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)
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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)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

‘Pseudo-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; 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 eponym
- Ciliopathies

- Abetalipoproteinemia
  --aka eponym-eponym

‘Pseudo-RP’
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

- Neuronal ceroid lipofuscinoses
  --aka ‘Batten dz’
- Ciliopathies

- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’

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DDx for an RP-like Fundus

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- Usher syndrome
- Peroxisomal disorders
- Abetalipoproteinemia
  --aka 'Bassen-Kornzweig dz'

‘Pseudo-RP’

Usher syndrome = Retinitis pigmentosa + [two words]
DDx for an RP-like Fundus

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Usher syndrome = *Retinitis pigmentosa* + sensorineural deafness

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

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Usher syndrome = *Retinitis pigmentosa* + sensorineural deafness

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

- Abetalipoproteinemia
  --aka 'Bassen-Kornzweig dz'
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(aka typical RP)

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(aka Complex RP; Syndromic RP)
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Where does Usher syndrome rank as a cause of deaf-blindness in the US?
It is the most common cause thereof

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It is the most common cause thereof

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

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It is the most common cause thereof

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

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Usher syndrome = Retinitis pigmentosa + sensorineural deafness

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

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

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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 

--**Type II**

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

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

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  --aka ‘Bassen-Kornzweig dz’
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Abetalipoproteinemia

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

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- Usher syndrome
- Peroxisomal disorders
- Neuronal ceroid

Peroxisomal disorders

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

Primary RP
(aka typical RP)

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

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

Primary RP
(aka typical RP)

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

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

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

Primary RP (aka typical RP)

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

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

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

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

What is the hallmark lab abnormality of the peroxisomal disorders that present with LCA?
(LCA = Leber’s congenital amaurosis, an age-related form of RP)
DDx for an RP-like Fundus

Primary RP (aka typical RP)

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

‘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)
**DDx for an RP-like Fundus**

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

---

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

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

**What is the hallmark lab abnormality of the peroxisomal disorders that present with LCA?**
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**What specific peroxisomal disorders can manifest an LCA-type presentation?**
--
--
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DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

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

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

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

Primary RP (aka typical RP)

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

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome? Cerebrohepatorenal syndrome

What is its inheritance pattern? AR

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

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

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      - Neonatal adrenoleukodystrophy
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- **‘Pseudo-RP’**
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Infantile Refsum dz
  - Alström syndrome
  - Bardet-Biedl syndrome
  - Joubert syndrome
  - Senior-Løken syndrome

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  - Abetalipoproteinemia

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

**What is its inheritance pattern?**
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**How do Zellweger syndrome pts present?**
In the *neonatal* period
DDx for an RP-like Fundus

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Zellweger syndrome facies: High forehead; hypertelorism
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How is Zellweger syndrome diagnosed?
By the constellation of findings (along with elevated levels of VLCFA in the blood)

How is Zellweger managed?
Supportively

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  --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
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
    - Neonatal adrenoleukodystrophy
  - Infantile Refsum 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

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
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?
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’

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
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 nonppt 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 its inheritance pattern?
AR

Note: Both are 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

‘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

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

- **Primary RP**
  - (aka typical RP)
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Neurovascular dizziness
  - Alström syndrome
  - Bardet-Biedl syndrome
  - Joubert syndrome
  - Senior-Løken syndrome

---

**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
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?
- 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
- -- LCA
- -- Deafness
- -- Hypotonia
- -- Seizures
- -- Abnormal facies
DDx for an RP-like Fundus

Secondary RP

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

Primary RP

(aka typical RP)

‘Pseudo-RP’

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

What is its inheritance pattern?
AR

How do NALD pts present?
In the late infancy period
- Hypotonia
- Seizures
- Abnormal facies

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

Note: NALD’s onset occurs a little later than does Zellweger’s

What is the nonpseudonymous 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
- Hypotonia
- Seizures
- Abnormal facies

What is the prognosis?
It is uniformly fatal by 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
  -- Infantile Refsum dz

‘Pseudo-RP’

What is the nonenonym 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:
-- ?
-- ?
-- ?
-- ?

What is the nonenonym 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
Pseudo-RP

DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

Primary RP
(aka typical RP)

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

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 its inheritance pattern?
AR

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

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, 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 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
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?
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

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

Secondary RP

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

Primary RP

(aka typical 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
- Hypotonia
- Seizures

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

---

**What is the nonesymnous 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

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

Primary RP
(aka typical RP)

‘Pseudo-RP’

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

What is its inheritance pattern?
AR

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

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.
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 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 (high forehead; hypertelorism)

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

*And albeit dreadful, the prognosis for NALD is better than that for Zellweger*

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

'Secondary RP'

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

What is its inheritance pattern?
AR

How do Z and NALD present?
In the neonatal period with:
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

--Later onset
--One fewer S/S (= no abnormal facies)
--Pts live a little longer

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:

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

What is the prognosis?
It is uniformly fatal by 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

‘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

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)

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?**
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

**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 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 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)
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?
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

Why does neonatal ADRENOleukodystrophy have adreno- in the name?
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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)

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
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

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)

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

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)
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
-- Neonatal adrenoleukodystrophy
-- Infantile Refsum dz

Primary RP
(aka typical 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 (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

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)
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?
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

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)

--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
- Ciliopathies
- Abetalipoproteinemia

‘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

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

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome
What is its inheritance pattern?
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How do Zellweger syndrome pts present?
In the neonatal period with:
-- LCA
-- Deafness
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-- Seizures
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What is the prognosis?
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- Usher syndrome
- Peroxisomal disorders
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  -- Infantile Refsum dz (aka…)

‘Pseudo-RP’

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

What is its inheritance pattern?
AR

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

What is the prognosis?
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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
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What is the non-eponymous name for infantile Refsum disease?
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
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What is the prognosis?
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**Is NALD the same condition as Zellweger syndrome?**
No, that is an X-linked condition that presents later in childhood.

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In the neonatal period with:
-- LCA
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-- Abnormal facies (high forehead; hypertelorism)

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

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

**What is its inheritance pattern?**
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**How do infantile Refsum dz pts present?**
In the early childhood period with:
-- LCA
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**What is the prognosis?**
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-- LCA
-- Deafness
-- Hypotonia
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What is its noneponymous name?
Infantile phytanic acid storage disease

What is the inheritance pattern?
AR

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

What is the prognosis?
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Primary RP
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Secondary RP
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- Usher syndrome
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  -- Zellweger syndrome
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  -- Infantine Refsum dz aka...Infantine phytic acid storage dz

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What is its inheritance pattern?
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How do Zellweger syndrome pts present?
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-- LCA
-- Deafness
-- Hypotonia
-- Seizures
What is the prognosis?
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Infantine phytic acid storage disease
What is its inheritance pattern?
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How do infantile Refsum disease 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)

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

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What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
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- LCA
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- Seizures
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Is NALD the same condition as infantile Refsum disease?
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What is its inheritance pattern?
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How do Zellweger syndrome pts present?
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-- LCA
-- Deafness
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-- Seizures
-- Abnormal facies (high forehead; hypertelorism)

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What is its inheritance pattern?
AR

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In the late infancy period with:
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AR

How do infantile Refsum dz pts present?
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-- LCA
-- Deafness
-- Hypotonia
-- Seizures

What is the prognosis?
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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

What is the prognosis?
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Primary RP
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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 prognosis?
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How do NALD pts present?
In the late infancy period with:
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How do infantile Refsum dz pts present?
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Primary RP (aka typical RP)

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‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
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How do Zellweger syndrome pts present?
In the neonatal period with:
-- LCA
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What is the prognosis?
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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

Just as NALD’s onset is a little later than Zellweger’s, infantile Refsum disease is a little later than that of NALD

What is the noneponymous name for infantile Refsum disease?
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
-- Seizures
-- Abnormal facies

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
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(aka typical RP)

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  -- Zellweger syndrome
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‘Pseudo-RP’

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- Usher syndrome
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What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
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How do Zellweger syndrome pts present?
In the *neonatal* period with:
-- LCA
-- Deafness
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-- Seizures
-- Abnormal facies (high forehead; hypertelorism)

What is the prognosis?
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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 its noneponymous name?
Infantile phytanic acid storage disease

What is the inheritance pattern?
AR

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

What is the noneponymous name?
Infantile phytanic acid storage disease

What is its inheritance pattern?
AR

How is infantile Refsum dyt pts present?
In the *early childhood* period with:
-- ?
-- ?
-- ?

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

Secondary RP

(aka Complex RP; Syndromic RP)

- Usher syndrome
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Primary RP

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‘Pseudo-RP’

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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 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?
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How do infantile Refsum dz pts present?
In the early childhood period with:
-- LCA
-- Deafness
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What is the prognosis?
It is uniformly fatal by **early adulthood** (if treatment is unsuccessful)
DDx for an RP-like Fundus

Secondary RP
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-- Deafness
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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 late childhood (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)

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‘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
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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

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**How do NALD pts present?**

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**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
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- Seizures

**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)

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- Usher syndrome
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‘Pseudo-RP’

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

What is the prognosis?
It is uniformly fatal by age \textit{1 year}

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

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How do NALD pts present?
In the \textbf{late infancy} period with:
-- LCA
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What is the noneponymous name for infantile Refsum disease?
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What is its inheritance pattern?
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How do infantile Refsum dz pts present?
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What is the prognosis?
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By what noneponymous name is infantile Refsum disease known?
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DDx for an RP-like Fundus

Primary RP
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‘Pseudo-RP’

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In the early childhood period with:
-- LCA
-- Deafness
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DDx for an RP-like Fundus

Secondary RP

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

Primary RP

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How do Zellweger syndrome pts present?
In the neonatal period with:
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Primary RP
(aka typical RP)

'Pseudo-RP'

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Cerebrohepatorenal syndrome

**What is its inheritance pattern?**
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**How do Zellweger syndrome pts present?**
In the *neonatal* period with:
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**What is the prognosis?**
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No, that is an X-linked condition that presents later in childhood

**What is its inheritance pattern?**
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**How do NALD pts present?**
In the *late infancy* period with:
-- LCA
-- Deafness
-- Hypotonia
-- Seizures

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

---

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

---

**What is the noneponymous name for infantile Refsum disease?**
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
-- Seizures

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

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‘Pseudo-RP’

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What is its inheritance pattern?
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How do Zellweger syndrome pts present?
In the neonatal period with:
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What is its inheritance pattern?
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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
-- Seizures

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

What is the non-epenymous 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?

How is infantile Refsum diagnosed?

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

Is Infantile phytanic acid storage dz known?

How is infantile Refsum diagnosed?

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)

By what non-epenymous name is Infantile Refsum disease known?
Infantile phytanic acid storage 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 *(aka Infantile phytanic acid storage dz)*

**‘Pseudo-RP’**

<table>
<thead>
<tr>
<th>What is the non euphemistic name for Zellweger syndrome?</th>
<th>Cerebrohepatorenal syndrome</th>
</tr>
</thead>
<tbody>
<tr>
<td>What is its inheritance pattern?</td>
<td>AR</td>
</tr>
<tr>
<td>How do Zellweger syndrome pts present?</td>
<td>In the <strong>neonatal</strong> period with:</td>
</tr>
<tr>
<td></td>
<td>--LCA</td>
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<td></td>
<td>--Deafness</td>
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<td></td>
<td>--Hypotonia</td>
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<td>--Seizures</td>
</tr>
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<td></td>
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<table>
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<tr>
<th>What is the non euphemistic name for Infantile Refsum dz?</th>
<th>Infantile phytanic acid storage dz</th>
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</tr>
<tr>
<td>How do Infantile Refsum dz pts present?</td>
<td>In the <strong>early childhood</strong> period with:</td>
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</table>

<table>
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<tr>
<th>What is the prognosis? It is uniformly fatal by?</th>
<th>age 1 year</th>
</tr>
</thead>
</table>

**Is Refsum disease known?**

- **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

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

**Is NALD the same condition as adrenoleukodystrophy?**

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

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

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

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

Primary RP
(aka typical 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 (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 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

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

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
--Seizures

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

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(aka typical RP)

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  -- Infantile Refsum dz (aka... Infantile phytanic acid storage dz)

‘Pseudo-RP’

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-- Abnormal facies (high forehead; hypertelorism)

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

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AR

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Dietary restriction of phytanic acid and phytol (a phytanic acid precursor), +/- plasmapheresis acutely

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(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

‘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
- 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

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

Primary RP
(aka typical RP)

‘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’

LCA
Deafness
Hypotonia
Seizures
Abnormal facies

Peroxisomal disorders tl;dr

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

Secondary RP

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

Primary RP

(aka typical RP)

‘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
- '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

Peroxisomal disorders tl;dr

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

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

‘Pseudo-RP’

Primary RP
(aka typical RP)

Peroxisomal disorders tl;dr

LCA
Deafness
Hypotonia
Seizures
Abnormal facies

Infantile Refsum
NALD
Zellweger syndrome

Best prognosis

(Rediu 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

‘Pseudo-RP’

Deafness
Hypotonia
Seizures
Abnormal facies

LCA

Peroxisomal disorders tl;dr

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

Abetalipoproteinemia

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

'Pseudo-RP'

LCA
Deafness
Hypotonia
Seizures
Abnormal facies

Infantile Refsum dz
NALD
Zellweger syndrome

All three are inherited AR

Peroxisomal disorders tl;dr

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

Secondary RP

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

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

‘Pseudo-RP’

What are the neuronal ceroid lipofuscinoses (NCLs; collectively referred to as Batten dz)?

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

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

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

'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,
depending on the form, it is fatal by late childhood to early adulthood.
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

What is the inheritance pattern?
AR

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

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

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

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

‘Pseudo-RP’

What 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?

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

‘Pseudo-RP’

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

What 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
(aka typical RP)

Secondary RP
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- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- 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?
AR

How do Batten dz pts present?

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

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

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

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

‘Pseudo-RP’

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

What is the inheritance pattern?
AR

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

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

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

- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- 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?
AR

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

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

Primary RP
(aka typical RP)

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

---

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

What is the inheritance pattern?
AR

How do Batten dz pts present?
In infancy to early childhood with:
  -- LCA
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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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- Usher syndrome
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  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
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- Ciliopathies
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

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

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

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

What is the inheritance pattern?
AR

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

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

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
DDx for an RP-like Fundus

Primary RP (aka typical RP)

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

What is the inheritance pattern?
AR

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

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

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  --Zellweger syndrome
  --Neonatal adrenoleukodystrophy
  --Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  --aka ‘Batten dz’
  --Neurodegenerative conditions stemming from the buildup of ceroid & lipofuscin in cells
- Peripheric neuropathy
  --Abetalipoproteinemia
  --Dyshidrotic epidermolysis bullosa (in the infantile forms)
- ‘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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DDx for an RP-like Fundus

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

What is the inheritance pattern?
AR

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

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

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
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

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Cilia are organelles found in the eyes, brain, and...
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What is a ciliopathy?
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Cilia are organelles found in the eyes, brain, and kidneys. The eyes??!! Which part of the eye contains cilia wiggling about?

None. Remember, cilia come in two basic flavors: Motile, and nonmotile. It is the nonmotile type which is ubiquitous in the eye.
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OK then, where in the eye are the nonmotile cilia, dysfunction of which leads to an RP-like fundus?
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‘Pseudo-RP’

Primary 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 one word disorder.

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

The eyes??!! Which part of the eye contains cilia wiggling about?
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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 brain and eyes.**

**The eyes??!! Which part of the eye contains cilia wiggling about?**

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‘Pseudo-RP’

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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).
Pseudo-RP

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  --?
  --?
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  --?

Which ciliopathies present with an RP-like fundus?

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‘Pseudo-RP’

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

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‘Pseudo-RP’

Got a mnemonic for remembering the ciliopathies?
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Got a mnemonic for remembering the ciliopathies? JABS. Imagine a cilia as it jabs someone in the eye
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What findings define the Bardet-Biedl complex?

The mnemonic is…
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‘Pseudo-RP’

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

(as in Simpson)
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Not surprisingly, the ‘R’ stands for RP-like fundus.
As for the rest…
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What findings define the Bardet-Biedl complex?
- Hypogonadism
- Obesity
- Mental retardation
- Extra fingers (polydactyly)
- RP-like fundus

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And yeah, I know, Homer only has four digits per hand--paucidactyly, not polydactyly. But the rest fits him pretty well.
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Alström syndrome has the same findings as Bardet-Biedl, with two additions:

What findings define the Bardet-Biedl complex?
- Hypogonadism
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The mnemonic is…
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How are BBS and AS managed?

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‘Pseudo-RP’

What are the main nonocular structures affected in JS?

The brainstem and cerebellum

What classic MRI finding is the hallmark of JS?

‘Molar tooth sign’

How do JS pts present?

In the late infancy period with:
- 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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Joubert syndrome: Molar-tooth sign
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Joubert syndrome: Facies. Note the large head, broad forehead
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DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

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

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

How is JS managed?
Supportively

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

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

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‘Pseudo-RP’

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

- **Ciliopathies**
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
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  -- aka ‘Bassen-Kornzweig dz’
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In three words (including syndromes), what sort of condition are these ciliopathies?
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How many familial oculorenal syndromes are there?
Including the four ciliopathies—six

The other two are Alport syndrome and Lowe syndrome
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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.

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Alport syndrome and Lowe syndrome
DDx for an RP-like Fundus

Primary RP
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‘Pseudo-RP’

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

**Primary RP**

- *aka* typical RP

**Secondary RP**

- *aka* Complex RP; Syndromic RP
  - **Usher syndrome**
  - Peroxisomal disorders
  - Neuronal ceroid lipofuscinoses
  - Ciliopathies
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  - Abetalipoproteinemia
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**‘Pseudo-RP’**

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*In three words (including syndromes), what sort of condition are these ciliopathies? They are familial oculorenal syndromes.*

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

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

Primary RP
(aka typical RP)

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

What lens pathologies occur with Alport and Lowe syndromes?
--
--
--

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

What are the other two?
Alport syndrome and Lowe syndrome

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

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

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

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

Are these associated with the ciliopathies?

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

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Alport syndrome and Lowe syndrome
DDx for an RP-like Fundus

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‘Pseudo-RP’

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

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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
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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
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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

![Diagram showing familial oculorenal syndromes and key features.](image)
Familial Oculorenal Syndromes *tl;dr*

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

**Not Ciliopathies**
- Alport syndrome
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**Key Features**
- Renal failure *without* hematuria
- Renal failure *with* hematuria
Familial Oculorenal Syndromes

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

Not Ciliopathies
- Alport syndrome
- Lowe syndrome

Renal failure
- without hematuria

Classic eye finding:

Renal failure
- with hematuria

Classic eye finding:
Familial Oculorenal Syndromes *tl;dr*

### Key Features

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

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

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

- Renal failure *with* hematuria
- Classic eye finding: *Lenticonus*
Familial Oculorenal Syndromes \textit{tl;dr}

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

- Not Ciliopathies

Key Features

- Renal failure \textit{without} hematuria
- Classic eye finding: \textit{Pigmentary retinopathy}
- Inheritance:

- Renal failure \textit{with} hematuria
- Classic eye finding: \textit{Lenticonus}
- Inheritance:
Familial Oculorenal Syndromes

Ciliopathies
- Joubert syndrome
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Key Features

Renal failure without hematuria

Classic eye finding:
Pigmentary retinopathy

Inheritance: AR

Renal failure with hematuria

Classic eye finding:
Lenticonus

Inheritance: X-linked
Familial Oculo-renal Syndromes \textit{tl};\textit{dr}

- Ciliopathies
  - Joubert syndrome
  - Alström syndrome
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  - Senior-Løken syndrome
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Renal failure
- \textit{without} hematuria
- Classic eye finding: \textit{Pigmentary retinopathy}
- Inheritance: \textit{AR}

Renal failure
- \textit{with} hematuria
- Classic eye finding: \textit{Lenticonus}
- Inheritance: \textit{X-linked}

\textit{For more info on Alport and Lowe syndromes, see slide-set L4}
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‘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

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.
DDx for an RP-like Fundus

What is the underlying problem in abetalipoproteinemia?
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--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.

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'

- Usher syndrome
- Peroxisomal disorders
- Neuronal ceroid lipofuscinoses
- Ciliopathies
- Abetalipoproteinemia
- --Zellweger syndrome
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How is it diagnosed?
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How is it treated?
With supplementary vitamins A & E.

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

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

<table>
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- **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’)
  - ‘Bassen-Kornzweig dz’
  - Bardet-Biedl syndrome
  - Alström syndrome
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**What is the most common cause of hypovitaminosis A? (It’s not abetalipoproteinemia)**
Malabsorption secondary to GI surgery (eg, gastric bypass; small-bowel resection)

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

**How is it treated?**
With supplementary vitamins A & E.

---

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

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(aka typical RP)

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  -- Joubert syndrome
  -- Senior-Løken syndrome
- **Abetalipoproteinemia**
  -- aka 'Bassen-Kornzweig dz'

‘Pseudo-RP’

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

Is hypobetalipoproteinemia a thing?

Indeed it is, via a condition called ‘familial hypobetalipoproteinemia,’ and it can affect the retina (it’s mentioned in the BCSC books, but not addressed in detail)
DDx for an RP-like Fundus

Primary RP

(aka typical RP)

Secondary RP

(aka Complex RP; Syndromic RP)

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  -- Alström syndrome
  -- Joubert syndrome
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alpha

A beta-lipoproteinemia
  -- aka 'Bassen-Kornzweig dz'

‘Pseudo-RP’

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

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  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- alpha A-beta lipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

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

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  -- Joubert syndrome
  -- Senior-Løken syndrome
- Alpha lipoproteinemia
  -- aka 'Bassen-Kornzweig dz'

‘Pseudo-RP’

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

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

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  --aka ‘Batten dz’
- Ciliopathies
  --Bardet-Biedl syndrome
  --Alström syndrome
  --Joubert syndrome
  --Senior-Løken syndrome
- Alpha- lipoproteinemia
  --aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

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

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

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

’Pseudo-RP’

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

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

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  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

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.

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

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(aka typical RP)

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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

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

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  --Joubert 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?

- LCAT deficiency
- Fish eye disease
- Tangier disease

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

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- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka 'Bassen-Korzweig 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

Low levels of α-lipoprotein are implicated in three corneal conditions. What are they?

- LCAT deficiency
- Fish eye disease
- Tangier disease

-- Abetalipoproteinemia (Bassen-Korzweig)
-- Familial hypobetalipoproteinemia
DDx for an RP-like Fundus

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  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
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- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

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

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

‘Pseudo-RP’
- Congenital syphilis
- Congenital rubella
Congenital rubella

Congenital syphilis
DDx for an RP-like Fundus

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‘Pseudo-RP’
- Congenital syphilis
- Congenital rubella
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  -- ?
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‘Pseudo-RP’
- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  --Toxoplasmosis
  --HSV
  --Others
Toxoplasmosis

HSV
DDx for an RP-like Fundus

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-associated retinopathy

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What is cancer-associated retinopathy (CAR)?

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

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- Congenital rubella
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  --HSV
  --Others
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
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Secondary RP

‘Pseudo-RP’

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

Kearns-Sayre syndrome

What is the classic triad of Kearns-Sayre syndrome?

- Pigmentary retinopathy
- CPEO
- Cardiac conduction abnormalities

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

A mitochondrial disease

What is the classic finding on muscle biopsy?

Ragged red fibers

At what age do symptoms begin occurring?

Usually shortly before age 10 years

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

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

Primary RP
(aka typical RP)

Secondary RP

‘Pseudo-RP’

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- Pigmentary retinopathy
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What does CPEO stand for?

Chronic progressive external ophthalmoplegia

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Usually shortly before age 10 years

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

What does CPEO stand for?

Chronic progressive external ophthalmoplegia

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

Primary RP
(aka typical RP)

Secondary RP

‘Pseudo-RP’

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Congenital rubella
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--Others
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Secondary RP

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

- Toxoplasmosis
- HSV
- Others

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.

Kearns-Sayre syndrome
CPEO
DDx for an RP-like Fundus

Primary RP (aka typical RP)

Secondary RP

‘Pseudo-RP’

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

Kearns-Sayre syndrome

What is the classic triad of Kearns-Sayre syndrome?
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Are the cardiac-conduction problems serious?

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

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

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

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

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Secondary RP

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

‘Pseudo-RP’

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-- ‘Batten dz’

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Kearns-Sayre syndrome

Toxoplasmosis
-- HSV
-- Others

Ciliopathies

Abetalipoproteinemia

Usher syndrome

Peroxisomal disorders

Neuronal ceroid lipofuscinoses

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-- HSV
-- Others

Ciliopathies

Abetalipoproteinemia

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Neuronal ceroid lipofuscinoses

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-- Senior-Løken syndrome
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DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
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  -- Joubert syndrome
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‘Pseudo-RP’
- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  -- Toxoplasmosis
  -- HSV
  -- Others
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
- Severe uveitis
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- Usher syndrome
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- Ciliopathies
  -- Bardet-Biedl syndrome
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- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  -- Toxoplasmosis
  -- HSV
  -- Others
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
- Severe uveitis

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

Primary RP
(aka typical RP)

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- Usher syndrome
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- Severe uveitis
- DUSN
DDx for an RP-like Fundus

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(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
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  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
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  -- aka 'Batten dz'
- Ciliopathies
  -- Bardet-Biedl syndrome

What does DUSN stand for?
- Diffuse unilateral subacute neuroretinitis

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

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
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
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What does DUSN stand for?
Diffuse unilateral subacute neuroretinitis
DDx for an RP-like Fundus

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

What does DUSN stand for?
- Diffuse unilateral subacute neuroretinitis

What is the cause?

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

Primary RP
(aka typical RP)

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

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

Primary RP (aka typical RP)

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

Primary RP
(aka typical RP)

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

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What does DUSN stand for?
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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

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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’

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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    -- Others
  - Cancer-associated retinopathy
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  - Severe uveitis
  - DUSN
  - CRAO
DDx for an RP-like Fundus

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  --Toxoplasmosis
  --HSV
  --Others
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
- Severe uveitis
- DUSN
- CRAO
- Drug toxicity
  --?
  --?
  --Others
DDx for an RP-like Fundus

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Secondary RP (aka Complex RP; Syndromic RP)
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  -- Toxoplasmosis
  -- HSV
  -- Others
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
- Severe uveitis
- DUSN
- CRAO
- Drug toxicity
  -- Hydroxychloroquine
  -- Thioridazine
  -- Others
Thioridazine: Pigmentary retinopathy

Hydroxychloroquine retinopathy
DDx for an RP-like Fundus

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(aka typical RP)

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

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

It is a phenothiazine

Their main use is as antipsychotics

Is thioridazine retinal toxicity dose-related?

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

How does thioridazine retinopathy present clinically?

The pt with c/o blurry vision; DFE will reveal pigment stippling in the macula

Does thioridazine use require periodic ophthalmic evaluations like hydroxychloroquine does?

Not so long as the dose is at or below 800 mg/day
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Drug toxicity
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  --aka ‘Batten dz’
- Ciliopathies
  --Bardet-Biedl syndrome
  --Alström syndrome
  --Joubert syndrome
  --Senior-Løken syndrome
- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’

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  --Others

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

--

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

--An ERG (if it’s not markedly abnormal, it’s not RP)

--**Kinetic** (ie, Goldmann, *not* Humphrey) VF testing. Again--if it’s not abnormal, it’s not RP.

--Dark adaptometry (ditto)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
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How do you rule-in pseudo-RP?

- Ciliopathies
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  --Alström syndrome
  --Joubert syndrome
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- Drug toxicity
  --Hydroxychloroquine
  --Thioridazine
  --Others

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

- Ciliopathies
  --Bardet-Biedl syndrome
  --Alström syndrome
  --Joubert syndrome
  --Senior-Løken syndrome
- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’
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  --Hydroxychloroquine
  --Thioridazine
  --Others

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

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

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

Primary RP
(aka typical RP)

Secondary RP
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- Usher syndrome
- Peroxisomal disorders
- Zellweger syndrome

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

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

- Ciliopathies
  --Bardet-Biedl syndrome
  --Alström syndrome
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- Abetalipoproteinemia
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**DDx for an RP-like Fundus**

- **Primary RP** *(aka typical RP)*
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  - Usher syndrome
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  - Hydroxychloroquine
  - Thioridazine
  - Others

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

- Ciliopathies
  - Bardet-Biedl syndrome
  - Alström syndrome
  - Joubert syndrome
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‘Pseudo-RP’

- Congenital syphilis

---

**How do you rule-in the causes of secondary RP?**
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- **Usher syndrome**
- Peroxisomal disorders
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  -- Neonatal adrenoleukodystrophy
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- Ciliopathies
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  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka 'Bassen-Kornzweig dz'

‘Pseudo-RP’
- Congenital syphilis

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

-- **Usher syndrome**: Check hearing

---

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

-- **Batten disease**: Punt to a geneticist

-- **Ciliopathies**: Primarily clinical (confirmatory genetics by a geneticist), but remember the following:
  ---- **Joubert syndrome**: Molar-tooth sign on brainstem MRI
  ---- **Senior-Løken**: Check renal

-- **Abetalipoproteinemia**: As discussed
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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  -- Zellweger syndrome
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- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
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‘Pseudo-RP’
- Congenital syphilis

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

-- Usher syndrome: Check hearing

-- The peroxisomal disorders:
  - Check serum levels of 'very long chain fatty acids' +/− phytanic acid levels
  - Batten disease: Punt to a geneticist
  - Ciliopathies: Primarily clinical (confirmatory genetics by a geneticist), but remember the following:
    - Joubert syndrome: Molar-tooth sign on brainstem MRI
  - Abetalipoproteinemia: As discussed

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

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

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‘Pseudo-RP’
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  ---- Joubert syndrome: Molar-tooth sign on brainstem MRI

-- 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

Secondary RP (aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
- Neuronal ceroid lipofuscinoses (aka Batten dz)
- Ciliopathies
- Abetalipoproteinemia (aka Bassen-Korzweig 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-Korzeig dz)

‘Pseudo-RP’

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

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(aka typical RP)

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- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  (aka Bassen-Korzyweig 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.
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-Korzeig dz)

‘Pseudo-RP’

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

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

‘Pseudo-RP’

Deaf/blind--
Usher syndrome

Elevated VLCFAs--
Peroxisomal disorders

Dreadful prognoses--
--Zellweger syndrome

Only Refsum treatable--
--Neonatal adrenoleukodystrophy

--Infantile Refsum dz

Progressive neuro decline--
Neuronal ceroid lipofuscinoses (aka Batten dz)

Death teens/early adult--

Ciliary disorders

Cilia JABS you in the eye--
--Bardet-Biedl syndrome

Relentless renal failure--
--Alström syndrome

‘Molar tooth sign’ in Joubert--
--Joubert syndrome

--Senior-Løken syndrome

Acanthocytosis of RBCs--
Abetalipoproteinemia
(aka Bassen-Korzyweig 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

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(aka typical RP)

Secondary RP
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‘Pseudo-RP’

- Deaf/blind--
  - Usher syndrome
  - Peroxisomal disorders
    - Zellweger syndrome
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- Elevated VLCFAs--
- Dreadful prognoses--
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- Ciliopathies
  - Bardet-Biedl syndrome
  - Alström syndrome
  - Joubert syndrome
  - Senior-Løken syndrome

- Acanthocytosis of RBCs--
  - Abetalipoproteinemia (aka Bassen-Korzyweig dz)

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

Speaking of guesses…
DDx for an RP-like Fundus

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(aka typical RP)

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‘Pseudo-RP’

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- 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

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(aka typical RP)

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(aka Complex RP; Syndromic RP)

‘Pseudo-RP’

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- Congenital rubella
- Infectious retinitis
  --Toxoplasmosis
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  --Others
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  --Hydroxychloroquine
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  --Others

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

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.