Leber's Congenital Amaurosis

- LCA is an age-related variant of abb.

Leber's Congenital Amaurosis

- LCA is an age-related variant of RP

Leber's Congenital Amaurosis

- **LCA is an age-related variant of RP**

Note that our understanding of both LCA and RP is evolving, and that the above assertion may be amended soon!

Leber's Congenital Amaurosis

- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the Snellen - not Snellen range

Leber's Congenital Amaurosis

- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range

Leber's Congenital Amaurosis

- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the **20/200 – LP** range

Which end of the spectrum is more typical in LCA--the 20/200, or the LP?

Leber's Congenital Amaurosis

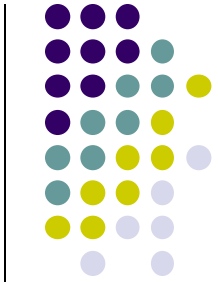
- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the **20/200 – LP** range

Which end of the spectrum is more typical in LCA--the 20/200, or the LP?
The LP. Most LCA pts have very, very poor vision

Leber's Congenital Amaurosis

- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age [ ], characterized as ['one word']

Leber's Congenital Amaurosis



- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'

Leber's Congenital Amaurosis

- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'

What is the most common presenting finding in hereditary retinal diseases?

Leber's Congenital Amaurosis

- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - **Nystagmus** by age 3 months, characterized as 'wandering'

What is the most common presenting finding in hereditary retinal diseases?
Nystagmus

Leber's Congenital Amaurosis

- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - **Nystagmus** by age 3 months, characterized as 'wandering'

What is the most common presenting finding in hereditary retinal diseases?
Nystagmus

What are some of the other classic hereditary retinal conditions that present with nystagmus?
--
--
--
--(There are many others)

Leber's Congenital Amaurosis

- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - **Nystagmus** by age 3 months, characterized as 'wandering'

What is the most common presenting finding in hereditary retinal diseases?
Nystagmus

What are some of the other classic hereditary retinal conditions that present with nystagmus?
--Albinism
--Achromatopsia
--Congenital stationary night blindness (CSNB)
--(There are many others)

Leber's Congenital Amaurosis

- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - **Nystagmus** by age 3 months, characterized as 'wandering'

What is the most common presenting finding in hereditary retinal diseases?
Nystagmus

What are some of the other classic hereditary retinal conditions that present with nystagmus?
--Albinism
--Achromatopsia
--Congenital stationary night blindness (CSNB)
--(There are many others)

And lest we forget:
--Any condition that interferes with vision from birth can cause a three words

Leber's Congenital Amaurosis

- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - **Nystagmus** by age 3 months, characterized as 'wandering'

What is the most common presenting finding in hereditary retinal diseases?
Nystagmus

What are some of the other classic hereditary retinal conditions that present with nystagmus?
--Albinism
--Achromatopsia
--Congenital stationary night blindness (CSNB)
--(There are many others)

And lest we forget:
--Any condition that interferes with vision from birth can cause a congenital *sensory* nystagmus

Leber's Congenital Amaurosis

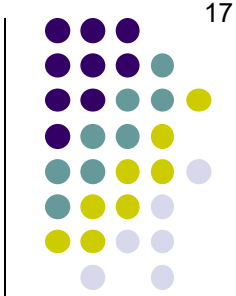
- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - **Nystagmus** by age 3 months, characterized as 'wandering'

What is the most common presenting finding in hereditary retinal diseases?
Nystagmus

What are some of the other classic hereditary retinal conditions that present with nystagmus?
--Albinism
--Achromatopsia
--Congenital stationary night blindness (CSNB)
--(There are many others)

And lest we forget:
--Any condition that interferes with vision from birth can cause a congenital *sensory* nystagmus
--Nystagmus from birth associated with normal vision is usually a three words

Leber's Congenital Amaurosis



- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - **Nystagmus** by age 3 months, characterized as 'wandering'

What is the most common presenting finding in hereditary retinal diseases?
Nystagmus

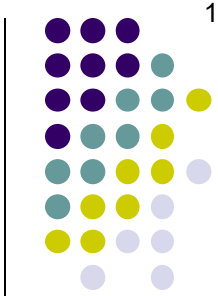
What are some of the other classic hereditary retinal conditions that present with nystagmus?
--Albinism
--Achromatopsia
--Congenital stationary night blindness (CSNB)
--(There are many others)

And lest we forget:
--Any condition that interferes with vision from birth can cause a congenital *sensory* nystagmus
--Nystagmus from birth associated with normal vision is usually a congenital *motor* nystagmus

Leber's Congenital Amaurosis

- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically one word in the # range (but can be one word

Leber's Congenital Amaurosis



- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically hyperopic in the +3 range (but can be myopic)

Leber's Congenital Amaurosis

- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically **hyperopic** in the +3 range (but can be myopic)

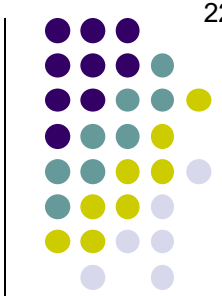
What is the significance of the hyperopia?

Leber's Congenital Amaurosis

- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically **hyperopic** in the +3 range (but can be myopic)

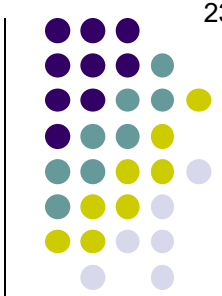
What is the significance of the hyperopia?
Probably nothing. Most infants and children are hyperopic until they start undergoing the process of emmetropization (which usually commences around age 7, and is completed by age 16). Because of their extremely poor vision, LCA eyes do not experience the stimulus needed to initiate and maintain the emmetropization process, and thus these eyes remain forever hyperopic.

Leber's Congenital Amaurosis



- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically hyperopic in the +3 range (but can be myopic)
  - Diagnosis of LCA requires a nearly-extinguished abb.

Leber's Congenital Amaurosis



- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically hyperopic in the +3 range (but can be myopic)
  - Diagnosis of LCA requires a nearly-extinguished ERG

Leber's Congenital Amaurosis

- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically hyperopic in the +3 range (but can be myopic)
  - Diagnosis of LCA requires a **nearly-extinguished ERG**

In what fundamental way does the ERG in LCA differ from that of most retinal dystrophies?

Leber's Congenital Amaurosis

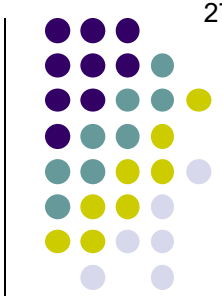
- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically hyperopic in the +3 range (but can be myopic)
  - Diagnosis of LCA requires a **nearly-extinguished ERG**

In what fundamental way does the ERG in LCA differ from that of most retinal dystrophies?
The ERG is almost undetectable from birth in infants with LCA. In contrast, the ERG in most retinal dystrophies will demonstrate slow, progressive diminution over time

Leber's Congenital Amaurosis

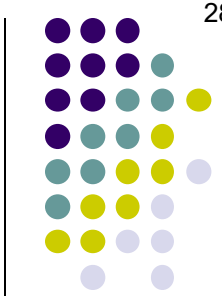
- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically hyperopic in the +3 range (but can be myopic)
  - Diagnosis of LCA requires a nearly-extinguished ERG
  - Pupils are [one word], and may be [one word]

Leber's Congenital Amaurosis



- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically hyperopic in the +3 range (but can be myopic)
  - Diagnosis of LCA requires a nearly-extinguished ERG
  - Pupils are sluggish, and may be paradoxical

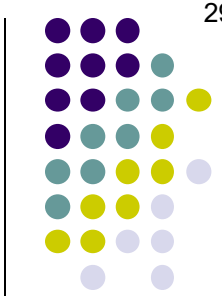
Leber's Congenital Amaurosis



- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically hyperopic in the +3 range (but can be myopic)
  - Diagnosis of LCA requires a nearly-extinguished ERG
  - Pupils are sluggish, and may be **paradoxical**

What are paradoxical pupils*?*

Leber's Congenital Amaurosis



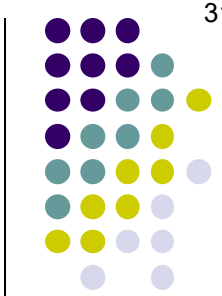
- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically hyperopic in the +3 range (but can be myopic)
  - Diagnosis of LCA requires a nearly-extinguished ERG
  - Pupils are sluggish, and may be **paradoxical**

What are paradoxical pupils*?*
Pupils that **dilate** in response to light

Leber's Congenital Amaurosis

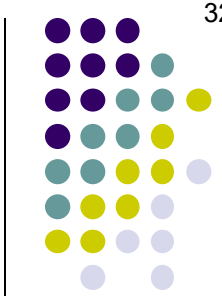
- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically hyperopic in the +3 range (but can be myopic)
  - Diagnosis of LCA requires a nearly-extinguished ERG
  - Pupils are sluggish, and may be paradoxical
  - Lenses are often one word

Leber's Congenital Amaurosis



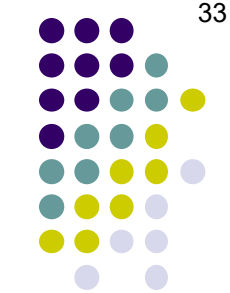
- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically hyperopic in the +3 range (but can be myopic)
  - Diagnosis of LCA requires a nearly-extinguished ERG
  - Pupils are sluggish, and may be paradoxical
  - Lenses are often cataractous

Leber's Congenital Amaurosis



- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically hyperopic in the +3 range (but can be myopic)
  - Diagnosis of LCA requires a nearly-extinguished ERG
  - Pupils are sluggish, and may be paradoxical
  - Lenses are often cataractous
  - The one word reflex may reflect an attempt to elicit some visual input via one word production

Leber's Congenital Amaurosis



- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically hyperopic in the +3 range (but can be myopic)
  - Diagnosis of LCA requires a nearly-extinguished ERG
  - Pupils are sluggish, and may be paradoxical
  - Lenses are often cataractous
  - The oculodigital reflex may reflect an attempt to elicit some visual input via phosphene production

Leber's Congenital Amaurosis

- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically hyperopic in the +3 range (but can be myopic)
  - Diagnosis of LCA requires a nearly-extinguished ERG
  - Pupils are sluggish, and may be paradoxical
  - Lenses are often cataractous
  - The oculodigital reflex may reflect an attempt to elicit some visual input via **phosphene** production

What are phosphenes*?*

Leber's Congenital Amaurosis

- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically hyperopic in the +3 range (but can be myopic)
  - Diagnosis of LCA requires a nearly-extinguished ERG
  - Pupils are sluggish, and may be paradoxical
  - Lenses are often cataractous
  - The oculodigital reflex may reflect an attempt to elicit some visual input via **phosphene** production

What are phosphenes*?*
The visual experience of 'shooting stars' that is produced by aggressive rubbing of the eyes

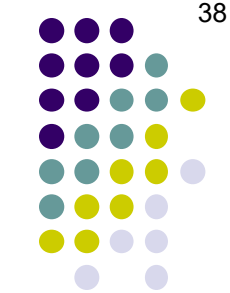
Leber's Congenital Amaurosis

- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically hyperopic in the +3 range (but can be myopic)
  - Diagnosis of LCA requires a nearly-extinguished ERG
  - Pupils are sluggish, and may be paradoxical
  - Lenses are often cataractous
  - The oculodigital reflex may reflect an attempt to elicit some visual input via phosphene production
  - Two classic corneal associations: abb. and

Leber's Congenital Amaurosis

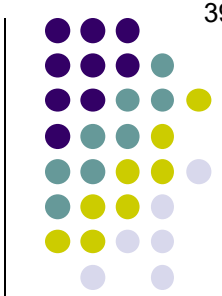
LCA: Oculodigital reflex

Leber's Congenital Amaurosis



- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically hyperopic in the +3 range (but can be myopic)
  - Diagnosis of LCA requires a nearly-extinguished ERG
  - Pupils are sluggish, and may be paradoxical
  - Lenses are often cataractous
  - The oculodigital reflex may reflect an attempt to elicit some visual input via phosphene production
  - Two classic corneal associations: KCN and keratoglobus

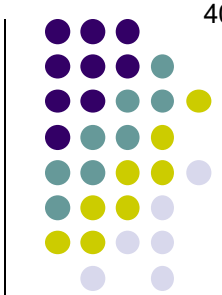
Leber's Congenital Amaurosis



- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically hyperopic in the +3 range (but can be myopic)
  - Diagnosis of LCA requires a nearly-extinguished ERG
  - Pupils are sluggish, and may be paradoxical
  - Lenses are often cataractous
  - The **oculodigital reflex** ma e visual input via phosphene
  - Two classic corneal associations: **KCN** and keratoglobus

Is there a relationship here?

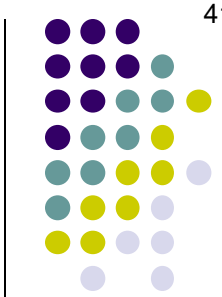
Leber's Congenital Amaurosis



- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically hyperopic in the +3 range (but can be myopic)
  - Diagnosis of LCA requires a nearly-extinguished ERG
  - Pupils are sluggish, and may be paradoxical
  - Lenses are often cataractous
  - The **oculodigital reflex** ma e visual input via phosphene p
  - Two classic corneal associations: **KCN** and keratoglobus

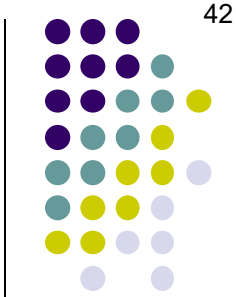
Is there a relationship here?
Possibly, in that the incessant eye-rubbing may contribute to the evelopment of KCN

Leber's Congenital Amaurosis



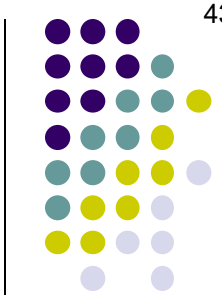
- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically hyperopic in the +3 range (but can be myopic)
  - Diagnosis of LCA requires a nearly-extinguished ERG
  - Pupils are sluggish, and may be paradoxical
  - Lenses are often cataractous
  - The oculodigital reflex may reflect an attempt to elicit some visual input via phosphene production
  - Two classic corneal associations: KCN and keratoglobus
  - The fundus appearance is highly variable, from one word to abb. -like to 'one word'

Leber's Congenital Amaurosis



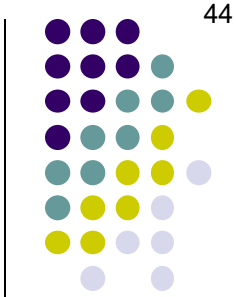
- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically hyperopic in the +3 range (but can be myopic)
  - Diagnosis of LCA requires a nearly-extinguished ERG
  - Pupils are sluggish, and may be paradoxical
  - Lenses are often cataractous
  - The oculodigital reflex may reflect an attempt to elicit some visual input via phosphene production
  - Two classic corneal associations: KCN and keratoglobus
  - The fundus appearance is highly variable, from normal to RP-like to 'marbleized'

Leber's Congenital Amaurosis



- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically hyperopic in the +3 range (but can be myopic)
  - Diagnosis of LCA requires a nearly-extinguished ERG
  - Pupils are sluggish, and may be paradoxical
  - Lenses are often cataractous
  - The oculodigital reflex may reflect an attempt to elicit some visual input via phosphene production
  - *The wide variation in fundus appearance has what implication re managing LCA?*
  - **The fundus appearance is highly variable, from normal to RP-like to 'marbleized'**

Leber's Congenital Amaurosis

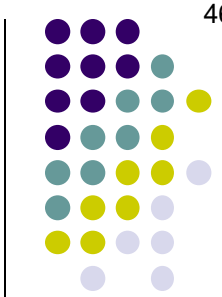


- LCA is an age-related variant of RP
- Presents with:
  - Severe ↓ VA in the 20/200 – LP range
  - Nystagmus by age 3 months, characterized as 'wandering'
  - Refractive error: Classically hyperopic in the +3 range (but can be myopic)
  - Diagnosis of LCA requires a nearly-extinguished ERG
  - Pupils are sluggish, and may be paradoxical
  - Lenses are often cataractous
  - The oculodigital reflex may reflect an attempt to elicit some visual input via phosphene production
  - *The wide variation in fundus appearance has what implication re managing LCA?*
    It implies that one cannot rely upon fundus appearance to make the diagnosis
  - **The fundus appearance is highly variable, from normal to RP-like to 'marbleized'**

Leber's Congenital Amaurosis

- LCA inheritance: abb.

Leber's Congenital Amaurosis

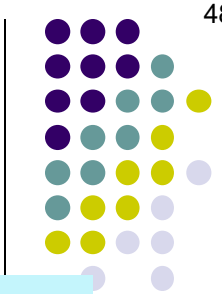


- LCA inheritance: AR

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals three words

Leber's Congenital Amaurosis

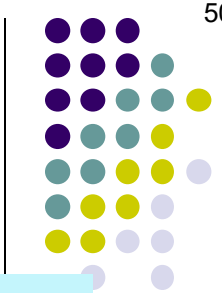


- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors

Leber's Congenital Amaurosis

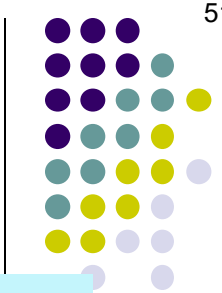
- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - two *classes* of disease
  -

Leber's Congenital Amaurosis



- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal disorders
  - Ciliopathies

Leber's Congenital Amaurosis

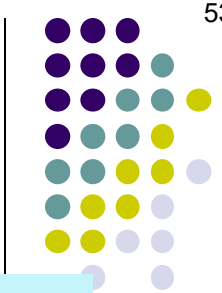


- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - Ciliopathies

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders*?*

Leber's Congenital Amaurosis



- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders*?*
    A heterogeneous group of disorders of peroxisome function

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders*?*
    A heterogeneous group of disorders of peroxisome function

    What is/are peroxisomes?

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *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

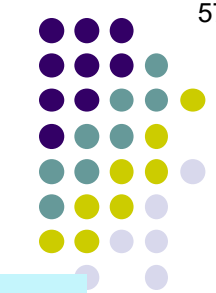
Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *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?

Leber's Congenital Amaurosis

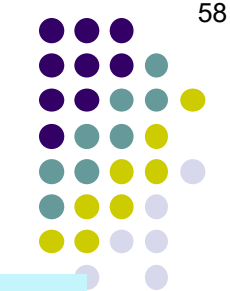


- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders*?*
    A heterogeneous group of disorders of peroxisome function

    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)

Leber's Congenital Amaurosis



- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  -
  -

What specific peroxisomal disorders can manifest an LCA-type presentation?
--
--
--

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

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

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

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

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

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**

What specific peroxisomal disorders can manifest an LCA-type presentation?

--**Zellweger syndrome**

--Neonatal adrenoleukodystrophy (NALD)

--Infantile Refsum dz

What are peroxisomal disorders?

A heterogeneous group of disorders of peroxisome function

n many aspects of cell metabolism

peroxisomal disorders that

chain fatty acids (VLCFA)

What is the noneponymous name for Zellweger syndrome?

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**

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

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

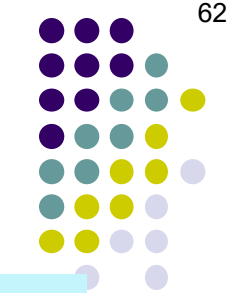
n many aspects of cell metabolism

peroxisomal disorders that

chain fatty acids (VLCFA)

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

Leber's Congenital Amaurosis



- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**

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

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

n many aspects of cell metabolism

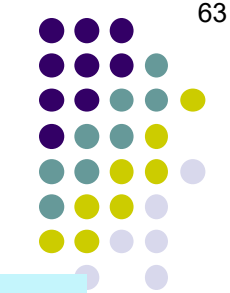
peroxisomal disorders that

chain fatty acids (VLCFA)

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?

Leber's Congenital Amaurosis



- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**

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

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

n many aspects of cell metabolism

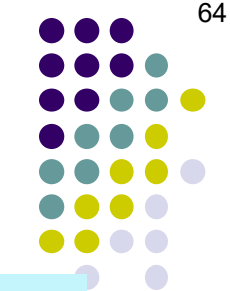
peroxisomal disorders that

chain fatty acids (VLCFA)

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

Leber's Congenital Amaurosis



- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**

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

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

n many aspects of cell metabolism

peroxisomal disorders that

chain fatty acids (VLCFA)

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**

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

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

n many aspects of cell metabolism

peroxisomal disorders that

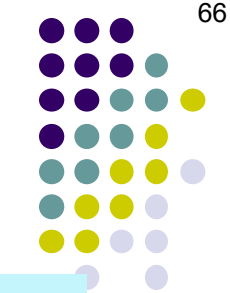
chain fatty acids (VLCFA)

What is the noneponymous name for Zellweger syndrome?
Cerebrohepatorenal syndrome

What is its inheritance pattern?
AR

How do Zellweger syndrome pts present?
In the [ ] period

Leber's Congenital Amaurosis



- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**

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

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

n many aspects of cell metabolism

peroxisomal disorders that

chain fatty acids (VLCFA)

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**

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

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

n many aspects of cell metabolism

peroxisomal disorders that

chain fatty acids (VLCFA)

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:
--***?***
--***?***
--***?***
--***?***
--***?***

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**

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

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

n many aspects of cell metabolism

peroxisomal disorders that

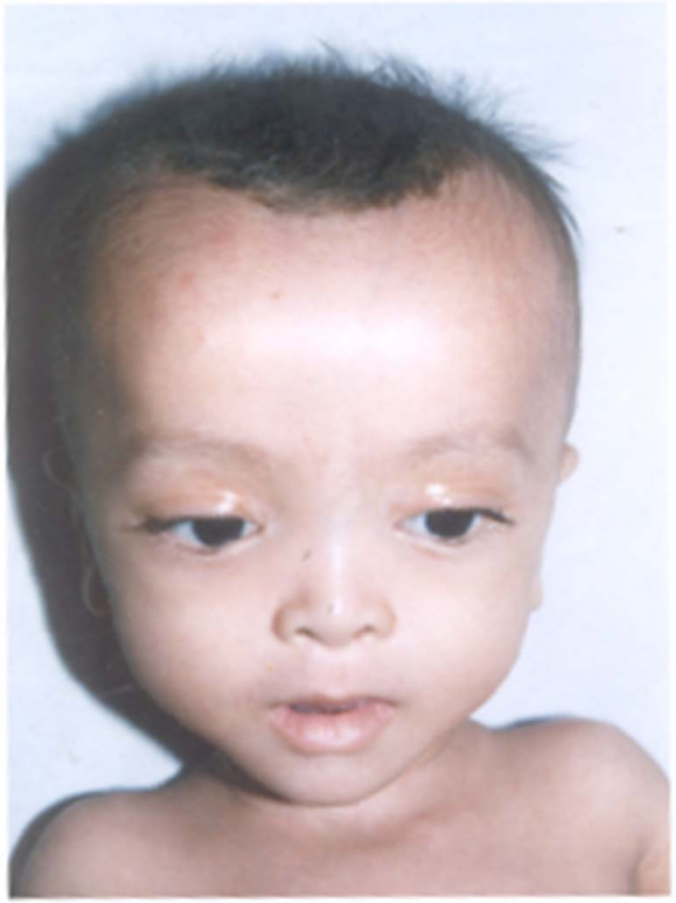
chain fatty acids (VLCFA)

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)

Leber's Congenital Amaurosis



Zellweger syndrome: Facies

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**

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

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

n many aspects of cell metabolism

peroxisomal disorders that

chain fatty acids (VLCFA)

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?

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**

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

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

n many aspects of cell metabolism

peroxisomal disorders that

chain fatty acids (VLCFA)

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

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

n many aspects of cell metabolism

peroxisomal disorders that

chain fatty acids (VLCFA)

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**

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**

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

What are peroxisomal disorders?
A heteroger

How is Zellweger syndrome diagnosed?

What is the noneponymou
Cerebrohepatorenal synd

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**

peroxisomal disorders that

chain fatty acids (VLCFA)

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**

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

What are peroxisomal disorders?
A heteroger

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

What is the noneponymou
Cerebrohepatorenal synd

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**

peroxisomal disorders that

chain fatty acids (VLCFA)

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**

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

What are peroxisomal disorders?
A heteroger

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

How is Zellweger managed?

What is the noneponymou
Cerebrohepatorenal synd

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**

peroxisomal disorders that

chain fatty acids (VLCFA)

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**

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

What are peroxisomal disorders?
A heteroger

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

How is Zellweger managed?
Supportively

What is the noneponymou
Cerebrohepatorenal synd

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**

peroxisomal disorders that

chain fatty acids (VLCFA)

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

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

Is NALD the same condition as adrenoleukodystrophy?

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

ll metabolism

s that

LCFA)

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

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

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

ll metabolism

s that

LCFA)

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
    - *What are* peroxisomal disorders?
      A heterogeneous group of disorders of peroxisome function

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

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

What is its inheritance pattern?

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the prognc
It is uniformly fatal by age **1 year**

ll metabolism

s that

LCFA)

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
    - *What are* peroxisomal disorders?
      A heterogeneous group of disorders of peroxisome function

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

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

What is its inheritance pattern?
AR

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the prognc
It is uniformly fatal by age **1 year**

ll metabolism

s that

LCFA)

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
    - *What are* peroxisomal disorders?
      A heterogeneous group of disorders of peroxisome function

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the prognc
It is uniformly fatal by age **1 year**

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

What is its inheritance pattern?
AR

Note: Both are inherited AR

ll metabolism

s that

LCFA)

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the prognc
It is uniformly fatal by age **1 year**

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

What is its inheritance pattern?
AR

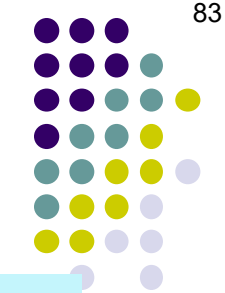
How do NALD pts present?

ll metabolism

s that

LCFA)

Leber's Congenital Amaurosis



- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

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

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

What is its inheritance pattern?
AR

How do NALD pts present?
In the [life period]

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the prognc
It is uniformly fatal by age **1 year**

ll metabolism

s that

LCFA)

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the prognc
It is uniformly fatal by age **1 year**

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

What is its inheritance pattern?
AR

How do NALD pts present?
In the **late infancy**

ll metabolism

s that

LCFA)

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
    - *What are* peroxisomal disorders?
      A heterogeneous group of disorders of peroxisome function

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal**
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the prognc
It is uniformly fatal by age **1 year**

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

What is its inheritance pattern?
AR

How do NALD pts present?
In the **late infancy**

ll metabolism

s that

LCFA)

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the prognc
It is uniformly fatal by age **1 year**

ll metabolism

s that

LCFA)

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:
--***?***
--***?***
--***?***
--***?***

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

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

What is its inheritance pattern?
AR

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

ll metabolism

s that

LCFA)

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--~~Abnormal facies~~

What is the prognc
It is uniformly fatal by age **1 year**

ll metabolism

s that

LCFA)

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

What is its inheritance pattern?
AR

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

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
    - *What are* peroxisomal disorders?
      A heterogeneous group of disorders of peroxisome function

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

ll metabolism

s that

LCFA)

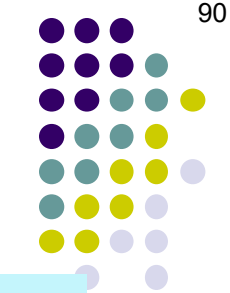
Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that presents later in childhood

What is its inheritance pattern?
AR

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

What is the prognosis?

Leber's Congenital Amaurosis



- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

ll metabolism

s that

LCFA)

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

What is its inheritance pattern?
AR

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

What is the prognosis?
It is uniformly fatal by life period

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**

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

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

ll metabolism

s that

LCFA)

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

What is its inheritance pattern?
AR

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

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
    - *What are* peroxisomal disorders?
      A heterogeneous group of disorders of peroxisome function

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hy
--Se
--Abnormal facies

What is the progno
It is uniformly **fatal by age 1 year**

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

What is its inheritance pattern?
AR

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

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

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

ll metabolism

s that

LCFA)

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
    - *What are* peroxisomal disorders?
      A heterogeneous group of disorders of peroxisome function

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do
In the
--LCA
--Deafn
--Hypot
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

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

What is its inheritance pattern?
AR

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

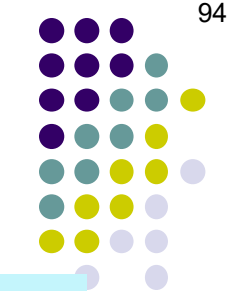
What is the prognosis?
It is uniformly fatal by **late childhood**

ll metabolism

s that

LCFA)

Leber's Congenital Amaurosis



- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heter

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

Why does neonatal ADRENOleukodystrophy have adreno- in the name?

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NAL
No, tha

What i
AR

How d
In the
--LCA
--Deaf
--Hypotonia
--Seizures

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heter

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

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

Is NAL
No, tha

What i
AR

How d
In the
--LCA
--Deaf
--Hypotonia
--Seizures

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

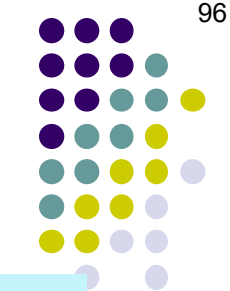
What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Leber's Congenital Amaurosis



- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heter

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

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?

Is NAL
No, tha

What i
AR

How d
In the
--LCA
--Deaf
--Hypotonia
--Seizures

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heter

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

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

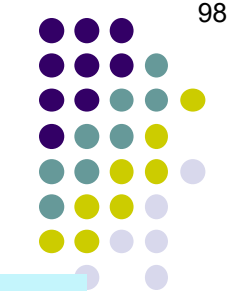
Is NAL
No, tha

What i
AR

How d
In the
--LCA
--Deaf
--Hypotonia
--Seizures

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

Leber's Congenital Amaurosis



- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heter

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

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?

Is NAL
No, tha

What i
AR

How d
In the
--LCA
--Deaf
--Hypotonia
--Seizures

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heter

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

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)

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NAL
No, tha

What i
AR

How d
In the
--LCA
--Deaf
--Hypotonia
--Seizures

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

What specific peroxisomal disorders can manifest an LCA-type presentation?
--Zellweger syndrome
--Neonatal adrenoleukodystrophy (NALD)
--**Infantile Refsum dz** = *adult Refsum dz?*

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

Is NALD the same condition as adrenoleukodystrophy?
No, that

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 nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

...ll metabolism

...*s that*

...LCFA)

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**

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

What specific peroxisomal disorders can manifest an LCA-type presentation?
--Zellweger syndrome
--Neonatal adrenoleukodystrophy (NALD)
--**Infantile Refsum dz** = *adult Refsum dz?* ***No!***

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

Is NALD the same condition as adrenoleukodystrophy?
No, that

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 nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

metabolism

s that

LCFA)

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

What specific peroxisomal disorders can manifest an LCA-type presentation?
--Zellweger syndrome
--Neonatal adrenoleukodystrophy (NALD)
--**Infantile Refsum dz** *aka...*

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the prognc
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that pr

What is its inheritance pattern?
AR

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

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

By what noneponymous name is infantile Refsum disease known?

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

What specific peroxisomal disorders can manifest an LCA-type presentation?
--Zellweger syndrome
--Neonatal adrenoleukodystrophy (NALD)
--**Infantile Refsum dz** *aka...infantile phytanic acid storage dz*

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that pr

What is its inheritance pattern?
AR

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

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

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

What specific peroxisomal disorders can manifest an LCA-type presentation?
--Zellweger syndrome
--Neonatal adrenoleukodystrophy (NALD)
--**Infantile Refsum dz** aka...infantile phytanic acid storage dz

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that pr

What is its inheritance pattern?
AR

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

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

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

What is its inheritance pattern?

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

What specific peroxisomal disorders can manifest an LCA-type presentation?
--Zellweger syndrome
--Neonatal adrenoleukodystrophy (NALD)
--**Infantile Refsum dz** aka...infantile phytanic acid storage dz

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that pr

What is its inheritance pattern?
AR

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

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

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

What is its inheritance pattern?
AR

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

What specific peroxisomal disorders can manifest an LCA-type presentation?
--Zellweger syndrome
--Neonatal adrenoleukodystrophy (NALD)
--**Infantile Refsum dz** aka...infantile phytanic acid storage dz

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the prognc
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that pr

What is its inheritance pattern?
AR

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

--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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

What specific peroxisomal disorders can manifest an LCA-type presentation?
--Zellweger syndrome
--Neonatal adrenoleukodystrophy (NALD)
--**Infantile Refsum dz** aka...infantile phytanic acid storage dz

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the prognc
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that pr

What is its inheritance pattern?
AR

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

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

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

What is its inheritance pattern?
AR

How do infantile Refsum dz pts present?

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

What specific peroxisomal disorders can manifest an LCA-type presentation?
--Zellweger syndrome
--Neonatal adrenoleukodystrophy (NALD)
--**Infantile Refsum dz** *aka...infantile phytanic acid storage dz*

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that pr

What is its inheritance pattern?
AR

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

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

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

What is its inheritance pattern?
AR

How do infantile Refsum dz pts present?
In the [redacted] period

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

What specific peroxisomal disorders can manifest an LCA-type presentation?
--Zellweger syndrome
--Neonatal adrenoleukodystrophy (NALD)
--**Infantile Refsum dz** aka...infantile phytanic acid storage dz

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that pr

What is its inheritance pattern?
AR

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

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

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

What is its inheritance pattern?
AR

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
    - *What are* peroxisomal disorders?
      A heterogeneous group of disorders of peroxisome function

What specific peroxisomal disorders can manifest an LCA-type presentation?
--Zellweger syndrome
--Neonatal adrenoleukodystrophy (NALD)
--**Infantile Refsum dz** aka...infantile phytanic acid storage dz

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafn
--Hypot
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that pr

What is its inheritance pattern?
AR

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

Just as NALD's onset is a little later than Zellweger's, infantile Refum's is a little later than that of NALD

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

What specific peroxisomal disorders can manifest an LCA-type presentation?
--Zellweger syndrome
--Neonatal adrenoleukodystrophy (NALD)
--**Infantile Refsum dz** aka...infantile phytanic acid storage dz

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that pr

What is its inheritance pattern?
AR

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

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

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

What is its inheritance pattern?
AR

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

What specific peroxisomal disorders can manifest an LCA-type presentation?
--Zellweger syndrome
--Neonatal adrenoleukodystrophy (NALD)
--**Infantile Refsum dz** *aka...infantile phytanic acid storage dz*

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that pr

What is its inheritance pattern?
AR

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

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

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

What is its inheritance pattern?
AR

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

What specific peroxisomal disorders can manifest an LCA-type presentation?
--Zellweger syndrome
--Neonatal adrenoleukodystrophy (NALD)
--**Infantile Refsum dz** aka...infantile phytanic acid storage dz

What is the nonep
Cerebrohepatoren

What is its inherita

In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--~~Seizures~~
--~~Abnormal facies~~

What is the prognc
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that pr

What is its inheritance pattern?

*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**)*

--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

nce pattern?

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

What specific peroxisomal disorders can manifest an LCA-type presentation?
--Zellweger syndrome
--Neonatal adrenoleukodystrophy (NALD)
--**Infantile Refsum dz** aka...infantile phytanic acid storage dz

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that pr

What is its inheritance pattern?
AR

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

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

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

What is its inheritance pattern?
AR

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

What is the prognosis?

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

What specific peroxisomal disorders can manifest an LCA-type presentation?
--Zellweger syndrome
--Neonatal adrenoleukodystrophy (NALD)
--**Infantile Refsum dz** *aka...infantile phytanic acid storage dz*

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that pr

What is its inheritance pattern?
AR

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

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

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

What is its inheritance pattern?
AR

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

What is the prognosis?
It is uniformly fatal by [redacted] (if treatment is unsuccessful)

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

What specific peroxisomal disorders can manifest an LCA-type presentation?
--Zellweger syndrome
--Neonatal adrenoleukodystrophy (NALD)
--**Infantile Refsum dz** aka...infantile phytanic acid storage dz

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that pr

What is its inheritance pattern?
AR

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

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

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

What is its inheritance pattern?
AR

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

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
    - *What are* peroxisomal disorders?
      A heterogeneous group of disorders of peroxisome function

What specific peroxisomal disorders can manifest an LCA-type presentation?
--Zellweger syndrome
--Neonatal adrenoleukodystrophy (NALD)
--**Infantile Refsum dz** aka...infantile phytanic acid storage dz

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that pr

What is its inheritance pattern?
AR

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

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

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

What is its inheritance pattern?
AR

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

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

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
  - *What are* peroxisomal disorders?
    A heterogeneous group of disorders of peroxisome function

What specific peroxisomal disorders can manifest an LCA-type presentation?
--Zellweger syndrome
--Neonatal adrenoleukodystrophy (NALD)
--**Infantile Refsum dz** aka...infantile phytanic acid storage dz

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linked condition that pr

What is its inheritance pattern?
AR

How do
In the **I**
--LCA
--Deafn
--Hypot
--Seizures

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

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

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

What is its inheritance pattern?

dz pts present?
eriod with:
--Deafness
--Hypotonia

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
    - *What are* peroxisomal disorders?
      A heterogeneous group of disorders of peroxisome function

What specific peroxisomal disorders can manifest an LCA-type presentation?
--Zellweger syndrome
--Neonatal adrenoleukodystrophy (NALD)
--**Infantile Refsum dz** aka...infantile phytanic acid storage dz

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linke

What is its inheritance pattern?
AR

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

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

How is infantile Refsum diagnosed?

ile Refsum
Infantile phytanic acid storage disease

What is its inheritance pattern?
AR

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

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
    - *What are* peroxisomal disorders?
      A heterogeneous group of disorders of peroxisome function

What specific peroxisomal disorders can manifest an LCA-type presentation?
--Zellweger syndrome
--Neonatal adrenoleukodystrophy (NALD)
--**Infantile Refsum dz** aka...infantile phytanic acid storage dz

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linke

What is its inheritance pattern?
AR

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

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

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

ile Refsum
Infantile phytanic acid storage disease

What is its inheritance pattern?
AR

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

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
    - *What are* peroxisomal disorders?
      A heterogeneous group of disorders of peroxisome function

What specific peroxisomal disorders can manifest an LCA-type presentation?
--Zellweger syndrome
--Neonatal adrenoleukodystrophy (NALD)
--**Infantile Refsum dz** aka...infantile phytanic acid storage dz

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linke

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 **I**

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

ile Refsum
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

Hold the phone--you can **treat** *this one??!! How is infantile Refsum treated?*

(<u>if treatment is unsuccessful</u>)

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
    - *What are* peroxisomal disorders?
      A heterogeneous group of disorders of peroxisome function

What specific peroxisomal disorders can manifest an LCA-type presentation?
--Zellweger syndrome
--Neonatal adrenoleukodystrophy (NALD)
--**Infantile Refsum dz** aka...infantile phytanic acid storage dz

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No, that is an X-linke

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 **I**

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

ile Refsum
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

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

(<u>if treatment is unsuccessful</u>)

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
    - *What are* peroxisomal disorders?
      A heterogeneous group of disorders of peroxisome function

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No,

Wh
AR

Hou
In t
--LC
--Deafness
--Hypotonia
--Seizures

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

LCA
Deafness } **Infantile Refsum**
Hypotonia
Seizures
Abnormal facies

Peroxismal disorders tl;dr

ame is infantile Refsum

age disease

tern?

lz pts present?
In the early childhood period with:
--Hypotonia

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
    - *What are* peroxisomal disorders?
      A heterogeneous group of disorders of peroxisome function

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No,

Wh
AR

Hou
In t
--LC
--Deafness
--Hypotonia
--Seizures

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

LCA
Deafness
Hypotonia
Seizures
Abnormal facies

Infantile Refsum
NALD

Peroxismal disorders tl;dr

ame is infantile Refsum

age disease

tern?

lz pts present?
In the early childhood period with:
--Hypotonia

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
    - *What are* peroxisomal disorders?
      A heterogeneous group of disorders of peroxisome function

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No,

Wh
AR

Hou
In t
--LC
--Deafness
--Hypotonia
--Seizures

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

LCA
Deafness
Hypotonia
Seizures
Abnormal facies

Infantile Refsum
NALD
Zellweger syndrome

Peroxismal disorders tl;dr

ame is infantile Refsum

age disease

lz pts present?
period with:
--Hypotonia

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
    - *What are* peroxisomal disorders?
      A heterogeneous group of disorders of peroxisome function

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No,

Wh
AR

Hou
In t
--LC
--Deafness
--Hypotonia
--Seizures

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

LCA
Deafness
Hypotonia
Seizures
Abnormal facies

Infantile Refsum
NALD
Zellweger syndrome

Presents earliest

ame is infantile Refsum

age disease

z pts present?
eriod with:
--Hypotonia

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

Peroxismal disorders tl;dr

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
    - *What are* peroxisomal disorders?
      A heterogeneous group of disorders of peroxisome function

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No,

Wh
AR

Hou
In t
--LC
--Deafness
--Hypotonia
--Seizures

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

LCA
Deafness
Hypotonia
Seizures
Abnormal facies

Infantile Refsum
NALD ← *Presents later*
Zellweger syndrome

Peroxismal disorders tl;dr

ame is infantile Refsum

age disease

tern?

lz pts present?
period with:
--Hypotonia

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
    - *What are* peroxisomal disorders?
      A heterogeneous group of disorders of peroxisome function

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No,

Wh
AR

Hou
In t
--LC
--Deafness
--Hypotonia
--Seizures

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

LCA
Deafness
Hypotonia
Seizures
Abnormal facies

Infantile Refsum ← *Presents latest*
NALD
Zellweger syndrome

ame is infantile Refsum

age disease

tern?

lz pts present?
period with:
--Hypotonia

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

Peroxismal disorders tl;dr

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
    - *What are* peroxisomal disorders?
      A heterogeneous group of disorders of peroxisome function

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No,

Wh
AR

Hou
In t
--LC
--Deafness
--Hypotonia
--Seizures

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

LCA
Deafness
Hypotonia
Seizures
Abnormal facies

Infantile Refsum ← *Best prognosis*
NALD
Zellweger syndrome

Peroxismal disorders tl;dr

ame is infantile Refsum

age disease

tern?

lz pts present?
eriod with:
--Hypotonia

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
    - *What are* peroxisomal disorders?
      A heterogeneous group of disorders of peroxisome function

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No,

Wh
AR

Hou
In t
--LC
--Deafness
--Hypotonia
--Seizures

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

LCA
Deafness
Hypotonia
Seizures
Abnormal facies

Infantile Refsum
NALD ← *Worse prognosis*
Zellweger syndrome

Peroxismal disorders tl;dr

ame is infantile Refsum

age disease

tern?

lz pts present?
In the early childhood period with:
--Hypotonia

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
    - *What are* peroxisomal disorders?
      A heterogeneous group of disorders of peroxisome function

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No,

Wh
AR

Hou
In t
--LC
--Deafness
--Hypotonia
--Seizures

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

LCA
Deafness
Hypotonia
Seizures
Abnormal facies

Infantile Refsum
NALD
Zellweger syndrome

Worst prognosis

Peroxismal disorders tl;dr

ame is infantile Refsum

age disease

z pts present?
eriod with:
--Hypotonia

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
    - *What are* peroxisomal disorders?
      A heterogeneous group of disorders of peroxisome function

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No,

Wh
AR

Hou
In t
--LC
--Deafness
--Hypotonia
--Seizures

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

LCA
Deafness
Hypotonia
Seizures
Abnormal facies

Infantile Refsum
NALD
Zellweger syndrome

All three are inherited

ame is infantile Refsum

age disease

tern?

z pts present?
period with:
--Hypotonia

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

Peroxismal disorders tl;dr

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
    - *What are* peroxisomal disorders?
      A heterogeneous group of disorders of peroxisome function

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No,

Wh
AR

Hou
In t
--LC
--Deafness
--Hypotonia
--Seizures

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

LCA
Deafness
Hypotonia
Seizures
Abnormal facies

Infantile Refsum
NALD
Zellweger syndrome

All three are inherited **AR**

ame is infantile Refsum

age disease

tern?

z pts present?
period with:
--Hypotonia

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

Peroxismal disorders tl;dr

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - **Peroxisomal disorders**
    - *What are* peroxisomal disorders?
      A heterogeneous group of disorders of peroxisome function

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

What is the nonep
Cerebrohepatoren

What is its inherita
AR

How do Zellweger
In the **neonatal** p
--LCA
--Deafness
--Hypotonia
--Seizures
--Abnormal facies

What is the progno
It is uniformly fatal by age **1 year**

Is NALD the same condition as adrenoleukodystrophy?
No,

Wh
AR

Hou
In t
--LC
--Deafness
--Hypotonia
--Seizures

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

LCA
Deafness
Hypotonia
Seizures
Abnormal facies

Infantile Refsum ← *Only infantile Refsum is treatable*

NALD
Zellweger syndrome

Peroxismal disorders tl;dr

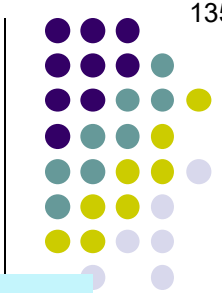
ame is infantile Refsum

tern?

z pts present?
period with:
--Hypotonia

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

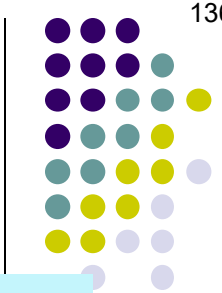
Leber's Congenital Amaurosis



- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal disorders
  - **Ciliopathies**

What is a ciliopathy?

Leber's Congenital Amaurosis



- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal disorders
  - **Ciliopathies**

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal disorders
  - **Ciliopathies**

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

Cilia are ubiquitous. That said, ciliopathies primarily affect three organs--what are they?

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal disorders
  - **Ciliopathies**

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

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

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal disorders
  - **Ciliopathies**

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

Cilia are ubiquit
The eyes, brain

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal disorders
  - **Ciliopathies**

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

Cilia are ubiquit
The eyes, brain

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal disorders
  - **Ciliopathies**

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

Cilia are ubiquit
The eyes, brain

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

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal disorders
  - **Ciliopathies**

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

Cilia are ubiquit
The eyes, brain

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

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal disorders
  - **Ciliopathies**

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

Cilia are ubiquit
The eyes, brain

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

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.

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal disorders
  - **Ciliopathies**

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

Cilia are ubiquit
The eyes, brain

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

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal diso
  - **Ciliopathies**

What specific ciliopathies can manifest an LCA-type presentation?
--?
--?
--?
--?

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal diso
  - **Ciliopathies**

What specific ciliopathies can manifest an LCA-type presentation?
--Bardet-Biedl syndrome
--Alström syndrome
--Joubert syndrome
--Senor-Løken syndrome

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal disorders
  - **Ciliopathies**

What specific ciliopathies can manifest an LCA-type presentation?

--**Bardet-Biedl syndrome**

--Alström syndrome

--Joubert syndrome

--Senior-Loken syndrome

What findings define the Bardet-Biedl complex?

--

--

--

--

--

The mnemonic is…

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal disorders
  - **Ciliopathies**

What specific ciliopathies can manifest an LCA-type presentation?

--**Bardet-Biedl syndrome**

--Alström syndrome

--Joubert syndrome

--S

What findings define the Bardet-Biedl complex?

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

(As in Simpson)

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal disorders
  - **Ciliopathies**

What specific ciliopathies can manifest an LCA-type presentation?

--**Bardet-Biedl syndrome**

--Alström syndrome

--Joubert syndrome

--S

What findings define the Bardet-Biedl complex?

--H

--O

--M

--E

--RP-like fundus

Not surprisingly, the 'R' *stands for* RP-like fundus.

As for the rest…

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal disorders
  - **Ciliopathies**

What specific ciliopathies can manifest an LCA-type presentation?
--**Bardet-Biedl syndrome**
--Alström syndrome
--Joubert syndrome
--Senior-Løken syndrome

What findings define the Bardet-Biedl complex?
--**H**ypogonadism
--**O**besity
--**M**ental retardation
--**E**xtra fingers (polydactyly)
--**R**P-like fundus

Not surprisingly, the 'R' *stands for* RP-like fundus.

As for the rest…

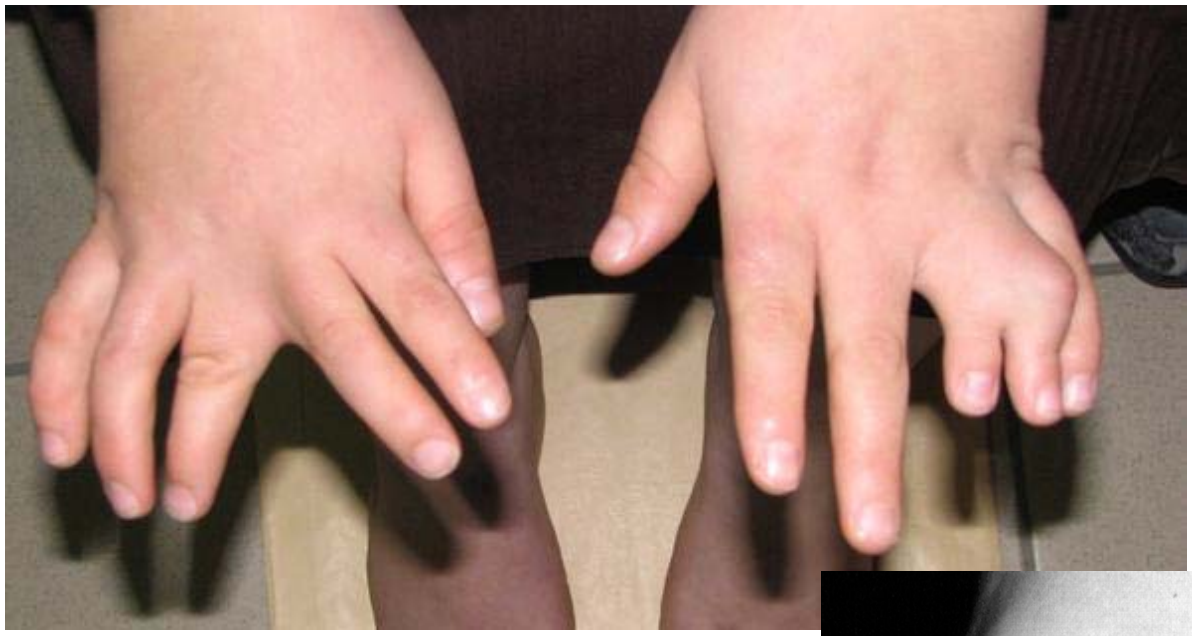
Leber's Congenital Amaurosis

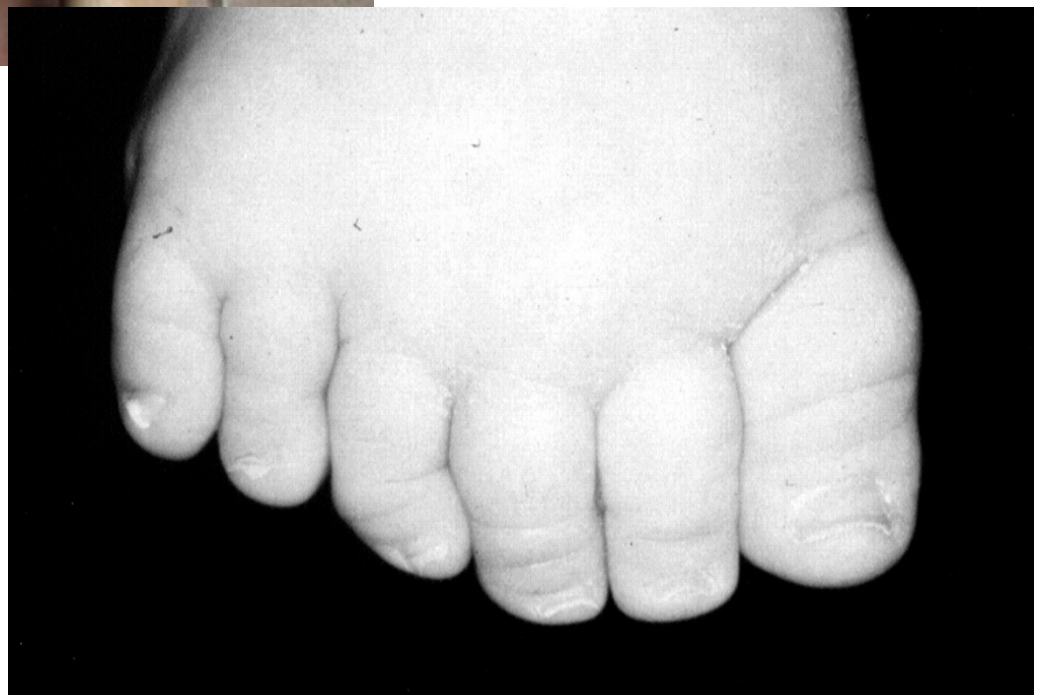
- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal disorders
  - **Ciliopathies**

What specific ciliopathies can manifest an LCA-type presentation?
--**Bardet-Biedl syndrome**
--Alström syndrome
--Joubert syndrome
--Senior-Loken syndrome

What findings define the Bardet-Biedl complex?
--**H**ypogonadism
--**O**besity
--**M**ental retardation
--**Extra fingers (polydactyly)**
--RP-like fundus

Take special note of the polydactyly finding in B-B!
My hunch is that an OKAP/Boards question would employ it as the key 'clue' to the dx.





Bardet-Biedl syndrome: Polydactyly

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal disorders
  - **Ciliopathies**

What specific ciliopathies can manifest an LCA-type presentation?
--**Bardet-Biedl syndrome**
--Alström syndrome
--Joubert syndrome
--Senior-Løken syndrome

What findings define the Bardet-Biedl complex?
--**H**ypogonadism
--**O**besity
--**M**ental retardation
--**Extra fingers (polydactyly)**
--**R**P-like fundus

Take special note of the polydactyly finding in B-B!
My hunch is that an OKAP/Boards question would employ it as the key 'clue' to the dx.

(And yeah, I know, Homer only has four digits per hand--paucidactyly, not polydactyly. But the rest of B-B fits him pretty well.)

Leber's Congenital Amaurosis

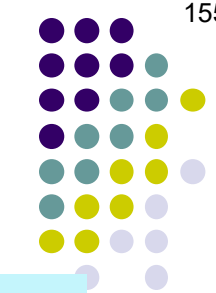
- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal diso
  - **Ciliopathies**

What specific ciliopathies can manifest an LCA-type presentation?
--Bardet Biedl syndrome
--**Alström syndrome**
--Jo
--S

What findings define the Bardet-Biedl complex?
*--**H**ypogonadism?*
*--**O**besity?*
*--**M**ental retardation?*
*--**E**xtra fingers (polydactyly)?*
--RP-like fundus

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

Leber's Congenital Amaurosis



- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal diso
  - **Ciliopathies**

What specific ciliopathies can manifest an LCA-type presentation?
--Bardet Biedl syndrome
--**Alström syndrome**
--Jo
--S

What findings define the Bardet-Biedl complex?
--**H**ypogonadism
--Obesity
--**M**ental retardation
--**E**xtra fingers (polydactyly)
--RP-like fundus

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal diso
  - **Ciliopathies**

What specific ciliopathies can manifest an LCA-type presentation?
--Bardet-Biedl syndrome
--**Alström syndrome**
--Jo
--S

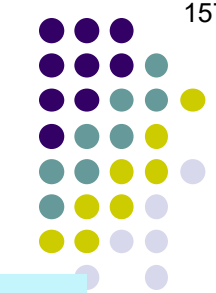
What findings define the Bardet-Biedl complex?
--**H**ypogonadism
--Obesity
--**M**ental retardation
--**E**xtra fingers (polydactyly)
--RP-like fundus
--?
--?

Both occur in childhood to teens

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

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

Leber's Congenital Amaurosis



- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal diso
  - **Ciliopathies**

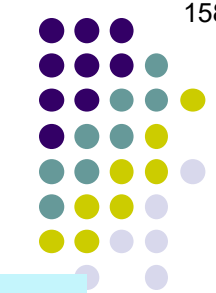
What specific ciliopathies can manifest an LCA-type presentation?
--Bardet-Biedl syndrome
--**Alström syndrome**
--Jo
--S

What findings define the Bardet-Biedl complex?
--**H**ypogonadism
--Obesity
--**M**ental retardation
--**E**xtra fingers (polydactyly)
--RP-like fundus
--Cardiomyopathy
--Early-onset Type 2 DM
} *Both occur in childhood to teens*

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

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

Leber's Congenital Amaurosis



- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal diso
  - **Ciliopathies**

What specific ciliopathies can manifest an LCA-type presentation?

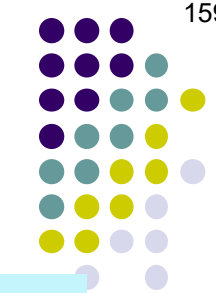
--**Bardet-Biedl syndrome**
--**Alström syndrome**

What findings define the Bardet-Biedl complex?

--**E**xtra fingers (polydactyly)
--**R**P-like fundus

How are Bardet-Biedl and Alström syndrome managed?

Leber's Congenital Amaurosis



- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal diso
  - **Ciliopathies**

What specific ciliopathies can manifest an LCA-type presentation?
--**Bardet-Biedl syndrome**
--**Alström syndrome**

What findings define the Bardet-Biedl complex?
--**E**xtra fingers (polydactyly)
--**R**P-like fundus

How are Bardet-Biedl and Alström syndrome managed?
Supportively

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absen
- DDx:
  - Peroxisomal diso
  - **Ciliopathies**

What specific ciliopathie
LCA-type presentation?
--Bardet-Biedl syndrome
--Alström syndrome
--**Joubert syndrome**
--Senior-Løken syndrom

What are the main nonocular structures affected in JS?

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absen
- DDx:
  - Peroxisomal diso
  - **Ciliopathies**

What specific ciliopathie
LCA-type presentation?
--Bardet-Biedl syndrome
--Alström syndrome
--**Joubert syndrome**
--Senior-Løken syndrom

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

Leber's Congenital Amaurosis

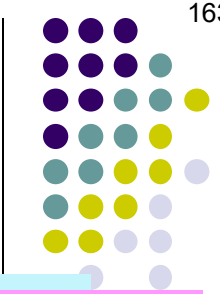
- LCA inheritance: AR
- Retinal histology reveals absen
- DDx:
  - Peroxisomal diso
  - **Ciliopathies**

What specific ciliopathie
LCA-type presentation?
--Bardet-Biedl syndrome
--Alström syndrome
--**Joubert syndrome**
--Senior-Løken syndrom

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

What classic MRI-brain finding is the hallmark of JS?

Leber's Congenital Amaurosis

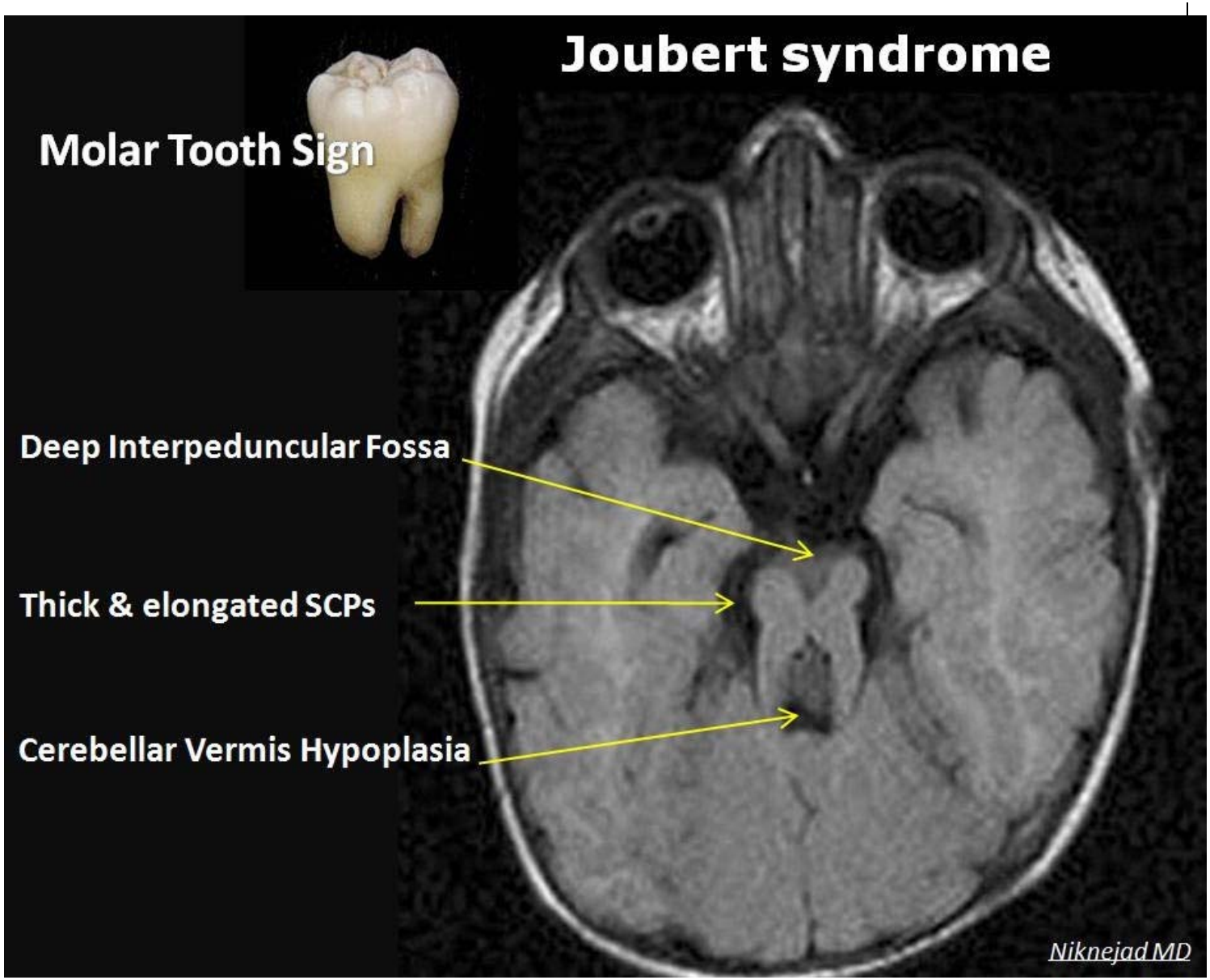


- LCA inheritance: AR
- Retinal histology reveals absen
- DDx:
  - Peroxisomal diso
  - **Ciliopathies**

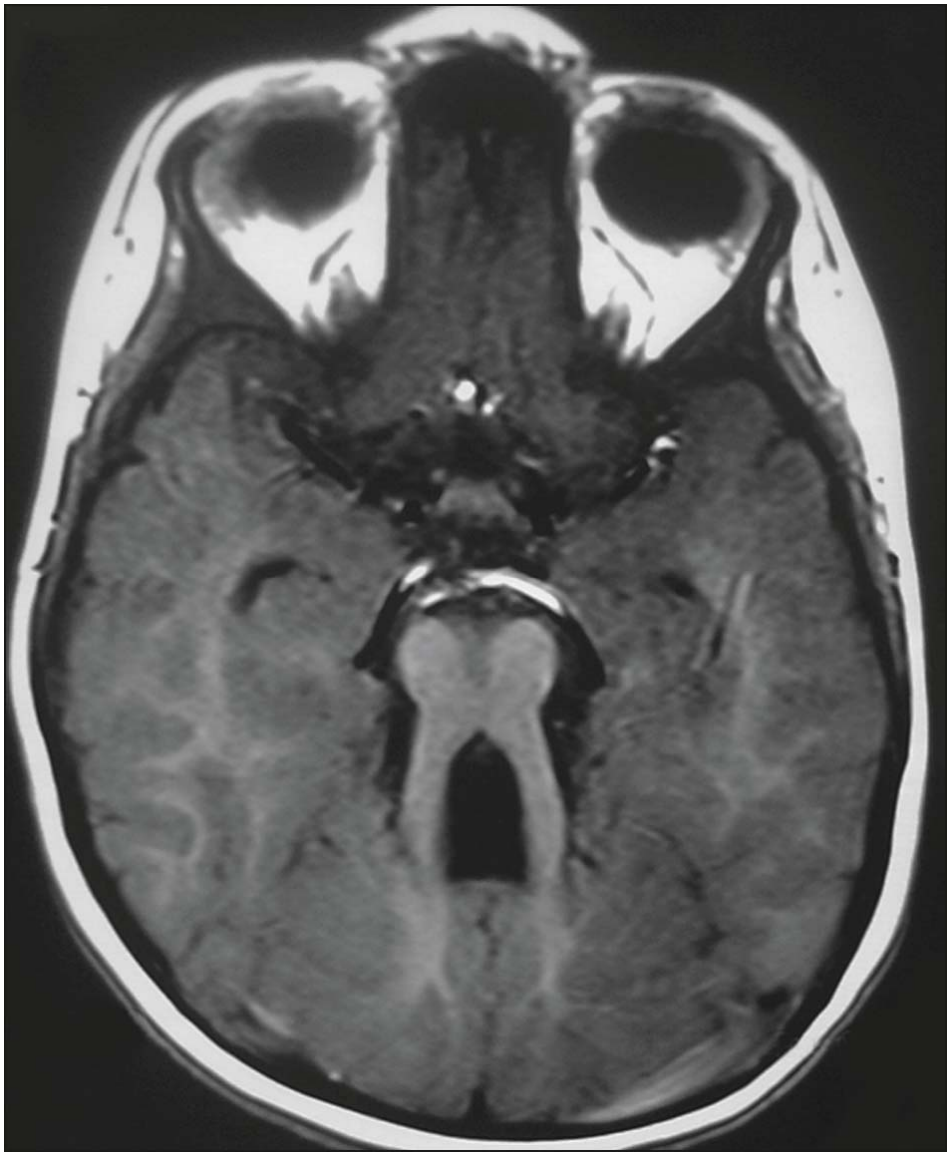
What specific ciliopathie
LCA-type presentation?
--Bardet-Biedl syndrome
--Alström syndrome
--**Joubert syndrome**
--Senior-Løken syndrom

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

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

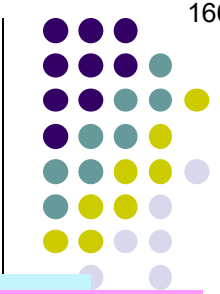


Joubert syndrome: Molar-tooth sign



Joubert syndrome: Molar-tooth sign

Leber's Congenital Amaurosis



- LCA inheritance: AR
- Retinal histology reveals absen
- DDx:
  - Peroxisomal diso
  - **Ciliopathies**

What specific ciliopathie
LCA-type presentation?
--Bardet-Biedl syndrome
--Alström syndrome
--Joubert syndrome
--Senior-Løken syndrom

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

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

How do JS pts present?

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absen
- DDx:
  - Peroxisomal diso
  - **Ciliopathies**

What specific ciliopathie
LCA-type presentation?
--Bardet-Biedl syndrome
--Alström syndrome
--**Joubert syndrome**
--Senior-Løken syndrom

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

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

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

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absen
- DDx:
  - Peroxisomal diso
  - **Ciliopathies**

What specific ciliopathie
LCA-type presentation?
--Bardet-Biedl syndrome
--Alström syndrome
--**Joubert syndrome**
--Senior-Løken syndrom

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

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

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

Leber's Congenital Amaurosis

Joubert syndrome: Facies. Note the large head, broad forehead

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absen
- DDx:
  - Peroxisomal diso
  - **Ciliopathies**

What specific ciliopathie
LCA-type presentation?
--Bardet-Biedl syndrome
--Alström syndrome
--**Joubert syndrome**
--Senior-Løken syndrom

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

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

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

What is the prognosis?

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absen
- DDx:
  - Peroxisomal diso
  - **Ciliopathies**

What specific ciliopathie
LCA-type presentation?
--Bardet-Biedl syndrome
--Alström syndrome
--**Joubert syndrome**
--Senior-Løken syndrom

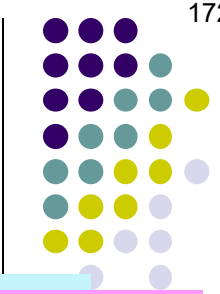
What are the main nonocular structures affected in JS?
The brainstem and cerebellum

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

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

What is the prognosis?
It is highly variable

Leber's Congenital Amaurosis



- LCA inheritance: AR
- Retinal histology reveals absen
- DDx:
  - Peroxisomal diso
  - **Ciliopathies**

What specific ciliopathie
LCA-type presentation?
--Bardet-Biedl syndrome
--Alström syndrome
--**Joubert syndrome**
--Senor-Løken syndrom

How is Joubert syndrome managed?

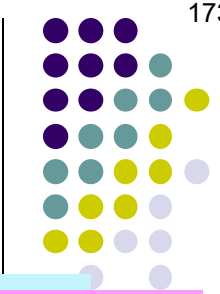
What are the main nonocular structures affected in JS?
The brainstem and cerebellum

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

How do JS pts present?
In the **late infancy** period with:
--Hypotonia
--Disordered breathing
--Intellectual and motor deficits
--Seizures
--Abnormal facies

What is the prognosis?
It is highly variable

Leber's Congenital Amaurosis



- LCA inheritance: AR
- Retinal histology reveals absen
- DDx:
  - Peroxisomal diso
  - **Ciliopathies**

What specific ciliopathie
LCA-type presentation?
--Bardet-Biedl syndrome
--Alström syndrome
--**Joubert syndrome**
--Senor-Løken syndrom

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

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

How do JS pts present?
In the **late infancy** period with:
--Hypotonia
--Disordered breathing
--Intellectual and motor deficits
--Seizures
--Abnormal facies

What is the prognosis?
It is highly variable

How is Joubert syndrome managed? **Supportively**

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal diso
  - **Ciliopathies**

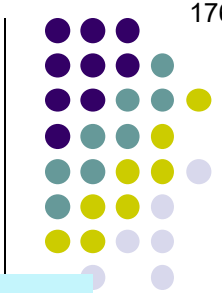
What specific ciliopathies can manifest an LCA-type presentation?
--Bardet-Biedl syndrome
--Alström syndrome
--Joubert syndrome
--**Senor-Løken syndrome**

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.**

Leber's Congenital Amaurosis

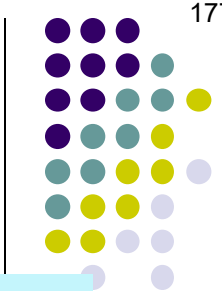
- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal disorders
  - Ciliopathies
- LCA should be thought of as a three words

Leber's Congenital Amaurosis



- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal disorders
  - Ciliopathies
- LCA should be thought of as a diagnosis of exclusion

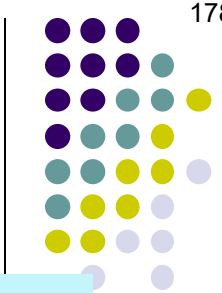
Leber's Congenital Amaurosis



- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal disorders
  - Ciliopathies
- LCA should be thought of as a **diagnosis of exclusion**

Why treat LCA as a diagnosis of exclusion?

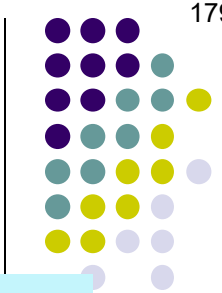
Leber's Congenital Amaurosis



- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal disorders
  - Ciliopathies
- LCA should be thought of as a **diagnosis of exclusion**

Why treat LCA as a diagnosis of exclusion?
Because while LCA is largely untreatable, a number of the entities on its DDx are, **if** they're caught in time. Thus, one **must** be certain a child does not have one of the treatable conditions before settling on the diagnosis that s/he has (untreatable) LCA.

Leber's Congenital Amaurosis



- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal disorders
  - Ciliopathies
- LCA should be thought of as a **diagnosis of exclusion**

Why treat LCA as a diagnosis of exclusion?
Because while LCA is largely untreatable, a number of the entities on its DDx are, **if** they're caught in time. Thus, one **must** be certain a child does not have one of the treatable conditions before settling on the diagnosis that s/he has (untreatable) LCA.

You said largely *untreatable. So, what treatment* **is** *available for LCA?*

Leber's Congenital Amaurosis

- LCA inheritance: AR
- Retinal histology reveals absence of photoreceptors
- DDx:
  - Peroxisomal disorders
  - Ciliopathies
- LCA should be thought of as a **diagnosis of exclusion**

Why treat LCA as a diagnosis of exclusion?
Because while LCA is largely untreatable, a number of the entities on its DDx are, **if** they're caught in time. Thus, one **must** be certain a child does not have one of the treatable conditions before settling on the diagnosis that s/he has (untreatable) LCA.

You said largely *untreatable. So, what treatment* **is** *available for LCA?*
There is no widely-available treatment. Recent research employing gene replacement therapy for **one** of the many genetic mutations causing LCA (there are at least 15; doubtless others have yet to be identified) has met with modest success.