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

Primary RP (aka typical RP)

Secondary RP (aka Complex RP; Syndromic RP)
- (the most common syndromic association)
- (one of the ‘inborn errors of metabolism’ families)
- (a family of neurodegenerative diseases)
- (a family of diseases related to dysfunction of a ubiquitous organelle)
- (a disorder of fat metabolism)

‘Pseudo-RP’
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘Pseudo-RP’
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

- Neuronal ceroid lipofuscinoses
  -- aka eponym
- Ciliopathies

- Abetalipoproteinemia
  -- aka eponym-eponym
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

  - Neuronal ceroid lipofuscinoses
    --aka ‘Batten dz’

  - Ciliopathies

  - Abetalipoproteinemia
    --aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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‘Pseudo-RP’

Usher syndrome = Retinitis pigmentosa +

Abetalipoproteinemia
--aka 'Bassen-Kornzweig dz'

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

There are three subtypes of Usher syndrome—what are they called? How do they differ?
- 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
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'

‘Pseudo-RP’
DDx for an RP-like Fundus

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

- Abetalipoproteinemia
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(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

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- Peroxisomal disorders

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

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

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DDx for an RP-like Fundus

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--Type I
--Type II
--Type III

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

Primary RP

(aka typical RP)

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

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

--- **Type I** manifests…in the first decade with profound hearing loss, RP and vestibular dysfunction
--- **Type II** manifests…in the ___ decade with ___ severity hearing loss, RP; vestibular function is ___ status
--- **Type III**

- Abetalipoproteinemia
  --aka 'Bassen-Kornzweig dz'
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- **Type III** has...

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  --aka ‘Bassen-Kornzweig dz’
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-- **Type III** has... hearing loss; the RP in severity; vestibular function is

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  --aka ‘Bassen-Kornzweig dz’
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--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

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‘Pseudo-RP’

Abetalipoproteinemia
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Primary RP
(aka typical RP)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

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

Primary RP  
(aka typical RP)

Secondary RP  
(aka Complex RP; Syndromic RP)

- Usher syndrome
- Peroxisomal disorders
- Neuronal ceroid

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

Primary RP (aka typical RP)

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

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

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

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- 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
DDx for an RP-like Fundus

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

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

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

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

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

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

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

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

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Intracellular organelles that play key roles in many aspects of cell metabolism

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

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

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(aka typical RP)
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(aka typical RP)

Secondary RP
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- Usher syndrome
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  --Zellweger syndrome
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‘Pseudo-RP’

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

What is it in the neonatal period with:
- LCA
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What is the prognosis?
It is uniformly fatal by age 1 year
DDx for an RP-like Fundus

Secondary RP

Primary RP

(aka Complex RP; Syndromic RP)

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

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

Primary RP
(aka typical RP)

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- Usher syndrome
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‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the ‘age’ period

DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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‘Pseudo-RP’

What is the noneponymous name for Zellweger syndrome?
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How do Zellweger syndrome pts present?
In the neonatal period with:
--?
--?
--?
--?
--?
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Zellweger syndrome facies: High forehead; hypertelorism
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Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

What is Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
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How do Zellweger syndrome pts present?
In the neonatal period with:
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- Usher syndrome
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  -- Zellweger syndrome
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‘Pseudo-RP’

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- Usher syndrome
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  -- Zellweger syndrome
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‘Pseudo-RP’

Primary RP
(aka typical RP)

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

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
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

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

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

Primary RP
(aka typical RP)

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?

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

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

Primary RP
(aka typical RP)

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

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Cerebrohepatorenal syndrome

What is its inheritance pattern?
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-- LCA
-- Deafness
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What is the prognosis?
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No, that is an X-linked condition that presents later in childhood

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Pseudo-RP

DDx for an RP-like Fundus

Primary RP (aka typical RP)

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Note: Both are inherited AR

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What is its inheritance pattern?
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How do NALD pts present?
In the late infancy period

Neonatal adrenoleukodystrophy's onset occurs a little later than does Zellweger’s
DDx for an RP-like Fundus

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(aka typical RP)

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

What is its inheritance pattern?
AR

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

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

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

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

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

Secondary RP

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

Primary RP
(aka typical RP)

‘Pseudo-RP’

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

What is its inheritance pattern?
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-- LCA
-- Deafness
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-- Seizures
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What is the prognosis?
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No, that is an X-linked condition that presents later in childhood

What is its inheritance pattern?
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In the late infancy period with:
-- LCA
-- Deafness
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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
- Neuronal ceroid lipofuscinoses
- Ciliopathies
- Abetalipoproteinemia

‘Pseudo-RP’

---

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
- LCA
- Deafness
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- Usher syndrome
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‘Pseudo-RP’

What is the non-epithelial condition?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
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-- LCA
-- Deafness
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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:
-- LCA
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What is the prognosis?
It is uniformly fatal by late childhood
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Primary RP
(aka typical RP)

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

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

What is its inheritance pattern?
AR

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

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
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Cerebrohepatorenal syndrome

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In the late infancy period with:
--LCA
--Deafness
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--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
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What is the noneponymous name for Zellweger syndrome?
CerebrohepatoRenal syndrome

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

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

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

Primary RP
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What is the prognosis?
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What is the prognosis for Neonatal adrenoleukodystrophy?
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Supportively (just like Zellweger pts)
DDx for an RP-like Fundus

**Primary RP** (aka typical RP)

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

**‘Pseudo-RP’**

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

**What is its inheritance pattern?**
AR

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

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

---

**What is the noneponymous name for Neonatal adrenoleukodystrophy?**
--Infantile Refsum dz

**How does Neonatal adrenoleukodystrophy have adreno-in the name?**
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Via its clinical presentation, coupled with MRI of the brain revealing gross white-matter abnormalities, and the elevated serum VLCFAs

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

---

**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’

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

What is its inheritance pattern?
AR

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

What is the prognosis?
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What is its inheritance pattern?
AR

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

What is the prognosis?
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Primary RP
(aka typical RP)

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

‘Pseudo-RP’

Primary RP

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

Is NALD the same condition as adrenoleukodystrophy?

What is its inheritance pattern?
AR

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the neonatal period with:
-- LCA
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-- Seizures
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  --Zellweger syndrome
  --Neonatal adrenoleukodystrophy
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‘Pseudo-RP’

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

What is the noneponymous name for Zellweger syndrome? Cerebrohepatorenal syndrome

What is its inheritance pattern? AR

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

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

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

What is its inheritance pattern? AR

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

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

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

What is its inheritance pattern? AR

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

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

What is the noneponymous name for NALD? Infantile Refsum disease

What is its inheritance pattern? AR

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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What is the noneponymous name for Zellweger syndrome?
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What is its inheritance pattern?
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How do Zellweger syndrome pts present?
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**How do Zellweger’s and NALD, infantile Refsum’s is inherited AR**

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**Like Zellweger’s and NALD, infantile Refsum’s is inherited AR**

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

In the neonatal period:
-- LCA
-- Deafness
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In the late infancy period:
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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.
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- LCA
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It is uniformly fatal by late childhood

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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
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Is infantile Refsum disease known?

How is infantile Refsum diagnosed?

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Primary RP

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

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‘Pseudo-RP’

### Primary RP

- **Secondary RP**
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It is uniformly fatal by **late childhood** (if treatment is unsuccessful)

---

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

**What is its inheritance pattern?**
AR

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

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

---

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

---

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

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

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

---

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

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

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

---

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

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

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

---

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

What is its inheritance pattern?
AR

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

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

---

HOW IS INFANTILE REFSUM DIAGNOSED?
Via elevated serum phytanic acid levels (and VLCFAs)

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How is infantile Refsum treated?
Dietary restriction of phytanic acid and phytol (a phytanic acid precursor), +/- plasmapheresis acutely

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

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DDx for an RP-like Fundus

Secondary RP

(aka Complex RP; Syndromic RP)

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

Peroxisomal disorders tl;dr

LCA

Deafness

Hypotonia

Seizures

Abnormal facies

Infantile Refsum

Signs/symptoms

Peroxisomal disorders tl;dr

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

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- Usher syndrome
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LCA
Deafness
Hypotonia
Seizures
Abnormal facies

NALD

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)
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  -- Infantile Refsum dz

‘Pseudo-RP’

LCA
Deafness
Hypotonia
Seizures
Abnormal facies

**Peroxisomal disorders tl;dr**

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

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  -- Infantile Refsum dz

Primary RP
(aka typical RP)

‘Pseudo-RP’

Peroxisomal disorders tl;dr

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

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‘Pseudo-RP’

Deafness
Hypotonia
Seizures
Abnormal facies

Peroxisomal disorders tl;dr

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

Secondary RP

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

LCA
- Deafness
- Hypotonia
- Seizures
- Abnormal facies

Peroxisomal disorders tl;dr

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

Primary RP
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  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
- Ciliopathies
- Abetalipoproteinemia

‘Pseudo-RP’

Peroxisomal disorders

LCA
Deafness
Hypotonia
Seizures
Abnormal facies

Infantile Refsum

Best prognosis

Infantile Refsum dz (NALD)

Zellweger syndrome

Peroxisomal disorders tl;dr

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

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(aka Complex RP; Syndromic RP)
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Primary RP
(aka typical RP)

‘Pseudo-RP’

Peroxisomal disorders tl;dr

LCA
Deafness
Hypotonia
Seizures
Abnormal facies

infantile Refsum
NALD → Zellweger syndrome
Worse prognosis

(Review slides--no questions)
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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  -- Neonatal adrenoleukodystrophy
  -- Infantile Refsum disease

‘Pseudo-RP’

- Abetalipoproteinemia

LCA
- Deafness
- Hypotonia
- Seizures
- Abnormal facies

Peroxisomal disorders tl;dr

(Worst prognosis)

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

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‘Pseudo-RP’

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

LCA
Deafness
Hypotonia
Seizures
Abnormal facies

Infantile Refsum NALD Zellweger syndrome

All three are inherited

Peroxisomal disorders tl;dr

(Review slides--no questions)
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‘Pseudo-RP’

LCA
Deafness
Hypotonia
Seizures
Abnormal facies

**Peroxisomal disorders tl;dr**

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

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‘Pseudo-RP’

LCA
Deafness
Hypotonia
Seizures
Abnormal facies

Peroxisomal disorders tl;dr

Only infantile Refsum is treatable

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

Primary RP
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- Usher syndrome
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  -- Zellweger syndrome
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  -- Infantile Refsum dz
- Neuronal ceroid lipofuscinoses
  -- aka ‘Batten dz’
- Ciliopathies
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

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

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

Neurodegenerative conditions stemming from the buildup of lipofuscin in cells

What is the inheritance pattern?
AR

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

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

Primary RP
(aka typical RP)

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

What are the neuronal ceroid lipofuscinoses (NCLs; collectively referred to as Batten dz)? Neurodegenerative conditions stemming from the buildup of two substances in cells depending on the form, it is fatal by late childhood to early adulthood.
DDx for an RP-like Fundus

Primary RP

(aka typical RP)

Secondary RP

(aka Complex RP; Syndromic RP)

- Usher syndrome
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- Abetalipoproteinemia
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‘Pseudo-RP’

---

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

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

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(aka typical RP)

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

What is the inheritance pattern?

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

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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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What is the inheritance pattern?
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In infancy to early childhood with:
--
--
--

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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

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

Primary RP (aka typical RP)

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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 found in the eye, brain, and kidneys

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

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Pseudo-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 found in the eyes, brain, and kidneys.

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

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

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Secondary RP

‘Pseudo-RP’

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

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

Primary RP (aka typical RP)

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

‘Pseudo-RP’

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

Cilia are organelles. That said, ciliopathies primarily affect three organs—what are they?
The eyes, brain, and kidneys

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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).
DDx for an RP-like Fundus

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  --?
  --?
  --?
  --?

Which ciliopathies present with an RP-like fundus?

What is a ciliopathy?
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Cilia are ubiquitous. That said, ciliopathies primarily affect three organs--what are they? The eyes, brain and kidneys
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Note that **all of the ciliopathies are marked by relentlessly progressive renal failure resulting in ESRD early in life!**

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

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

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‘Pseudo-RP’

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

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

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

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

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

Alström syndrome has the same findings as Bardet-Biedl, with two additions:

The mnemonic is…
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- Mental retardation
- Extra fingers (polydactyly)
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The mnemonic is…’Double D’s’
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- DM type 2 (early onset)

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How are BBS and AS managed?

Alström syndrome has the same findings as Bardet-Biedl, with two additions:

What findings define the Bardet-Biedl complex?
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How are BBS and AS managed? Supportively

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‘Pseudo-RP’

What are the main nonocular structures affected in JS?

The brainstem and cerebellum

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

How do JS pts present?
In the late infancy period with:
- LCA
- Hypotonia
- Abnormal breathing (hyperpnea or apnea)
- Intellectual and motor deficits
- Seizures
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What is the prognosis?
It is highly variable
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Joubert syndrome: Molar-tooth sign
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Joubert syndrome: Facies. Note the large head, broad forehead.
**DDx for an RP-like Fundus**

1. **Primary RP** (aka typical RP)
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   - Neonatal adrenoleukodystrophy
   - Infantile Refsum dz (aka ‘Batten dz’)

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3. **‘Pseudo-RP’**

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Supportively

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  -- Joubert syndrome
  -- Senior-Løken syndrome

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

Primary RP (aka typical RP)

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

‘Pseudo-RP’

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

- **Ciliopathies**
  -- Bardet-Biedl syndrome
  -- Alström syndrome
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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

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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
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

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

- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

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

Primary RP (aka typical RP)

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

‘Pseudo-RP’

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

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

What are the other two?
- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’
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

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

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

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

What are the other two?
Alport syndrome and Lowe syndrome

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

Primary RP
(aka typical RP)

Secondary RP
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- Usher syndrome
- Peroxisomal disorders
  -- Zellweger syndrome
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‘Pseudo-RP’

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

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

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

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

Primary RP
(aka typical RP)

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

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

- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

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

- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

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

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

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)
- Usher syndrome
- Peroxisomal disorders
- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
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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.)

In three words (including syndromes), what are these ciliopathies?
They are familial oculorenal syndromes
**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
      - -- Bardet-Biedl syndrome
      - -- Alström syndrome
      - -- Joubert syndrome
      - -- Senior-Løken syndrome
    - Abetalipoproteinemia
      - *aka* ‘Bassen-Kornzweig dz’

- **‘Pseudo-RP’**

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

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

---

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

In three words (including syndromes), what are these ciliopathies? They are familial oculorenal syndromes.
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

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

- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’

\[\text{Are Alport and Lowe syndrome in the DDx for an RP-like fundus?} \]
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.

\[\text{What are the other two?} \]
Alport syndrome and Lowe syndrome

\[\text{Abnormalities of what intraocular structure are associated with Alport and Lowe syndromes?} \]
DDx for an RP-like Fundus

**Primary RP**
(aka typical RP)

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

**‘Pseudo-RP’**

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

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

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

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

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

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

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

Abetalipoproteinemia
- aka ‘Bassen-Kornzweig dz’

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

What are the other two?
Alport syndrome and Lowe syndrome

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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.

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

What are the other two?
Alport syndrome and Lowe syndrome

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

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

Are these associated with the ciliopathies?
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
- Neuronal ceroid lipofuscinoses
- Ciliopathies
  -- Bardet-Biedl syndrome
  -- Alström syndrome
  -- Joubert syndrome
  -- Senior-Løken syndrome
- Abetalipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

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

What are the other two?
Alport syndrome and Lowe syndrome

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

Are these associated with the ciliopathies?
No

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

What lens pathologies occur with Alport and Lowe syndromes?
- Lenticonus
- Cataracts
- Microspherophakia
Familial Oculorenal Syndromes *tl;dr*

- One sort
- The other sort
Familial Oculorenal Syndromes \textit{tl;dr}

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

- Ciliopathies
- Not Ciliopathies

-?
-?
-?
-?
Familial Oculorenal Syndromes *tl;dr*

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

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

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

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

**Key Features**
- Renal failure
- *hematuria*
Familial Oculorenal Syndromes *tl;dr*

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

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

**Key Features**
- Renal failure *without* hematuria
- Renal failure *with* hematuria
Familial Oculorenal Syndromes *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:

...
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 *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:

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- 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 *tl;dr*

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

Renal failure *without* hematuria

Classic eye finding: *Pigmentary retinopathy*

Inheritance: *AR*

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

Renal failure *with* hematuria

Classic eye finding: *Lenticonus*

Inheritance: *X-linked*

*For more info on Alport and Lowe syndromes, see slide-set L4*
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

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

- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’

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

Abetalipoproteinemia
--aka 'Bassen-Kornzweig dz'

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DDx for an RP-like Fundus

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

How is abetalipoproteinemia inherited?

--Senior-Løken syndrome

- Abetalipoproteinemia
  --aka 'Bassen-Kornzweig dz'

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

How is it treated?
With supplementary vitamins A & E
What is the underlying problem in abetalipoproteinemia? 
One of the lipoproteins (ApoB) is not synthesized by the body

How is abetalipoproteinemia inherited? 
AR

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Checking vitamin A levels is a good start

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

How is abetalipoproteinemia inherited?
AR

How does the absence of ApoB lead to secondary RP?

--Senior-Løken syndrome

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

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

How is abetalipoproteinemia inherited?
AR

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

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

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- Abetalipoproteinemia
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DDx for an RP-like Fundus

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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).

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

Acanthocytosis of the RBCs

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

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

How is it treated?
With supplementary vitamins A & E

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

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

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

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

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

What does acanthocytosis mean?
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*Abetalipoproteinemia*
—aka ‘Bassen-Kornzweig dz’
Acanthocytosis
DDx for an RP-like Fundus

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

How is it treated?

--Senior-Løken syndrome

- Abetalipoproteinemia
  --aka ‘Bassen-Kornzweig dz’
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One of the lipoproteins (ApoB) is not synthesized by the body

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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

--Senior-Løken syndrome

- Abetalipoproteinemia
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DDx for an RP-like Fundus

What is the underlying problem in abetalipoproteinemia?
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How is abetalipoproteinemia inherited?
AR

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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).

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

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

How is it treated?
With supplementary vitamins A & E

- Senior-Løken syndrome

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

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

**How is abetalipoproteinemia inherited?**
AR

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

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

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

---Senior-Løken syndrome

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

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

**What is the most common cause of hypovitaminosis A?** *(It’s not abetalipoproteinemia)*
Malabsorption secondary to GI surgery (eg, gastric bypass; small-bowel resection)
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP
(aka Complex RP; Syndromic RP)

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

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

Primary RP
(aka typical RP)

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

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

Primary RP
(aka typical RP)

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

‘Pseudo-RP’

Is \textit{a-alpha-lipoproteinemia} a thing?
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘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
DDx for an RP-like Fundus

Primary RP (aka typical RP)

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

alpha

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?
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

alpha

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 α-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
- \textbf{alpha}\textsuperscript{-}lipoproteinemia
  -- aka ‘Bassen-Kornzweig dz’

‘Pseudo-RP’

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

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

Primary RP
(aka typical RP)

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- Usher 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

‘Pseudo-RP’

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

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

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

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’

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

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

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

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

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

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?
DDx for 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’

‘Pseudo-RP’

So, the hypolipoproteinemias can be divided into those involving \textbf{α-lipoproteins}, which lead to pathology of the cornea…

…and \textbf{β-lipoproteins}, which lead to pathology of the retina

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

- \textit{LCAT} deficiency
- Fish eye disease
- Tangier disease

\textit{Abetalipoproteinemia} (Bassen-Korzweig)
-- Familial hypobetalipoproteinemia
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘Pseudo-RP’
- Congenital infection
- Congenital infection

Congenital syphilis
Congenital rubella
DDx for 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
Congenital rubella

Congenital syphilis
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

‘Pseudo-RP’
- Congenital syphilis
- Congenital rubella
- Infectious retinitis
  -- '?
  -- '?
  -- Others
DDx for 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
Toxoplasmosis

HSV
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

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

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

Primary RP
*(aka typical RP)*

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

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

What is cancer-associated retinopathy (CAR)?

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

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

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

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

Primary RP
(aka typical RP)

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

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

Primary RP
(aka typical RP)

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

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

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

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

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

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

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

Primary RP
(aka typical RP)

Secondary RP

‘Pseudo-RP’

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

What is the classic triad of Kearns-Sayre syndrome?
-- Pigmentary retinopathy
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The ophthalmoplegia (usually the ptosis)
Kearns-Sayre syndrome: Pigmentary retinopathy
What is the classic triad of Kearns-Sayre syndrome?
- Pigmentary retinopathy
- CPEO
- Cardiac conduction abnormalities

What does CPEO stand for?
Chronic progressive external ophthalmoplegia

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 does CPEO stand for?
Chronic progressive external ophthalmoplegia

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

DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP

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

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

Primary RP
(aka typical RP)

Secondary RP

‘Pseudo-RP’

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

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

What does CPEO stand for?
Chronic progressive external ophthalmoplegia

-aka ‘Bassen-Kornzweig dz’

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

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Ragged red fibers

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The ophthalmoplegia (usually the ptosis)

What does CPEO stand for?
Chronic progressive external ophthalmoplegia

Kearns-Sayre syndrome
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP

‘Pseudo-RP’

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

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?

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

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The ophthalmoplegia (usually the ptosis)

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Chronic progressive external ophthalmoplegia

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

Primary RP (aka typical RP)

Secondary RP

‘Pseudo-RP’

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

Primary RP (aka typical RP)

- Vasculitis
- Toxoplasmosis
- HSV
- Others

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

---

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

**What does CPEO stand for?**
Chronic progressive external ophthalmoplegia

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

---

**Kearns-Sayre syndrome**

---

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

---

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

---

**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)
239

1998

1999

2000

2002

2003

2004

CPEO
DDx for an RP-like Fundus

Primary RP
(aka typical RP)

Secondary RP

‘Pseudo-RP’

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

Kears-Sayre syndrome

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

Are the cardiac-conduction problems serious?

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

Primary RP
(aka typical RP)

Secondary RP

‘Pseudo-RP’

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

Kearns-Sayre syndrome

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

--aka ‘Bassen-Kornzweig dz’

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

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

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

Kearsns-Sayre syndrome

What is the classic triad of Kearsn-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'
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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-- Cardiac conduction abnormalities

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A mitochondrial disease

-- aka ‘Bassen-Kornzweig dz’

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

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

What is the classic finding on muscle biopsy?

--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?
‘Ragged red fibers’

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

Primary RP (aka typical RP)

Secondary RP

‘Pseudo-RP’

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At what age do symptoms begin occurring?

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-- aka 'Zellweger syndrome'

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-- Infantile Refsum dz

-- Bardet-Biedl syndrome

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

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Toxoplasmosis
HSV
Others
DDx for an RP-like Fundus

Primary RP
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The ophthalmoplegia (usually the ptosis)

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Secondary RP

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

Why is the order important?
Because in making the proper diagnosis, the astute ophthalmologist can refer the pt to a cardiologist before s/he has a fatal dysrhythmia

Kearns-Sayre syndrome
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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four-letter abb. for infectious cause
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What does DUSN stand for?
Diffuse unilateral subacute neuroretinitis

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

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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

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(aka typical RP)

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  What does DUSN stand for?
  Diffuse unilateral subacute neuroretinitis

What is the cause?
Infestation by a type of bug (most commonly, specific bug)

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What does DUSN stand for?
Diffuse unilateral subacute neuroretinitis

What is the cause?
Infestation by a worm (most commonly, Baylisacaris)
DUSN (circle indicates the worm’s location)
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Note: DUSN is addressed in detail in its own slide-set
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- Drug toxicity
  -- Hydroxychloroquine
  -- Thioridazine
  -- Others
Thioridazine: Pigmentary retinopathy

Hydroxychloroquine retinopathy
DDx for an RP-like Fundus

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

Drug toxicity
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Drug toxicity
-- Hydroxychloroquine
-- Thioridazine
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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?
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
What class of medicine is thioridazine?
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What are the phenothiazines used to treat?

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

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

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

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

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

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

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

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How do you rule-in pseudo-RP?
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Some by their unilaterality...

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Some by their unilaterality...
Some by history...

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

'Pseudo-RP'

Congenital syphilis

---

--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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Joubert syndrome: Molar-tooth sign on brainstem MRI

---

Senior-Løken: Check renal

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Abetalipoproteinemia: As discussed
How do you rule-in the causes of secondary RP?

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