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

My advice: In prepping for the OKAP, try to run through this slide-set once or twice a month. When you reach the final, frantic few weeks of cramming, don’t wade through the whole thing—just do the \textit{tl;dr} at the end (it starts around slide 364).

Finally: This is a big topic, and big topics beget big slide-sets. There’s a natural break around slide 236; I placed a \textit{Break time!} slide at that point to mark it.

Now, let’s get* this bread!

*\textit{Bake} this bread? \textit{Make} this bread? I can never remember.
First things first. An RP-like fundus has three characteristic attributes. What are they?
--?
--?
--?
First things first. An RP-like fundus has three characteristic attributes. 
What are they?

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

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

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

What two vision issues are the defining characteristics of RP?
--Nyctalopia
--Visual field loss
Typical pattern of VF loss in RP: Mid-peripheral scotomata → coalesce into partial rings → coalesce into complete ring → expand rapidly outward → expand slowly inward

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

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

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

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

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

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

--Bone spicules
--Waxy disc pallor
--Arteriolar narrowing

What two vision issues are the defining characteristics of RP?

--Nyctalopia
--Visual field loss

Results of what specialized testing modality are always abnormal in RP?

Electroretinogram (ERG)
These next few slides lay out the overall way you should think about an RP-like fundus (especially on the OKAP/Boards)
When a pt’s fundi have an RP-like appearance, one of three things is going on:
1) 
2) 
3) 

When a pt’s fundi have an RP-like appearance, one of three things is going on:
1) ?
2) ?
3) ?
When a pt's fundi have an RP-like appearance, one of three things is going on:
1) The pt has RP (duh)
2) ?
3) ?
When a pt’s fundi have an RP-like appearance, one of three things is going on:
1) The pt has RP; or
2) s/he has a systemic condition in which retinal manifestations c/w RP occur
3) ?
When a pt’s fundi have an RP-like appearance, one of three things is going on:  
1) The pt has RP; or 
2) s/he has a systemic condition in which retinal manifestations c/w RP occur; or 
3) s/he has a condition that has nothing to do with RP—it just happens to have a similar appearance
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

‘Pseudo-RP’

For more on RP itself, see slide-set R38
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

The secondary/syndromic RP conditions can be grouped into five categories based on the underlying issue—what are these categories?

(Hints forthcoming on the next slide)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- (the most common syndromic association)
- ?
- ?
- ?
- ?

‘Pseudo-RP’

The secondary/syndromic RP conditions are can be grouped into five categories based on the underlying issue—what are these categories?
The secondary/syndromic RP conditions can be grouped into five categories based on the underlying issue—what are these categories?
The 2ndry/syndromic RP conditions are can be grouped into five categories based on the underlying issue—what are these categories?
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

The secondary/syndromic RP conditions can be grouped into five categories based on the underlying issue—what are these categories?
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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
- Neuronal ceroid lipofuscinoses

‘Pseudo-RP’

The 2ndry/syndromic RP conditions are can be grouped into five categories based on the underlying issue—what are these categories?
The 2ndry/syndromic RP conditions are grouped into five categories based on the underlying issue—what are these categories?
The secondary/syndromic RP conditions can be grouped into five categories based on the underlying issue—what are these categories?
The 2ndry/syndromic RP conditions can be grouped into five categories based on the underlying issue—what are these categories?
The 2ndry/syndromic RP conditions are can be grouped into five categories based on the underlying issue—what are these categories?
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

‘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

‘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

‘Pseudo-RP’
- Abetalipoproteinemia
  --aka eponym-eponym
DDx for an RP-like Fundus

Primary RP (aka typical RP)

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

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

- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

- Usher syndrome
- Peroxisomal disorders

Usher syndrome = Retinitis pigmentosa + two words

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

Primary RP
(aka typical RP)

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

Usher syndrome = *Retinitis pigmentosa* + sensorineural deafness

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

Primary RP
(aka typical RP)

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

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

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

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

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

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

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

- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’
Usher syndrome = *Retinitis pigmentosa* + sensorineural deafness

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

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

--?

--?

--?

Abetalipoproteinemia

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

Primary RP
(aka typical RP)

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

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

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

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

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

Primary RP (aka typical RP)

Secondary RP (aka Complex RP; Syndromic RP)

- Usher syndrome
- Peroxisomal disorders

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

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

There are three types of Usher syndrome--what are they called? How do they manifest?

--Type I manifests...
--Type II
--Type III

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

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

**‘Pseudo-RP’**

### DDx for an RP-like Fundus

- **Primary RP**
- **Secondary RP**
- **‘Pseudo-RP’**

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

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

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

--**Type I** manifests... in the [ ] decade with [ severity ] hearing loss, RP and vestibular dysfunction
--**Type II**
--**Type III**

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

Primary RP
(aka typical RP)

Secondary RP
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‘Pseudo-RP’

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

There are three types of Usher syndrome--what are they called? How do they manifest?
--Type I manifests…in the first decade with profound hearing loss, RP and vestibular dysfunction
--Type II
--Type III

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

Primary RP
(aka typical RP)

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

There are three types of Usher syndrome--what are they called? How do they manifest?
--**Type I** manifests...in the first decade with profound hearing loss, RP and vestibular dysfunction
--**Type II** manifests...
--**Type III**

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

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

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

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

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

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

- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’
**Usher syndrome** = *Retinitis pigmentosa* + sensorineural deafness

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

There are three types of Usher syndrome -- what are they called? How do they manifest?

-- **Type I** manifests...in the first decade with profound hearing loss, RP and vestibular dysfunction.

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

-- **Type III** manifests...in the third decade with mild hearing loss, RP; vestibular function is intact.

**DDx for an RP-like Fundus**

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

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

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

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

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

Primary RP (aka typical RP)
- Usher syndrome
- Peroxisomal disorders
- Abetalipoproteinemia
  --aka 'Bassen-Kornzweig dz'

'Pseudo-RP'

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

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

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

---**Type I** manifests...in the first decade with profound hearing loss, RP and vestibular dysfunction

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

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

DDx for an RP-like Fundus
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

- Usher syndrome
- Peroxisomal disorders

‘Pseudo-RP’

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

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

There are three types of Usher syndrome--what are they called? How do they manifest?

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

Abetalipoproteinemia
--aka ‘Bassen-Kornzweig dz’

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

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

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

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

Peroxisomal disorders are a heterogeneous group of disorders of peroxisome function. Peroxisomes are intracellular organelles that play key roles in many aspects of cell metabolism. The hallmark lab abnormality of the peroxisomal disorders that present with LCA is abnormally high serum levels of very long chain fatty acids (VLCFA). Specific peroxisomal disorders that can manifest an LCA-type presentation include:

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

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

What is/are peroxisomes?
What are peroxisomal disorders?
A heterogeneous group of disorders of peroxisome function

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

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

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

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

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

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

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

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

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

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

---

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

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

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

**What 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
- Peroxisomal disorders
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Infantile Refsum dz
- Neuronal ceroid lipofuscinoses

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?

What is its inheritance pattern?
AR

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

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

Primary RP
(aka typical RP)

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

'Pseudo-RP'

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

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Cerebrohepatorenal syndrome

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Secondary RP
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  - Usher syndrome
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    -- Zellweger syndrome
      -- Neonatal adrenoleukodystrophy
      -- Infantile Refsum dz
  - Neuronal ceroid lipofuscinoses
  - Ciliopathies

‘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 ‘age’ period
What is the noneponymous name for Zellweger syndrome? Cerebrohepatorenal syndrome

What is its inheritance pattern? AR

How do Zellweger syndrome pts present? In the neonatal period with:
--RP-like fundus
--?
--?
--?
--?
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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Cerebrohepatorenal syndrome

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‘Pseudo-RP’

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Cerebrohepatorenal syndrome

What is its inheritance pattern?
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In the neonatal period with:
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-- Deafness
-- Hypotonia
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What is the prognosis?
It is uniformly fatal by age 1 year

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

How is Zellweger syndrome 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

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

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

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

**Primary RP**
(aka typical RP)

**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?**
AR

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

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

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

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(aka typical RP)

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‘Pseudo-RP’

What is Zellweger syndrome diagnosed? 
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How is Zellweger managed? 
Supportively

What is its inheritance pattern?
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In the neonatal period with:
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-- Deafness
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-- Seizures
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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)
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- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz

‘Pseudo-RP’

What is the nonepithelial name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

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

What is its inheritance pattern?
AR

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

What is the prognosis?
It is uniformly fatal by late childhood
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 nonpseudonym for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

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DDx for an RP-like Fundus

Primary RP
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-- Abnormal facies (high forehead; hypertelorism)

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

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

What is its inheritance pattern?
AR

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

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

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
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  -- Zellweger syndrome
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-- Seizures
-- Abnormal facies

What is the prognosis?
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What is its inheritance pattern?
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Primary RP
(aka typical RP)

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  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
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‘Pseudo-RP’

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

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

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

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

What is its inheritance pattern?
AR

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

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

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:
- RP-like fundus
- Deafness
- Hypotonia
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- Abnormal facies (high forehead; hypertelorism)

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

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

**What is its inheritance pattern?**
AR

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

What is its inheritance pattern?
AR

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

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

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

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

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

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 non-pseudonymous name for Zellweger syndrome?**

- Cerebrohepatorenal syndrome

**What is its inheritance pattern?**

- AR

**How do Zellweger syndrome pts present?**

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

**What is the prognosis?**

- It is uniformly fatal by age 1 year

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

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

**What is its inheritance pattern?**

- AR

**How do NALD pts present?**

- In the late infancy period

- -- Seizures

**What is the non-pseudonymous name for Infantile Refsum Disease?**

- [Specific name not provided in the image]
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 pts present?
In the neonatal period with:
-- RP-like fundus
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What is the prognosis?
It is uniformly fatal by age 1 year

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

What is its inheritance pattern?
AR

How do NALD pts present?
In the late infancy period

What is NALD’s onset?
In the late infancy period

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 non-epithelial name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
-- RP-like fundus
-- Deafness
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-- Seizures
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What is the prognosis?
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Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that presents later in childhood

What is its inheritance pattern?
AR

How do NALD pts present?
In the late infancy period with:
-- RP-like fundus
-- ?
-- ?
-- ?
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

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

What is the prognosis?
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What is its inheritance pattern?
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In the late infancy period with:
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Primary RP
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What is the prognosis?
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What is its inheritance pattern?
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How do NALD pts present?
In the late infancy period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures

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

Note that NALD has the same S/S as Zellweger, except it’s missing the last one on the list
DDx for an RP-like Fundus

Primary RP (aka typical RP)

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

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(aka typical RP)

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‘Pseudo-RP’

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-- Deafness
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-- Seizures

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

Primary RP
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  -- Zellweger syndrome
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‘Pseudo-RP’

What is the nonɵnymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
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What is the prognosis?
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Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that presents later in childhood

What is its inheritance pattern?
AR

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

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

Primary RP
(aka typical RP)

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‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
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-- 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?
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What is its inheritance pattern?
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How do NALD pts present?
In the late infancy period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures
-- Abnormal facies

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

And albeit dreadful, the prognosis for NALD is better than that for Zellweger
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?
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-- Deafness
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What is the prognosis? It is uniformly fatal by age 1 year

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

What is its inheritance pattern? AR

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

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
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In the neonatal period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures
-- Abnormal facies (high forehead; hypertelorism)

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

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

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Why does neonatal ADRENOleukodystrophy have adreno- in the name?
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:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures
-- Abnormal facies (high forehead; hypertelorism)

What is the prognosis?
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-- RP-like fundus
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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
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  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz

‘Pseudo-RP’

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

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
-- RP-like fundus
-- Deafness
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What is the prognosis?
It is uniformly fatal by age 1 year

---Neonatal adrenoleukodystrophy
---Infantile Refsum dz

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Supportively (just like Zellweger pts)

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Supportively (just like Zellweger pts)

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
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‘Pseudo-RP’

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

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

Why does neonatal ADRENOleukodystrophy have adreno- in the name?
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How is NALD managed?
Supportively (just like Zellweger pts)
DDx for an RP-like Fundus

Primary RP (aka typical RP)
- Usher syndrome
- Peroxisomal disorders
  --Zellweger syndrome
  --Neonatal adrenoleukodystrophy
  --Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
- Ciliopathies
- Abetalipoproteinemia

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:
--RP-like fundus
--Deafness
--Hypotonia
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What is the prognosis?
It is uniformly fatal by age 1 year

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

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AR

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

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

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

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

What is its inheritance pattern?
AR

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

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(aka typical RP)

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

'Secondary RP'

'Pseudo-RP'

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

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
-- RP-like fundus
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-- Seizures
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What is the prognosis?
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Before we get into it: Is infantile Refsum dz the same as adult-onset Refsum dz?

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

Before we get into it: Is infantile Refsum dz the same as adult-onset Refsum dz?
DDx for an RP-like Fundus

Primary RP (aka typical RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Abetalipoproteinemia

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

Pseudo-RP

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

What is its inheritance pattern?
AR

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

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

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

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

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In the late infancy period with:
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-- Seizures

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

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

What is its inheritance pattern?
AR

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

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

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

‘Pseudo-RP’

---

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

**What is its inheritance pattern?**
AR

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

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

---

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

**What is its inheritance pattern?**
AR

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

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

---

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

---

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

---

**What is its inheritance pattern?**
AR

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

**What is the prognosis?**
It is uniformly fatal by **early adulthood**

---

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

---

**What is its inheritance pattern?**
AR

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

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

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

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

- **‘Pseudo-RP’**

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Cerebrohepatorenal syndrome

**What is its inheritance pattern?**
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**How do Zellweger syndrome pts present?**
In the neonatal period with:
-- RP-like fundus
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-- Hypotonia
-- Seizures
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**What is the prognosis?**
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**What is its inheritance pattern?**
AR

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In the late infancy period with:
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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:
-- RP-like fundus
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**What is the prognosis?**
It is uniformly fatal by late childhood (if treatment is unsuccessful)
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Primary RP
(aka typical RP)

Secondary RP
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- Usher syndrome
- Peroxisomal disorders
  --Zellweger syndrome
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  --Infantile Refsum dz aka Infantile phytanic acid storage dz

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
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How do Zellweger syndrome pts present?
In the neonatal period with:
--RP-like fundus
--Deafness
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--Seizures
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What is the prognosis?
It is uniformly fatal by age 1 year

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

What is its inheritance pattern?
AR

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

What is its noneponymous name?
Infantile phytanic acid storage disease

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

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

What is its inheritance pattern?
AR

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

What is the prognosis?
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PPseudo-RP

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

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

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

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

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

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

Like Zellweger's and NALD, infantile Refsum's is inherited AR

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

What is its inheritance pattern?
AR

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

What is the prognosis?
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(aka typical RP)

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

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
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-- Seizures
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In the *late infancy* period with:
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Infantile phytanic acid storage disease

What is its inheritance pattern?
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How do infantile Refsum dz pts present?
In the *late childhood* period

What is the prognosis?
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‘Pseudo-RP’

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What is its inheritance pattern?
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In the late infancy period with:
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-- Deafness
-- Hypotonia
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Infantile phytanic acid storage disease

What is its inheritance pattern?
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‘Pseudo-RP’

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What is the noneponymous name for infantile Refsum disease?
Infantile phytanic 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:
-- RP-like fundus
-- 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 Refum’s is a little later than that of NALD
Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

What is its inheritance pattern?
* AR

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

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

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

What is its inheritance pattern?
* AR

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

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

What is its inheritance pattern?
* AR

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

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

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

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

- Abetalipoproteinemia

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures
-- Abnormal facies
What is the prognosis?
It is uniformly fatal by age 1 year

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

What is its inheritance pattern?
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What is the prognosis?
It is uniformly fatal by late childhood

By what noneponymous name is infantile Refsum disease known?
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What is its inheritance pattern?
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In the early childhood period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures

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

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

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

‘Pseudo-RP’

Usher syndrome

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

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?
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--RP-like fundus
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What is its inheritance pattern?
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What is the prognosis?
It is uniformly fatal by late childhood

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

What is its inheritance pattern?
AR

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

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

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

Primary RP
(aka typical RP)

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

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

What is its inheritance pattern?
AR

How do NALD pts present?
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What is the prognosis?
It is uniformly fatal by age 1 year

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What is the prognosis?
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‘Pseudo-RP’

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AR

How do Zellweger syndrome pts present?
In the neonatal period with:
-- RP-like fundus
-- Deafness
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What is the prognosis?
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Is NALD the same condition as Zellweger syndrome?
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What is its inheritance pattern?
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How do NALD pts present?
In the late infancy period with:
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What is the prognosis?
It is uniformly fatal by late childhood (if treatment is unsuccessful)

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

What is its inheritance pattern?
AR

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

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

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

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

---

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

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

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

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

---

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

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

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

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

Primary RP
(aka typical RP)

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

Pseudo-RP

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

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

What is its inheritance pattern?
AR

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

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

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

What is its inheritance pattern?
AR

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

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

---

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

**What is its inheritance pattern?**
AR

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

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

---

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

**What is its inheritance pattern?**
AR

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

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

---

**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

---

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

**What is its inheritance pattern?**
AR

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

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

What is its inheritance pattern?
AR

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

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

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

What is its inheritance pattern?
AR

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

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

Is Refsum disease known?
Yes

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

What is its inheritance pattern?
AR

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

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

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

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

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

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

---

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

Cerebrohepatorenal syndrome

**What is its inheritance pattern?**

AR

**How do Zellweger syndrome pts present?**

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

**What is the prognosis?**

It is uniformly fatal by age **1 year**

---

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

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

**What is its inheritance pattern?**

AR

**How do NALD pts present?**

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

**What is the prognosis?**

It is uniformly fatal by **late childhood**

---

**By what noneponymous name is infantile Refsum disease known?**

Infantile phytanic acid storage disease

**What is its inheritance pattern?**

AR

**How do infantile Refsum dz pts present?**

In the **early childhood** period with:
- RP-like fundus
- Deafness
- Hypotonia
- Seizures

**What is the prognosis?**

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

---

**How is infantile Refsum diagnosed?**

Via elevated serum phytanic acid levels (and VLCFAs)

**How is infantile Refsum treated?**

Dietary restriction of phytanic acid and phytol (a phytanic acid precursor); plasmapheresis may be employed acutely

---

**Hol up—you can treat this one??!!**

**How is infantile Refsum treated?**

Dietary restriction of phytanic acid and phytol (a phytanic acid precursor); plasmapheresis may be employed acutely
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

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

What is its inheritance pattern?
AR

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

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

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

What is its inheritance pattern?
AR

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

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

How is infantile Refsum disease treated?
Dietary restriction of phytanic acid and phytol (a phytanic acid precursor); plasmapheresis may be employed acutely

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

Infantile Refsum

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

Peroxisomal disorders tl;dr

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

Primary RP (aka typical RP)

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

‘Pseudo-RP’

RP-like fundus

Deafness

Hypotonia

Seizures

Abnormal facies

Peroxisomal disorders tl;dr

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

Peroxisomal disorders tl;dr

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

Peroxisomal disorders tl;dr

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

Peroxisomal disorders tl;dr

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

- Abetalipoproteinemia

**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
- Abetalipoproteinemia

‘Pseudo-RP’

RP-like fundus
Deafness
Hypotonia
Seizures
Abnormal facies

Infantile Refsum
Best prognosis

Peroxisomal disorders tl;dr

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

Primary RP (aka typical RP)

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

‘Pseudo-RP’

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

Peroxisomal disorders tl;dr

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

Abnormal facies

Deafness

Hypotonia

Seizures

RP-like fundus

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
- NALD

‘Pseudo-RP’

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

Peroxisomal disorders tl;dr

All three are inherited
DDx for an RP-like Fundus

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

RP-like fundus

- Deafness
- Hypotonia
- Seizures
- Abnormal facies

Infantile Refsum

NALD

Zellweger syndrome

All three are inherited AR

Peroxisomal disorders tl;dr
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

Peroxisomal disorders tl;dr

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

What is the inheritance pattern?

AR

### DDx for an RP-like Fundus

**Primary RP**

(aka typical RP)

**Secondary RP**

(aka Complex RP; Syndromic RP)

- Usher syndrome
- Peroxisomal disorders
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  - aka ‘Batten dz’
- Ciliopathies
- Abetalipoproteinemia
  - aka ‘Bassen-Kornzweig dz’
What are the neuronal ceroid lipofuscinoses (NCLs; collectively referred to as Batten dz)?
Neurodegenerative conditions stemming from the buildup of ceroid & lipofuscin in cells

What is the inheritance pattern?
AR

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

What is the prognosis?
Depending on the form, it is fatal by late childhood to early adulthood
Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

What is the inheritance pattern?
AR

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

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

What is the inheritance pattern?
AR

How do Batten dz pts present?
In infancy to early childhood with:
--RP-like fundus
--Seizures
--Myoclonus
--Microcephaly (in the infantile forms)
--Relentlessly progressive neurologic and cognitive decline
What are the neuronal ceroid lipofuscinoses (NCLs; collectively referred to as Batten dz)? Neurodegenerative conditions stemming from the buildup of ceroid & lipofuscin in cells.

What is the inheritance pattern? AR

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

What is the prognosis?

DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

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

‘Pseudo-RP’

‘Primary RP’

‘Secondary RP’
Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

What is the inheritance pattern?
AR

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

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

What is the inheritance pattern?
AR

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

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

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

How is Batten dz managed?
Supportively

DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

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

Neuronal ceroid lipofuscinoses
--aka 'Batten dz'

Abetalipoproteinemia
--aka 'Bassen-Kornzweig dz'
--Infantile Refsum dz

Zellweger syndrome
Neonatal adrenoleukodystrophy
Infantile Refsum dz

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

What is the inheritance pattern?
AR

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

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

What is the inheritance pattern?
AR

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

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

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

How is Batten dz managed?
Supportively

DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

What is the inheritance pattern?
AR

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

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

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

How is Batten dz managed?
Supportively
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
--RP-like fundus
--Seizures
--Myoclonus
--Microcephaly (in the infantile forms)
--Relentlessly progressive neurologic and cognitive decline

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

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

How is Batten dz managed?
Supportively

DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

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

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

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

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

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

‘Pseudo-RP’

What is a ciliopathy?

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

Cilia are ubiquitous. That said, ciliopathies primarily affect three organs—what are they?
Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
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The eyes, brain and kidneys
What is a ciliopathy?
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Cilia are organelles. That said, ciliopathies primarily affect three organs—what are they?
The eyes, brain and kidneys

The eyes??!! Which part of the eye contains cilia wiggling about?
What is a ciliopathy?
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Cilia are organelles. That said, ciliopathies primarily affect three organs—what are they?
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The eyes??!! Which part of the eye contains cilia wiggling about?
None. Remember, cilia come in two basic flavors: Motile, and nonmotile. It is the nonmotile type which is ubiquitous in the eye.

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‘Pseudo-RP’
What is a ciliopathy?
An inherited condition marked by abnormal structure and/or function of cilia. Cilia are organelles.

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

Cilia are organelles present in organs:
- The eyes
- Brain

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

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

DDx for an RP-like Fundus

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What is a ciliopathy?

An inherited condition marked by abnormal structure and/or function of cilia.

Cilia are organelles.

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

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

None. Remember, cilia come in two basic flavors: Motile, and nonmotile. It is the nonmotile type which is ubiquitous in the eye.

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

Cilia are organelles that are found in the eye, brain, and kidneys. The nonmotile type is ubiquitous in the eye.

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

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As for where in the PR complex the cilia is located: Let’s review PR structure

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.

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.
First things first: Which is a cone, and which a rod?
First things first: Which is a cone, and which a rod?
First things first: Which is a cone, and which a rod?

How can you tell?
First things first: Which is a cone, and which a rod?

How can you tell?
By the shape of their two words
First things first: Which is a cone, and which a rod?

How can you tell?
By the shape of their outer segments
First things first: Which is a cone, and which a rod?

How can you tell?
By the shape of their outer segments

Which portion constitutes their outer segments?
First things first: Which is a cone, and which a rod?

How can you tell?
By the shape of their outer segments

Which portion constitutes their outer segments?
First things first: Which is a cone, and which a rod?

How can you tell?
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Which portion constitutes their outer segments?

The presence of an outer segment implies the existence of an inner segment. Is this the case?
First things first: Which is a cone, and which a rod?

How can you tell?
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Which portion constitutes their outer segments?

The presence of an outer segment implies the existence of an inner segment. Is this the case?
Indeed it is
First things first: Which is a cone, and which a rod?

How can you tell?
By the shape of their outer segments

Which portion constitutes their outer segments?

The presence of an outer segment implies the existence of an inner segment. Is this the case?
Indeed it is

Where is the inner segment?
First things first: Which is a cone, and which a rod?

How can you tell?
By the shape of their outer segments

Which portion constitutes their outer segments?

The presence of an outer segment implies the existence of an inner segment. Is this the case? Indeed it is

Where is the inner segment?
First things first: Which is a cone, and which a rod?

How can you tell?
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Which portion constitutes their outer segments?

The presence of an outer segment implies the existence of an inner segment. Is this the case?
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Where is the inner segment?

What connects the inner and outer segments?
First things first: Which is a cone, and which a rod?

How can you tell?
By the shape of their outer segments

Which portion constitutes their outer segments?

The presence of an outer segment implies the existence of an inner segment. Is this the case? Indeed it is

Where is the inner segment?

What connects the inner and outer segments?
The cilium. **This is the cilia component of the PR!**
First things first: Which is a cone, and which a rod?

How can you tell?
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For more on PR and retinal histology, see slide-set R17
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‘Pseudo-RP’

- Which ciliopathies present with an RP-like fundus?

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

**‘Pseudo-RP’**

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

Cilia are ubiquitous. That said, ciliopathies primarily affect three organs--what are they? The eyes, brain and **kidneys**
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‘Pseudo-RP’

Got a mnemonic for remembering the ciliopathies?
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‘Pseudo-RP’

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

The mnemonic is…
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‘Pseudo-RP’

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

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

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

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

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

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‘Pseudo-RP’

What findings define the Bardet-Biedl complex?
- Hypogonadism
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- Mental retardation
- Extra fingers (polydactyly)
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And yeah, I know, Homer only has four digits per hand--paucidactyly, not polydactyly. But the rest fits him pretty well.
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Alström syndrome includes an RP-like fundus (duh), but shares only one other finding with the B-B complex— which one?

What findings define the Bardet-Biedl complex?
-- Hypogonadism?
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DDx for an RP-like Fundus

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

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

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

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

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

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

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

Both occur in childhood to teens
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‘Pseudo-RP’

How are BBS and AS managed?
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How are BBS and AS managed?
Supportively

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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' (look it up)

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

What is the prognosis?
It is highly variable
What are the main nonocular structures affected in JS?

The brainstem and cerebellum
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Joubert syndrome: Molar-tooth sign (look at the brainstem)
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Joubert syndrome: Facies. Note the large head, broad forehead
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  --aka ‘Batten dz’
- Ciliopathies
  --Bardet-Biedl syndrome
  --Alström syndrome
  --Joubert syndrome
  --Senior-Løken syndrome
- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

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

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

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

How is JS managed?
Supportively

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

Primary RP
(aka typical RP)

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

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

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

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‘Pseudo-RP’

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

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

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(aka typical RP)

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

‘Pseudo-RP’

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

How many familial oculorenal syndromes are there?

Including the four ciliopathies—six

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

Primary RP
(aka typical RP)

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

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

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

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How many familial oculorenal syndromes are there? Including the four ciliopathies—six
DDx for an RP-like Fundus

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- Usher syndrome
- Peroxisomal disorders
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  --Neonatal adrenoleukodystrophy

‘Pseudo-RP’

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

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

What are the other two?

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

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

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

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‘Pseudo-RP’

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

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

Are Alport and Lowe syndromes ciliopathies?

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

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

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They are familial oculorenal syndromes

- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
  -- Alport syndrome? No!
  -- Lowe syndrome? No!

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

Primary RP
(aka typical RP)

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

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

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

What are the other two?
Alport syndrome and Lowe syndrome

Are Alport and Lowe syndromes ciliopathies? No

Note: The latest (at the time I’m writing this—August 2023) copy of the Peds book asserts that Lowe syndrome is a ciliopathy. However, I’m pretty certain it’s wrong on this score. FWIW, Dr Google seems to agree with me. Still, caveat emptor.
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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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 oculorenal syndromes**

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

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

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

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

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

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‘Pseudo-RP’

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

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?
- Alport syndrome and Lowe syndrome

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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 oculorenal syndromes.

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What are the other two?
Alport syndrome and Lowe syndrome

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

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What are the other two?
Alport syndrome and Lowe syndrome

Abnormalities of what intraocular structure are associated with Alport and Lowe syndromes?
The lens
Primary RP
(aka typical RP)

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

‘Pseudo-RP’

- Abetalipoproteinemia
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- Bardet-Biedl syndrome
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For completeness’ sake:
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**What are the other two?**
Alport syndrome and Lowe syndrome

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

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

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic)
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- Peroxisomal disorders
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For completeness’ sake:
In three words (including syndromes), what sort of condition are these ciliopathies?
They are familial oculorenal syndromes.

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

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

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

Primary RP
(aka typical RP)

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

Pseudo-RP

- ‘Bassen-Kornzweig dz’
- Bardet-Biedl syndrome
- Alström syndrome
- Joubert syndrome
- Senior-Løken syndrome

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

Ciliopathies
- Bardet-Biedl syndrome
- Alström syndrome
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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**

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

For completeness' sake:

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

They are familial oculorenal syndromes

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

**‘Pseudo-RP’**

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

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*What are the other two?*

**Alport syndrome and Lowe syndrome**

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

**The lens**

What lens pathologies occur with Alport and Lowe syndromes?

- **Lenticonus!**
- Cataracts
- Microspherophakia

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

Lenticonus
Anterior lenticonus

Posterior lenticonus
Primary RP
(aka typical RP)

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

‘Pseudo-RP’

For completeness’ sake:
In three words (including syndromes), what sort of condition are these ciliopathies?
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What are the other two?
Alport syndrome and Lowe syndrome

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

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

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

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic)
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  -- Zellweger syndrome
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For completeness’ sake:
In three words (including syndromes), what sort of condition are these ciliopathies?
They are familial oculorenal syndromes

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

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

Are these associated with the ciliopathies?
No

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

What are the other two?
Alport syndrome and Lowe syndrome
Familial Oculorenal Syndromes *tl;dr*

- One sort
- The other sort
Familial Oculo-renal Syndromes *tl;dr*

- Ciliopathies
- Not Ciliopathies
Familial Oculoorenal Syndromes \textit{tl;dr}

- Ciliopathies
  - ?
  - ?
  - ?
  - ?

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

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

- Not Ciliopathies
  - Alport syndrome
  - Lowe syndrome
Familial Oculorenal Syndromes *tl;dr*

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

**Key Features**

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

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

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

---

**Key Features**

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

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

Not Ciliopathies
- Alport syndrome
- Lowe syndrome

**Key Features**

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

- Renal failure *with* hematuria
  - Classic eye finding: Lenticonus
Familial Oculorenal Syndromes

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

Not Ciliopathies
- Alport syndrome
- Lowe syndrome

Key Features

Renal failure without hematuria
Classic eye finding: Pigmentary retinopathy

Renal failure with hematuria
Classic eye finding: Lenticonus
Familial Oculorenal Syndromes

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

Not Ciliopathies
- Alport syndrome
- Lowe syndrome

Renal failure 
*without* hematuria

Classic eye finding:
*Pigmentary retinopathy*

Inheritance:

Renal failure 
*with* hematuria

Classic eye finding:
*Lenticonus*

Inheritance:
Familial Oculorenal Syndromes *tl;dr*

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

Not Ciliopathies
- Alport syndrome
- Lowe syndrome

**Key Features**

Renal failure
*without* hematuria

Classic eye finding:
*Pigmentary retinopathy*

Inheritance:
*AR*

Renal failure
*with* hematuria

Classic eye finding:
*Lenticonus*

Inheritance:
*X-linked*
Familial Oculorenal Syndromes \textit{tl;dr}

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

- Alport syndrome
- Lowe syndrome

Renal failure
- without hematuria

Classic eye finding:
- Pigmentary retinopathy

Inheritance:
- AR

Key Features

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

Renal failure
- with hematuria

Classic eye finding:
- Lenticonus

Inheritance:
- X-linked
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

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

--Senior-Løken syndrome

- Abetalipoproteinemia
  --aka 'Bassen-Kornzweig dz'
What is the underlying problem in abetalipoproteinemia?
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How is abetalipoproteinemia inherited?

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How does the absence of ApoB lead to secondary RP?

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**DDx for an RP-like Fundus**

- *What is the underlying problem in abetalipoproteinemia?*
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In addition to low vitamin A levels, there is a classic finding on peripheral blood smear—what is it?
Acanthocytosis of the RBCs

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

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- **Abetalipoproteinemia**
  --aka 'Bassen-Kornzweig dz'

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

Malabsorption secondary to GI surgery (e.g., gastric bypass; small-bowel resection)
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?
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With supplementary vitamins A & E

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

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

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

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

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  --Zellweger syndrome
  --Neonatal adrenoleukodystrophy
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  --aka 'Batten dz'
- Ciliopathies
  --Bardet-Biedl syndrome
  --Alström syndrome
  --Joubert syndrome
  --Senior-Løken syndrome
- A-beta lipoproteinemia
  --aka 'Bassen-Kornzweig dz'

‘Pseudo-RP’

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

Primary RP
(aka typical RP)

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

alpha

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

Is a-alpha-lipoproteinemia a thing?
Indeed it is
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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  --aka ‘Batten dz’
- Ciliopathies
  --Bardet-Biedl syndrome
  --Alström syndrome
  --Joubert syndrome
  --Senior-Løken syndrome
- α-lipoproteinemia
  --aka ‘Bassen-Kornzweig dz’

Is a-α-lipoproteinemia a thing? Indeed it is

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

Primary RP
(aka typical RP)

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

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

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
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- A-beta-lipoproteinemia
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Is a-alpha-lipoproteinemia a thing?
Indeed it is

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

Primary RP
(aka typical RP)

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

Is a-α-lipoprotein a thing?
Indeed it is

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?

These are some serious zebras, so don’t trip if you don’t know them.
Is α-lipoproteinemia a thing?

Indeed it is

Does the absence of α-lipoprotein affect the retina?

No, but it does affect the cornea

'Pseudo-RP'

262

Secondary RP

(aka Complex RP; Syndromic RP)

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

Primary RP

(aka typical RP)

DDx for an RP-like Fundus

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

-- LCAT deficiency
-- Fish eye disease
-- Tangier disease

Is α-α-lipoproteinemia a thing?

Indeed it is

Does the absence of α-lipoprotein affect the retina?

No, but it does affect the cornea
Is alpha-lipoproteinemia a thing?

Indeed it is.

Does the absence of $\alpha$-lipoprotein affect the retina?

No, but it does affect the cornea.

'Pseudo-RP'

Secondary RP (aka Complex RP; Syndromic RP)

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

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

- LCAT deficiency
- Fish eye disease
- Tangier disease

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

Does the absence of $\alpha$-lipoprotein affect the retina?

No, but it does affect the cornea.
Is α-lipoproteinemia a thing? Indeed it is. Does the absence of α-lipoprotein affect the retina? No, but it does affect the cornea.

'Pseudo-RP'

Secondary RP

(aka Complex RP; Syndromic RP)

- Usher syndrome
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  -- Alström syndrome
  -- Joubert syndrome
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- Abetalipoproteinemia
  -- aka 'Bassen-Korzystewicz dz'

Primary RP

(aka typical RP)

'Secondary RP'

- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
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  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka 'Bassen-Korzystewicz dz'

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

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

DDx for an RP-like Fundus

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

-- LCAT deficiency
-- Fish eye disease
-- Tangier disease

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

Primary RP (aka typical RP)

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

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

Primary RP (aka typical RP)
- Congenital syphilis
- Congenital rubella

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

‘Pseudo-RP’
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DDx for an RP-like Fundus

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  --Alström syndrome
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  --Senior-Løken syndrome
- Abetalipoproteinemia
  --aka 'Bassen-Kornzweig dz'

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

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

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269
DDx for an RP-like Fundus

Toxoplasmosis

HSV
DDx for an RP-like Fundus

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(aka typical RP)

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

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

Cancer-associated retinopathy

--aka 'Bassen-Kornzweig dz'
What is cancer-associated retinopathy (CAR)?
A paraneoplastic process in which, by unhappy coincidence, retinal cells possess surface proteins that cross-react with antigens found on cancer cells. If/when the immune system becomes sensitized to the cancer-cell antigens, it will subsequently target the same proteins in the retina, with devastating results.

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How does CAR present?
The pt will c/o progressive bilateral loss of VA and color vision, as well as nyctalopia.
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How does CAR present? What is the appearance of the retina on DFE?
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How does CAR present? What is the appearance of the retina on DFE?
The pt will c/o progressive bilateral loss of VA and color vision, as well as nyctalopia. Initially the retina’s appearance is unremarkable, leaving the physician at a loss to explain the pt’s symptoms. Eventually, the progressive retinal degeneration leads to arteriolar narrowing, RPE mottling, and ONH atrophy. (Sound familiar?)

Cancer-associated retinopathy

--aka 'Bassen-Kornzweig dz'
DDx for an RP-like Fundus
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How does CAR present? What is the appearance of the retina on DFE? What does testing reveal? The pt will c/o progressive bilateral loss of VA and color vision, as well as nyctalopia. Initially the retina’s appearance is unremarkable, leaving the physician at a loss to explain the pt’s symptoms. Eventually, the progressive retinal degeneration leads to arteriolar narrowing, RPE mottling, and ONH atrophy. (Sound familiar?)
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Cancer-associated retinopathy

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

CAR-associated VF loss
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Which cancer is most likely to produce CAR?

---
--aka ‘Bassen-Kornzweig dz’
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Which cancer is most likely to produce CAR? lung cancer is far and away the biggest culprit

Cancer-associated retinopathy --aka ‘Bassen-Kornzweig dz’
What is cancer-associated retinopathy (CAR)?
A paraneoplastic process in which, by unhappy coincidence, retinal cells possess surface proteins that cross-react with antigens found on cancer cells. If/when the immune system becomes sensitized to the cancer-cell antigens, it will subsequently target the same proteins in the retina, with devastating results.

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(287)
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**Is CAR treatable?**
Steroids, plasmapheresis, and/or IVIG have been thrown at it, but the visual prognosis is dismal

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’
- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  -- Toxoplasmosis
  -- HSV
  -- Others
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- Kearns-Sayre syndrome
What is the classic triad of Kearns-Sayre syndrome?
--?
--?
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--aka 'Bassen-Kornzweig dz'

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

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

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

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

DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP

‘Pseudo-RP’

Congenital syphillis
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

Kearns-Sayre syndrome
DDx for an RP-like Fundus

Kearns-Sayre syndrome: Pigmentary retinopathy
What is the classic triad of Kearns-Sayre syndrome?
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- CPEO
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What does CPEO stand for?

CPEO stands for Chronic progressive external ophthalmoplegia.

At what age do symptoms begin occurring?
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Of the classic triad, which is the first to occur?
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What does CPEO stand for?
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Which EOMs are typically affected first?
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What does it mean to say an ophthalmoplegia is 'external'?

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- CPEO
- Cardiac conduction abnormalities

What does CPEO stand for?
Chronic progressive external ophthalmoplegia

What does it mean to say an ophthalmoplegia is ‘external’?
It means the iris and CB are not affected/involved

Kearns-Sayre syndrome

DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP

‘Pseudo-RP’

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Congenital rubella
Infectious retinitis
- Toxoplasmosis
- HSV
- Others
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However, the disease is relentlessly progressive, and eventually all of the EOMs are paralyzed.

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(aka typical RP)

Secondary RP

‘Pseudo-RP’

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Congenital rubella
Infectious retinitis
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Kearns-Sayre syndrome

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

Primary RP (aka typical RP)

Secondary RP

‘Pseudo-RP’

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

- Toxoplasmosis
- HSV
- Others
- Neurological and developmental disorders
  - Usher syndrome
  - Peroxisomal disorders
  - Neuronal ceroid lipofuscinoses
  - Ciliopathies
  - Abetalipoproteinemia

Primary RP (aka typical RP)

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

- aka ‘Batten dz’
- aka ‘Bassen-Kornzweig dz’

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- Chronic progressive external ophthalmoplegia (CPEO)

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

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

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

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

Are the cardiac-conduction problems serious?

--aka 'Bassen-Kornzweig dz'

--aka typical RP

DDx for an RP-like Fundus

Primary RP

Secondary RP

‘Pseudo-RP’

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

Primary RP
(aka typical RP)

Secondary RP

‘Pseudo-RP’

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- Congenital rubella
- Infectious retinitis
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Kearns-Sayre syndrome

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‘Pseudo-RP’
- Toxoplasmosis
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--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’

--aka ‘Bassen-Kornzweig dz’

--aka typical RP

DDx for an RP-like Fundus

Primary RP

Secondary RP

‘Pseudo-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
Cancer-associated retinopathy

Congenital syphilis
Congenital rubella
Infectious retinitis

--aka typical RP

Secondary RP

‘Pseudo-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
Cancer-associated retinopathy

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

Primary RP
(aka typical RP)

Secondary RP

‘Pseudo-RP’

Congenital syphilis
Congenital rubella
Infectious retinitis
--Toxoplasmositis
--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?

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

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

Secondary RP
- Usher syndrome
- Peroxisomal disorders
- Neuronal ceroid lipofuscinoses
- Ciliopathies
- Abetalipoproteinemia
- Toxoplasmosis
- HSV
- Others

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

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

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

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

At what age do symptoms begin occurring?
Usually around 10 years

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

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

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

At what age do symptoms begin occurring? Usually around 10 years

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

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

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

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

At what age do symptoms begin occurring?
Usually around 10 years

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

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

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

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

At what age do symptoms begin occurring?
Usually around 10 years

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

Why is the order important?

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
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

Secondary RP

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

‘Pseudo-RP’

- Toxoplasmosis
- HSV
- Others
- Cancer-associated retinopathy

**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 around 10 years

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

*Why is the order important?*
Because in making the proper diagnosis, the astute eye dentist can refer the pt to a cardiologist before s/he has a fatal dysrhythmia
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

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

Next pseudo-RP condition (no question)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

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

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

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

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

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

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

#### ‘Pseudo-RP’
- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  - Toxoplasmosis
  - HSV
  - Others
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
- Severe uveitis
- DUSN
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

What does DUSN stand for?

- Diffuse unilateral subacute neuroretinitis

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
  --Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  --aka ‘Batten dz’
- Ciliopathies
  --Bardet-Biedl syndrome
- Abetalipoproteinemia

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

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

Primary RP
(aka typical RP)

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

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

What does DUSN stand for?
Diffuse unilateral subacute neuroretinitis

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
  - Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  - aka ‘Batten dz’
- Ciliopathies
  - Bardet-Biedl syndrome

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

What does DUSN stand for?
Diffuse unilateral subacute neuroretinitis

What is the cause?
Infestation by a type of bug
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

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

What does DUSN stand for?
Diffuse unilateral subacute neuroretinitis

What is the cause?
Infestation by a worm
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

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

What does DUSN stand for?
Diffuse unilateral subacute neuroretinitis

What is the cause?
Infestation by a worm (most commonly, specific worm)
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

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

What does DUSN stand for?
Diffuse unilateral subacute neuroretinitis

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

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

Primary RP
(aka typical RP)

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

What does DUSN stand for?
Diffuse unilateral subacute neuroretinitis

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

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

Primary RP
(aka typical RP)

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

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

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

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

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

For more on DUSN see slide-set R15

--aka ‘Batten dz’
- Ciliopathies
  --Bardet-Biedl syndrome
- DUSN

What does DUSN stand for?
Diffuse unilateral subacute neuroretinitis

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

Primary RP
(aka typical RP)

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

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

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

Primary RP
(aka typical RP)

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’
- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  -- Toxoplasmosis
  -- HSV
  -- Others
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
- Severe uveitis
- DUSN
- CRAO
- Drug toxicity
  -- ?
  -- ?
  -- Others
DDx for an RP-like Fundus

Primary RP (aka typical RP)
- congenital syphilis
- congenital rubella
- infectious retinitis
- cancer-associated retinopathy
- severe uveitis
- DUSN
- CRAO
- drug toxicity
- toxoplasmosis
- HSV
- others

Secondary RP (aka Complex RP; Syndromic RP)
- Usher syndrome
- peroxisomal disorders
  -- Zellweger syndrome
  -- neonatal adrenoleukodystrophy
  -- infantile Refsum dz
- neuronal ceroid lipofuscinoses
  -- aka ‘Batten dz’
- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’
- congenital syphilis
- congenital rubella
- infectious retinitis
  -- toxoplasmosis
  -- HSV
  -- others
- cancer-associated retinopathy
- Kearns-Sayre syndrome
- severe uveitis
- DUSN
- CRAO
- drug toxicity
  -- hydroxychloroquine
  -- thioridazine
  -- others
DDx for an RP-like Fundus

Thioridazine: Pigmentary retinopathy

Hydroxychloroquine retinopathy
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

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

Hydroxychloroquine (Plaquenil) toxicity is addressed in detail in R25
What class of medicine is thioridazine?

Drug toxicity

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

Primary RP  
(aka typical RP)  

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

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

Drug toxicity  
-- Hydroxychloroquine  
-- Thioridazine  
-- Others  

What class of medicine is thioridazine?  
It is a phenothiazine  

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  

337
DDx for an RP-like Fundus

Primary RP (aka typical RP)
- Congenital syphilis
- Congenital rubella
- Infectious retinitis
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
- Severe uveitis
- DUSN
- CRAO
- Drug toxicity
  -- Hydroxychloroquine
  -- Others

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

‘Pseudo-RP’
- Hydroxychloroquine
- HSV
- Others

What class of medicine is thioridazine?
It is a phenothiazine

What are the phenothiazines used to treat?

Drug toxicity
-- Hydroxyzine
-- Others
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

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

What class of medicine is thioridazine?
It is a phenothiazine

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

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

**Primary RP**
(aka typical RP)

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

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

---

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

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

**Is thioridazine retinal toxicity dose-related?**

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

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

Not so long as the dose is at or below 800 mg/day.
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

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

What class of medicine is thioridazine?
It is a phenothiazine

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

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

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

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

- Usher syndrome
- Peroxisomal disorders
  --Zellweger syndrome
  --Neonatal adrenoleukodystrophy

‘Pseudo-RP’

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

Drug toxicity
--Hydroxychloroquine
--Thioridazine
--Others

What class of medicine is thioridazine?
It is a phenothiazine

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

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

Primary RP
(aka typical RP)

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

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

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How does thioridazine retinopathy present clinically?

Drug toxicity
--Hydroxychloroquine
--Thioridazine
--Others
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The pt with c/o blurry vision; DFE will reveal pigment stippling in the macula

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**Does thioridazine use require periodic ophthalmic evaluations like hydroxychloroquine does?**
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- Severe uveitis
- DUSN
- CRAO
- Drug toxicity
-- Toxoplasmosis
-- HSV
-- Others

Secondary RP
(aka Complex RP; Syndromic RP)
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- Peroxisomal disorders
-- Zellweger syndrome
-- Neonatal adrenoleukodystrophy
- Hydroxychloroquine
- Thioridazine
- Others

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

Does thioridazine use require periodic ophthalmic evaluations like hydroxychloroquine does?
Not so long as the dose is at or below 800 mg/day
What tests should be run to determine whether a pt has RP?

- An ERG (if it's not markedly abnormal, it's not RP)
- Kinetic (ie, Goldmann, not Humphrey) VF testing. Again—if it's not abnormal, it's not RP
- Dark adaptometry (ditto)

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- Ciliopathies
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How do you rule-in pseudo-RP?

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

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

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- ( +/- Infectious retinitis)
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  --Others
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- Kearns-Sayre syndrome
- ( +/- Severe uveitis)
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- Drug toxicity
  --Hydroxychloroquine
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How do you rule-in pseudo-RP?
Some causes can be identified by the presence (or absence) of certain signs/symptoms...
Some via pertinent lab results...
Some by their unilaterality...

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Some via pertinent lab results...
Some by their unilaterality...
Some by history...

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‘Pseudo-RP’
- Congenital syphilis

---

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

**Usher syndrome**:

---

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

**Batten disease**: Punt to a geneticist

**Ciliopathies**: Primarily clinical (confirmatory genetics by a geneticist), but remember the following:

---

**Joubert syndrome**: Molar-tooth sign on brainstem MRI

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

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---

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

--Usher syndrome: Check hearing

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

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

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-- The peroxisomal disorders: Check serum levels of ‘very long chain fatty acids’ +/- phytanic acid levels
How do you rule-in the causes of secondary RP?

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

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- **Abetalipoproteinemia**: As discussed
tl;dr starts on the next slide
(When you hear *RP-like fundus*…

DDx for an RP-like Fundus

? ? ?
DDx for an RP-like Fundus

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

(When you hear *RP-like fundus*…these three categories should instantly spring to mind—make sure they do!)
Next, make sure you can name the *five categories of secondary/syndromic RP*.
Next, make sure you can name the **five categories of secondary/syndromic RP**. Toggle back and forth between this slide and the previous one until you can name all five with ease!
Then, make sure you can name the three *peroxisomal disorders* and the four *ciliopathies*. 

DDx for an RP-like Fundus

Primary RP
*(aka typical RP)*

Secondary RP
*(aka Complex RP; Syndromic RP)*
- Usher syndrome
- Peroxisomal disorders
  - ?
  - ?
  - ?
- Neuronal ceroid lipofuscinoses *(aka Batten dz)*
- Ciliopathies
  - ?
  - ?
  - ?
  - ?
- Abetalipoproteinemia
  *(aka Bassen-Korzeig dz)*
Then, make sure you can name the three **peroxisomal disorders** and the four **ciliopathies**. Again, toggle back and forth between this slide and the previous one until you’ve mastered them all.
These are my best guesses (emphasis on guesses) regarding factoids that should be kept in mind for each.
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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‘Pseudo-RP’

Deaf/blind--
Usher syndrome

Elevated VLCFA---
Zellweger syndrome

Dreadful prognoses--
Neonatal adrenoleukodystrophy

Only Refsum treatable--
Infantile Refsum dz

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

Death teens/early adult--

Ciliopathies

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-Korfsweig dz)

These are my best guesses (emphasis on guesses) regarding factoids that should be kept in mind for each. Toggle!
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Elevated VLCFAs--

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Progressive neuro decline--

Death teens/early adult--

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Others

Cancer-associated retinopathy

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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)

- Deaf/blind--
- Elevated VLCFAs--
- Dreadful prognoses--
- Only Refsum treatable--
- Progressive neuro decline--
- Death teens/early adult--
- Cilia JABS you in the eye--
- Relentless renal failure--
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Secondary RP
(aka Complex RP; Syndromic RP)

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

Speaking of guesses… These are the causes of pseudo-RP I would pay particular attention to.
Note: I’m not suggesting the other conditions are low-yield topics for the OKAP. Rather, I think they are unlikely to be the correct answer to a question intended to test your knowledge of RP imitators.

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