lowe syndrome
Familial Oculorenal Syndromes *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 \textit{tl;dr}

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

Not Ciliopathies
- Alport syndrome
- Lowe syndrome

**Key Features**

Renal failure \textit{without} hematuria

Classic eye finding:

Renal failure \textit{with} hematuria

Classic eye finding:
Familial Oculorenal Syndromes \textit{tl;dr}

Ciliopathies

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

Not Ciliopathies

- Alport syndrome
- Lowe syndrome

Key Features

Renal failure \textit{without} hematuria

Classic eye finding: \textit{Pigmentary retinopathy}

Renal failure \textit{with} hematuria

Classic eye finding: \textit{Lenticonus}
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:**
- *Pigmentary retinopathy*

**Inheritance:**
- [Yellow box]

---

**Renal failure**
- *with* hematuria

**Classic eye finding:**
- *Lenticonus*

**Inheritance:**
- [Yellow box]
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
  - *with* hematuria

- **Classic eye finding:**
  - *Pigmentary retinopathy*
  - *Lenticonus*

- **Inheritance:**
  - *AR*
  - *X-linked*
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:
- Lenticular

Inheritance:
- X-linked

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

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.
DDx for an RP-like Fundus

What is the underlying problem in abetalipoproteinemia?

- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’

--Senior-Løken syndrome

What is the underlying problem in abetalipoproteinemia?

- One of the lipoproteins (ApoB) is not synthesized by the body

How is abetalipoproteinemia inherited?

AR

How does the absence of ApoB lead to secondary RP?

Without ApoB, fact cannot be properly absorbed. Without proper fat absorption, adequate vitamin A (a fat-soluble vitamin) levels cannot be maintained. And absent adequate vitamin A levels, the retina will cease functioning (as will other aspects of the CNS).

How is it diagnosed?

Checking vitamin A levels is a good start

How is it treated?

With supplementary vitamins A & E
DDx for an RP-like Fundus

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

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

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

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

**What is the underlying problem in abetalipoproteinemia?**
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What is the underlying problem in abetalipoproteinemia?
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--Senior-Løken syndrome

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

*How is it diagnosed?*

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

In addition to low vitamin A levels, there is a classic finding on peripheral blood smear—what is it?

Acanthocytosis of the RBCs

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

How is it treated?
With supplementary vitamins A & E

---Senior-Løken syndrome

- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’
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In addition to low vitamin A levels, there is a classic finding on peripheral blood smear--what is it?
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What does acanthocytosis mean?

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Checking vitamin A levels is a good start

How is it treated?
With supplementary vitamins A & E

--Senior-Løken syndrome

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

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

How is abetalipoproteinemia inherited?
AR

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

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

How is it treated?
With supplementary vitamins A & E

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

--Senior-Løken syndrome

● Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’
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How is it diagnosed?
Checking vitamin A levels is a good start

How is it treated?
With supplementary vitamins A & E

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

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

Primary RP (aka typical RP)

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

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

Primary RP
(aka typical RP)

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

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

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(aka typical RP)

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  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- alpha-lipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

Is a-alpha-lipoproteinemia a thing?
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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  -- Senior-Løken syndrome
- 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
DDx for an RP-like Fundus

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(aka typical RP)

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  --Alström syndrome
  --Joubert syndrome
  --Senior-Løken syndrome
- Aβ-lipoproteinemia
  --aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

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

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

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‘Pseudo-RP’

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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
DDx for an RP-like Fundus

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No, but it does affect the cornea
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'Pseudo-RP'

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

Is a-$\alpha$-lipoprotein 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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- 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?
- 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
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- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

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

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

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- Fish eye disease
- Tangier disease

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

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

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

‘Pseudo-RP’

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

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(aka typical RP)

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- Usher syndrome
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  -- Zellweger syndrome
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- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

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

Primary RP
(aka typical RP)

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

Congenital syphilis
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
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- Ciliopathies
  --Bardet-Biedl syndrome
  --Alström syndrome
  --Joubert syndrome
  --Senior-Løken syndrome
- Abetalipoproteinemia
  --aka 'Bassen-Kornzweig dz'

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

Primary RP
(aka typical RP)

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

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*aka* Complex RP; Syndromic RP
- Usher syndrome
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- Abetalipoproteinemia
  -- *aka* ‘Bassen-Kornzweig dz’

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

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- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’
- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  --Toxoplasmosis
  --HSV
  --Others
- Cancer-associated retinopathy
What is cancer-associated retinopathy (CAR)?

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

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

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

- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’
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‘Pseudo-RP’
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‘Pseudo-RP’
- Congenital syphilis
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- Infectious retinitis
  -- Toxoplasmosis
  -- HSV
  -- Others
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
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

- A mitochondrial disease

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
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Secondary RP

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Infectious retinitis
--Toxoplasmosis
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Kearns-Sayre syndrome

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Primary RP (aka typical RP)

Secondary RP

Pseudo-RP

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Infectious retinitis
Cancer-associated retinopathy
Kearns-Sayre syndrome

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

What does CPEO stand for?

Chronic progressive external ophthalmoplegia

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

What is the classic finding on muscle biopsy?
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Congenital rubella
Infectious retinitis
Cancer-associated retinopathy
--aka ‘Bassen-Kornzweig dz’

Ciliopathies
Abetalipoproteinemia

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(aka typical RP)

Secondary RP

‘Pseudo-RP’

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- Congenital rubella
- Infectious retinitis
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
- Usher syndrome
- Peroxisomal disorders
- Neuronal ceroid lipofuscinoses
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Primary RP
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- --Zellweger syndrome
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- --Infantile Refsum dz
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- --Alström syndrome
- --Joubert syndrome
- --Senior-Løken syndrome

--aka ‘Batten dz’

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CPEO
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Primary RP
(aka typical RP)

Secondary RP

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

Are the cardiac-conduction problems serious?

‘Pseudo-RP’

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

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

Primary RP
(aka typical RP)

Secondary RP

‘Pseudo-RP’

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- CPEO
- Cardiac conduction abnormalities

What is the classic triad of Kearns-Sayre syndrome?

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

Kearns-Sayre syndrome

--aka ‘Bassen-Kornzweig dz’

At what age do symptoms begin occurring?

Usually shortly before age 10 years

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

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

Primary RP
(aka typical RP)

Secondary RP

‘Pseudo-RP’

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

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

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

--aka ‘Bassen-Kornzweig dz’

--aka ‘Batten dz’
--Zellweger syndrome
--Neonatal adrenoleukodystrophy
--Infantile Refsum dz
--Bardet-Biedl syndrome
--Alström syndrome
--Joubert syndrome
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Kearns-Sayre syndrome

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

What is the classic finding on muscle biopsy?

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

Secondary RP

Pseudo-RP

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

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In very general terms, what sort of disease is K-SS?
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What is the classic finding on muscle biopsy?
‘Ragged red fibers’

For Primary RP
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For Secondary RP

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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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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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Kearns-Sayre syndrome
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(aka typical RP)

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(aka Complex RP; Syndromic RP)
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- four-letter abb. for infectious cause
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- Severe uveitis
- DUSN

What does DUSN stand for?
- Diffuse unilateral subacute neuroretinitis

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

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
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- Ciliopathies
  --Bardet-Biedl syndrome
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- Congenital syphilis
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- **DUSN**

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

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‘Pseudo-RP’
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- **DUSN**

What is the cause?

What does DUSN stand for?

Diffuse unilateral subacute neuroretinitis
DDx for an RP-like Fundus

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*(aka typical RP)*

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- Usher syndrome
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- DUSN

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

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(aka typical RP)

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

What is the cause?
Infestation by a worm (most commonly, Baylisacaris)
DUSN (circle indicates the worm’s location)
DDx for an RP-like Fundus

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(aka typical RP)

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

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Note: DUSN is addressed in detail in its own slide-set
DDx for an RP-like Fundus

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four-letter abb. for a vascular cause
DDx for an RP-like Fundus

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*(aka typical RP)*

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- Drug toxicity
  --?
  --?
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- Drug toxicity
  -- Hydroxychloroquine
  -- Thioridazine
  -- Others
Thioridazine: Pigmentary retinopathy

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

Drug toxicity
-- Hydroxychloroquine
  -- Thioridazine
  -- Others

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

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

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

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

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Very much so. At doses of # mg/day or less, retinopathy is very rare. However, at higher doses, it can develop within a matter of weeks
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

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

What class of medicine is thioridazine?
It is a phenothiazine

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

Is thioridazine retinal toxicity dose-related?
Very much so. At doses of 800 mg/day or less, retinopathy is very rare. However, at higher doses, it can develop within a matter of weeks
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  - Drug toxicity
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    -- Thioridazine
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**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
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What tests should be run to determine whether a pt has RP?
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- Drug toxicity
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  --Thioridazine
  --Others

What tests should be run to determine whether a pt has RP?
--An ERG (if it’s not markedly abnormal, it’s not RP)
--Kinetic (ie, Goldmann, not Humphrey) VF testing. Again--if it’s not abnormal, it’s not RP.
--Dark adaptometry (ditto)
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How do you rule-in pseudo-RP?
- 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…

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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…

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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...

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

-- Usher syndrome: Check hearing

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

-- Batten disease: Punt to a geneticist

-- Ciliopathies: Primarily clinical (confirmatory genetics by a geneticist), but remember the following:-- Joubert syndrome: Molar-tooth sign on brainstem MRI-- Abetalipoproteinemia: As discussed

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*Usher syndrome:* Check hearing

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--Abetalipoproteinemia: As discussed
tl;dr starts on the next slide
DDx for an RP-like Fundus

(When you hear *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!)
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-Kor兹weig dz)

‘Pseudo-RP’

Next, make sure you can name the five categories of secondary/syndromic RP. Toggle back and forth between this slide and the previous one until you can name all five with ease!
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-Korzweig dz)

Then, make sure you can name the three *peroxisomal disorders* and the four *ciliopathies*. 
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- Abetalipoproteinemia
  (aka Bassen-Korzweig dz)

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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- Abetalipoproteinemia
  (aka Bassen-Kor兹weig 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)
- Deaf/blind--
- Elevated VLCFAs--
- Dreadful prognoses--
- Only Refsum treatable--
- Progressive neuro decline--
- Death teens/early adult--

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

These are my best guesses (emphasis on guesses) regarding factoids that should be kept in mind for each. Toggle!
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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Deaf/blind--
- Usher syndrome
- Peroxisomal disorders
  --Zellweger syndrome
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  --Infantile Refsum dz

Elevated VLCFAs--
Dreadful prognoses--
Only Refsum treatable--

Progressive neuro decline--
Death teens/early adult--

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Acanthocytosis of RBCs--

Abetalipoproteinemia
(aka Bassen-Korzweig dz)

‘Pseudo-RP’

- Congenital syphilis
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- Drug toxicity
  --Hydroxychloroquine
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  --Others

Speaking of guesses…
DDx for an RP-like Fundus

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Secondary RP
(aka Complex RP; Syndromic RP)

‘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…These are the causes of pseudo-RP I would pay particular attention to.
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

‘Pseudo-RP’

- Congenital syphilis
- Congenital rubella
- Infectious retinitis
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  --Others
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- CRAO
- Drug toxicity
  --Hydroxychloroquine
  --Thioridazine
  --Others

Deaf/blind-- Usher syndrome
Elevated VLCFAs-- Peroxisomal disorders
  --Zellweger syndrome
  --Neonatal adrenoleukodystrophy
  --Infantile Refsum dz
Progressive neuro decline-- Neuronal ceroid lipofuscinoses (aka Batten dz)
Death teens/early adult--
Cilia JABS you in the eye--
Relentless renal failure--
‘Molar tooth sign’ in Joubert--
Acanthocytosis of RBCs-- Abetalipoproteinemia
(aka Bassen-Korzweig dz)

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.