In my teaching experience, this is one of the more challenging topics in ophthalmology. (The corneal dystrophies are up there too.) While I won’t say this set makes learning the material easy—there’s simply too much esoterica for that—I do think it makes it easier.

My point here is not to toot my own horn, but rather to give hope and encouragement—*you can master this topic!*

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

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

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

What two vision issues are the defining characteristics of RP?

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

What two vision issues are the defining characteristics of RP?
--Nyctalopia
--Visual field loss
DDx for an RP-like Fundus

Typical pattern of VF loss in RP: Mid-peripheral scotomata \(\rightarrow\) coalesce into *partial* rings \(\rightarrow\) coalesce into *complete* ring \(\rightarrow\) expand rapidly *outward* \(\rightarrow\) expand slowly *inward*
First things first. An RP-like fundus has three characteristic attributes. What are they?
--Bone spicules
--Waxy disc pallor
--Arteriolar narrowing

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

Results of what specialized testing modality are always abnormal in RP?
First things first. An RP-like fundus has three characteristic attributes. *What are they?*
--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)
Characterisic 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) The pt has RP (duh)
2) Primary RP (aka typical RP)
3) ?
4) ?

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

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

‘Pseudo-RP’

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

Primary RP
(aka typical RP)

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

- (a family of neurodegenerative diseases)

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

- (a disorder of fat metabolism)

‘Pseudo-RP’
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘Pseudo-RP’
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

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

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

Pseudo-RP

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'
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?

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

Usher syndrome = Retinitis pigmentosa + sensorineural deafness

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

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

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

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

Primary RP (aka typical RP)
- Zellweger syndrome
- Neonatal adrenoleukodystrophy
- Infantile Refsum dz (aka 'Batten dz')
- 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…
-- Type II
-- Type III

DDx for an RP-like Fundus

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

Primary RP
(aka typical RP)

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- 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 [severity] hearing loss, RP and vestibular dysfunction
— Type II
— 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**
--**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…
---**Type III**

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

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

It is the most common cause thereof

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

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

- 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

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

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

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It is the most common cause thereof

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-- **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'
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... hearing loss; the RP severity?
  severity
  severity

- 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...progressive hearing loss; the RP varies in severity; vestibular function is sporadic

- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’
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--Type I manifests...in the first decade with profound hearing loss, RP and vestibular dysfunction
--Type II manifests...in the second decade with moderate hearing loss, RP; vestibular function is intact
--Type III has...progressive hearing loss; the RP varies in severity; vestibular function is sporadic

Abetalipoproteinemia
--aka 'Bassen-Kornzweig dz'

DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

Primary RP
(aka typical RP)

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

What are peroxisomal disorders?
Pseudo-RP

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

What are peroxisomal disorders?
A heterogeneous group of disorders of peroxisome function
What 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)
Primary RP (aka typical RP)

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

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

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

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

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

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

What specific peroxisomal disorders can manifest an LCA-type presentation?
--
--
--
What are peroxisomal disorders?
A heterogeneous group of disorders of peroxisome function

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

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

What specific peroxisomal disorders can manifest an LCA-type presentation?
--Zellweger syndrome
--Neonatal adrenoleukodystrophy (NALD)
--Infantile Refsum dz
What is the noneponymous name for Zellweger syndrome?

Cerebrohepatorenal syndrome

What is its inheritance pattern?

AR

How do Zellweger syndrome pts present?

In the neonatal period with:

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

What is the prognosis?

It is uniformly fatal by age 1 year
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

What is the non-eponymous 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
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  -- Zellweger syndrome
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‘Pseudo-RP’

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What is its inheritance pattern?
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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
- Neuronal ceroid lipofuscinoses

Pseudo-RP

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Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR
What is the noneponymous name for Zellweger syndrome? Cerebrohepatorenal syndrome

What is its inheritance pattern? AR

How do Zellweger syndrome pts present?
Pseudo-RP

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

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

DDx for an RP-like Fundus

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
DDx for 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:
--?
--?
--?
--?
--?
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
    -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses

‘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.
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Cerebrohepatorenal syndrome

What is its inheritance pattern?
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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?
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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

‘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?
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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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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

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

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

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

How is Zellweger managed?
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 managed?
Supportively

How is Zellweger syndrome diagnosed?
By the constellation of findings (along with elevated levels of VLCFA in the blood)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

What is the nonempirical 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
DDx for 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
-- 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
DDx for 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 nonepithelial condition associated with Zellweger syndrome?**
Cerebrohepatorenal syndrome

**What is its inheritance pattern?**
AR

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

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

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

**What is its inheritance pattern?**
AR

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

**What is the prognosis?**
It is uniformly fatal by late childhood
DDx for 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
-- Seizures
-- Abnormal facies

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

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

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

Primary RP
(aka typical RP)

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

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

What is its inheritance pattern?
AR

Note: Both are inherited AR
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

DDx for an RP-like Fundus

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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

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What is its inheritance pattern?
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In the late infancy period with:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures

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

Primary RP
(aka typical RP)

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

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

What is its inheritance pattern?
AR

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

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

How do NALD pts present?
In the ‘age’ period
DDx for 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
  --Seizures
  --Abnormal facies

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

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

What is its inheritance pattern?
AR

How do NALD pts present?
In the late infancy period

What is the noneponymous name for infantile Refsum dz?
Neonatal adrenoleukodystrophy
DDx for 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 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

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

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

Primary RP
(aka typical RP)

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

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

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

What is its inheritance pattern?
AR

How do NALD pts present?
In the late infancy period with:
-- ?
-- ?
-- ?
-- ?
DDx for 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 nonetymological name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

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

What is its inheritance pattern?
AR

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

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

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

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

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

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

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

What is its inheritance pattern?
AR

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

What is the prognosis?
It is uniformly fatal by late childhood
DDx for 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
- Seizures
- Abnormal facies

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

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

What is its inheritance pattern?
AR

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

What is the prognosis?
It is uniformly fatal by late childhood
DDx for 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
-- Seizures
-- Abnormal facies

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

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

What is its inheritance pattern?
AR

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

What is the prognosis?
It is uniformly fatal by late childhood
DDx for 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
-- Seizures
-- Abnormal facies

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

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

What is its inheritance pattern?
AR

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

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

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

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
Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

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

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

And albeit dreadful, the prognosis for NALD is better than that for Zellweger
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

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

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What is its inheritance pattern?
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--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?
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
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- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz

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What is its inheritance pattern?
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(aka typical RP)

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

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’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

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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

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?
In NALD, the VLCFAs impair the function of adrenal glands by compromising the integrity of adrenal-cell membranes

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

How is NALD managed?
Supportively (just like Zellweger pts)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

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

How is NALD managed?
Supportively (just like Zellweger pts)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

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

How is NALD managed?
Supportively (just like Zellweger)

--Hypotonia
--Seizures

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

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

What is its inheritance pattern?
AR

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

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
- Abetalipoproteinemia

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

‘Pseudo-RP’

Is NALD the same condition as adrenoleukodystrophy? No!

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

What is its inheritance pattern? AR

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

It is uniformly fatal by age 1 year
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

'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?
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How do infantile Refsum dz pts present?
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-- RP-like fundus
-- Deafness
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Primary RP (aka typical RP)

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

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

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In the early childhood period

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

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

What is the prognosis?
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- Deafness
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- 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.
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Primary RP (aka typical RP)

Secondary RP (aka Complex RP; Syndromic RP)
- Usher syndrome
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  -- Zellweger syndrome
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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:
-- RP-like fundus
-- Deafness
-- Hypotonia
-- Seizures

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

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

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How do infantile Refsum dz pts present?
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What is the prognosis?
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-- Deafness
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In the early childhood period with:
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What is the prognosis?
It is uniformly fatal by late adulthood (if treatment is unsuccessful)

What is the noneponymous name for Infantile phytanic acid storage disease?
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What is its inheritance pattern?
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How do Infantile phytanic acid storage disease pts present?
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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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In the late infancy period with:
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What is the prognostic outcome?
It is uniformly fatal by late childhood

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

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

What is its inheritance pattern?
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What is the prognosis?
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- RP-like fundus
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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

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

What is NALD?
Neonatal adrenoleukodystrophy

What is its inheritance pattern?
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-- Seizures
-- Abnormal facies

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

fatal by age 1 year
fatal by late childhood
fatal by early adulthood
DDx for an RP-like Fundus

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-- Seizures

What is the prognosis?
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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
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-- 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)
Pseudo-RP

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

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

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

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)

Refsum disease known?
Yes

What is its inheritance pattern?
AR

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

What is the prognosis?
It is uniformly fatal by 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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‘Pseudo-RP’

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-- Seizures

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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)

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

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

Hol up—you can treat this one??!! How is infantile Refsum treated?
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
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It is uniformly fatal by early adulthood (if treatment is unsuccessful)

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Pseudo-RP

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

Signs/symptoms
- 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)

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‘Pseudo-RP’

RP-like fundus

Deafness

Hypotonia

Seizures

Abnormal facies

Peroxisomal disorders tl;dr

(Review slides--no questions)
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  -- 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)
Primary RP
(aka typical RP)

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‘Pseudo-RP’

RP-like fundus
Deafness
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Abnormal facies

Peroxisomal disorders tl;dr

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‘Pseudo-RP’

RP-like fundus
Deafness
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Seizures
Abnormal facies

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

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‘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)

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‘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
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- Abetalipoproteinemia

‘Pseudo-RP’

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

Worse prognosis

Peroxisomal disorders tl;dr

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

- Abetalipoproteinemia
- Peroxismal disorders tl;dr

Peroxisomal disorders tl;dr
(Review slides--no questions)
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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  -- Zellweger syndrome
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‘Pseudo-RP’

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

Peroxisomal disorders tl;dr

All three are inherited

(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
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  -- Infantile Refsum dz

'Pseudo-RP'

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

Infantile Refsum
NALD
Zellweger syndrome

All three are inherited AR

Peroxisomal disorders tl;dr

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

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

Only infantile Refsum is treatable
What are the neuronal ceroid lipofuscinoses (NCLs; collectively referred to as Batten dz)?
What are the neuronal ceroid lipofuscinoses (NCLs; collectively referred to as Batten dz)? Neurodegenerative conditions stemming from the buildup of two substances in cells.
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

What is the inheritance pattern?
AR

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

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

What is the inheritance pattern?

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

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

What is the inheritance pattern?

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

What is the inheritance pattern?
AR

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

DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

Usher syndrome

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

Neuronal ceroid lipofuscinoses
  -- aka ‘Batten dz’

Ciliopathies

Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

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

Secondary RP
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- Usher syndrome
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'Pseudo-RP'

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

What is the inheritance pattern?
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DDx for an RP-like Fundus
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--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
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?
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  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka 'Batten 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?
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How do Batten dz pts present?
In infancy to early childhood with:
- RP-like fundus
- Seizures
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- 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?
- Acetyl-CoA transferase deficiency (aka 'Bassen-Kornzweig dz')
- Relentlessly progressive neurologic and cognitive decline

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?
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DDx for an RP-like Fundus

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  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka ‘Batten dz’
- Ciliopathies

‘Pseudo-RP’

What is a ciliopathy?

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

The eyes, brain and kidneys.
What is a ciliopathy?
An inherited condition marked by abnormal structure and/or function of cilia
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- Ciliopathies

‘Pseudo-RP’

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

Cilia are organelles that move within the eyes, brain, kidneys, etc.

The eyes??!! Which part of the eye contains cilia wiggling about?
Primary RP
(aka typical RP)

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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 that are abundant in the eye, brain, kidney...

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

Primary RP

 Secondary RP

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

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**DDx for an RP-like Fundus**

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  - aka ‘Batten dz’
- **Ciliopathies**

**‘Pseudo-RP’**

---

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

Recall that, fundamentally, RP is a **one word** disorder.

**Cilia are organelles in the brain.**

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

None. Remember, cilia come in two basic flavors: Motile, and nonmotile. It is the **non**motile type which is ubiquitous in the eye.
DDx for an RP-like Fundus

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‘Pseudo-RP’

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

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- Ciliopathies

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

Cilia are organelles with a whip-like lobe that protrudes into the surrounding area. 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.

The eyes are part of the brain.
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

Which ciliopathies present with an RP-like fundus?

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

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DDx for an RP-like Fundus

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

Which ciliopathies present with 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 ubiquitous. That said, ciliopathies primarily affect three organs--what are they?
The eyes, brain and kidneys

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

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

Got a mnemonic for remembering the ciliopathies?
DDx for an RP-like Fundus

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

The mnemonic is…
DDx for an RP-like Fundus

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

‘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

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

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

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  -- aka ‘Batten dz’
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  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome

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

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

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  -- Joubert syndrome

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

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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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

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

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What are they?

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How are BBS and AS managed?

‘Pseudo-RP’
DDx for an RP-like Fundus

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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?

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

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

What is the prognosis?
It is highly variable
What are the main nonocular structures affected in JS?
The brainstem and cerebellum
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(aka typical RP)

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‘Pseudo-RP’

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The brainstem and cerebellum

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DDx for an RP-like Fundus

Joubert syndrome: Molar-tooth sign (look at the brainstem)
DDx for an RP-like Fundus

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-- Disordered breathing (hyperpnea or apnea)
-- Intellectual and motor deficits
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DDx for an RP-like Fundus

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

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How is JS managed?
Supportively

What is the prognosis?
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  -- Senior-Løken syndrome

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

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(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
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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 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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  - \textit{aka} typical RP

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      - Zellweger syndrome
      - Neonatal adrenoleukodystrophy

- **‘Pseudo-RP’**

---

**For completeness’ sake:**

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

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  - Bardet-Biedl 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?

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

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

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

- Ciliopathies
  -- Bardet-Biedl 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?

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

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They are familial oculorenal syndromes

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

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

Primary RP
(aka typical RP)

Secondary RP
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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?
DDx for an RP-like Fundus

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‘Pseudo-RP’

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

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
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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.

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

What are the other two?
Alport syndrome and Lowe syndrome

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

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

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

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What are the other two?
Alport syndrome and Lowe syndrome

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

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(aka typical RP)

Secondary RP
(aka Complex RP, Syndromic RP)
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‘Pseudo-RP’

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

What are the other two?
Alport syndrome and Lowe syndrome

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

For completeness’ sake:
In three words (including syndromes), what sort of condition are these ciliopathies?
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Abnormalities of what intraocular structure are associated with Alport and Lowe syndromes?

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‘Pseudo-RP’

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

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

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

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

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

What is the lens?

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

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The lens

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- Cataracts?
- Microspherophakia?

Which is the classic association with Alport and Lowe syndromes? Which should come first to mind?
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Abnormalities of what intraocular structure are associated with Alport and Lowe syndromes?
The lens

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

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

What are the other two?
Alport syndrome and Lowe syndrome
Anterior lenticonus

Posterior lenticonus
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Alport syndrome and Lowe syndrome

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What lens pathologies occur with Alport and Lowe syndromes?
- Lenticonus
- Cataracts
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Are these associated with the ciliopathies?
No

Are these associated with the ciliopathies?
No
Familial Oculorenal Syndromes 

- One sort
- The other sort
Familial Oculorenal Syndromes

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

- Ciliopathies
  - ?
  - ?
  - ?
  - ?

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

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

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

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

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

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

### Ciliopathies
- Joubert syndrome
- Alström syndrome
- Bardet-Biedl syndrome
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### 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
- Alport syndrome
- Lowe syndrome

**Not Ciliopathies**
- Renal failure *without* hematuria
  - Classic eye finding:
  - Pigmentary retinopathy
  - Lenticonus

**Renal failure *with* hematuria**
  - Classic eye finding:
Familial Oculorenal Syndromes *tl;dr*

Ciliopathies

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

Not Ciliopathies

- Alport syndrome
- Lowe syndrome

**Key Features**

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:

Key Features

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

Renal failure \textit{without} hematuria

Classic eye finding: \textit{Pigmentary retinopathy}

Inheritance: \textit{AR}

Not Ciliopathies

- Alport syndrome
- Lowe syndrome

Renal failure \textit{with} hematuria

Classic eye finding: \textit{Lenticonus}

Inheritance: \textit{X-linked}

\textbf{For more info on Alport and Lowe syndromes, see slide-set L4}
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‘Pseudo-RP’

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

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

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

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

How is it treated?
With supplementary vitamins A & E

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

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- **Abetalipoproteinemia**
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What is the most common cause of hypovitaminosis A? (It’s not abetalipoproteinemia)
Malabsorption secondary to GI surgery (eg, gastric bypass; small-bowel resection)
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- Abetalipoproteinemia
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In abetalipoproteinemia, β-lipoprotein is absent (that’s what the prefix -a- indicates). Is hypobetalipoproteinemia a thing?
In abetalipoproteinemia, \( \beta \)-lipoprotein is absent (that’s what the prefix -a- indicates). Is hypobetalipoproteinemia a thing?
Indeed it is, via a condition called ‘familial hypobetalipoproteinemia,’ and it can affect the retina (it’s mentioned in the BCSC books, but not addressed in detail)
DDx for an RP-like Fundus

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alpha

A beta lipoproteinemia

-- aka 'Bassen-Kornzweig dz'

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

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*aka typical RP*

**Secondary RP**  
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- Neuronal ceroid lipofuscinoses  
  -- *aka* 'Batten dz'
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  -- Bardet-Biedl syndrome  
  -- Alström syndrome  
  -- Joubert syndrome  
  -- Senior-Løken syndrome
- **alpha**  
  *A*-**beta** lipoproteinemia  
  -- *aka* 'Bassen-Kornzweig dz'

---

*Is a-***alpha***-lipoproteinemia a thing?*
Indeed it is, but it's not called that, for obvious reasons
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- Ciliopathies
  --Bardet-Biedl syndrome
  --Alström syndrome
  --Joubert syndrome
  --Senior-Løken syndrome
- alpha-lipoproteinemia
  --aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

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

*Does the absence of alpha-lipoprotein affect the retina?*
Is \textit{a-alpha-lipoproteinemia} a thing?
Indeed it is, but it’s not called that, for obvious reasons

Does the absence of \textit{\alpha-lipoprotein} affect the retina?
No, but it does affect the
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‘Pseudo-RP’

alpha

A beta-lipoproteinemia
-- aka ‘Bassen-Kornzweig dz’

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

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

Primary RP (aka typical RP)

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

alpha

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

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Does the absence of \( \alpha \)-lipoprotein affect the retina?
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  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

Low levels of α-lipoprotein are implicated in three corneal conditions. What are they?
-- LCAT deficiency
-- Fish eye disease
-- Tangier disease

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

Is a-α-lipoprotein a thing?
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(aka typical RP)

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‘Pseudo-RP’

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

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

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- Abetalipoproteinemia
  -- aka 'Bassen-Korzywieg dz'

‘Pseudo-RP’

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

…and \( \beta \)-lipoproteins, which lead to pathology of the retina

Low levels of \( \alpha \)-lipoprotein are implicated in three corneal conditions. What are they?
- LCAT deficiency
- Fish eye disease
- Tangier disease

---Abetalipoproteinemia (Bassen-Korzywieg dz)
--- Familial hypobetalipoproteinemia
**DDx for an RP-like Fundus**

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*(aka typical RP)*

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

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

Primary RP  
(aka typical RP)

Secondary RP  
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
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  -- Alström syndrome
  -- Joubert syndrome
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  -- aka ‘Bassen-Kornzweig dz’

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

Congenital rubella

Congenital syphilis
DDx for an RP-like Fundus

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(aka typical RP)

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  -- Joubert syndrome
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- Abetalipoproteinemia
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‘Pseudo-RP’
- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  --?
  --?
  -- Others
DDx for an RP-like fundus

Primary RP

(aka typical RP)

Secondary RP

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‘Pseudo-RP’

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

Toxoplasmosis

HSV
DDx for an RP-like Fundus

Primary RP (aka typical RP)
- Congenital syphilis
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- Infectious retinitis
- Cancer-associated retinopathy
- Pseudo-RP

Secondary RP (aka Complex RP; Syndromic RP)
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‘Pseudo-RP’
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‘Pseudo-RP’
- Congenital syphilis
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- Infectious retinitis
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  -- HSV
  -- Others
- Cancer-associated retinopathy
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.

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?) VF testing will reveal scotoma, often in a ring-like pattern. ERG will be abnormal early on—an important clue that something is seriously amiss.

Which cancer is most likely to produce CAR?

Small-cell lung cancer is far and away the biggest culprit.

How is CAR diagnosed?

Retinal Ab panels are available and should be ordered, but do not possess particularly impressive sensitivity and/or specificity. Given a significant index of suspicion, the ophthalmologist should initiate a workup in search of an undiagnosed cancer.

Is CAR treatable?

Steroids, plasmapheresis, and/or IVIG have been thrown at it, but the visual prognosis is dismal.
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---**Cancer-associated retinopathy**

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CAR-associated VF loss
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--aka ‘Bassen-Kornzweig dz’
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  --Alström syndrome
  --Joubert syndrome
  --Senior-Løken syndrome
- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’

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

Primary RP
(aka typical RP)

Secondary RP

‘Pseudo-RP’

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

What is the classic triad of Kearns-Sayre syndrome?
--
--
--

-- aka ‘Bassen-Kornzweig dz’

What is the classic finding on muscle biopsy?

Ragged red fibers

At what age do symptoms begin occurring?

Usually shortly before age 10 years

Of the classic triad, which is the first to occur?
The ophthalmoplegia (usually the ptosis)
What is the classic triad of Kearns-Sayre syndrome?

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

Kearns-Sayre syndrome

--aka 'Bassen-Kornzweig dz'

DDx for an RP-like Fundus

Primary RP (aka typical RP)

Secondary RP

'Secondary RP'

--Zellweger syndrome
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--aka 'Batten dz'

--aka 'Bassen-Kornzweig dz'

Congenital syphilis
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What is the classic triad of Kearns-Sayre syndrome?
Kearns-Sayre syndrome: Pigmentary retinopathy
DDx for an RP-like Fundus

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Kearns-Sayre syndrome

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uciones
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CPEO

What does CPEO stand for?

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

What does CPEO stand for?
Chronic progressive external ophthalmoplegia

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

--aka ‘Bassen-Kornzweig dz’

--aka ‘Batten dz’

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--Neonatal adrenoleukodystrophy
--Infantile Refsum dz
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Primary RP
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- Senior-Løken syndrome
- 'Batten dz'
- 'Bassen-Kornzweig dz'

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

**'Pseudo-RP'**

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

What does CPEO stand for?
Chronic progressive external ophthalmoplegia

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

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

CPEO
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

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’

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

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

DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP

‘Pseudo-RP’

- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  --Toxoplasmosis
  --HSV
  --Others
- Cancer-associated retinopathy
  Kearns-Sayre syndrome
  --aka 'Bassen-Kornzweig dz'

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

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

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

Primary RP (aka typical RP)

Secondary RP

‘Pseudo-RP’

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

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

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

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

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

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

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

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

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

Primary RP
(aka typical RP)

Secondary RP

‘Pseudo-RP’

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

Kearns-Sayre syndrome

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

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

What is the classic finding on muscle biopsy?

- aka 'Bassen-Kornzweig dz'
- aka 'Batten dz'
- aka 'Bassen-Kornzweig dz'

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

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

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‘Ragged red fibers’
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--Pigmentary retinopathy
--CPEO
--Cardiac conduction abnormalities

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

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

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

Of the classic triad, which is the first to occur?
The ophthalmoplegia (usually the ptosis)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP

‘Pseudo-RP’

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

Kearns-Sayre syndrome

What is the classic triad of Kearns-Sayre syndrome?
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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’

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

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

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

Secondary RP

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

Primary RP

(aka typical RP)

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

DDx for an RP-like Fundus

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

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

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

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

**Of the classic triad, which is the first to occur?**
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A mitochondrial disease

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

--aka ‘Bassen-Kornzweig dz’

Kearns-Sayre syndrome
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP

‘Pseudo-RP’

- Congenital syphilis
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  -- Toxoplasmosis
  -- HSV
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Kearns-Sayre syndrome

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A mitochondrial disease

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

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

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

Why is the order important?

Kearns-Sayre syndrome
DDx for an RP-like Fundus

Primary RP (aka typical RP)
- What is the classic triad of Kearns-Sayre syndrome?
  - Pigmentary retinopathy
  - CPEO
  - Cardiac conduction abnormalities

Secondary RP
- 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

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

Kearns-Sayre syndrome

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

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(aka typical RP)

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  --aka ‘Bassen-Kornzweig dz’

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  --HSV
  --Others
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- Severe uveitis

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

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(aka typical RP)

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(aka Complex RP; Syndromic RP)
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- Severe uveitis
- DUSN
DDx for an RP-like Fundus

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(aka typical RP)

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(aka Complex RP; Syndromic RP)

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  --aka ‘Batten dz’
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  --Bardet-Biedl syndrome
  --Others

‘Pseudo-RP’

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

What does DUSN stand for?

Diffuse unilateral subacute neuroretinitis

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
- Toxoplasmosis
- HSV
- Others

‘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

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(aka typical RP)

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(aka Complex RP; Syndromic RP)
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‘Pseudo-RP’
- Congenital syphilis
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  -- Toxoplasmosis
  -- HSV
  -- Others
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
- Severe uveitis
- DUSN

What does DUSN stand for?
Diffuse unilateral subacute neuroretinitis

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

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
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- Neuronal ceroid 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 type of bug (most commonly, specific bug)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

What does DUSN stand for?
Diffuse unilateral subacute neuroretinitis

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

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
- Toxoplasmosis
- HSV
- Others

‘Pseudo-RP’
- Congenital syphilis
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- 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
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  -- Neonatal adrenoleukodystrophy
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What does DUSN stand for?
Diffuse unilateral subacute neuroretinitis

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

Primary RP
(aka typical RP)

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

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

For more on DUSN, see slide-set R15. If it’s not available, an abbreviated coverage of DUSN can be found in the White Dot Syndrome set (R16). Or, email me and I’ll send you a copy of R15.
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
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- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  -- Toxoplasmosis
  -- HSV
  -- Others
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
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Note: 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
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- Congenital syphilis
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  -- 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
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  - Infantile Refsum dz
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  - Others
- Cancer-associated retinopathy
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- DUSN
- CRAO
- Drug toxicity
  - ?
  - ?
  - Others
DDx for an RP-like Fundus

Primary RP

(aka typical RP)

Secondary RP

(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
  -- Neonatal adrenoleukodystrophy
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- Congenital syphilis
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  -- 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
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- Drug toxicity
  -- Hydroxychloroquine
  -- Thioridazine
  -- Others

Note: Hydroxychloroquine (Plaquenil) toxicity is addressed in detail in its own slide-set
Primary RP
*(aka typical RP)*

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

‘Pseudo-RP’
- Congenital syphilis
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- Kearns-Sayre syndrome
- Severe uveitis
- DUSN
- CRAO

What class of medicine is thioridazine?

Drug toxicity
-- Hydroxychloroquine
-- Thioridazine
-- Others
Primary RP (aka typical RP)

Secondary RP (aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
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‘Pseudo-RP’
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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.
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

‘Pseudo-RP’

- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  --Toxoplasmosis
  --HSV
  --Others
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- 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?**
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
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‘Pseudo-RP’
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**What class of medicine is thioridazine?**
It is a phenothiazine

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

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

**Drug toxicity**
-- Hydroxychloroquine
-- Thioridazine
-- Others
What class of medicine is thioridazine?
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Their main use is as antipsychotics

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

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- 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
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How does thioridazine retinopathy present clinically?

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

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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

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

Does thioridazine use require periodic ophthalmic evaluations like hydroxychloroquine does?

Drug toxicity
--Hydroxychloroquine
--Thioridazine
--Others
What class of medicine is thioridazine?
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What are the phenothiazines used to treat?
Their main use is as antipsychotics

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

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

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

DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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  --Others
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
- Severe uveitis
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- CRAO

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

Primary RP

(aka typical RP)

Secondary RP

(aka Complex RP; Syndromic RP)

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

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

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(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

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

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

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(aka typical RP)

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

How do you rule-in pseudo-RP?
- Ciliopathies
  --Bardet-Biedl syndrome
  --Alström syndrome
  --Joubert syndrome
  --Senior-Løken syndrome
- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’
DDx for an RP-like Fundus

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

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

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

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

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

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

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

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

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

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(aka typical RP)

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(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
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- Infectious retinitis
  -- Toxoplasmosis
  -- HSV
  -- Others
- Cancer-associated retinopathy
- Kearns-Sayre syndrome
- Severe uveitis
- DUSN
- CRAO
- Drug toxicity
  -- Hydroxychloroquine
  -- Thioridazine
  -- Others

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

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

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

‘Pseudo-RP’
- Congenital syphilis

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

- Usher syndrome:

-- Check hearing

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

- Batten disease:
  - Punt to a geneticist

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

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

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

‘Pseudo-RP’
- Congenital syphilis

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

-- Usher syndrome: Check hearing

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

-- Neuronal ceroid lipofuscinoses: Punt to a geneticist

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

-- Abetalipoproteinemia: As discussed

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

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

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

‘Pseudo-RP’

- Congenital syphilis
  -- *Usher syndrome*: Check hearing
  -- The peroxisomal disorders:

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

**Primary RP**
(aka typical RP)

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

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**DDx for an RP-like Fundus**

- **Usher syndrome**: Check hearing
- **The peroxisomal disorders**: Check serum levels of ‘very long chain fatty acids’ +/- phytanic acid levels
How do you rule-in the causes of secondary RP?

**Primary RP**
(aka typical RP)

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(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

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-- **Usher syndrome**: Check hearing

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

-- **Batten disease**: Check serum levels of ‘very long chain fatty acids’ +/- phytanic acid levels
DDx for an RP-like Fundus

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

‘Pseudo-RP’
- Congenital syphilis

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

-- Usher syndrome: Check hearing

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

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

Primary RP
(aka typical RP)

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(aka Complex RP; Syndromic RP)

DDx for an RP-like Fundus

'Pseudo-RP'

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

Neuronal ceroid lipofuscinoses
- Zellweger syndrome
- Neonatal adrenoleukodystrophy
- Infantile Refsum disease

- "Bassen-Kornzweig dz"
- "Batten dz" aka 'Batten disease'

Peroxisomal disorders
- Abetalipoproteinemia
- Peroxisomal disorders
- The peroxisomal disorders: Check serum levels of 'very long chain fatty acids', phytanic acid levels.

Usher syndrome
- Neonatal adrenoleukodystrophy
- "Bassen-Kornzweig dz"
- "Batten dz"

Ciliopathies
- The peroxisomal disorders: Check serum levels of 'very long chain fatty acids', phytanic acid levels.

- "Bassen-Kornzweig dz"
- "Batten dz"

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  -- Infantile Refsum dz
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  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’
- Congenital syphilis

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

-- Usher syndrome: Check hearing

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

-- Batten disease: Punt to a geneticist

-- Ciliopathies: Primarily clinical (confirmatory genetics by a geneticist), but remember the following:
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(aka typical RP)

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

‘Pseudo-RP’
- Congenital syphilis

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

--Usher syndrome: Check hearing
--The peroxisomal disorders: Check serum levels of ‘very long chain fatty acids’ +/- phytanic acid levels
--Batten disease: Punt to a geneticist
--Ciliopathies: Primarily clinical (confirmatory genetics by a geneticist), but remember the following:
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  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’
- Congenital syphilis

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

-- Usher syndrome: Check hearing

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

-- Batten disease: Punt to a geneticist

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

-- Abetalipoproteinemia: As discussed
tl;dr starts on the next slide
(When you hear *RP-like fundus*…
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

‘Pseudo-RP’

(When you hear RP-like fundus…these three categories should instantly spring to mind—make sure they do!)
Next, make sure you can name the *five categories of secondary/syndromic RP*. 
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
  -- 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
- Abetalipoproteinemia
  (aka Bassen-Korzyweig dz)

‘Pseudo-RP’

Then, make sure you can name the three *peroxisomal disorders* and the four *ciliopathies*. Again, toggle back and forth between this slide and the previous one until you’ve mastered them all.
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

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

- Ciliopathies
  - Bardet-Biedl syndrome
  - Alström syndrome
  - Joubert syndrome
  - Senior-Løken syndrome
- Abetalipoproteinemia
  (aka Bassen-Korzweig dz)

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

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

Deaf/blind--

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

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

Usher syndrome
Peroxisomal disorders

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

Neuronal ceroid lipofuscinoses (aka Batten dz)

Ciliopathies

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

Abetalipoproteinemia
(aka Bassen-Korzyweig dz)

Deaf/blind--

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

Primary RP (aka typical RP)

Secondary RP (aka Complex RP; Syndromic RP)
- Deaf/blind--
- Elevated VLCFAs--
- Dreadful prognoses--
- Only Refsum treatable--
- Progressive neuro decline--
- Death teens/early adult--

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

- Neuronal ceroid lipofuscinoses (aka Batten dz)

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

Speaking of guesses…

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

- Acanthocytosis of RBCs--

- Abetalipoproteinemia (aka Bassen-Korzwieg dz)
Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

‘Pseudo-RP’

- Congenital syphilis
- Congenital rubella
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  --Toxoplasmosis
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  --Others
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- Kearns-Sayre syndrome
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- CRAO
- 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.