Selected Systemic Diseases of Childhood with Corneal Findings

*Wilson's disease*

--Corneal finding:

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

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

**Systemic diseases of childhood with corneal findings**
Selected 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**
- It can also be **golden-brown**
- **Ruby red**

What layer of the cornea is it located in?
- **Descemet's**

Where does it start and how does it proceed?
- Starts superiorly, and proceeds inferiorly from there

Does all Wilson's disease patients manifest the ring?
- No, so its absence does not rule out the condition
Wilson's disease
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What is its course; ie, where does it start, and how does it proceed?
Wilson's disease
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*In which layer of the cornea is it located?*  
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**What is its course; ie, where does it start, and how does it proceed?**  
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I’ve heard of something called a Fleisher ring. Is that just shorthand for Kayser-Fleischer ring?

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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, an iron line on the cornea secondary to keratoconus.
Wilson's disease
--Corneal finding: Kayser-Fleischer ring

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

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

What is its course; i.e., where does it start, and how does it proceed?
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OK then, what is a Fleisher ring?
An iron line on the cornea secondary to keratoconus.
### 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?**

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Wilson’s disease
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Selected Systemic Diseases of Childhood with Corneal Findings

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

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

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

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

In what order does copper deposition occur in Wilson’s disease?
-- 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.

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-- Yes, in two ways:
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By what appearance-related moniker are copper-induced cataracts known?
‘Sunflower cataracts’

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

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’

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

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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 degeneration'
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

What are the CNS manifestations in Wilson’s?
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.
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--Systemic findings:

Is a Kayser-Fleischer ring pathognomonic for Wilson's disease?
Selected Systemic Diseases of Childhood with Corneal Findings

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

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

**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
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--Systemic findings:
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Is a Kayser-Fleischer ring pathognomonic for Wilson's disease?  
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Which other liver conditions are particularly likely to produce a K-F ring?

*other liver disorders*
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--Corneal finding: Kayser-Fleischer ring
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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.)
Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease
--Corneal finding: Kayser-Fleischer ring
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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.
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**Systemic Findings:**

- Liver and renal damage stigmata
- CNS signs/symptoms

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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?

- 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
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**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
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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:
--Serum free copper levels:
--Urinary copper levels:

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

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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’.
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

What is the treatment for Wilson's dz?

Penicillamine

Does the K-F ring regress with treatment?
Yes
Selected Systemic Diseases of Childhood with Corneal Findings

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--Corneal finding: Kayser-Fleischer ring
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--Corneal finding: Kayser-Fleischer ring
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**Riley-Day syndrome**
--Corneal finding:
Selected Systemic Diseases of Childhood with Corneal Findings

**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--Systemic findings:
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**Riley-Day syndrome**
--Corneal finding: keratitis/ulcers
Selected Systemic Diseases of Childhood with Corneal Findings

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  --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
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*

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

**Wilson's disease**
- Corneal finding: Kayser-Fleischer ring
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- Systemic findings:
  - Liver and renal damage stigmata
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**Riley-Day syndrome**
- Corneal finding: keratitis/ulcers
- aka... *familial dysautonomia*

Keratitis 2º to decreased corneal sensitivity is called...?

Decreased corneal sensitivity

(Riley-Day is one cause of congenital corneal anesthesia) and abnormal lacrimation
Wilson's disease
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Systemic diseases of childhood with corneal findings

What are the two problems underlying the propensity for keratitis and corneal ulcers that characterize Riley-Day?
Decreased corneal sensitivity
Keratitis 2° to decreased corneal sensitivity is called...
Neurotrophic keratitis

(Riley-Day is one cause of congenital corneal anesthesia) and abnormal lacrimation
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**Riley-Day syndrome**
--Corneal finding: keratitis/ulcers
--aka... *familial dysautonomia*
--Systemic findings:
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--Disease of *copper* metabolism
--Systemic findings:
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  --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**
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What ethnic group is most commonly affected in Riley-Day syndrome?

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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
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In severe cases, before age 1 year

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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)

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The rule is: The earlier the onset, the more severely affected is the pt
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Systemic, or topical?  
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--aka familial dysautonomia
--Systemic findings:
  --Temperature instability
  --Pain insensitivity
  --Labile BP
  --Excess sweating

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

What is the treatment for cystinosis?

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

Cysteamine smells terrible
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

**Systemic diseases**

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

**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: needed to treat systemic manifestations, but do nothing for the corneal crystals, which require **topical cysteamine**

- **Topical cysteamine** has a notorious downside that renders compliance difficult. What is it? Cysteamine smells terrible
Selected Systemic Diseases of Childhood with Corneal Findings

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--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

**Congenital syphilis**
--Corneal finding:
Selected Systemic Diseases of Childhood with Corneal Findings

**Wilson's disease**
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
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  --Liver and renal damage stigmata
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**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

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

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

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

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

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

--
--
Other than IK, what are the stigmata of congenital syphilis?

- Circumoral scars
- Hutchinson teeth
- Saddle nose
- Saber shins

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

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

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

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
- 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

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?

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
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