Wilson's disease
--Corneal finding: ?
Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease
--Corneal finding: Kayser-Fleischer ring

Systemic diseases of childhood with corneal findings
Wilson's disease
--Corneal finding: Kayser-Fleischer ring

What color is the Kayser-Fleischer ring?

The classic color is green, but it can also be golden-brown or even ruby red.

In which layer of the cornea is it located?
Descemet's

What is its course; ie, where does it start, and how does it proceed?
It starts superiorly, and proceeds inferiorly from there.

Do all Wilson's dz pts manifest the ring?
No, so its absence does not rule out the condition.
Wilson's disease
--Corneal finding: Kayser-Fleischer ring

What color is the Kayser-Fleischer ring?
The classic color is **green**, but it can also be **golden-brown** or even **ruby red**.
Selected Systemic Diseases of Childhood with Corneal Findings

Kayser-Fleischer ring
Wilson's disease
--Corneal finding: Kayser-Fleischer ring

What color is the Kayser-Fleischer ring?
The classic color is green, but it can also be golden-brown or even ruby red.

In which layer of the cornea is it located?
Wilson's disease
--Corneal finding: Kayser-Fleischer ring

What color is the Kayser-Fleischer ring?
The classic color is green, but it can also be golden-brown or even ruby red

In which layer of the cornea is it located?
Descemet's
Wilson's disease
--Corneal finding: Kayser-Fleischer ring

**What color is the Kayser-Fleischer ring?**
The classic color is **green**, but it can also be **golden-brown** or even **ruby red**

*In which layer of the cornea is it located?*
Descemet’s

*What is its course; ie, where does it start, and how does it proceed?*
Wilson's disease
--Corneal finding: Kayser-Fleischer ring

What color is the Kayser-Fleischer ring?
The classic color is green, but it can also be golden-brown or even ruby red.

In which layer of the cornea is it located?
Descemet's

What is its course; ie, where does it start, and how does it proceed?
It starts superiorly, and proceeds inferiorly from there.
Wilson's disease
--Corneal finding: Kayser-Fleischer ring

What color is the Kayser-Fleischer ring?
The classic color is green, but it can also be golden-brown or even ruby red

In which layer of the cornea is it located?
Descemet’s

What is its course; ie, where does it start, and how does it proceed?
It starts superiorly, and proceeds inferiorly from there

Do all Wilson’s dz pts manifest the ring?
Wilson's disease
--Corneal finding: Kayser-Fleischer ring

What color is the Kayser-Fleischer ring?
The classic color is green, but it can also be golden-brown or even ruby red

In which layer of the cornea is it located?
Descemet's

What is its course; ie, where does it start, and how does it proceed?
It starts superiorly, and proceeds inferiorly from there

Do all Wilson's dz pts manifest the ring?
No, so its absence does not rule out the condition
Wilson's disease
--Corneal finding: Kayser-Fleischer ring

- Disease of copper metabolism
- Systemic findings:
  - Liver and renal damage stigmata
  - CNS signs/symptoms

What color is the Kayser-Fleischer ring?
The classic color is green, but it can also be golden-brown or even ruby red.

In which layer of the cornea is it located?
Descemet's

What is its course; ie, where does it start, and how does it proceed?
It starts superiorly, and proceeds inferiorly from there.

Do all Wilson's disease patients manifest the ring?
No, so its absence does not rule out the condition.

I've heard of something called a Fleischer ring. Is that just shorthand for Kayser-Fleischer ring?
**Wilson’s disease**

--Corneal finding: Kayser-Fleischer ring

The classic color is **green**, but it can also be **golden-brown** or even **ruby red**.

In which layer of the cornea is it located?

**Descemet’s**

What is its course; i.e., where does it start, and how does it proceed?

It starts superiorly, and proceeds inferiorly from there.

Do all Wilson’s dz pts manifest the ring?

No, so its absence does not rule out the condition.

I’ve heard of something called a Fleisher ring. Is that just shorthand for Kayser-Fleischer ring?

No, a Fleisher ring is something completely different.
Wilson's disease
--Corneal finding: Kayser-Fleischer ring

- What color is the Kayser-Fleischer ring?
  - The classic color is green, but it can also be golden-brown or even ruby red.

- In which layer of the cornea is it located?
  - Descemet's

- What is its course; ie, where does it start, and how does it proceed?
  - It starts superiorly, and proceeds inferiorly from there.

- Do all Wilson's disease pts manifest the ring?
  - No, so its absence does not rule out the condition.

*I've heard of something called a Fleisher ring. Is that just shorthand for Kayser-Fleischer ring?*

No, a Fleisher ring is something completely different.

**OK then, what is a Fleisher ring?**

*What is its course; ie, where does it start, and how does it proceed?*

*Do all Wilson's disease pts manifest the ring?*

No, so its absence does not rule out the condition.
Wilson’s disease
---Corneal finding: Kayser-Fleischer ring

**What color is the Kayser-Fleischer ring?**

*The classic color is green, but it can also be golden-brown or even ruby red.*

**In which layer of the cornea is it located?**

*Descemet’s*

**What is its course; i.e., where does it start, and how does it proceed?**

*It starts superiorly, and proceeds inferiorly from there.*

**Do all Wilson’s dz pts manifest the ring?**

*No, so its absence does not rule out the condition.*

---

**Fleischer ring**

*I’ve heard of something called a Fleisher ring. Is that just shorthand for Kayser-Fleischer ring?*

*No, a Fleischer ring is something completely different.*

**OK then, what is a Fleisher ring?**

*A line on the cornea secondary to keratoconus element condition.*

**What is its course; i.e., where does it start, and how does it proceed?**

*It starts superiorly, and proceeds inferiorly from there.*

**Do all Wilson’s dz pts manifest the ring?**

*No, so its absence does not rule out the condition.*
Wilson's disease
--Corneal finding: Kayser-Fleischer ring

What color is the Kayser-Fleischer ring?
The classic color is green, but it can also be golden-brown or even ruby red.

In which layer of the cornea is it located?
Descemet's

What is its course; ie, where does it start, and how does it proceed?
It starts superiorly, and proceeds inferiorly from there.

Do all Wilson's disease pts manifest the ring?
No, so its absence does not rule out the condition.

I've heard of something called a Fleischer ring. Is that just shorthand for Kayser-Fleischer ring?
No, a Fleischer ring is something completely different.

OK then, what is a Fleischer ring?
An iron line on the cornea secondary to keratoconus.

Selected Systemic Diseases of Childhood with Corneal Findings
Fleischer ring in keratoconus
Wilson’s disease
--Corneal finding: Kayser-Fleischer ring
--Disease of element metabolism

Systemic diseases of childhood with corneal findings
**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --
  --
Selected Systemic Diseases of Childhood with Corneal Findings

**Wilson’s disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Systemic diseases of childhood with corneal findings
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

In Wilson’s, copper deposition occurs in a particular order:
--First in the
--Next in the
--Later, in the
**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

*In Wilson’s, copper deposition occurs in a particular order:*
--First in the liver
--Next in the kidneys
--Later, in the CNS

**Selected Systemic Diseases of Childhood with Corneal Findings**

Systemic diseases of childhood with corneal findings
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

In Wilson's, copper deposition occurs in a particular order:
--First in the liver
--Next in the kidneys
--Later, in the CNS

At what point does the eye become involved?
Wilson’s disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

In Wilson’s, copper deposition occurs in a particular order:
--First in the liver
--Next in the kidneys
--Later, in the CNS

At what point does the eye become involved?
It really gets going in the CNS phase
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

In Wilson's, copper deposition occurs in a particular order:
--First in the liver
--Next in the kidneys
--Later, in the CNS

At what point does the eye become involved?

Does eye involvement extend beyond the K-F ring?

--
--
--

At what point does the eye become involved?

Selected Systemic Diseases of Childhood with Corneal Findings
**Wilson’s disease**
-- Corneal finding: Kayser-Fleischer ring
-- Disease of copper metabolism
-- Systemic findings:
  -- Liver and renal damage stigmata
  -- CNS signs/symptoms

At what point does the eye become involved?
It really gets going in the CNS phase.

Does eye involvement extend beyond the K-F ring?
-- Yes, in two ways:
  --
  --

Selected Systemic Diseases of Childhood with Corneal Findings
**Wilson’s disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

**Does eye involvement extend beyond the K-F ring?**
--Yes, in two ways:
  --A copper-induced cataract is common
  --Retinal function (as measured by ERG) can be affected

**At what point does the eye become involved?**
It really gets going in the CNS phase.
Wilson’s disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

In Wilson’s, copper deposition occurs in a particular order:
--First in the liver
--Next in the kidneys
--Later, in the CNS

At what point does the eye become involved?
--It really gets going in the CNS phase

Does eye involvement extend beyond the K-F ring?
--Yes, in two ways:
  --A copper-induced cataract is common
  --Retinal function (as measured by ERG) can be affected

By what appearance-related moniker are copper-induced cataracts known?
--‘Sunflower cataracts’
**Wilson’s disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

In Wilson’s, copper deposition occurs in a particular order:
--First in the liver
--Next in the kidneys
--Later, in the CNS

At what point does the eye become involved?

It really gets going in the CNS phase.

Does eye involvement extend beyond the K-F ring?
Yes, in two ways:
  --A **copper-induced cataract** is common
  --Retinal function (as measured by ERG) can be affected

By what appearance-related moniker are copper-induced cataracts known? ‘Sunflower cataracts’
‘Sunflower cataract’ in Wilson’s dz

A sunflower for comparison
**Wilson’s disease**

--Corneal finding: Kayser-Fleischer ring

--Disease of copper metabolism

--Systemic findings:

--Liver and renal damage stigmata

--CNS signs/symptoms

--- Does eye involvement extend beyond the K-F ring?

--- Yes, in two ways:

--- A **copper-induced cataract** is common

--- Retinal function (as measured by ERG) can be affected

--- By what appearance-related moniker are copper-induced cataracts known?

--- ‘Sunflower cataracts’

--- BTW: What is the **non**eponymous name of Wilson’s dz?
**Wilson's disease**
- Corneal finding: Kayser-Fleischer ring
- Disease of copper metabolism
- Systemic findings:
  - Liver and renal damage stigmata
  - CNS signs/symptoms

Does eye involvement extend beyond the K-F ring?
- Yes, in two ways:
  - A copper-induced cataract is common
  - Retinal function (as measured by ERG) can be affected

By what appearance-related moniker are copper-induced cataracts known?
'Sunflower cataracts'

It really gets going in the CNS phase

BTW: What is the non-eponymous name of Wilson’s dz?
'Hepato-'
**Wilson’s disease**
-- Corneal finding: Kayser-Fleischer ring
-- Disease of copper metabolism
-- Systemic findings:
  -- Liver and renal damage stigmata
  -- CNS signs/symptoms

In Wilson’s, copper deposition occurs in a particular order:
-- First in the liver
-- Next in the kidneys
-- Later, in the CNS

At what point does the eye become involved?
It really gets going in the CNS phase

Does eye involvement extend beyond the K-F ring?
Yes, in two ways:
-- A copper-induced cataract is common
-- Retinal function (as measured by ERG) can be affected

By what appearance-related moniker are copper-induced cataracts known?
‘Sunflower cataracts’

BTW: What is the *non* eponymous name of Wilson’s dz?
‘Hepato-lenticular’
**Wilson’s disease**
- Corneal finding: Kayser-Fleischer ring
- Disease of copper metabolism
- Systemic findings:
  - Liver and renal damage stigmata
  - CNS signs/symptoms

---

In Wilson’s disease, copper deposition occurs in a particular order:
- First in the liver
- Next in the kidneys
- Later, in the CNS

At what point does the eye become involved?
- It really gets going in the CNS phase

Does eye involvement extend beyond the K-F ring?
- Yes, in two ways:
  - A **copper-induced cataract** is common
  - Retinal function (as measured by ERG) can be affected

BTW: What is the **non-eponymous name of Wilson’s dz**?
- ‘Hepato-lenticular degeneration’

By what appearance-related moniker are copper-induced cataracts known?
- ‘Sunflower cataracts’
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

What are the CNS manifestations in Wilson’s?
Motor function is Parkinson’s-like: Rigidity, tremor, unintelligible speech, and involuntary movements. Frank dementia may occur.
**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

*What are the CNS manifestations in Wilson's?*
Motor function is disease-like: Rigidity, tremor, unintelligible speech, and involuntary movements. Frank dementia may occur.
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Selected Systemic Diseases of Childhood with Corneal Findings

What are the CNS manifestations in Wilson’s?
Motor function is Parkinson's-like: Rigidity, tremor, unintelligible speech, and involuntary movements. Frank dementia may occur.
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:

Is a Kayser-Fleischer ring pathognomonic for Wilson’s disease?
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:

Is a Kayser-Fleischer ring pathognomonic for Wilson's disease?
No, it may be present in other liver disorders, or exogenous chalcosis
**Selected Systemic Diseases of Childhood with Corneal Findings**

**Wilson's disease**
- Corneal finding: Kayser-Fleischer ring
- Disease of copper metabolism
- Systemic findings:
  - Liver and renal damage stigmata
  - CNS signs/symptoms

*Is a Kayser-Fleischer ring pathognomonic for Wilson's disease?*
No, it may be present in other liver disorders, or exogenous chalcosis.

*Which other liver conditions are particularly likely to produce a K-F ring?*
Primary biliary cirrhosis and chronic active hepatitis. (BTW, if you're tempted to dismiss this as low-yield minutiae, take note: This factoid was considered important enough to be a chapter-leading “Highlight” in the BCSC Cornea book.)
**Wilson’s disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism

Systemic findings:

Is a Kayser-Fleischer ring pathognomonic for Wilson’s disease?
No, it may be present in other liver disorders, or exogenous chalcosis.

Does the absence of a Kayser-Fleischer ring rule out Wilson’s disease?
No, because (as mentioned earlier) it generally doesn’t appear until late in the disease course.

Which other liver conditions are particularly likely to produce a K-F ring?
Primary biliary cirrhosis and chronic active hepatitis. (BTW, if you’re tempted to dismiss this as low-yield minutiae, take note: This factoid was considered important enough to be a chapter-leading “Highlight” in the BCSC Cornea book.)
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Is a Kayser-Fleischer ring pathognomonic for Wilson's disease?
No, it may be present in other liver disorders or exogenous chalcosis.

Does the absence of a Kayser-Fleischer ring rule out Wilson's disease?
No, because (as mentioned earlier) it generally doesn't appear until late in the disease course.

Which other liver conditions are particularly likely to produce a K-F ring?
Primary biliary cirrhosis and chronic active hepatitis. (BTW, if you're tempted to dismiss this as low-yield minutiae, take note: This factoid was considered important enough to be a chapter-leading “Highlight” in the BCSC Cornea book.)

Kayser-Fleischer ring DDx:
- Wilson’s disease
- Primary biliary cirrhosis
- Chronic active hepatitis

No question—proceed when ready.
**Wilson's disease**
- Corneal finding: Kayser-Fleischer ring
- Disease of copper metabolism
- Systemic findings:
  - Liver and renal damage stigmata
  - CNS signs/symptoms

Is a Kayser-Fleischer ring pathognomonic for Wilson’s disease?
No, it may be present in other liver disorders, or exogenous chalcosis

Does the absence of a Kayser-Fleischer ring rule out Wilson's disease?

**Selected Systemic Diseases of Childhood with Corneal Findings**

of childhood with corneal findings
Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease
-- Corneal finding: Kayser-Fleisher ring
-- Disease of copper metabolism
-- Systemic findings:

Is a Kayser-Fleisher ring pathognomonic for Wilson's disease?
No, it may be present in other liver disorders, or exogenous chalcosis

Does the absence of a Kayser-Fleisher ring rule out Wilson's disease?
No, because (as mentioned earlier) it generally doesn't appear until late in the disease course

of childhood with corneal findings
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  - Liver and renal damage stigmata
  - CNS signs/symptoms

Is a Kayser-Fleischer ring pathognomonic for Wilson's disease?
No, it may be present in other liver disorders, or exogenous chalcosis

Does the absence of a Kayser-Fleischer ring rule out Wilson's disease?
No, because (as mentioned earlier) it generally doesn’t appear until late in the disease course

Clearly, the K-F ring is of limited utility in the diagnosis of Wilson’s. So in addition to checking for K-F rings, what other steps should be taken to make the diagnosis?

Three lab tests make for a good start:
--Serum ceruloplasmin levels
--Serum free copper levels
--Urinary copper levels
Wilson's disease

--Corneal finding: Kayser-Fleischer ring

--Disease of copper metabolism

Systemic findings:

Liver and renal damage stigmata

CNS signs/symptoms

Is a Kayser-Fleischer ring pathognomonic for Wilson's disease?

No, it may be present in other liver disorders, or exogenous chalcosis.

Does the absence of a Kayser-Fleischer ring rule out Wilson's disease?

No, because (as mentioned earlier) it generally doesn't appear until late in the disease course.

Clearly, the K-F ring is of limited utility in the diagnosis of Wilson's. So in addition to checking for K-F rings, what other steps should be taken to make the diagnosis?

Three lab tests make for a good start:

--Serum ceruloplasmin levels
--Serum free copper levels
--Urinary copper levels
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism

Systemic findings:
--Liver and renal damage stigmata
--CNS signs/symptoms

Is a Kayser-Fleischer ring pathognomonic for Wilson's disease?
No, it may be present in other liver disorders, or exogenous chalcosis.

Does the absence of a Kayser-Fleischer ring rule out Wilson's disease?
No, because (as mentioned earlier) it generally doesn't appear until late in the disease course.

Clearly, the K-F ring is of limited utility in the diagnosis of Wilson's. So in addition to checking for K-F rings, what other steps should be taken to make the diagnosis?

Three lab tests make for a good start:
--Serum ceruloplasmin levels
--Serum free copper levels
--Urinary copper levels
Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease

- Corneal finding: Kayser-Fleischer ring
- Disease of copper metabolism

Systemic findings:
- Liver and renal damage stigmata
- CNS signs/symptoms

Is a Kayser-Fleischer ring pathognomonic for Wilson's disease?
No, it may be present in other liver disorders, or exogenous chalcosis.

Does the absence of a Kayser-Fleischer ring rule out Wilson's disease?
No, because (as mentioned earlier) it generally doesn't appear until late in the disease course.

Clearly, the K-F ring is of limited utility in the diagnosis of Wilson's. So in addition to checking for K-F rings, what other steps should be taken to make the diagnosis?
Three lab tests make for a good start: And their expected values are:

- Serum ceruloplasmin levels:
  - For each, indicate 'High' or 'Low'
- Serum free copper levels:
- Urinary copper levels:
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
Systemic findings:
--Liver and renal damage stigmata
--CNS signs/symptoms

Is a Kayser-Fleischer ring pathognomonic for Wilson's disease?
No, it may be present in other liver disorders, or exogenous chalcosis.

Does the absence of a Kayser-Fleischer ring rule out Wilson's disease?
No, because (as mentioned earlier) it generally doesn't appear until late in the disease course.

Clearly, the K-F ring is of limited utility in the diagnosis of Wilson's. So in addition to checking for K-F rings, what other steps should be taken to make the diagnosis?
Three lab tests make for a good start: And their expected values are:
--Serum ceruloplasmin levels: LOW
--Serum free copper levels: HIGH
--Urinary copper levels: HIGH

For each, indicate ‘High’ or ‘Low’
**Wilson’s disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

What is the treatment for Wilson’s dz?
Penicillamine

Does the K-F ring regress with treatment?
Yes
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

What is the treatment for Wilson’s disease? Penicillamine

Penicillamine
**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

*What is the treatment for Wilson's dz?*
Penicillamine

*Does the K-F ring regress with treatment?*
**Wilson’s disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

*What is the treatment for Wilson’s dz?*
Penicillamine

*Does the K-F ring regress with treatment?*
Yes
**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

**Riley-Day syndrome**
--Corneal finding: ?
Wilson's disease
--Corneal finding: **Kayser-Fleischer ring**
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

**Riley-Day syndrome**
--Corneal finding: **keratitis/ulcers**
Wilson’s disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka…
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka…familial dysautonomia
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka…familial dysautonomia

What are the two problems underlying the propensity for keratitis and corneal ulcers that characterize Riley-Day?
**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

**Riley-Day syndrome**
--Corneal finding: keratitis/ulcers
--aka…*familial dysautonomia*

What are the two problems underlying the propensity for keratitis and corneal ulcers that characterize Riley-Day? Decreased corneal sensitivity (Riley-Day is one cause of *congenital corneal anesthesia*) and abnormal lacrimation.
**Wilson’s disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

**Riley-Day syndrome**
--Corneal finding: keratitis/ulcers
--aka…*familial dysautonomia*

*Keratitis* $\angle$ to decreased corneal sensitivity is called…?

**Decreased corneal sensitivity**

Underlying the propensity for keratitis and corneal ulceration?

(Riley-Day is one cause of congenital corneal anesthesia) and abnormal lacrimation
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Systemic diseases of childhood with corneal findings

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka… familial dysautonomia

Keratitis 2° to decreased corneal sensitivity is called…? Neurotrophic keratitis

Decreased corneal sensitivity (Riley-Day is one cause of congenital corneal anesthesia) and abnormal lacrimation

What are the two problems underlying the propensity for keratitis and corneal ulcers that characterize Riley-Day?
Decreased corneal sensitivity and abnormal lacrimation
**Wilson’s disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

**Riley-Day syndrome**
--Corneal finding: keratitis/ulcers
--aka…*familial dysautonomia*
--Systemic findings:
  --
  --
  --
Selected Systemic Diseases of Childhood with Corneal Findings

Wilson’s disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka…familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excessive sweating
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka… familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excessive sweating

What ethnic group is most commonly affected in Riley-Day syndrome?

What does it mean to say a Jewish person is Ashkenazi?

To oversimplify a bit, it means s/he is of European descent

Why should this matter to an American ophthalmologist?

Many American Jews are of Ashkenazi descent

Selected Systemic Diseases of Childhood with Corneal Findings
**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

**Riley-Day syndrome**
--Corneal finding: keratitis/ulcers
--aka familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excessive sweating

What ethnic group is most commonly affected in Riley-Day syndrome?
Ashkenazi Jews
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excessive sweating

What ethnic group is most commonly affected in Riley-Day syndrome?
Ashkenazi Jews

What does it mean to say a Jewish person is Ashkenazi?
Selected Systemic Diseases of Childhood with Corneal Findings

**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

**Riley-Day syndrome**
--Corneal finding: keratitis/ulcers
--aka...
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excessive sweating

What ethnic group is most commonly affected in Riley-Day syndrome? Ashkenazi Jews

What does it mean to say a Jewish person is Ashkenazi? To oversimplify a bit, it means s/he is of European descent
Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka... familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excessive sweating

What ethnic group is most commonly affected in Riley-Day syndrome?
Ashkenazi Jews

What does it mean to say a Jewish person is Ashkenazi?
To oversimplify a bit, it means s/he is of European descent

Why should this matter to an American ophthalmologist?

Systemic diseases of childhood with corneal findings
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excessive sweating

What ethnic group is most commonly affected in Riley-Day syndrome?
Ashkenazi Jews

What does it mean to say a Jewish person is Ashkenazi?
To oversimplify a bit, it means s/he is of European descent

Why should this matter to an American ophthalmologist?
Many American Jews are Ashkenazi
**Selected Systemic Diseases of Childhood with Corneal Findings**

**Wilson's disease**  
--Corneal finding: Kayser-Fleischer ring  
--Disease of copper metabolism  
--Systemic findings:  
  --Liver and renal damage stigmata  
  --CNS signs/symptoms

**Riley-Day syndrome**  
--Corneal finding: keratitis/ulcers  
--aka... familial dysautonomia  
--Systemic findings:  
  --Temperature instability  
  --Pain insensitivity  
  --Labile BP  
  --Excessive sweating

**How is Riley-Day managed?**  
--ATs, punctal plugs, and tarsorrhaphy are first line. Scleral CLs may be useful. If more aggressive tx is indicated, amniotic membrane placement or conj flaps might be needed.
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excessive sweating

How is Riley-Day managed?
ATs, punctal plugs, and tarsorrhaphy are first line. Scleral CLs may be useful. If more aggressive tx is indicated, amniotic membrane placement or conj flaps might be needed.

--Labile BP
--Excessive sweating
**Selected Systemic Diseases of Childhood with Corneal Findings**

- **Wilson’s disease**
  -- Corneal finding: **Kayser-Fleischer ring**
  -- Disease of copper metabolism
  -- Systemic findings:
    -- Liver and renal damage stigmata
    -- CNS signs/symptoms

- **Cystinosis**
  -- Corneal finding: ?

- **Riley-Day syndrome**
  -- Corneal finding: **keratitis/ulcers**
  -- aka… **familial dysautonomia**
  -- Systemic findings:
    -- Temperature instability
    -- Pain insensitivity
    -- Labile BP
    -- Excess sweating

- **Systemic diseases of childhood with corneal findings**
Selected Systemic Diseases of Childhood with Corneal Findings

**Wilson’s disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

**Cystinosis**
--Corneal finding: Iridescent crystals

**Systemic diseases of childhood with corneal findings**

**Riley-Day syndrome**
--Corneal finding: keratitis/ulcers
--aka…familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating
Corneal crystals in cystinosis
Cystinosis
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of…Fanconi syndrome
--c/o severe…photophobia
--Treatment: Topical cysteamine

Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka…familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

At what age do the crystals appear?
In severe cases, before age 1 year
Are the corneal crystal clinically significant?
Yes
In what way are the significant?
The produce disabling photophobia
Can the photophobia be so severe as to warrant PK?
Yes
Can the crystal recur in the graft?
Yes

Selected Systemic Diseases of Childhood with Corneal Findings
Selected Systemic Diseases of Childhood with Corneal Findings

**Wilson’s disease**
- Corneal finding: Kayser-Fleischer ring
- Disease of copper metabolism
- Systemic findings:
  - Liver and renal damage stigmata
  - CNS signs/symptoms

**Cystinosis**
- Corneal finding: Iridescent crystals
- Usual ethnicity: French-Canadiens
- Component of Fanconi syndrome
- c/o severe photophobia
- Treatment: Topical cysteamine

**Wilson’s disease**
- Corneal finding: Kayser-Fleischer ring
- Disease of copper metabolism
- Systemic findings:
  - Liver and renal damage stigmata
  - CNS signs/symptoms

**Cystinosis**
- Corneal finding: Iridescent crystals
- Usual ethnicity: French-Canadiens
- Component of Fanconi syndrome
- c/o severe photophobia
- Treatment: Topical cysteamine

**Riley-Day syndrome**
- Corneal finding: Keratitis/ulcers
- aka…familial dysautonomia
- Systemic findings:
  - Temperature instability
  - Pain insensitivity
  - Labile BP
  - Excess sweating

At what age do the crystals appear? In severe cases, before age 1 year

Can the photophobia be so severe as to warrant PK? Yes

Can the crystal recur in the graft? Yes
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

At what age do the crystals appear?
In severe cases, before age 1 year

Are the corneal crystal clinically significant?
Yes

The crystals produce disabling photophobia

Can the photophobia be so severe as to warrant PK?
Yes

Can the crystal recur in the graft?
Yes
Cystinosis
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of Fanconi syndrome
--c/o severe photophobia
--Treatment: Topical cysteamine

Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Short stature
  --Rickets
  --ESRD

At what age do the crystals appear?
In severe cases, before age 1 year

Are the corneal crystal clinically significant?
Yes

Riley-Day syndrome
--Corneal finding: Keratitis/ulcers
--aka...familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating
Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease
- Corneal finding: Kayser-Fleischer ring
- Disease of copper metabolism
- Systemic findings:
  - Short stature
  - Rickets
  - ESRD

Cystinosis
- Corneal finding: Iridescent crystals
- Usual ethnicity: French-Canadiens
- Component of Fanconi syndrome
- c/o severe photophobia
- Treatment: Topical cysteamine

Wilson's disease
- Corneal finding: Kayser-Fleischer ring
- Disease of copper metabolism
- Systemic findings:
  - Liver and renal damage stigmata
  - CNS signs/symptoms

Riley-Day syndrome
- Corneal finding: keratitis/ulcers
- aka...familial dysautonomia
- Systemic findings:
  - Temperature instability
  - Pain insensitivity
  - Labile BP
  - Excess sweating

At what age do the crystals appear?
In severe cases, before age 1 year

Are the corneal crystal clinically significant?
Yes

In what way are the significant?
The produce disabling photophobia
Can the photophobia be so severe as to warrant PK?
Yes
Can the crystal recur in the graft?
Yes
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Cystinosis
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of Fanconi syndrome
--c/o severe photophobia
--Treatment: Topical cysteamine

Riley-Day syndrome
--Corneal finding: Keratitis/ulcers
  --aka familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

At what age do the crystals appear?
In severe cases, before age 1 year

Are the corneal crystal clinically significant?
Yes

In what way are the significant?
The produce disabling photophobia
Selected Systemic Diseases of Childhood with Corneal Findings

**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

**Cystinosis**
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of Fanconi syndrome
--c/o severe photophobia
--Treatment: Topical cysteamine

**Riley-Day syndrome**
--Corneal finding: keratitis/ulcers
--aka...familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

---

At what age do the crystals appear?
In severe cases, before age 1 year

Are the corneal crystal clinically significant?
Yes

In what way are the significant?
The production of disabling photophobia

Take note of this, as it makes for a great factoid upon which to base a test question!
Selected Systemic Diseases of Childhood with Corneal Findings

**Wilson's disease**
- Corneal finding: Kayser-Fleischer ring
- Disease of copper metabolism
- Systemic findings:
  - Short stature
  - Rickets
  - ESRD

**Cystinosis**
- Corneal finding: Iridescent crystals
- Usual ethnicity: French-Canadiens
- Component of Fanconi syndrome
- c/o severe photophobia
- Treatment: Topical cysteamine

**Riley-Day syndrome**
- Corneal finding: Keratitis/ulcers
- aka familial dysautonomia
- Systemic findings:
  - Temperature instability
  - Pain insensitivity
  - Labile BP
  - Excess sweating

At what age do the crystals appear?
In severe cases, before age 1 year

Are the corneal crystal clinically significant?
Yes

In what way are they significant?
The produce disabling photophobia

Can the photophobia be so severe as to warrant PK?
Yes

Can the crystal recur in the graft?
Yes
Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Cystinosis
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of Fanconi syndrome
--c/o severe photophobia
--Treatment: Topical cysteamine

At what age do the crystals appear?
In severe cases, before age 1 year

Are the corneal crystal clinically significant?
Yes

In what way are the significant?
The produce disabling photophobia

Can the photophobia be so severe as to warrant PK?
Yes

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka...familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Short stature
  --Rickets
  --ESRD

At what age do the crystals appear?
In severe cases, before age 1 year

Are the corneal crystal clinically significant?
Yes

In what way are the significant?
The produce disabling photophobia

Can the photophobia be so severe as to warrant PK?
Yes

Can the crystal recur in the graft?
Yes
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Cystinosis
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of Fanconi syndrome
--c/o severe photophobia
--Treatment: Topical cysteamine

At what age do the crystals appear?
In severe cases, before age 1 year

Are the corneal crystal clinically significant?
Yes

In what way are the significant?
The produce disabling photophobia

Can the photophobia be so severe as to warrant PK?
Yes

Can the crystal recur in the graft?
Yes

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka...familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

Selected Systemic Diseases of Childhood with Corneal Findings
Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Cystinosis
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of Fanconi syndrome
--c/o severe photophobia
--Treatment: Topical cysteamine

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka…familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

At what age do the crystals appear?
In severe cases, before age 1 year

Are the corneal crystal clinically significant?
Yes

In what way are the significant?
The produce disabling photophobia

Can the photophobia be so severe as to warrant PK?
Yes

Can the crystal recur in the graft?
Yes
Cystinosis
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of…Fanconi syndrome
--c/o severe…photophobia
--Treatment: Topical cysteamine

Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:

In one word, what sort of condition is cystinosis?
A metabolic disorder

Cystinosis
--Corneal finding: Iridescent crystals

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka…familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating
Cystinosis
--Corneal finding: Iridescence crystals
--Usual ethnicity: French-Canadiens
--Component of Fanconi syndrome
--c/o severe photophobia
--Treatment: Topical cysteamine

Wilson’s disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Short stature
  --Rickets
  --ESRD

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka…familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

Cystinosis
--Corneal finding: Iridescence crystals

In one word, what sort of condition is cystinosis?
A metabolic disorder

Selected Systemic Diseases of Childhood with Corneal Findings
Selected Systemic Diseases of Childhood with Corneal Findings

**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  - Short stature
  - Rickets
  - ESRD

**Cystinosis**
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of…Fanconi syndrome
--c/o severe…photophobia
--Treatment: Topical cysteamine

**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  - Liver and renal damage stigmata
  - CNS signs/symptoms

**Riley-Day syndrome**
--Corneal finding: Keratitis/ulcers
--aka…familial dysautonomia
--Systemic findings:
  - Temperature instability
  - Pain insensitivity
  - Labile BP
  - Excess sweating

In one word, what sort of condition is cystinosis?
A metabolic disorder

Metabolism of what substrate is involved?

**Cystinosis**
--Corneal finding: Iridescent crystals
**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:

**In one word, what sort of condition is cystinosis?**
A metabolic disorder

**Metabolism of what substrate is involved?**
Amino acids (‘protein’ is also correct)

**Cystinosis**
--Corneal finding: Iridescent crystals

**Riley-Day syndrome**
--Corneal finding: keratitis/ulcers
--aka...familial dysautonomia
--Systemic findings:
   --Temperature instability
   --Pain insensitivity
   --Labile BP
   --Excess sweating
Cystinosis
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of...Fanconi syndrome
--c/o severe...photophobia
--Treatment: Topical cysteamine

Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  In one word, what sort of condition is cystinosis?
  A metabolic disorder

  Metabolism of what substrate is involved?
  Amino acids ('protein' is also correct)

  Which amino acid is involved, and in general terms, what is the metabolic issue?

Cystinosis
--Corneal finding: Iridescent crystals

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka...familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating
**Cystinosis**
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of...Fanconi syndrome
--C/o severe...photophobia
--Treatment: Topical cysteamine

**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:

In one word, what sort of condition is cystinosis?
A metabolic disorder

Metabolism of what substrate is involved?
Amino acids ('protein' is also correct)

Which amino acid is involved, and in general terms, what is the metabolic issue?
The AA cystine cannot be properly disposed of, and accumulates in organs

**Riley-Day syndrome**
--Corneal finding: keratitis/ulcers
--aka...familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating
**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:

*In one word, what sort of condition is cystinosis?*
A metabolic disorder

*Metabolism of what substrate is involved?*
Amino acids ('protein' is also correct)

*Which amino acid is involved, and in general terms, what is the metabolic issue?*
The AA cystine cannot be properly disposed of, and accumulates in organs

*Which organ is most profoundly affected? (It’s not the eye)*

**Cystinosis**
--Corneal finding: Iridescent crystals

**Riley-Day syndrome**
--Corneal finding: keratitis/ulcers
--aka...familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

---

**Selected Systemic Diseases of Childhood with Corneal Findings**
Cystinosis
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of…Fanconi syndrome
--c/o severe…photophobia
--Treatment: Topical cysteamine

Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
In one word, what sort of condition is cystinosis?
A metabolic disorder

Metabolism of what substrate is involved?
Amino acids ('protein' is also correct)

Which amino acid is involved, and in general terms, what is the metabolic issue?
The AA cystine cannot be properly disposed of, and accumulates in organs

Which organ is most profoundly affected? (It’s not the eye)
The kidneys

Cystinosis
--Corneal finding: Iridescent crystals

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka…familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating
**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  - Short stature
  - Rickets
  - ESRD

**Cystinosis**
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of…Fanconi syndrome
--c/o severe…photophobia
--Treatment: Topical cysteamine

**Systemic diseases of childhood with corneal findings**

1. **Cystinosis**
   - Corneal finding: Iridescent crystals
   - Usual ethnicity: French-Canadiens
   - Component of…Fanconi syndrome
   - c/o severe…photophobia
   - Treatment: Topical cysteamine

2. **Wilson's disease**
   - Corneal finding: Kayser-Fleischer ring
   - Disease of copper metabolism
   - Systemic findings:
     - Short stature
     - Rickets
     - ESRD

3. **Riley-Day syndrome**
   - Corneal finding: keratitis/ulcers
   - aka…familial dysautonomia
   - Systemic findings:
     - Temperature instability
     - Pain insensitivity
     - Labile BP
     - Excess sweating

---

**In one word, what sort of condition is cystinosis?**
A metabolic disorder

**Metabolism of what substrate is involved?**
Amino acids (‘protein’ is also correct)

**Which amino acid is involved, and in general terms, what is the metabolic issue?**
The AA cystine cannot be properly disposed of, and accumulates in organs

---

**Renal damage in cystinosis results in what eponymous syndrome?**
Fanconi's syndrome

---

**Which organ is most profoundly affected? (It’s not the eye)**
The kidneys

---

**Which organ is most profoundly affected?**

---

**Excess sweating**
Cystinosis
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of…Fanconi syndrome
--c/o severe…photophobia
--Treatment: Topical cysteamine

Wilson’s disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Short stature
  --Rickets
  --ESRD

Wilson’s disease
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka…
familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

Selected Systemic Diseases of Childhood with Corneal Findings

In one word, what sort of condition is cystinosis?
A metabolic disorder

Metabolism of what substrate is involved?
Amino acids (‘protein’ is also correct)

Which amino acid is involved, and in general terms, what is the metabolic issue?
The AA cystine cannot be properly disposed of, and accumulates in organs

Which organ is most profoundly affected? (It’s not the eye)
The kidneys

Renal damage in cystinosis results in what eponymous syndrome?
Fanconi’s syndrome

--Excess sweating
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  - Short stature
  - Rickets
  - ESRD

In one word, what sort of condition is cystinosis?
A metabolic disorder

Metabolism of what substrate is involved?
Amino acids (‘protein’ is also correct)

Which amino acid is involved, and in general terms, what is the metabolic issue?
The AA cystine cannot be properly disposed of, and accumulates in organs

Which organ is most profoundly affected? (It's not the eye)
The kidneys

Fanconi’s syndrome…that’s where pts get pancytopenia, skin findings, short stature and increased risk of malignancy, right?

Renal damage in cystinosis results in what eponymous syndrome?
Fanconi’s syndrome

Cystinosis
--Corneal finding: Iridescent crystals

--Excess sweating

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka…familial dysautonomia
--Systemic findings:
  - Temperature instability
  - Pain insensitivity
  - Labile BP
  - Excess sweating
In one word, what sort of condition is cystinosis? A metabolic disorder

Metabolism of what substrate is involved? Amino acids ('protein' is also correct)

Which amino acid is involved? The AA cystine cannot be properly disposed of and accumulates in organs

Which organ is most profoundly affected? The kidneys

Renal damage in cystinosis results in what eponymous syndrome? Fanconi’s syndrome

Fanconi’s syndrome…that’s where pts get pancytopenia, skin findings, short stature and increased risk of malignancy, right? No, that’s Fanconi’s anemia, an inherited form of bone-marrow failure

Selected Systemic Diseases of Childhood with Corneal Findings

Wilson’s disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
   --Short stature
   --Rickets
   --ESRD

Cystinosis
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of Fanconi syndrome
--c/o severe photophobia
--Treatment: Topical cysteamine

Wilson’s disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
   --Liver and renal damage stigmata
   --CNS signs/symptoms

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka familial dysautonomia
--Systemic findings:
   --Temperature instability
   --Pain insensitivity
   --Labile BP
   --Excess sweating

Cystinosis
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of Fanconi syndrome
--c/o severe photophobia
--Treatment: Topical cysteamine

Systemic diseases of childhood with corneal findings
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
    In one word, what sort of condition is cystinosis?
    A metabolic disorder
    Metabolism of what substrate is involved?
    Amino acids (‘protein’ is also correct)
    Which amino acid is involved, and in general terms, what is the metabolic issue?
    The AA cystine cannot be properly disposed of, and accumulates in organs
    Which organ is most profoundly affected?
    The kidneys
    Renal damage in cystinosis results in what eponymous syndrome?
    Fanconi’s syndrome
    What is the fundamental renal problem in Fanconi’s syndrome?
--Excess sweating

Cystinosis
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of Fanconi syndrome
--c/o severe photophobia
--Treatment: Topical cysteamine

Riley-Day syndrome
--Corneal finding: Keratitis/ulcers
--aka familial dysautonomia
--Systemic findings:
    Temperature instability
    Pain insensitivity
    Labile BP
    Excess sweating

Selected Systemic Diseases of Childhood with Corneal Findings
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  - Short stature
  - Rickets
  - ESRD

Wilson's disease is a disease of copper metabolism. The systemic findings include short stature, rickets, and end-stage renal disease (ESRD).

Cystinosis
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of Fanconi syndrome
--'c/o' severe photophobia
--Treatment: Topical cysteamine

Cystinosis is a metabolic disorder involving the metabolism of amino acids, particularly cystine. The corneal finding is iridescent crystals, and the disease is associated with Fanconi syndrome and photophobia. Treatment involves topical cysteamine.

Riley-Day syndrome
--Corneal finding: Keratitis/ulcers
--aka familial dysautonomia
--Systemic findings:
  - Temperature instability
  - Pain insensitivity
  - Labile BP
  - Excess sweating

Riley-Day syndrome is characterized by corneal keratitis or ulcers and systemic findings such as temperature instability, pain insensitivity, labile blood pressure, and excess sweating.

Selected Systemic Diseases of Childhood with Corneal Findings

In one word, what sort of condition is cystinosis?
A metabolic disorder

Metabolism of what substrate is involved?
Amino acids ('protein' is also correct)

Which amino acid is involved, and in general terms, what is the metabolic issue?
The AA cystine cannot be properly disposed of, and accumulates in organs

Which organ is most profoundly affected? (It’s not the eye)
The kidneys

Renal damage in cystinosis results in what eponymous syndrome?
Fanconi’s syndrome

What is the fundamental renal problem in Fanconi’s syndrome?
Failure of the proximal tubules to resorb a broad range of substances
Selected Systemic Diseases of Childhood with Corneal Findings

**Wilson's disease**
-- Corneal finding: Kayser-Fleischer ring
-- Disease of copper metabolism
-- Systemic findings:

In one word, what sort of condition is cystinosis?
A metabolic disorder

Metabolism of what substrate is involved?
Amino acids (‘protein’ is also correct)

Which amino acid is involved, and in general terms, what is the metabolic issue?
The AA cystine cannot be properly disposed of, and accumulates in organs

Which organ is most profoundly affected? (It's not the eye)
The kidneys

Renal damage in cystinosis results in what eponymous syndrome?
Fanconi's syndrome

What is the fundamental renal problem in Fanconi’s syndrome?
Failure of the proximal tubules to resorb a broad range of substances

Resorption failure produces what short and/or long-term problems?
--
--
--

-- Excess sweating

Cystinosis
-- Corneal finding: Iridescent crystals
-- Systemic findings:

--
**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:

In one word, what sort of condition is cystinosis?
A metabolic disorder

Metabolism of what substrate is involved?
Amino acids (‘protein’ is also correct)

Which amino acid is involved, and in general terms, what is the metabolic issue?
The AA cystine cannot be properly disposed of, and accumulates in organs

Which organ is most profoundly affected?
The kidneys

Renal damage in cystinosis results in what eponymous syndrome?
Fanconi’s syndrome

What is the fundamental renal problem in Fanconi’s syndrome?
Failure of the proximal tubules to resorb a broad range of substances

Resorption failure produces what short and/or long-term problems?
--Failure to thrive→growth failure→short stature/dwarfism
--Rickets
--Progressive renal failure→ESRD

--Excess sweating
Selected Systemic Diseases of Childhood with Corneal Findings

**Wilson's disease**
- Corneal finding: Kayser-Fleischer ring
- Disease of copper metabolism
- Systemic findings:
  - Liver and renal damage stigmata
  - CNS signs/symptoms

**Cystinosis**
- Corneal finding: Iridescent crystals
- Systemic findings:
  - Short stature
  - Rickets
  - ESRD

**Riley-Day syndrome**
- Corneal finding: keratitis/ulcers
- aka... familial dysautonomia
- Systemic findings:
  - Temperature instability
  - Pain insensitivity
  - Labile BP
  - Excess sweating

In one word, what sort of condition is cystinosis?
A metabolic disorder

Metabolism of what substrate is involved?
Amino acids (‘protein’ is also correct)

Which amino acid is involved, and in general terms, what is the metabolic issue?
The AA cystine cannot be properly disposed of, and accumulates in organs

Which organ is most profoundly affected?
The kidneys

Renal damage in cystinosis results in what eponymous syndrome?
Fanconi’s syndrome

What is rickets?
A condition in which inadequate mineralization of long-bone growth plates leads to disruption in plate function, resulting in impaired bone growth as well as the characteristic bone ‘bowing’

**Rickets**
- Failure to thrive → growth failure → short stature/dwarfism
- Progressive renal failure → ESRD

--Excess sweating
Cystinosis
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of…Fanconi syndrome
--c/o severe…photophobia
--Treatment: Topical cysteamine

Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka…
  --familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Cystinosis
--Corneal finding: Iridescent crystals
--Systemic findings:
  --Short stature
  --Rickets
  --ESRD

Renal damage in cystinosis results in what eponymous syndrome?
  Fanconi's syndrome

What is rickets?
A condition in which inadequate mineralization of long-bone growth plates leads to disruption in plate function, resulting in impaired bone growth as well as the characteristic bone ‘bowing’

--Failure to thrive→growth failure→short stature/dwarfism
--ESRD
  --Excess sweating
Systemic drugs and ocular toxicity: Matching

Rickets
Cystinosis
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of…Fanconi syndrome
--c/o severe…photophobia
--Treatment: Topical cysteamine

Wilson’s disease
--Corneal finding: Kayser-Fleisher ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka…familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

Systemic diseases of childhood with corneal findings

In one word, what sort of condition is cystinosis?
A metabolic disorder

Metabolism of what substrate is involved?
Amino acids (‘protein’ is also correct)

Can other substances besides cystine result in Fanconi syndrome?

Which organ is most profoundly affected?
The kidneys

Renal damage in cystinosis results in what syndromic condition?
Fanconi’s syndrome

What is the fundamental renal problem in Fanconi’s syndrome?
Failure of the proximal tubules to resorb a broad range of substances

Resorption failure produces what short and/or long-term problems?
--Failure to thrive → growth failure → short stature/dwarfism
--Rickets
--Progressive renal failure → ESRD

--Excess sweating
**Selected Systemic Diseases of Childhood with Corneal Findings**

**Wilson's disease**
- Corneal finding: Kayser-Fleischer ring
- Disease of copper metabolism
- Systemic findings:
  - In one word, what sort of condition is cystinosis?
    - A metabolic disorder
  - Metabolism of what substrate is involved?
    - Amino acids (‘protein’ is also correct)
  - Can other substances besides cystine result in Fanconi syndrome?
    - Indeed they can--the list of such substances is very long
  - Which organ is most profoundly affected?
    - The kidneys

**Cystinosis**
- Corneal finding: Iridescent crystals
- Systemic findings:
  - Renal damage in cystinosis results in what syndromic condition?
    - Fanconi’s syndrome
  - What is the fundamental renal problem in Fanconi’s syndrome?
    - Failure of the proximal tubules to resorb a broad range of substances
  - Resorption failure produces what short and/or long-term problems?
    - Failure to thrive → growth failure → short stature/dwarfism
    - Excess sweating

**Other systemic diseases**
- Wilson’s disease
- Cystinosis
- Other systemic diseases of childhood with corneal findings
Selected Systemic Diseases of Childhood with Corneal Findings

**Wilson’s disease**
- Corneal finding: Kayser-Fleischer ring
- Disease of copper metabolism

Speaking of the list of such substances...One of these is copper, which is why Wilson’s dz is also associated with Fanconi syndrome (Note: Neither the Peds nor Cornea book mentions Fanconi syndrome in association with Wilson’s, so continue to associate Fanconi’s with cystinosis in your test prep)

Amino acids (‘protein’ is also correct)

Can other substances besides cystine result in Fanconi syndrome? Indeed they can -- the list of such substances is very long

Which organ is most profoundly affected? The kidneys

Renal damage in cystinosis results in what syndromic condition? Fanconi’s syndrome

What is the fundamental renal problem in Fanconi’s syndrome? Failure of the proximal tubules to resorb a broad range of substances

Resorption failure produces what short and/or long-term problems?
- Failure to thrive → growth failure → short stature/dwarfism
- Rickets
- Progressive renal failure → ESRD

-- Excess sweating
Selected Systemic Diseases of Childhood with Corneal Findings

**Wilson's disease**
- Corneal finding: Kayser-Fleischer ring
- Disease of copper metabolism
- Systemic findings:
  - Liver and renal damage stigmata
  - CNS signs/symptoms

**Cystinosis**
- Corneal finding: Iridescent crystals

**Riley-Day syndrome**
- Corneal finding: keratitis/ulcers
- aka...familial dysautonomia
- Systemic findings:
  - Labile BP
  - Excess sweating

With regard to eye involvement: Are the crystals limited to the cornea?
Selected Systemic Diseases of Childhood with Corneal Findings

**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

**Cystinosis**
--Corneal finding: Iridescent crystals

**Riley-Day syndrome**
--Corneal finding: keratitis/ulcers
--aka…familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

With regard to eye involvement: Are the crystals limited to the cornea?
No, they can be found throughout the anterior segment–conj, TM, iris, etc, as well as in the retina.
Crystalline retinopathy in cystinosis
Selected Systemic Diseases of Childhood with Corneal Findings

Wilson’s disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Cystinosis
--Corneal finding: Iridescent crystals

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
  aka…familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

With regard to eye involvement: Are the crystals limited to the cornea?
No, they can be found throughout the anterior segment—conj, TM, iris, etc, as well as in the retina

The Retina book identifies three general causes of crystalline retinopathy—what are they?

Systemic diseases of childhood with corneal findings

Drug-induced Ocular dx
The Retina book identifies three general causes of crystalline retinopathy—what are they?

Systemic dz
--Liver and renal damage stigmata
--CNS signs/symptoms

Drug-induced Ocular dx
--Liver and renal damage stigmata
--CNS signs/symptoms
Cystinosis
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of Fanconi syndrome
--c/o severe photophobia
--Treatment: Topical cysteamine

Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
  aka... familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

Systemic diseases of childhood with corneal findings

With regard to eye involvement: Are the crystals limited to the cornea?
No, they can be found throughout the anterior segment—conj, TM, iris, etc, as well as in the retina

Ocular dz

Selected Systemic Diseases of Childhood with Corneal Findings

The Retina book identifies three general causes of crystalline retinopathy—what are they?

Systemic dz

Drug-induced

Cystinosis
--Corneal finding: Iridescent crystals

With regard to eye involvement: Are the crystals limited to the cornea?
Selected Systemic Diseases of Childhood with Corneal Findings

**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

**Cystinosis**
--Corneal finding: Iridescent crystals
--Component of Fanconi syndrome
--c/o severe photophobia
--Treatment: Topical cysteamine

**Systemic diseases of childhood with corneal findings**

**Drug-induced**

**Ocular dz**

**Cystinosis**
--Corneal finding: Iridescent crystals

At this juncture, we are well familiar with the systemic condition, **cystinosis**.

With regard to eye involvement: Are the crystals limited to the cornea?
No, they can be found throughout the anterior segment—conj, TM, iris, etc, as well as in the retina.

**Systemic dz**

Corneal finding: keratitis/ulcers
--aka... familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

**Riley-Day syndrome**
--Corneal finding: keratitis/ulcers
--aka... familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

**Drug-induced**

**Ocular dz**

**Cystinosis**
--Corneal finding: Iridescent crystals

At this juncture, we are well familiar with the systemic condition, **cystinosis**.

With regard to eye involvement: Are the crystals limited to the cornea?
No, they can be found throughout the anterior segment—conj, TM, iris, etc, as well as in the retina.

**Systemic dz**

Corneal finding: keratitis/ulcers
--aka... familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

**Riley-Day syndrome**
--Corneal finding: keratitis/ulcers
--aka... familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating
Cystinosis
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of…Fanconi syndrome
--c/o severe…photophobia
--Treatment: Topical cysteamine

Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Systemic diseases of childhood with corneal findings

Drug-induced
('<')?
('<')?
('<')?

Three drugs are well-known causes—what are they?

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka…familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

With regard to eye involvement: Are the crystals limited to the cornea?
No, they can be found throughout the anterior segment–conj, TM, iris, etc, as well as in the retina

Cystinosis
--Corneal finding: Iridescent crystals

Ocular dz

Selected Systemic Diseases of Childhood with Corneal Findings
**Selected Systemic Diseases of Childhood with Corneal Findings**

**Wilson's disease**
- Corneal finding: Kayser-Fleischer ring
- Disease of copper metabolism
- Systemic findings:
  - Liver and renal damage stigmata
  - CNS signs/symptoms

**Cystinosis**
- Corneal finding: Iridescent crystals
- Usual ethnicity: French-Canadiens
- Component of Fanconi syndrome
- c/o severe photophobia
- Treatment: Topical cysteamine

**Systemic diseases of childhood with corneal findings**

**Drug-induced**
- Tamoxifen
- Canthaxanthine
- Talc

**Riley-Day syndrome**
- Corneal finding: keratitis/ulcers
- aka...familial dysautonomia
- Systemic findings:
  - Temperature instability
  - Pain insensitivity
  - Labile BP
  - Excess sweating

**With regard to eye involvement: Are the crystals limited to the cornea?**
No, they can be found throughout the anterior segment—conj, TM, iris, etc, as well as in the retina.

**Three drugs are well-known causes—what are they?**

**Ocular dz**

**Cystinosis**
- Corneal finding: Iridescent crystals
Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka...familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

Cystinosis
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of...Fanconi syndrome
--c/o severe...photophobia
--Treatment: Topical cysteamine

Drug-induced
  --Tamoxifen
  --Canthaxanthine
  --Talc

With regard to eye involvement: Are the crystals limited to the cornea?
No, they can be found throughout the anterior segment—conj, TM, iris, etc, as well as in
  --the retina

Systemic dz
  -- Cystinosis

Ocular dz
  ?

One ocular-only condition makes the cut—what is it?
Selected Systemic Diseases of Childhood with Corneal Findings

**Wilson's disease**
- Corneal finding: Kayser-Fleischer ring
- Disease of copper metabolism
- Systemic findings:
  - Liver and renal damage stigmata
  - CNS signs/symptoms

**Cystinosis**
- Corneal finding: Iridescent crystals
- Usual ethnicity: French-Canadiens
- Component of Fanconi syndrome
- c/o severe photophobia
- Treatment: Topical cysteamine

**Drug-induced**
- Tamoxifen
- Canthaxanthine
- Talc

**Systemic dz**
- Cystinosis

**Systemic diseases of childhood with corneal findings**

**Ocular dz**
- Bietti crystalline dystrophy

**Riley-Day syndrome**
- Corneal finding: keratitis/ulcers
- aka...familial dysautonomia
- Systemic findings:
  - Temperature instability
  - Pain insensitivity
  - Labile BP
  - Excess sweating

With regard to eye involvement: Are the crystals limited to the cornea?
No, they can be found throughout the anterior segment—conj, TM, iris, etc, as well as in the retina.

One ocular-only condition makes the cut—what is it?

Selected Systemic Diseases of Childhood with Corneal Findings

Drug-induced
- Tamoxifen
- Canthaxanthine
- Talc

With regard to eye involvement: Are the crystals limited to the cornea?
No, they can be found throughout the anterior segment—conj, TM, iris, etc, as well as in the retina.

One ocular-only condition makes the cut—what is it?
Selected Systemic Diseases of Childhood with Corneal Findings

**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

**Systemic diseases of childhood with corneal findings**

**Cystinosis**
--Corneal finding: Iridescent crystals
--Systemic findings:
  --Short stature
  --Rickets
  --ESRD

---

**Cystinosis comes in three forms--what are they?**

---

**Riley-Day syndrome**
--Corneal finding: keratitis/ulcers
--aka...familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

---

---

---
Selected Systemic Diseases of Childhood with Corneal Findings

**Wilson's disease**
- Corneal finding: Kayser-Fleischer ring
- Disease of copper metabolism
- Systemic findings:
  - Liver and renal damage stigmata
  - CNS signs/symptoms

**Systemic diseases of childhood with corneal findings**

**Cystinosis comes in three forms--what are they?**
- Infantile onset
- Juvenile onset
- Adult onset

**Cystinosis**
- Corneal finding: Iridescent crystals
- Systemic findings:
  - Short stature
  - Rickets
  - ESRD

**Riley-Day syndrome**
- Corneal finding: keratitis/ulcers
- aka...familial dysautonomia
- Systemic findings:
  - Temperature instability
  - Pain insensitivity
  - Labile BP
  - Excess sweating
Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Cystinosis
--Corneal finding: Iridescent crystals
--Systemic findings:
  --Short stature
  --Rickets
  --ESRD

Cystinosis comes in three forms--what are they? Which manifest corneal crystals?
--Infantile onset.
--Juvenile onset.
--Adult onset.

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka...familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating
Selected Systemic Diseases of Childhood with Corneal Findings

**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

**Cystinosis**
--Corneal finding: Iridescent crystals
--Systemic findings:
  --Short stature
  --Rickets
  --ESRD

**Cystinosis comes in three forms--what are they?** Which manifest corneal crystals?
--Infantile onset. Corneal crystals+.
--Juvenile onset. Corneal crystals+.
--Adult onset. Corneal crystals+.

**Systemic diseases of childhood with corneal findings**

**Riley-Day syndrome**
--Corneal finding: keratitis/ulcers
--aka...familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating
**Selected Systemic Diseases of Childhood with Corneal Findings**

**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

**Cystinosis**
--Corneal finding: Iridescent crystals
--Systemic findings:
  --Short stature
  --Rickets
  --ESRD

**Riley-Day syndrome**
--Corneal finding: keratitis/ulcers
--aka...familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

**Cystinosis comes in three forms--what are they? Which manifest corneal crystals? Which develop features of Fanconi syndrome?**
--Infantile onset. Corneal crystals+. Severe Fanconi syndrome.
--Adult onset. Corneal crystals+. No Fanconi syndrome.
Cystinosis
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of…Fanconi syndrome
--c/o severe…photophobia
--Treatment: Topical cysteamine

Wilson’s disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Systemic diseases of childhood with corneal findings

Cystinosis comes in three forms--what are they? Which manifest corneal crystals? Which develop features of Fanconi syndrome?
--Infantile onset. Corneal crystals+. **Severe** Fanconi syndrome.
--Juvenile onset. Corneal crystals+. **Mild** Fanconi syndrome.
--Adult onset. Corneal crystals+. **No** Fanconi syndrome.

Cystinosis
--Corneal finding: Iridescent crystals
--Systemic findings:
  --Short stature
  --Rickets
  --ESRD

**Wilson’s disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

**Riley-Day syndrome**
--Corneal finding: keratitis/ulcers
  --aka…**familial dysautonomia**
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating
Selected Systemic Diseases of Childhood with Corneal Findings

**Cystinosis**
--Corneal finding: Iridescent crystals
--Systemic findings:
  --Short stature
  --Rickets
  --ESRD

**Wilson’s disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

**Riley-Day syndrome**
--Corneal finding: keratitis/ulcers
--aka…familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

Cystinosis comes in three forms--what are they? Which manifest corneal crystals? Which develop features of Fanconi syndrome?
--Infantile onset. Corneal crystals+. Severe
--Juvenile onset. Corneal crystals+. MMD
--Adult onset. Corneal crystals+. No Fanconi syndrome

Note that all three types manifest corneal crystals!
**Cystinosis**
--Corneal finding: *Iridescent crystals*
--Systemic findings:
  --Short stature
  --Rickets
  --ESRD

*Wilson's disease*
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

*Systemic diseases of childhood with corneal findings*

Cystinosis comes in three forms--what are they? Which manifest corneal crystals? Which develop features of Fanconi syndrome?
--Adult onset. Corneal crystals+. *No Fanconi syndrome.*

*Note also that the earlier the onset, the worse the dz*

*Riley-Day syndrome*
--Corneal finding: keratitis/ulcers
--aka...*familial dysautonomia*
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating
Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

A word on dz classification:
Be forewarned that the three relevant BCSC books (*Peds*, *Cornea* and *Retina*) do not employ a consistent schema for classifying the cystinoses. To wit:

Cystinosis comes in three forms—what are they? Which manifest corneal crystals? Which develop features of Fanconi syndrome?

--**Infantile onset**. Corneal crystals+. Severe Fanconi syndrome.
--**Juvenile onset**. Corneal crystals+. Mild Fanconi syndrome.
--**Adult onset**. Corneal crystals+. No Fanconi syndrome.

Cystinosis
--Corneal finding: Iridescent crystals
--Systemic findings:
  --Short stature
  --Rickets
  --ESRD

(No question—proceed when ready)
**Selected Systemic Diseases of Childhood with Corneal Findings**

**Wilson's disease**
- Corneal finding: Kayser-Fleischer ring
- Disease of copper metabolism
- Systemic findings:
  - Liver and renal damage stigmata
  - CNS signs/symptoms

**Cystinosis**
- Corneal finding: Iridescent crystals
- Systemic findings:
  - Short stature
  - Rickets
  - ESRD

**A word on dz classification:**
Be forewarned that the three relevant BCSC books (*Peds*, *Cornea* and *Retina*) do not employ a consistent schema for classifying the cystinoses. To wit:
- The *Retina* book divvies them into *Nephropathic, Late-onset and Benign*.

---

**(No question—proceed when ready)**
Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Cystinosis comes in three forms--what are they? Which manifest corneal crystals? Which develop features of Fanconi syndrome?

--Infantile onset. Corneal crystals+. Severe Fanconi syndrome.
--Adult onset. Corneal crystals+. No Fanconi syndrome.

A word on dz classification:
Be forewarned that the three relevant BCSC books (Peds, Cornea and Retina) do not employ a consistent schema for classifying the cystinoses. To wit:
--The Retina book divvies them into Nephropathic, Late-onset and Benign.
--The Cornea book divides them into two groups: Nephropathic and Nonnephropathic. It then subdivides Nephropathic into Infantile (aka Classic) and Intermediate (aka Juvenile) forms.
Cystinosis
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of…Fanconi syndrome
--c/o severe…photophobia
--Treatment: Topical cysteamine

Wilson’s disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka…familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

Systemic diseases
of childhood with
corneal findings

Cystinosis comes in three forms—what are they? Which manifest corneal crystals? Which develop features of Fanconi syndrome?
--Infantile onset. Corneal crystals+. Severe Fanconi syndrome.
--Adult onset. Corneal crystals+. No Fanconi syndrome.

A word on dz classification:
Be forewarned that the three relevant BCSC books (Peds, Cornea and Retina) do not employ a consistent schema for classifying the cystinoses. To wit:
--The Retina book divvies them into Nephropathic, Late-onset and Benign.
--The Cornea book divides them into two groups: Nephropathic and Nonnephropathic. It then subdivides Nephropathic into Infantile (aka Classic) and Intermediate (aka Juvenile) forms.
--The Peds book refers to an infantile form (thereby implying the existence of other forms), but addresses the issue no further.

(No question—proceed when ready)
Selected Systemic Diseases of Childhood with Corneal Findings

A word on dz classification:

Be forewarned that the three relevant BCSC books (Peds, Cornea and Retina) do not employ a consistent schema for classifying the cystinoses. To wit:

--The Retina book divvies them into Nephropathic, Late-onset and Benign.
--The Cornea book divides them into two groups: Nephropathic and Nonnephropathic. It then subdivides Nephropathic into Infantile (aka Classic) and Intermediate (aka Juvenile) forms.
--The Peds book refers to an infantile form (thereby implying the existence of other forms), but addresses the issue no further.

The terms Infantile Onset, Juvenile Onset and Adult Onset are my attempt at a compromise nomenclature. Caveat emptor.

(No question—proceed when ready)
Cystinosis
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of…Fanconi syndrome
--c/o severe…photophobia
--Treatment: ?

Wilson’s disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
  --aka…familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

What is the treatment for cystinosis?

Cystinosis
--Corneal finding: Iridescent crystals
--Systemic findings:
  --Short stature
  --Rickets
  --ESRD
--Treatment: ?

Systemic diseases of childhood with corneal findings
**Cystinosis**

--Corneal finding: Iridescent crystals
--Systemic findings:
  --Short stature
  --Rickets
  --ESRD
--Treatment: cysteamine

**Wilson's disease**

--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

**Riley-Day syndrome**

--Corneal finding: keratitis/ulcers
  --aka...familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

**What is the treatment for cystinosis?**

Cysteamine

**Systemic diseases**

Selected Systemic Diseases of Childhood with Corneal Findings
**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

**Cystinosis**
--Corneal finding: Iridescent crystals
--Systemic findings:
  --Short stature
  --Rickets
  --ESRD
--Treatment: cysteamine

**Riley-Day syndrome**
--Corneal finding: keratitis/ulcers
  --aka...familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

**Systemic diseases of childhood with corneal findings**

---

What is the treatment for cystinosis?
Cysteamine

Systemic, or topical?
Cystinosis
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of…Fanconi syndrome
--c/o severe…photophobia
--Treatment: Topical cysteamine

Wilson’s disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
  --aka…familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

Selected Systemic Diseases of Childhood with Corneal Findings

What is the treatment for cystinosis?
Cysteamine

Systemic, or topical?
Both. Systemic cysteamine is needed to treat the systemic manifestations, but do nothing for the corneal crystals, which require topical cysteamine.

Cystinosis
--Corneal finding: Iridescent crystals
--Systemic findings:
  --Short stature
  --Rickets
  --ESRD
--Treatment: Topical cysteamine
**Cystinosis**
--Corneal finding: Iridescent crystals
--Systemic findings:
  --Short stature
  --Rickets
  --ESRD
--Treatment: Topical cysteamine

**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

**Systemic diseases**

---

What is the treatment for cystinosis?

*Topical cysteamine has a notorious downside that renders compliance difficult. What is it?*

---

**Cystinosis**
--Corneal finding: Iridescent crystals
--Systemic findings:  
  --Short stature
  --Rickets
  --ESRD
--Treatment: Topical cysteamine

**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:  
  --Liver and renal damage stigmata
  --CNS signs/symptoms

**Riley-Day syndrome**
--Corneal finding: keratitis/ulcers
--aka...familial dysautonomia
--Systemic findings:  
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating
**Selected Systemic Diseases of Childhood with Corneal Findings**

---

**Cystinosis**
- Corneal finding: Iridescent crystals
- Systemic findings:
  - Short stature
  - Rickets
  - ESRD
- Treatment: Topical cysteamine

---

**Wilson's disease**
- Corneal finding: Kayser-Fleischer ring
- Disease of copper metabolism
- Systemic findings:
  - Liver and renal damage stigmata
  - CNS signs/symptoms

---

**Riley-Day syndrome**
- Corneal finding: keratitis/ulcers
- aka…familial dysautonomia
- Systemic findings:
  - Temperature instability
  - Pain insensitivity
  - Labile BP
  - Excess sweating

---

**Systemic diseases**

**What is the treatment for cystinosis?**
- **Topical cysteamine** has a notorious downside that renders compliance difficult. **What is it?**
- Cysteamine smells **terrible**

---

**Cysteamine**
- Short stature
- Rickets
- ESRD

---

**Systemic, or topical?**
- Both. Systemic cysteamine is needed to treat the systemic manifestations, but do nothing for the corneal crystals, which require **topical cysteamine**.
**Wilson’s disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

**Riley-Day syndrome**
--Corneal finding: keratitis/ulcers
--aka…familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

**Cystinosis**
--Corneal finding: Iridescent crystals
--Systemic findings:
  --Short stature
  --Rickets
  --ESRD
--Treatment: Topical cysteamine

**Congenital syphilis**
--Corneal finding: ?

Selected Systemic Diseases of Childhood with Corneal Findings
Selected Systemic Diseases of Childhood with Corneal Findings

**Wilson's disease**
--Corneal finding: *Kayser-Fleischer ring*
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

**Riley-Day syndrome**
--Corneal finding: *keratitis/ulcers*
--aka... *familial dysautonomia*
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

**Congenital syphilis**
--Corneal finding: *Interstitial keratitis*

**Cystinosis**
--Corneal finding: *Iridescent crystals*
--Usual ethnicity: French-Canadiens
--Component of... *Fanconi syndrome*
--c/o severe... *photophobia*
--Treatment: Topical cysteamine

**Systemic diseases of childhood with corneal findings**
Corneal scar/haze

Ghost vessels

Congenital syphilis: Interstitial keratitis
Wilson’s disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
  --Liver and renal damage stigmata
  --CNS signs/symptoms

Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--aka…familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

Congenital syphilis
--Corneal finding: Interstitial keratitis
--Systemic findings: usual stigmata

Cystinosis
--Corneal finding: Iridescent crystals
--Usual ethnicity: French-Canadiens
--Component of…Fanconi syndrome
--c/o severe…photophobia
--Treatment: Topical cysteamine

Systemic diseases of childhood with corneal findings
Selected Systemic Diseases of Childhood with Corneal Findings

- **Wilson's disease**
  - Corneal finding: Kayser-Fleischer ring
  - Disease of copper metabolism
  - Systemic findings:
    - Liver and renal damage stigmata
    - CNS signs/symptoms

- **Congenital syphilis**
  - Corneal finding: Interstitial keratitis
  - Systemic findings: usual stigmata

- **Cystinosis**
  - Corneal finding: Iridescent crystals
  - Usual ethnicity: French-Canadiens
  - Component of Fanconi syndrome
  - c/o severe photophobia
  - Treatment: Topical cysteamine

- **Riley-Day syndrome**
  - Corneal finding: keratitis/ulcers
  - aka...familial dysautonomia
  - Systemic findings:
    - Temperature instability
    - Pain insensitivity
    - Labile BP
    - Excess sweating

**Systemic diseases of childhood with corneal findings**

What are the ‘usual stigmata’ of congenital syphilis?
Other than IK, what are the stigmata of congenital syphilis?
Other than IK, what are the stigmata of congenital syphilis?

- Circumoral scars
- Hutchinson teeth
- Saddle nose
- Saber shins
- MR (Mental retardation)
- CN8 deafness
Other than IK, what are the stigmata of congenital syphilis?

- **Circumoral scars**
- Hutchinson teeth
- Saddle nose
- Saber shins
- MR
- CN8 deafness

What is the formal term for these circumoral scars?
Other than IK, what are the stigmata of congenital syphilis?

- **Circumoral scars**
- Hutchinson teeth
- Saddle nose
- Saber shins
- MR
- CN8 deafness

What is the formal term for these circumoral scars? ‘Rhagades’
Congenital syphilis: Circumoral scars
Other than IK, what are the stigmata of congenital syphilis?
- Circumoral scars
- **Hutchinson teeth**
- Saddle nose
- Saber shins
- MR
- CN8 deafness

What is the classic description of Hutchinson teeth?
Other than IK, what are the stigmata of congenital syphilis?

- Circumoral scars
- **Hutchinson teeth**
- Saddle nose
- Saber shins
- MR
- CN8 deafness

*What is the classic description of Hutchinson teeth? 'Peg-shaped'*
Congenital syphilis: Teephus
Other than IK, what are the stigmata of congenital syphilis?
- Circumoral scars
- Hutchinson teeth
- Saddle nose
- Saber shins
- MR
- CN8 deafness

Speaking of abnormal teeth…Name two other congenital eye syndromes that are associated with abnormal dentition in some form or fashion:
- 
- CN8 deafness
Other than IK, what are the stigmata of congenital syphilis?

- Circumoral scars
- Hutchinson teeth
- Saddle nose
- Saber shins
- MR CN8 deafness

Speaking of abnormal teeth... Name two other congenital eye syndromes that are associated with abnormal dentition in some form or fashion:

- Axenfeld-Rieger syndrome
- Incontinentia pigmenti
Other than IK, what are the stigmata of congenital syphilis?
- Circumoral scars
- Hutchinson teeth
- Saddle nose
- Saber shins
- MR
- CN8 deafness

Speaking of abnormal teeth…Name two other congenital eye syndromes that are associated with abnormal dentition in some form or fashion:
- Axenfeld-Rieger syndrome
- Incontinentia pigmenti

In three words, what sort of condition is Axenfeld-Rieger?
An…
Other than IK, what are the stigmata of congenital syphilis?

- Circumoral scars
- Hutchinson teeth
- Saddle nose
- Saber shins
- MR
- CN8 deafness

Speaking of abnormal teeth…Name two other congenital eye syndromes that are associated with abnormal dentition in some form or fashion:

--Axenfeld-Rieger syndrome
--Incontinentia pigmenti

In three words, what sort of condition is Axenfeld-Rieger?
An…anterior-segment dysgenesis
Other than IK, what are the stigmata of congenital syphilis?

- Circumoral scars
- Hutchinson teeth
- Saddle nose
- Saber shins
- MR
- CN8 deafness

Speaking of abnormal teeth... Name two other congenital eye syndromes that are associated with abnormal dentition in some form or fashion:

- Axenfeld-Rieger syndrome
- Incontinentia pigmenti

In three words, what sort of condition is Axenfeld-Rieger?
An... anterior-segment dysgenesis

If limited to one word, what sort of condition is Axenfeld-Rieger?
A...
Other than IK, what are the stigmata of congenital syphilis?

- Circumoral scars
- Hutchinson teeth
- Saddle nose
- Saber shins
- MR
- CN8 deafness

Speaking of abnormal teeth… Name two other congenital eye syndromes that are associated with abnormal dentition in some form or fashion:

- Axenfeld-Rieger syndrome
- Incontinentia pigmenti

In three words, what sort of condition is Axenfeld-Rieger?

An... anterior-segment dysgenesis

If limited to one word, what sort of condition is Axenfeld-Rieger?

A... neurocristopathy
Other than IK, what are the stigmata of congenital syphilis?

- Circumoral scars
- Hutchinson teeth
- Saddle nose
- Saber shins
- MR
- CN8 deafness

Speaking of abnormal teeth…Name two other congenital eye syndromes that are associated with abnormal dentition in some form or fashion:

--Axenfeld-Rieger syndrome
--Incontinentia pigmenti

In one word, what sort of condition is incontinentia pigmenti?

A…
Other than IK, what are the stigmata of congenital syphilis?
- Circumoral scars
- Hutchinson teeth
- Saddle nose
- Saber shins
- MR
- CN8 deafness

Speaking of abnormal teeth... Name two other congenital eye syndromes that are associated with abnormal dentition in some form or fashion:
-- Axenfeld-Rieger syndrome
-- Incontinentia pigmenti

In one word, what sort of condition is incontinentia pigmenti?
A... phakomatosis
Other than IK, what are the stigmata of congenital syphilis?

- Circumoral scars
- Hutchinson teeth
- Saddle nose
- Saber shins
- MR
- CN8 deafness

In the present context, to what does the term Hutchinson’s triad refer?
Other than IK, what are the stigmata of congenital syphilis?

- Circumoral scars
- Hutchinson teeth
- Saddle nose
- Saber shins
- MR
- CN8 deafness

In the present context, to what does the term Hutchinson’s triad refer? To the three stigmata of congenital syphilis that are especially common.
Other than IK, what are the stigmata of congenital syphilis?
- Circumoral scars
- Hutchinson teeth
- Saddle nose
- Saber shins
- MR
- CN8 deafness

Which three comprise Hutchinson’s triad?
- Interstitial keratitis
- Deafness
- Hutchinson teeth

In the present context, to what does the term Hutchinson’s triad refer?
To the three stigmata of congenital syphilis that are especially common
Other than **IK**, what are the stigmata of congenital syphilis?
- Circumoral scars
- **Hutchinson teeth**
- Saddle nose
- Saber shins
- MR
- **CN8 deafness**

*Which three comprise Hutchinson’s triad?*
- Interstitial keratitis
- Deafness
- Hutchinson teeth

*In the present context, to what does the term Hutchinson’s triad refer? To the three stigmata of congenital syphilis that are especially common*
Other than IK, what are the stigmata of congenital syphilis?
- Circumoral scars
- Hutchinson teeth
- Saddle nose
- Saber shins
- MR
- CN8 deafness

*What is the classic retinal finding in congenital lues?*
Other than IK, what are the stigmata of congenital syphilis?
- Circumoral scars
- Hutchinson teeth
- Saddle nose
- Saber shins
- MR
- CN8 deafness

What is the classic retinal finding in congenital lues?
Salt-and-pepper retinitis
Congenital syphilis: Salt-and-pepper retinitis
Other than IK, what are the stigmata of congenital syphilis?
- Circumoral scars
- Hutchinson teeth
- Saddle nose
- Saber shins
- MR
- CN8 deafness

What is the classic retinal finding in congenital lues?
*Salt-and-pepper retinitis*

What congenital condition is most strongly associated with ‘salt-and-pepper’ retinitis?
Other than IK, what are the stigmata of congenital syphilis?

- Circumoral scars
- Hutchinson teeth
- Saddle nose
- Saber shins
- MR
- CN8 deafness

What is the classic retinal finding in congenital lues?
Salt-and-pepper retinitis

What congenital condition is most strongly associated with ‘salt-and-pepper’ retinitis?
Rubella
Congenital rubella: Salt-and-pepper retinitis