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?

Selected Systemic Diseases of Childhood with Corneal Findings

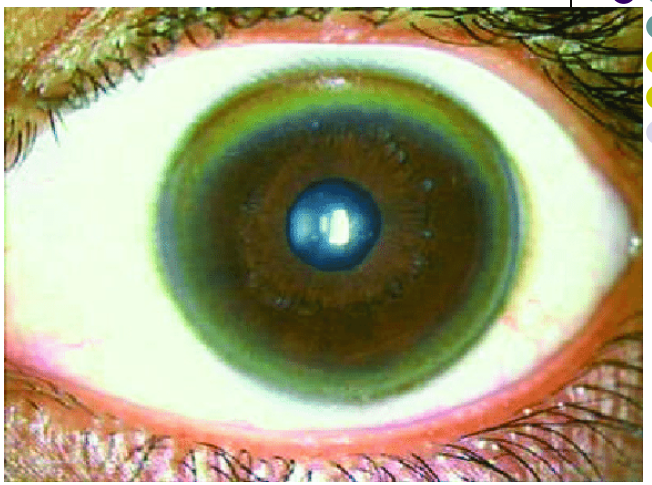
Wilson's disease

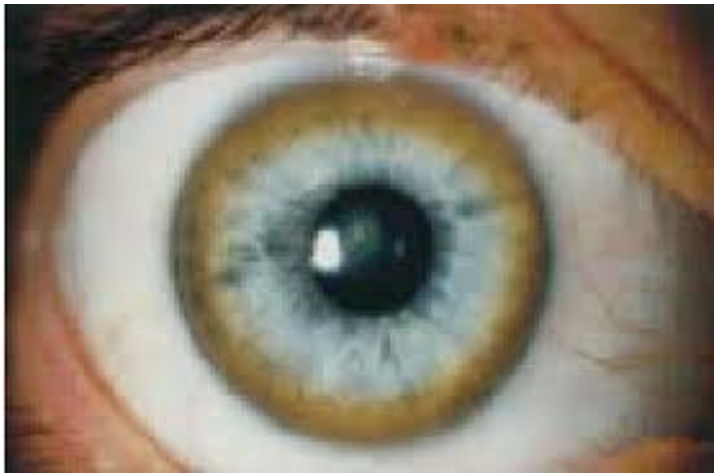
--Corneal finding: Kayser-Fleischer ring

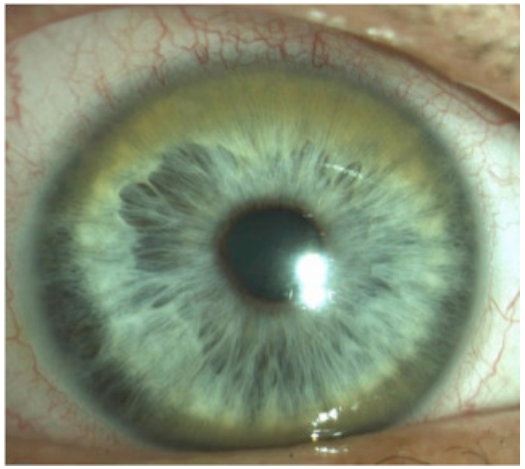
What color is the Kayser-Fleischer ring?

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

Selected Systemic Diseases of Childhood with Corneal Findings







Kayser-Fleischer ring

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** , but it can also be **golden-brown** or even **ruby red**

In which layer of the cornea is it located?

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** , but it can also be **golden-brown** or even **ruby red**

In which layer of the cornea is it located?

Descemet's

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** , 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?

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** , 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

Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease

--Corneal finding: Kayser-Fleischer ring

What color is the Kayser-Fleischer ring?
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It starts superiorly, and proceeds inferiorly from there

Do all Wilson's dz pts manifest the ring?

Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease

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

Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease

--Corneal finding: Kayser-Fleischer ring

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

-brown or even

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

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Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease

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OK then, what is a Fleischer ring?

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Wilson's disease

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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 Fleischer ring?
An ele-ment line on the cornea secondary to condition

-brown or even

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

Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease

--Corneal finding: Kayser-Fleischer ring

What color is the Kayser-Fleischer ring?

-brown or even

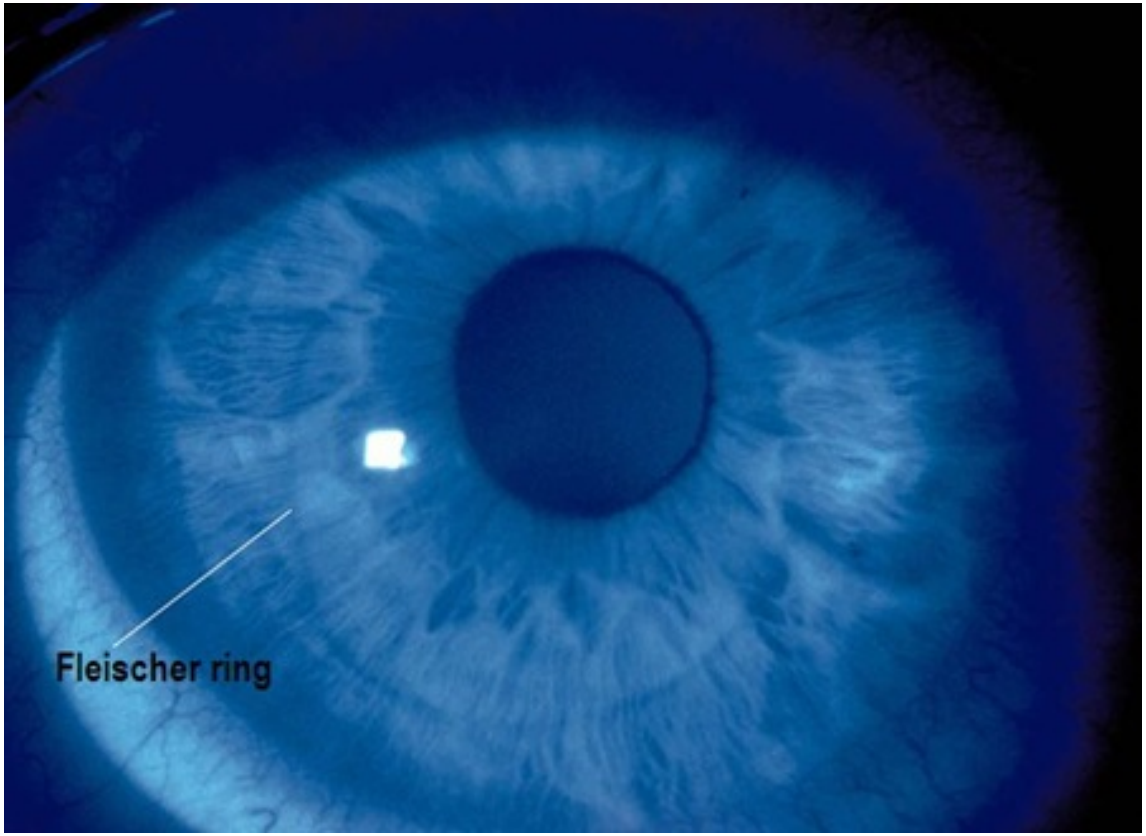
I've heard of something called a Fleisher ring*. Is that just shorthand for Kayser-Fleischer ring?*
No, a Fleischer ring is something completely different

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

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

Selected Systemic Diseases of Childhood with Corneal Findings



Fleischer ring in keratoconus

Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of element metabolism

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 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:
--
--

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

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

In Wilson's, copper deposition occurs in a particular order:

--First in the

--Next in the

--Later, in the

Syste
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

In Wilson's, copper deposition occurs in a particular order:

--First in the liver

--Next in the kidneys

--Later, in the CNS

Syste
of childhood with
corneal findings

Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease

--Corneal finding: Kayser-Fleischer ring

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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?

Syste
of childhood with
corneal findings

Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease

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--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

Syste
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:

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--CNS signs/symptoms

Does eye involvement extend beyond the K-F ring?

*At what point does the **eye become involved**?*

It really gets going in the CNS phase

Syste
of childhood with corneal findings

Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease
--Corneal finding: Kayser-Fleischer ring
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Does eye involvement extend beyond the K-F ring?
Yes, in two ways:
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At what point does the ***eye become involved****?*
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Syste
of childhood with corneal findings

Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease

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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

Syste
of childhood with corneal findings

Selected Systemic Diseases of Childhood with Corneal Findings

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

It really gets going in the CNS phase

Syste of childhood with corneal findings

Selected Systemic Diseases of Childhood with Corneal Findings

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--Disease of copper metabolism

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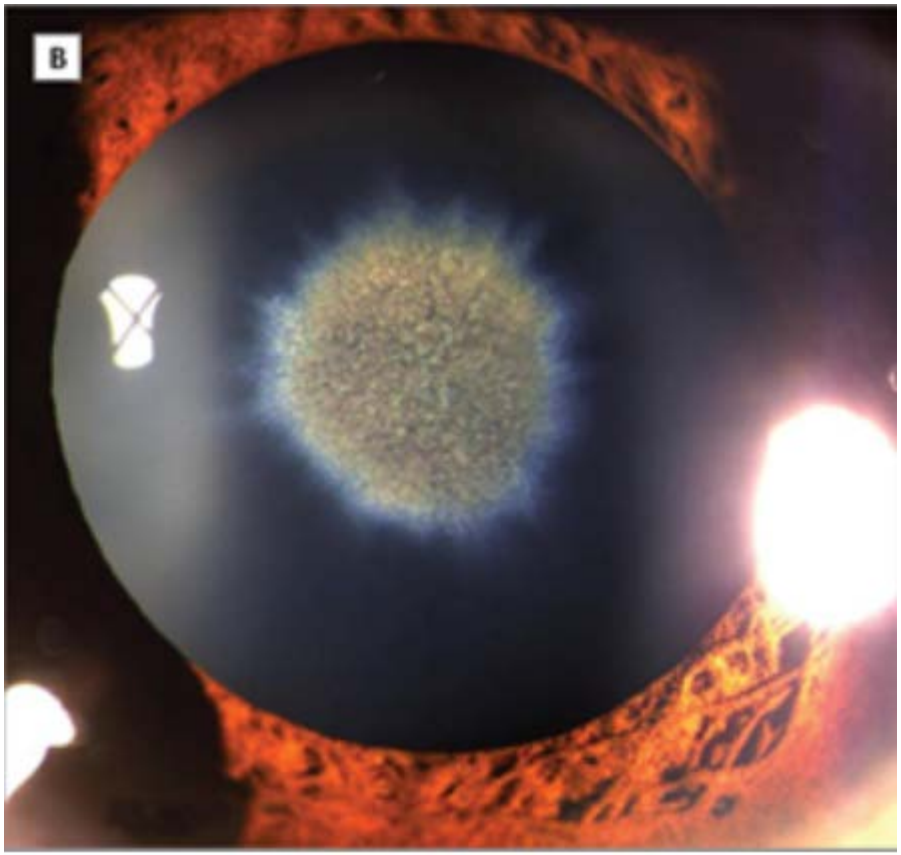
By what appearance-related moniker are copper-induced cataracts known?

'Sunflower cataracts'

It really gets going in the CNS phase

Syste
of childhood with corneal findings

Selected Systemic Diseases of Childhood with Corneal Findings



A sunflower for comparison

‘Sunflower cataract’ in Wilson’s dz

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

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It really gets going in the CNS phase

*BTW: What is the **non**eponymous name of Wilson's dz?*

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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*BTW: What is the **non**eponymous name of Wilson's dz?*

'Hepato-

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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--CNS signs/symptoms

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Yes, in two ways:

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'Sunflower cataracts'

It really gets going in the CNS phase

*BTW: What is the **non**eponymous name of Wilson's dz?*

'Hepato-lenticular

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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--CNS signs/symptoms

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Yes, in two ways:

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It really gets going in the CNS phase

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?

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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

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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corneal findings

Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease

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--Systemic findings:

--Liver and renal damage stigmata

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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.

ses
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corneal findings

Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease

--Corneal finding: Kayser-Fleischer ring

--Disease of copper metabolism

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

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

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

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

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:

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?

Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease

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--Disease of copper metabolism

--Systemic findings:

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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

--Disease of copper metabolism

--Systemic findings:

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? **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

Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease

--Corneal finding: Kayser-Fleischer ring

--Disease of copper metabolism

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?

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:

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

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

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?

corneal findings

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

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 [redacted] levels

--Serum [redacted] levels

--Urinary [redacted] levels

corneal findings

Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease

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--Disease of copper metabolism

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

corneal findings

Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease

--Corneal finding: Kayser-Fleischer ring

--Disease of copper metabolism

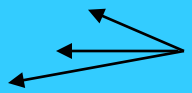
Clearly, the K-F ring is of limited utility in the diagnosis of Wilson's. So in addition to checking for K-F rings, what other steps should be taken to make the diagnosis?

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'

corneal findings

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

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'

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

What is the treatment for Wilson's dz?

ic diseases
dhood with
al findings

Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease
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Penicillamine

ic diseases
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Does the K-F ring regress with treatment?

ic diseases
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What is the treatment for Wilson's dz?
Penicillamine

Does the K-F ring regress with treatment?
Yes

ic diseases
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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

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

Systemic diseases of childhood with corneal findings

Riley-Day syndrome
--Corneal finding: keratitis/ulcers

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

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

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

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
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--Systemic findings:
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--CNS signs/symptoms

Systemic diseases of childhood with corneal findings

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?

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

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
--Disease of copper metabolism
--Systemic findings:
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Systemic diseases of childhood with corneal findings

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

Keratitis 2º to decreased corneal sensitivity is called…?

…erlying the propensity for keratitis and corneal …ay?

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
--CNS signs/symptoms

Systemic diseases of childhood with corneal findings

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

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

erlying the propensity for keratitis and corneal
ay?
Decreased corneal sensitivity (Riley-Day is one cause of *congenital corneal anesthesia*) and abnormal lacrimation

Selected Systemic Diseases of Childhood with Corneal Findings

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Riley-Day syndrome
--Corneal finding: keratitis/ulcers
--Hypotonia

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

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Systemic diseases of childhood with corneal findings

Riley-Day syndrome
...tis/ulcers
...mia

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

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

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Why should this matter to an American ophthalmologist?

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Many American Jews are Ashkenazi

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--Corneal finding: keratitis/ulcers
--Labile BP
--Excessive sweating

How is Riley-Day managed?

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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.

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Cystinosis
--Corneal finding: *?*

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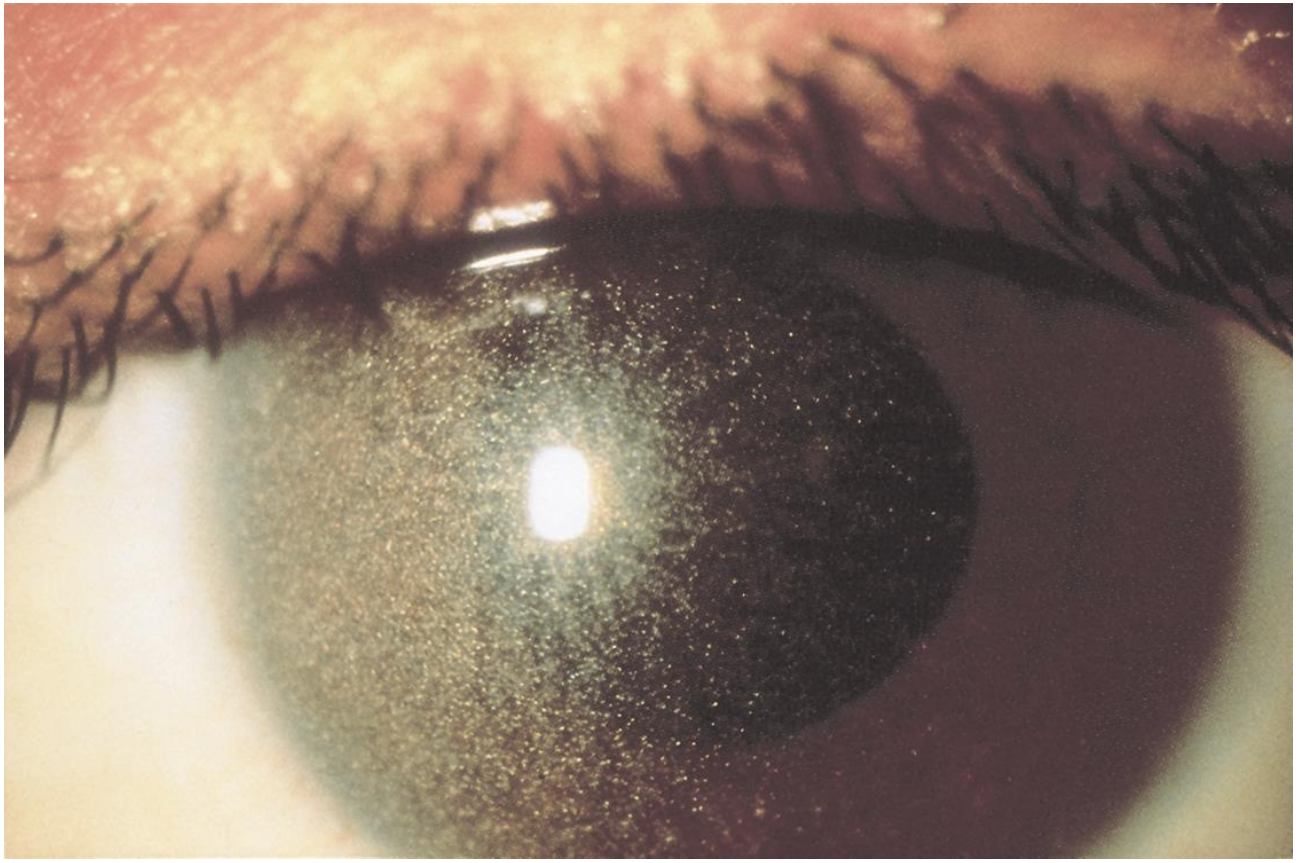

Corneal crystals in cystinosis

Selected Systemic Diseases of Childhood with Corneal Findings

Wilson's disease
--Corneal finding: Kayser-Fleischer ring
--Disease of copper metabolism
--System
--Liver
--CNS

At what age do the crystals appear?

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

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Are the corneal crystal clinically significant?

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The produce disabling photophobia

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Take note of this, as it makes for a great factoid upon which to base a test question!

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Can the photophobia be so severe as to warrant PK?

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In one word, what sort of condition is cystinosis?
A [redacted] disorder

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Metabolism of what substrate is involved?

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Renal damage in cystinosis results in what eponymous syndrome?

eponym

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--Corneal finding: Irid

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No, that's Fanconi's ***anemia***, an inherited form of bone-marrow failure

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Cystinosis
--Corneal finding: Iric
--Systemic findings:
--Short stature
--Rickets
--ESRD

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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

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What is rickets*?*

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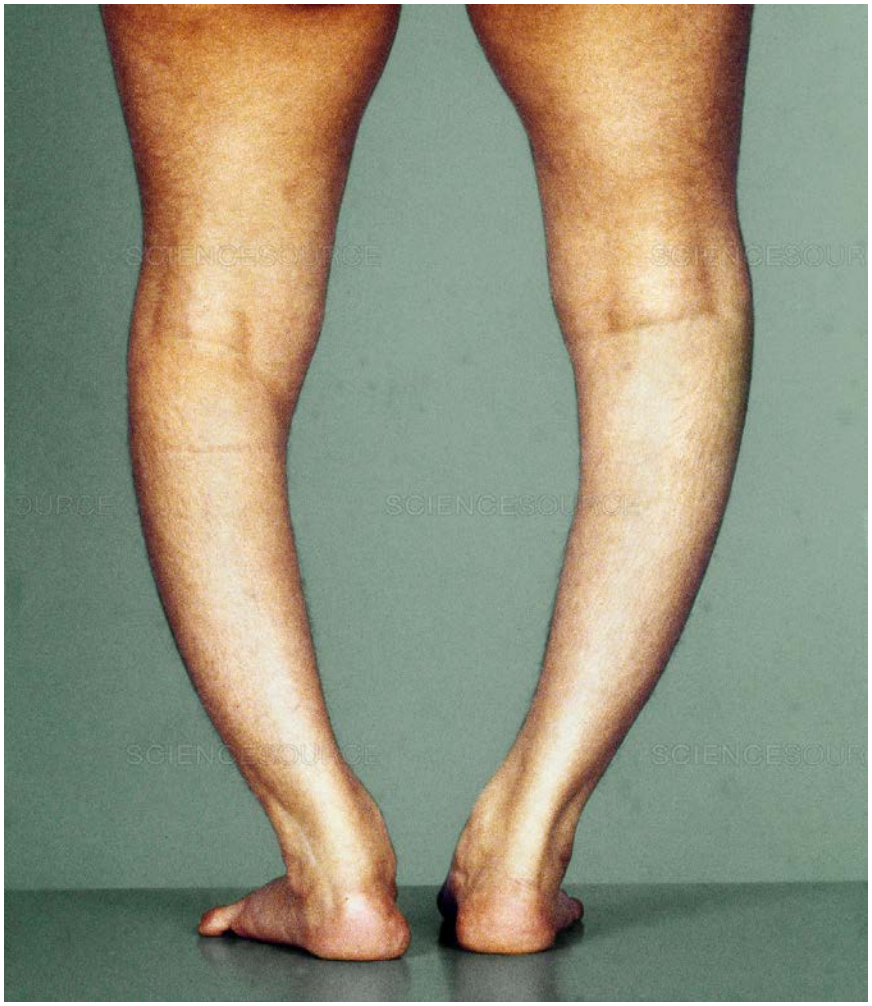
What is rickets*?*
A condition in which inadequate mineralization of long-bone growth plates leads to disruption in plate function, resulting in impaired bone growth as well as the characteristic bone 'bowing'

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

--Excess sweating

Systemic drugs and ocular toxicity: Matching



Rickets

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?

Which organ is most profoun
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

Cystinosis
--Corneal finding: Iric
--Systemic findings:
--Short stature
--Rickets
--ESRD

--Excess sweating

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Selected Systemic Diseases of Childhood with Corneal Findings

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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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Systemic diseases of childhood with corneal findings

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?

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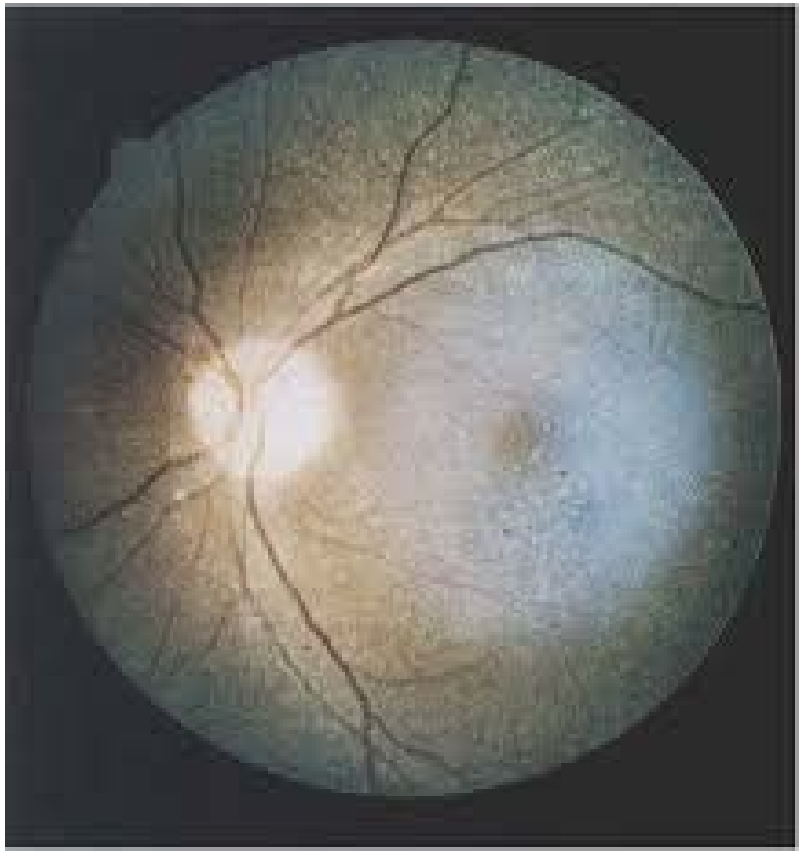

Crystalline retinopathy in cystinosis

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Systemic diseases of childh... corneal...

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

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Systemic diseases of childh... corneal

Systemic dz

Drug-induced

Ocular dz

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Drug-induced

Systemic diseases of childh... corneal...

Systemic dz
└ Cystinosis

Ocular dz

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

Cystinosis
--Corneal finding: Iridescent crystals

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--aka: *familial dysautonomia*
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Systemic diseases of childh... corneal...

Drug-induced
└ ?
└ ?
└ ?

Systemic dz
└ Cystinosis

Ocular dz

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

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Systemic diseases of childh... corneal...

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- Tamoxifen
- Canthaxanthine
- Talc

Systemic dz
- Cystinosis

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Systemic diseases of childh... corneal...

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Systemic dz
- Cystinosis

Ocular dz
- *?*

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

Cystinos...
--Cornea...

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Systemic diseases of childh... corneal...

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Systemic dz
- Cystinosis

Ocular dz
- **Bietti crystalline dystrophy**

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Systemic diseases of childhood with

Cystinosis comes in three forms--what are they?

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

Note that **all three types** *manifest corneal crystals!*

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

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--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)

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Systemic diseases

What is the treatment for cystinosis?

Cystinosis
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--ESRD
--Treatment: ***?***

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Systemic, or topical?

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Systemic diseases

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:
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--Treatment: Topical cysteamine

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Systemic diseases

What is the treatment for cystinosis?
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Topical cysteamine has a notorious downside that renders compliance difficult. What is it?

S
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Cysteamine smells **terrible**

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--CNS signs/symptoms

Congenital syphilis
--Corneal finding: *?*

Systemic diseases of childhood with corneal findings

Cystinosis
--Corneal finding: Iridescent crystals
--Systemic findings:
--Short stature
--Rickets
--ESRD
--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:
--Liver and renal damage stigmata
--CNS signs/symptoms

Congenital syphilis
--Corneal finding: Interstitial keratitis

Systemic diseases of childhood with corneal findings

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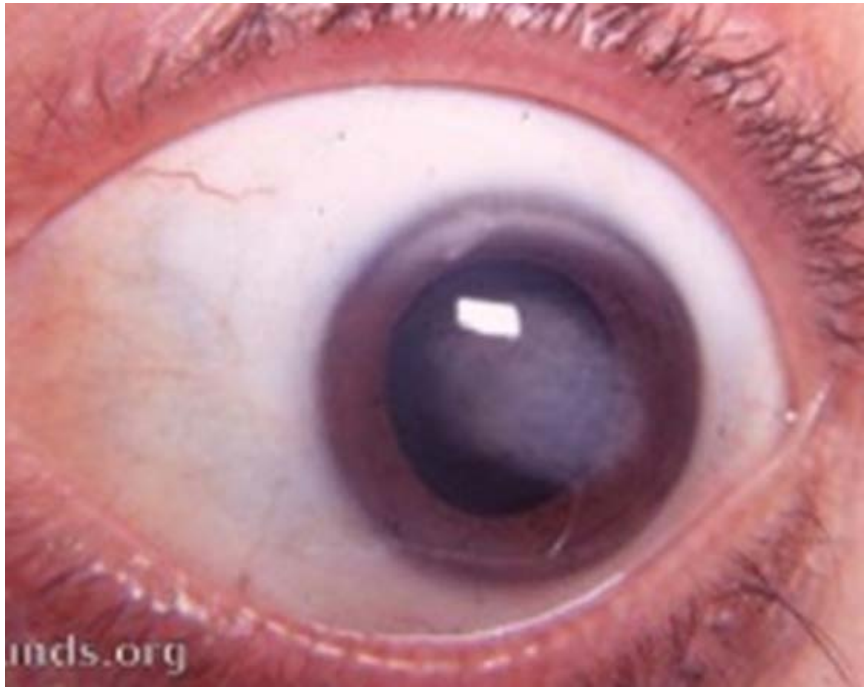
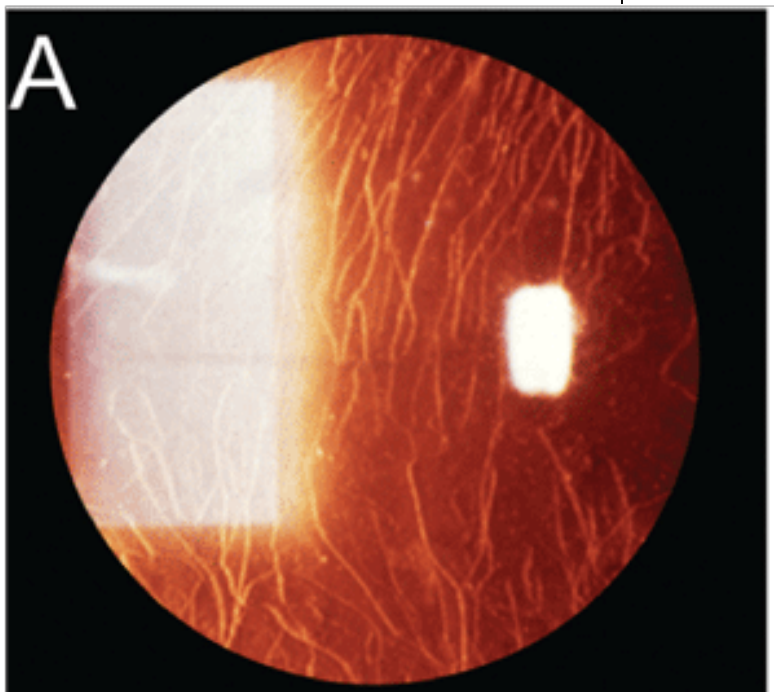
Selected Systemic Diseases of Childhood with Corneal Findings



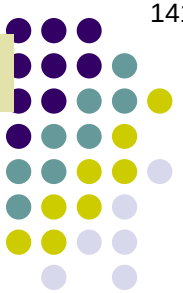
Corneal scar/haze



Ghost vessels

Congenital syphilis: Interstitial keratitis

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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--CNS signs/symptoms

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

Systemic diseases of childhood with corneal findings

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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**

What are the 'usual stigmata' of congenital syphilis?

Systemic diseases of childhood with corneal findings

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

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

Q

Selected Systemic Diseases of Childhood with Corneal Findings

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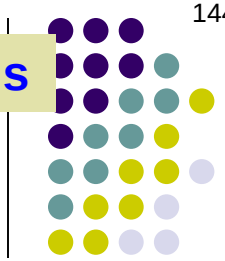
  -
  -

A

Selected Systemic Diseases of Childhood with Corneal Findings



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

Q

Selected Systemic Diseases of Childhood with Corneal Findings

- 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?

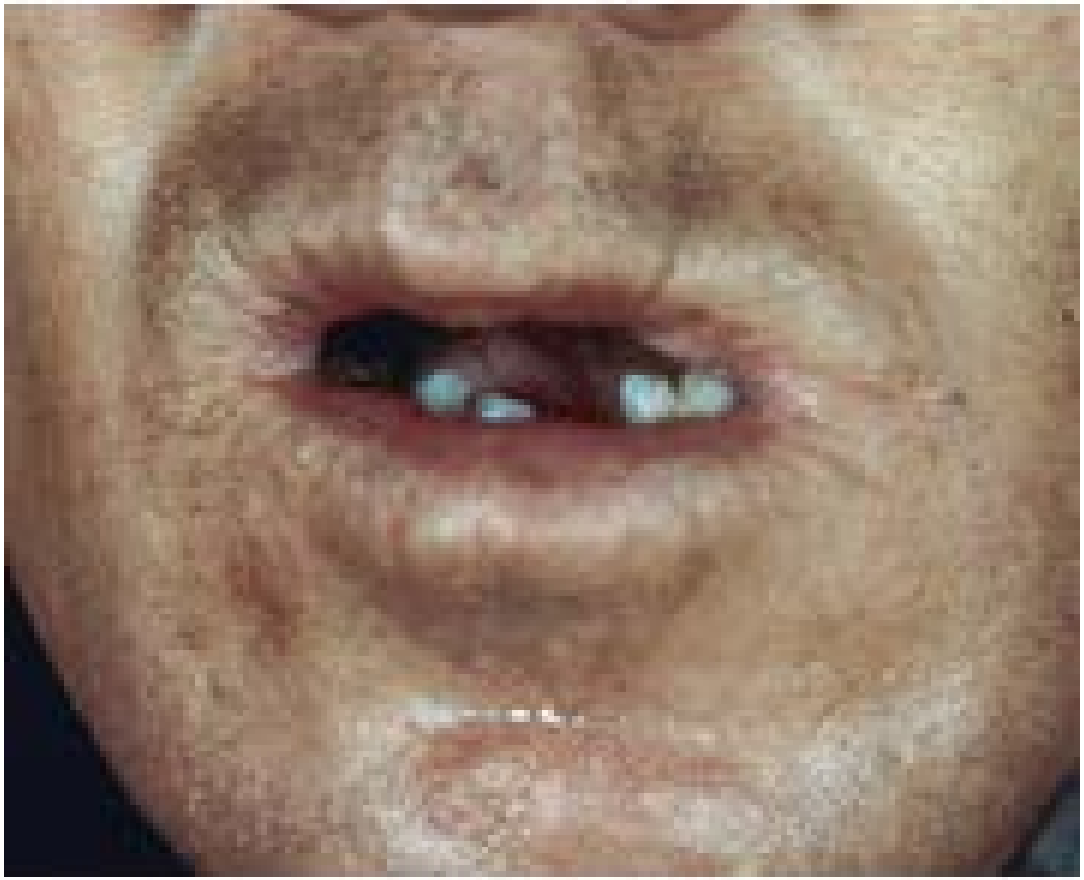
A

Selected Systemic Diseases of Childhood with Corneal Findings

- 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'

Selected Systemic Diseases of Childhood with Corneal Findings



Congenital syphilis: Circumoral scars

Q

Selected Systemic Diseases of Childhood with Corneal Findings

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

What is the classic description of Hutchinson teeth*?*

A

Selected Systemic Diseases of Childhood with Corneal Findings

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  - **Hutchinson teeth**
  - Saddle nose
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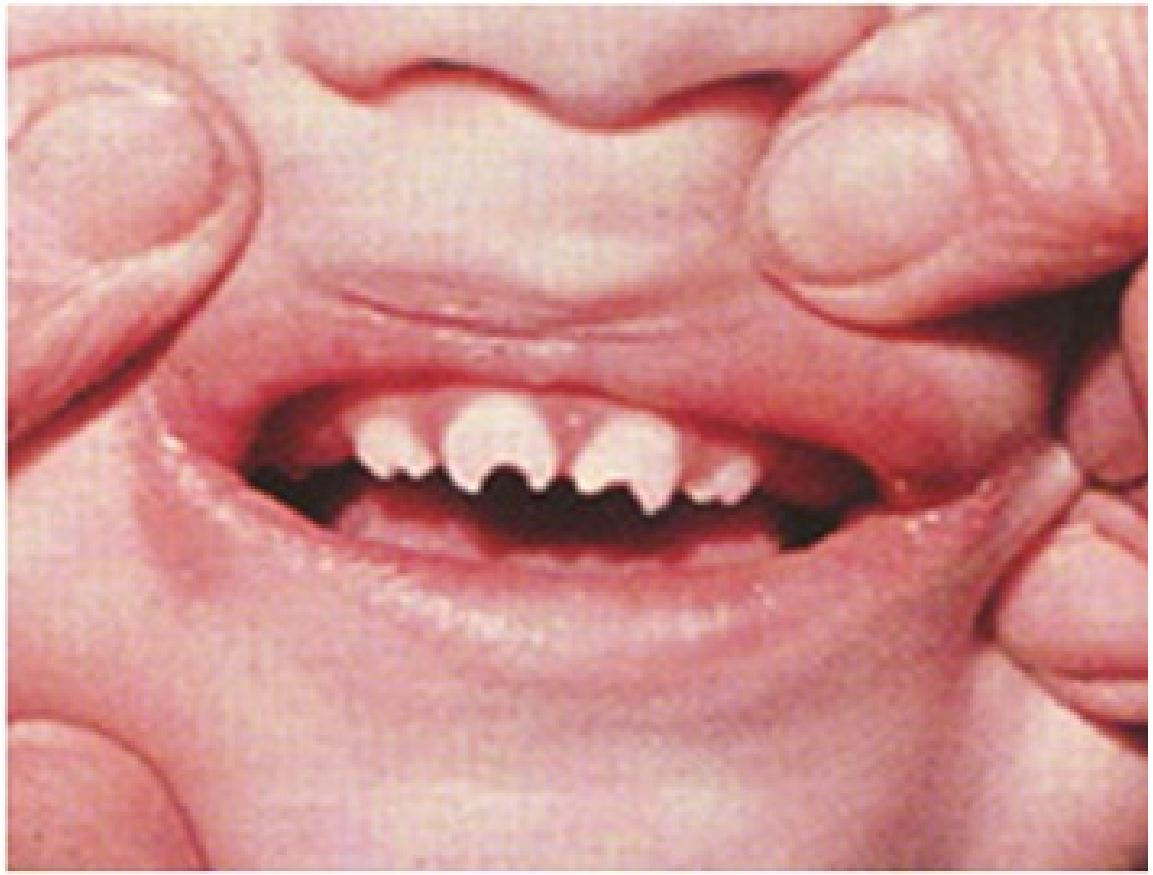
What is the classic description of Hutchinson teeth*?*
'Peg-shaped'

Selected Systemic Diseases of Childhood with Corneal Findings



Congenital syphilis: Teephus

Q

Selected Systemic Diseases of Childhood with Corneal Findings

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  - Circumoral scars
  - Hutchinson teeth
  - Saddle nose
  - Sa
  - M
  - CN8 deafness

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

--

--

A

Selected Systemic Diseases of Childhood with Corneal Findings

- Other than IK, what are the stigmata of congenital syphilis?
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  - Hutchinson teeth
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  - M
  - 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**

Q

Selected Systemic Diseases of Childhood with Corneal Findings

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  - (

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--**Axenfeld-Rieger syndrome**

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

A

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

Q

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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…

A

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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

Q

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  - Saddle nose
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In one word, what sort of condition is incontinentia pigmenti?
A…

A

Selected Systemic Diseases of Childhood with Corneal Findings

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

Q

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In the present context, to what does the term Hutchinson's triad *refer?*

A

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In the present context, to what does the term Hutchinson's triad *refer?*
To the three stigmata of congenital syphilis that are especially common

Q

Selected Systemic Diseases of Childhood with Corneal Findings

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Which three comprise Hutchinson's triad?
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  - **CN8 deafness**

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

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To **the three stigmata of congenital syphilis that are especially common**

Q

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What is the classic retinal *finding in congenital lues?*

A

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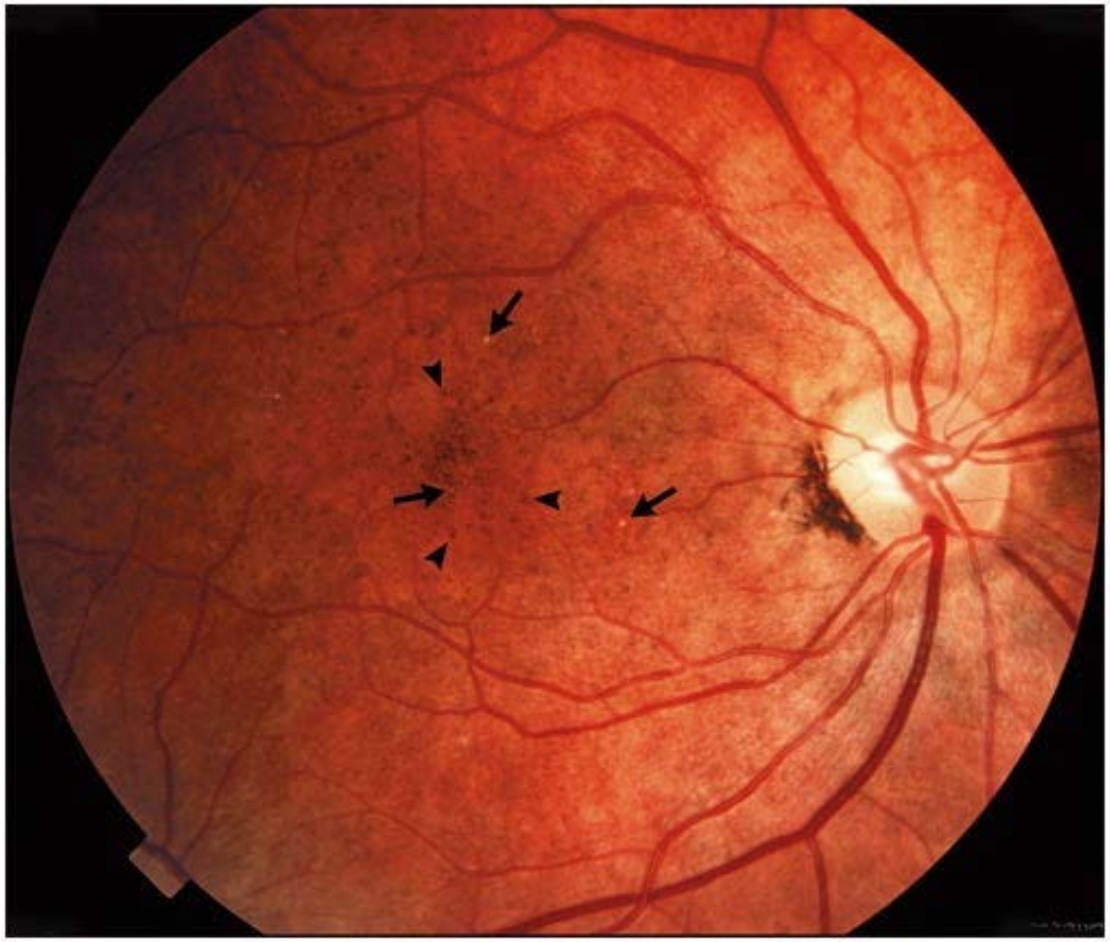
What is the classic retinal *finding in congenital lues?*
***Salt-and-pepper* retinitis**

Selected Systemic Diseases of Childhood with Corneal Findings



Congenital syphilis: Salt-and-pepper retinitis

Q

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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?

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Rubella

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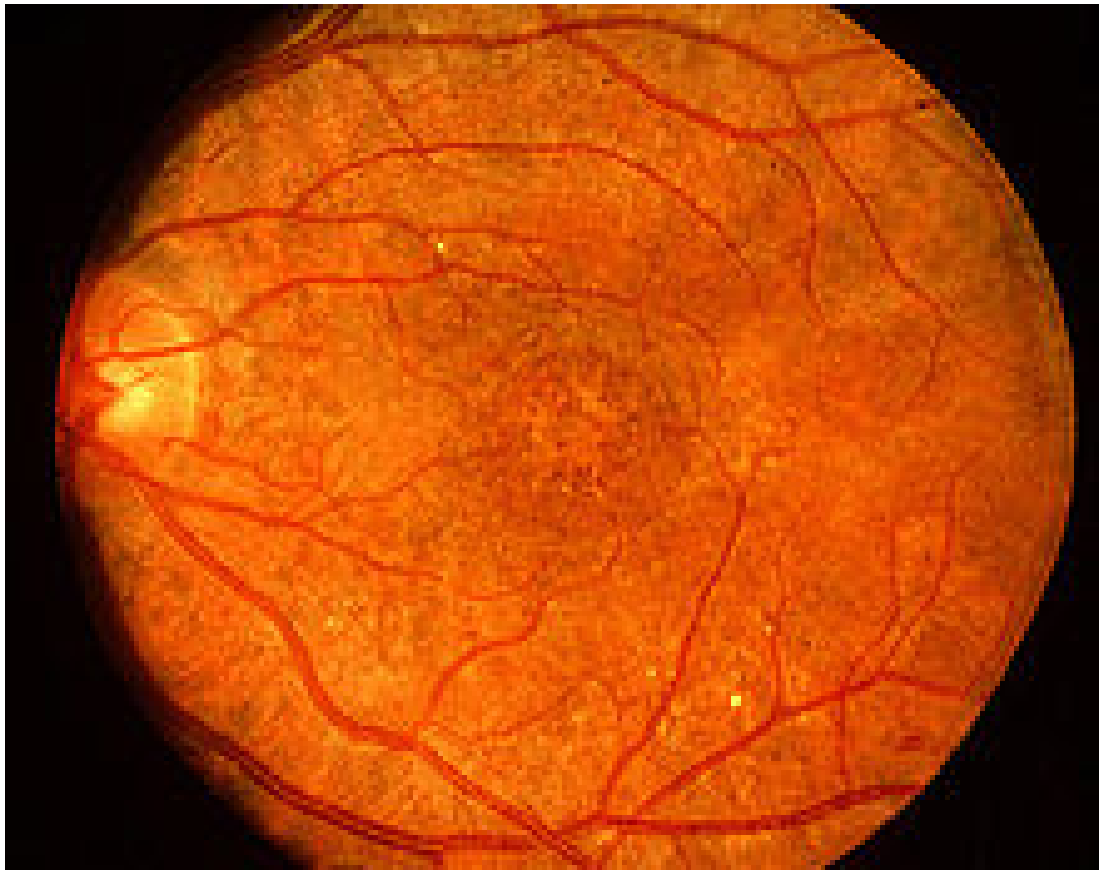

Congenital **rubella**: Salt-and-pepper retinitis