Hereditary Vitreoretinopathies

specific disease

specific disease

specific disease

specific disease

group of diseases

specific disease

Hereditary Vitreoretinopathies

Goldmann-Favre Syndrome

Norrie Disease

X-Linked Juvenile Retinoschisis

Familial Exudative Vitreoretinopathy

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

Hereditary Vitreoretinopathies

X-Linked Juvenile Retinoschisis

--Named for peripheral changes, but present in only % of cases

Goldmann-Favre Syndrome

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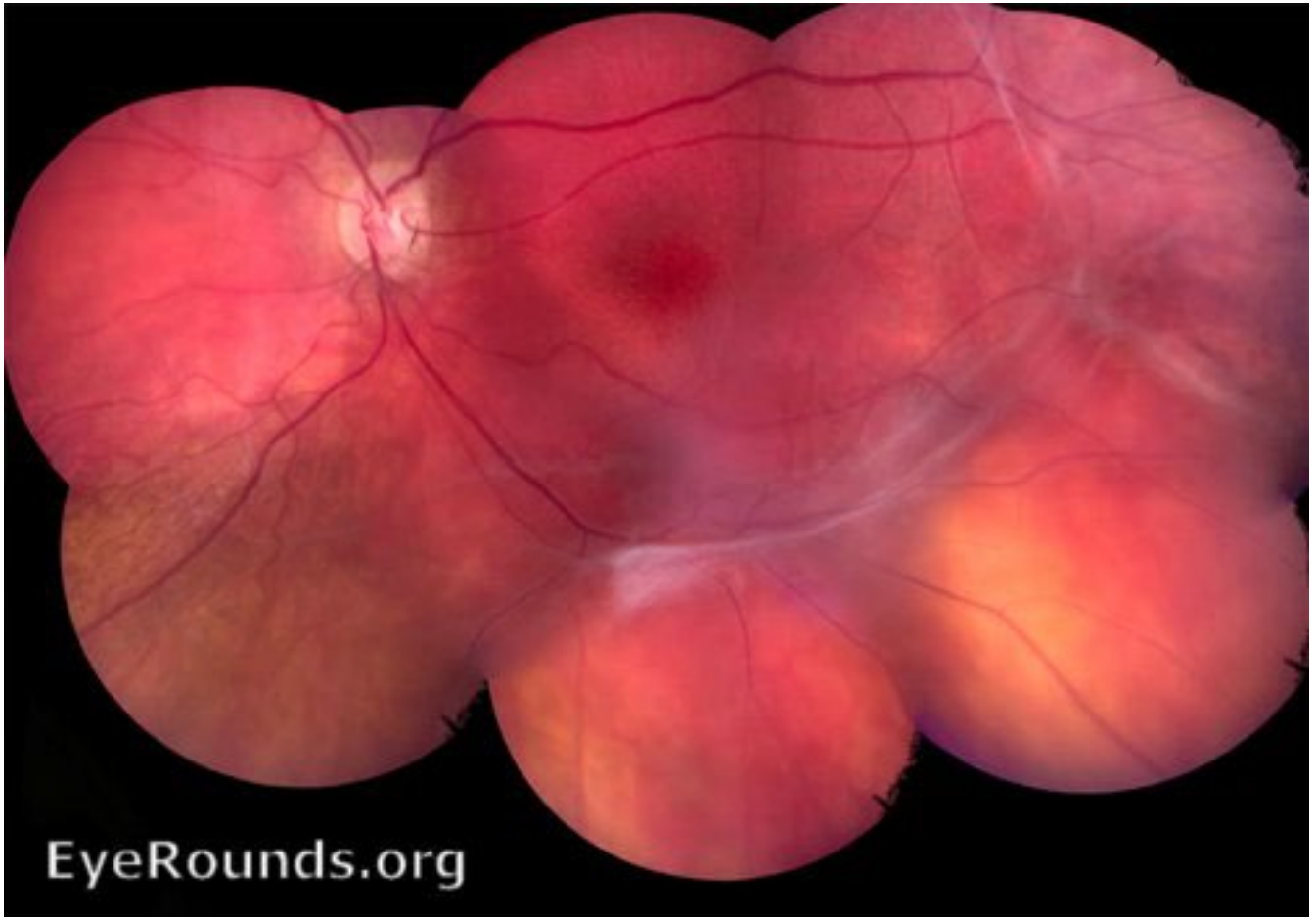

X-linked juvenile retinoschisis: Peripheral retinoschisis

Hereditary Vitreoretinopathies

X-Linked Juvenile Retinoschisis

--Named for peripheral changes, but present in only 50% of cases
--Foveal schisis present in…[%]

Goldmann-Favre Syndrome

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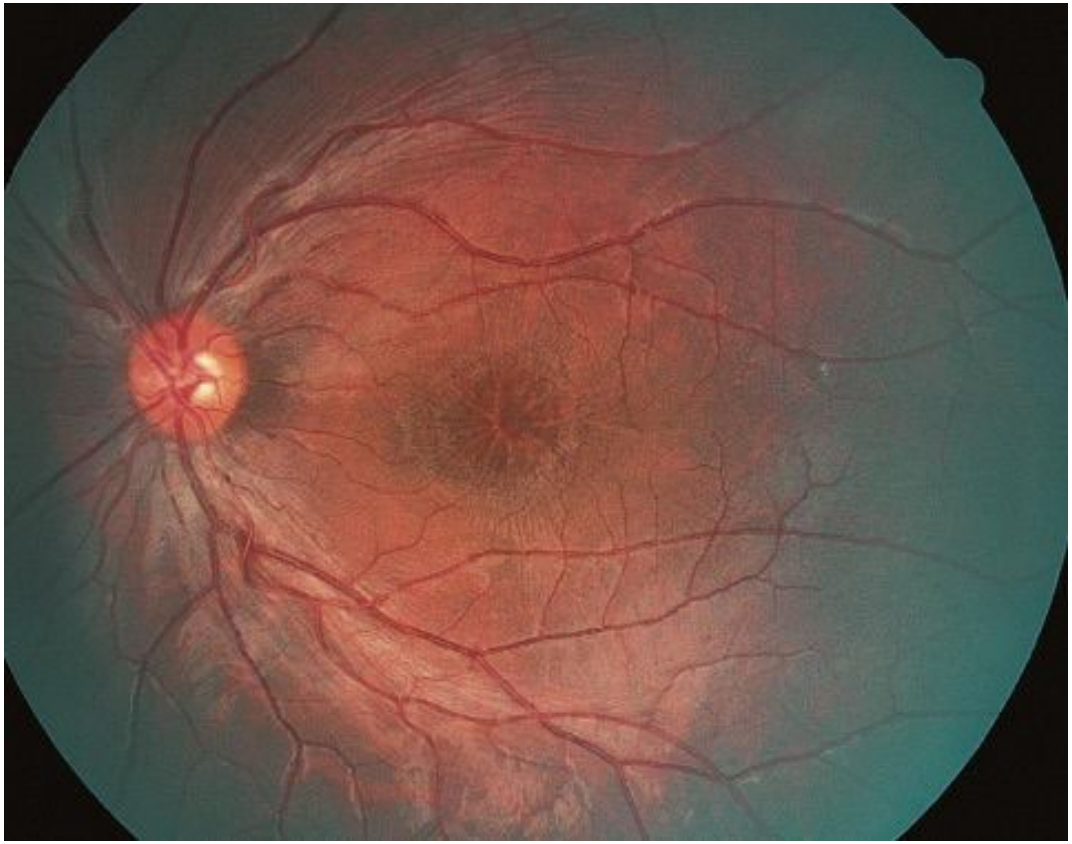
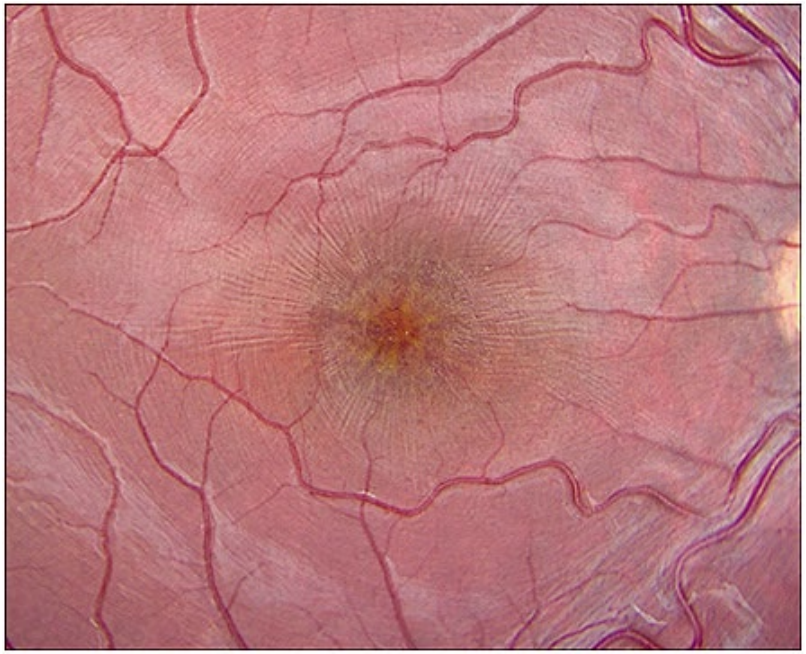
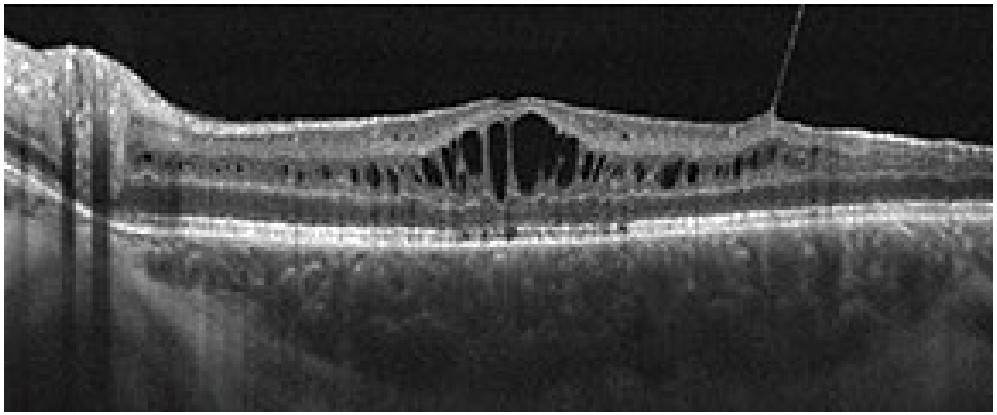


X-linked juvenile retinoschisis: Foveal cysts

Hereditary Vitreoretinopathies





X-linked juvenile retinoschisis: Foveal cysts

Hereditary Vitreoretinopathies

X-Linked Juvenile Retinoschisis

--Named for peripheral changes, but present in only 50% of cases
--Foveal schisis present in…100%
--Looks like CME, but is **dry** dz

(CME = cystoid macular edema)

Goldmann-Favre Syndrome

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--Looks like CME, but is **dry** dz
--Split is in…[*retinal layer*]

Goldmann-Favre Syndrome

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--Foveal schisis present in…100%
--Looks like CME, but is **dry** dz
--Split is in…RNFL

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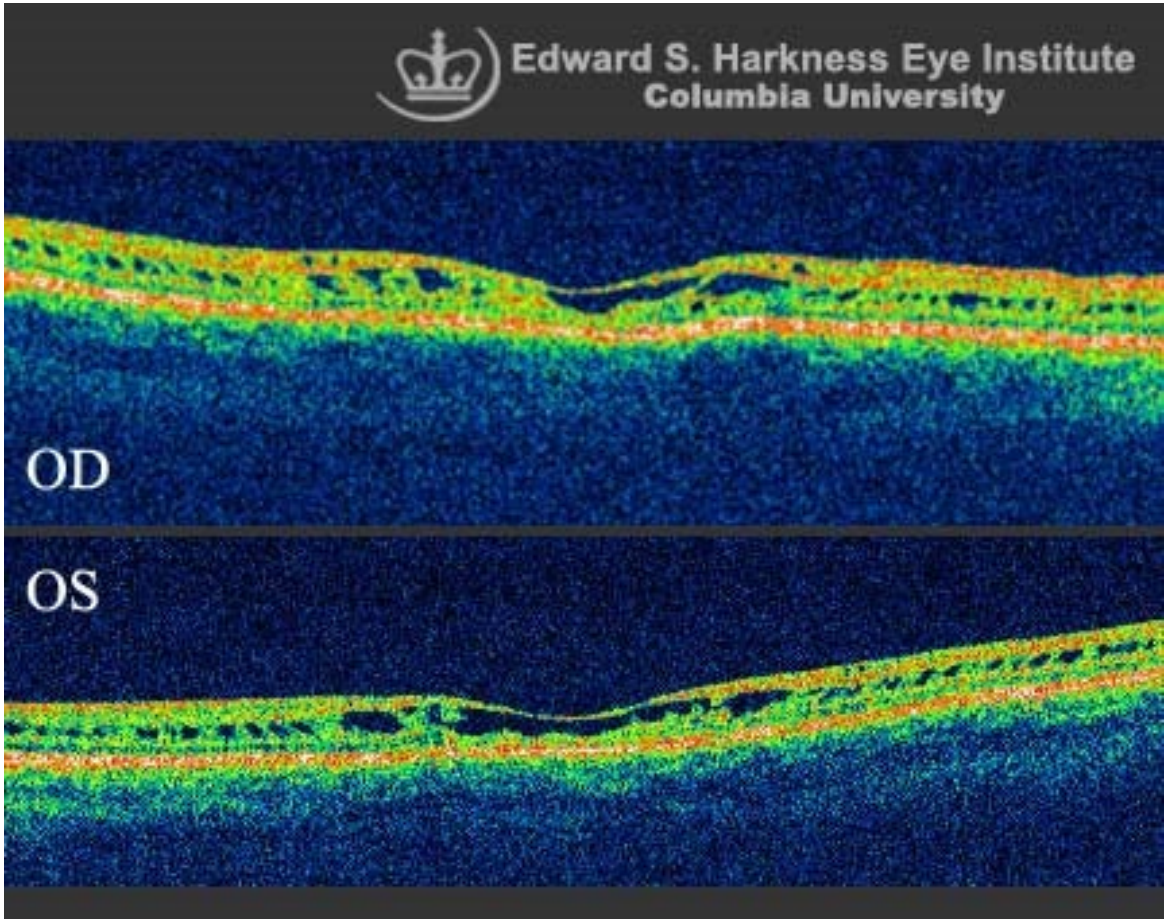

X-linked juvenile retinoschisis: RNFL schisis

Hereditary Vitreoretinopathies

Goldmann-Favre Syndrome

Norrie Disease

X-Linked Juvenile Retinoschisis

--Named for peripheral changes, but present in only 50% of cases
--Foveal schisis present
--Looks like CME, but is
--Split is in…RNFL

My mnemonic for recalling this fact is to remember that young (ie, 'juvenile') men play in the NFL.
On the other hand, the schisis commonly seen in the elderly occurs in the outer plexiform and/or outer nuclear layers of the retina.

kudative opathy

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

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--Named for peripheral changes, but present in only 50% of cases
--Foveal schisis present in…100%
--Looks like CME, but is **dry** dz
--Split is in…RNFL
--Abnormal cells:

Goldmann-Favre Syndrome

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--Split is in…RNFL
--Abnormal cells: Müller cells

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--Split is in…RNFL
--Abnormal cells: Müller cells
--ERG: Diminution of the…[a-*wave* *vs* b-*wave* *vs both]*

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What is the term for an ERG in which the a-wave is preserved but the b-wave is diminished?

--Abnormal cells: Müller cells
--**ERG: Diminution of the…*b*-wave**

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What is the term for an ERG in which the a-wave is preserved but the b-wave is diminished?
Such an ERG is said to demonstrate a 'negative' or 'electronegative' waveform

--Abnormal cells: Müller cells
--**ERG: Diminution of the…*b*-wave**

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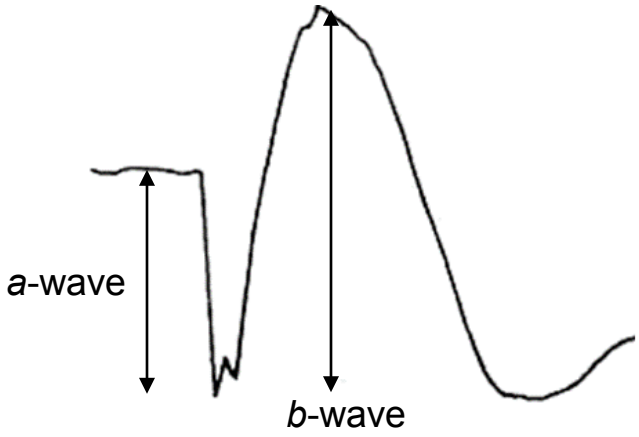
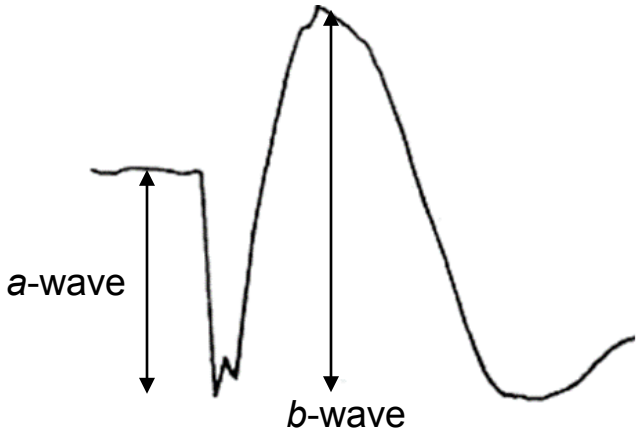
Normal ERG



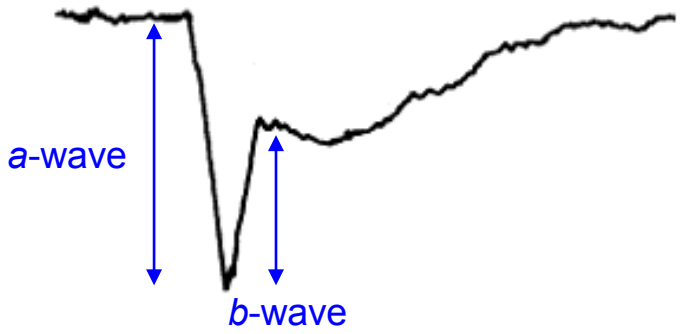
In a normal full-field ERG, the *b*-wave is substantially larger than the *a*-wave

Hereditary Vitreoretinopathies

Normal ERG



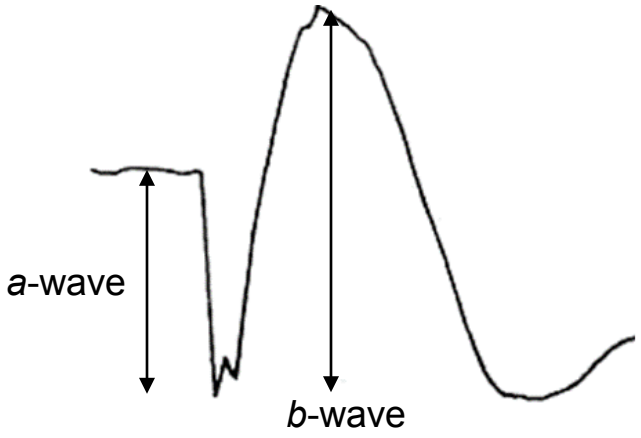
ERG in XLJR



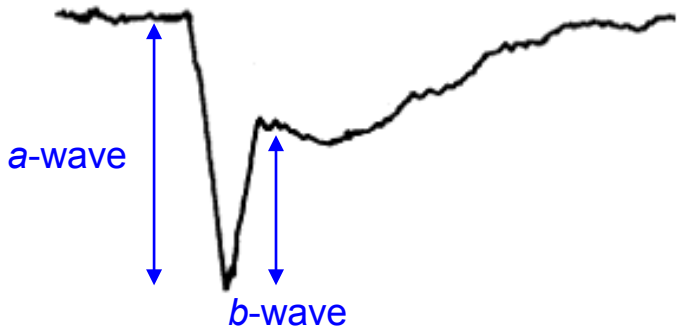
However, in XLJR the *b*-wave is substantially **smaller** than the *a*-wave, which is preserved in size

Hereditary Vitreoretinopathies

Normal ERG



ERG in XLJR



However, in XLJR the *b*-wave is substantially **smaller** than the *a*-wave, which is preserved in size

This constitutes a 'negative' or ('electronegative') ERG

Hereditary Vitreoretinopathies

Goldmann-Favre Syndrome

Norrie Disease

X-Linked Juvenile Retinoschisis

Is a negative ERG pathognomonic for XLJR?

Such an ERG is said to demonstrate a
'negative' or 'electronegative' waveform

--Abnormal cells: Müller cells
--**ERG: Diminution of the…*b*-wave**

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Is a negative ERG pathognomonic for XLJR?
No, several conditions can cause it

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Is a negative ERG pathognomonic for XLJR?
No, several conditions can cause it. That said, the only other one likely to present in *childhood* is abb,

Such an ERG is said to demonstrate a
'negative' or 'electronegative' waveform

--Abnormal cells: Müller cells
--**ERG: Diminution of the…*b*-wave**

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Is a negative ERG pathognomonic for XLJR?
No, several conditions can cause it. That said, the only other one likely to present in *childhood* is CSNB.

Such an ERG is said to demonstrate a
'negative' or 'electronegative' waveform

--Abnormal cells: Müller cells
--**ERG: Diminution of the…*b*-wave**

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Norrie Disease

What does CSNB *stand for in this context?*

X-Linked … Retinosch…

Is a negative ER…
No, several conditions can cause it. That said, the only other one likely to present in *childhood* is **CSNB**.

Such an ERG is said to demonstrate a
'negative' or 'electronegative' waveform

--Abnormal cells: Müller cells
--**ERG: Diminution of the…*b*-wave**

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Norrie Disease

What does CSNB *stand for in this context?*
Congenital stationary night blindness

X-Linked … Retinosch…

Is a negative ER…
No, several conditions can cause it. That said, the only other one likely to present in *childhood* is **CSNB**.

Such an ERG is said to demonstrate a
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Familial Exudative Vitreoretinopathy

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Knobloch Syndrome

Hereditary Vitreoretinopathies

Goldmann-Favre Syndrome

Norrie Disease

What does CSNB *stand for in this context?*
Congenital stationary night blindness

In a nutshell, what's CSNB?

X-Linked J… Retinosch…

Is a negative ER…
No, several conditions can cause it. That said, the only other one likely to present in *childhood* is **CSNB**.

Such an ERG is said to demonstrate a
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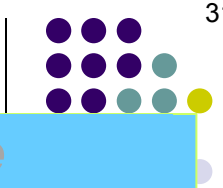
Hereditary Vitreoretinopathies



Goldmann-Favre Syndrome

Norrie Disease

What does CSNB *stand for in this context?*
Congenital stationary night blindness

In a nutshell, what's CSNB?
A congenital condition in which a dearth of functioning [rods vs cones] leads to [ ], [ ], and variably decreased [abb.]

X-Linked J… Retinosch…

Is a negative ER…
No, several conditions can cause it. That said, the only other one likely to present in *childhood* is **CSNB**.

Such an ERG is said to demonstrate a
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Goldmann-Favre Syndrome

Norrie Disease

X-Linked J… Retinosch…

What does CSNB *stand for in this context?*
Congenital stationary night blindness

In a nutshell, what's CSNB?
A congenital condition in which a dearth of functioning rods leads to nyctalopia , nystagmus , and variably decreased VA

Is a negative ER…
No, several conditions can cause it. That said, the only other one likely to present in *childhood* is **CSNB**.

Such an ERG is said to demonstrate a
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Hereditary Vitreoretinopathies

X-Linked Juvenile Retinoschisis

--Named for peripheral changes, but present in only 50% of cases
--Foveal schisis present in…100%
--Looks like CME, but is **dry** dz
--Split is in…RNFL
--Abnormal cells: Müller cells
--ERG: Diminution of the…*b*-wave
--If severe, peripheral changes can lead to…[*two problems*]

Next question

Goldmann-Favre Syndrome

Norrie Disease

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--If severe, peripheral changes can lead to…RD, vitreous heme

Goldmann-Favre Syndrome

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Goldmann-Favre Syndrome

--Macula looks like.. one dz
but periphery looks like.. diff dz

Norrie Disease

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Goldmann-Favre Syndrome

--Macula looks like…XLJR,
but periphery looks like…RP *(RP = retinitis pigmentosa)*

Norrie Disease

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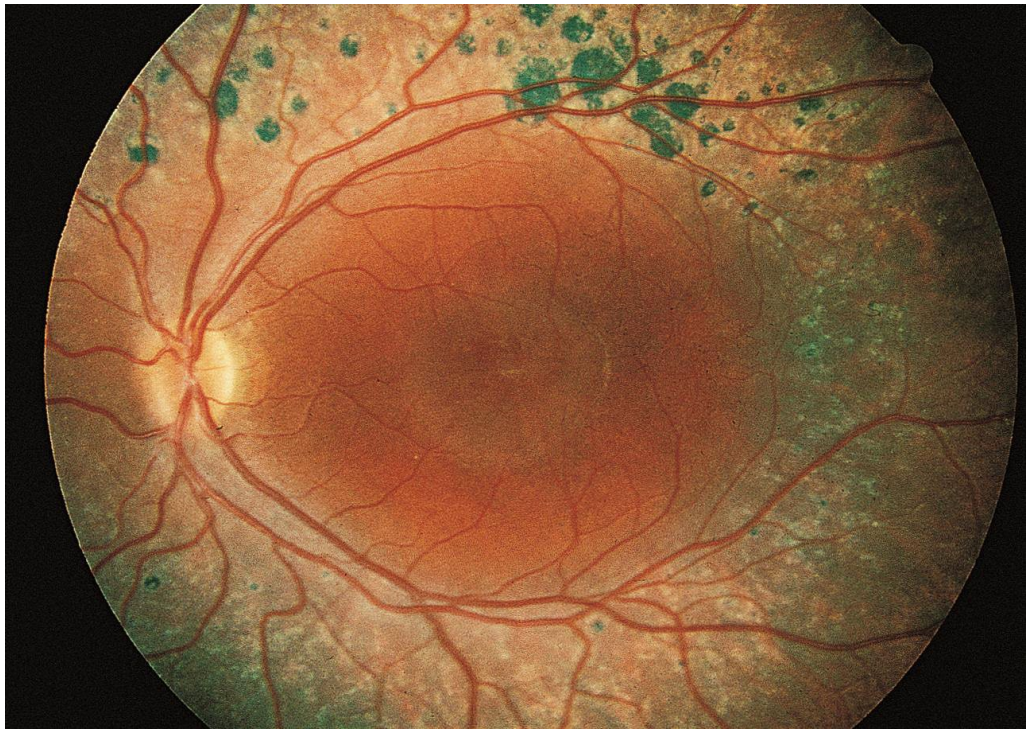

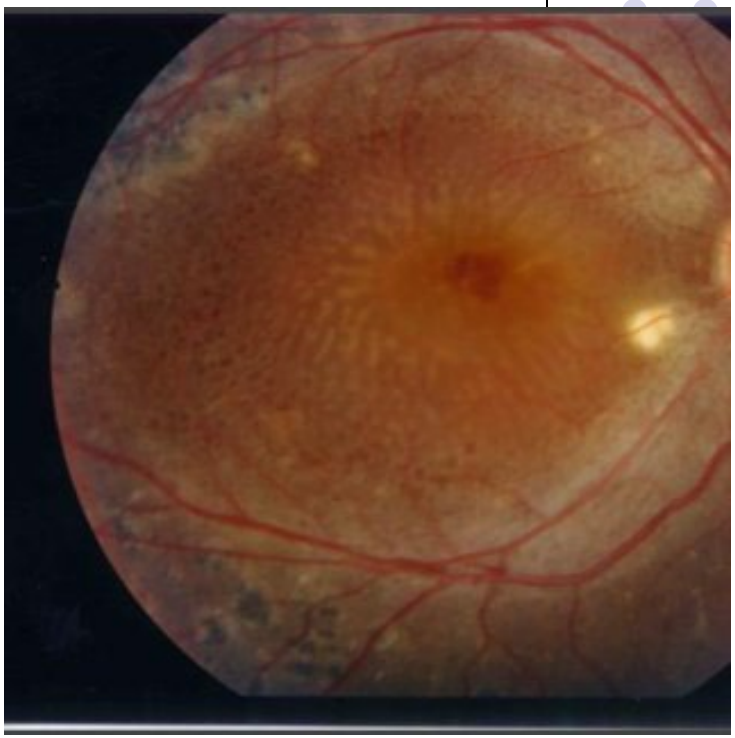

Goldmann-Favre syndrome

Hereditary Vitreoretinopathies

X-Linked Juvenile Retinoschisis

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--Split is in…RNFL
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Goldmann-Favre Syndrome

--Macula looks like…XLJR, but periphery looks like…RP
--Vitreous is… two words

Norrie Disease

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Goldmann-Favre Syndrome

--Macula looks like…XLJR, but periphery looks like…RP
--Vitreous is…optically empty

Norrie Disease

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--If severe, peripheral changes can lead to…RD, vitreous heme

Goldmann-Favre Syndrome

--Macula looks like…XLJR, but periphery looks like…RP
--Vitreous is…optically empty
Presents with… one word

Norrie Disease

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Goldmann-Favre Syndrome

--Macula looks like…XLJR, but periphery looks like…RP
--Vitreous is…optically empty
Presents with…nyctalopia

Norrie Disease

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Goldmann-Favre Syndrome

--Macula looks like…XLJR, but periphery looks like…RP
--Vitreous is…optically empty
Presents with…nyctalopia
--Is also known as

something some-thing somethings

Norrie Disease

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Goldmann-Favre Syndrome

--Macula looks like…XLJR, but periphery looks like…RP
--Vitreous is…optically empty
Presents with…nyctalopia
--Is also known as *enhanced S-cone dz/syndrome*

Norrie Disease

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--Macula looks like…XLJR,
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--Vitreous is…optically empty
Presents with…nyctalopia
--Is also known as
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Norrie Disease

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--Split is in…RNFL
--Abnormal cells: Müller ce
--ERG: Diminution of the…
--If severe, peripheral char
lead to…RD, vitreous he

The BCSC Retina *book calls Goldmann-Favre a "diffuse photoreceptor dystrophy." In what ways are photoreceptors affected?*
--**Rods**:

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

Hereditary Vitreoretinopathies

Goldmann-Favre Syndrome

--Macula looks like…XLJR,
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--**Rods**: Non-functioning

…l Exudative …etinopathy

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--Vitreous is…optically empty
Presents with…nyctalopia
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Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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--Rod response: Undetectable
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Hereditary Hyalo
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--Blue cones: **Enhanced** (hence the name of the syndrome)

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What's up with the disease/syndrome ambiguity?

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What's up with the disease/syndrome ambiguity?
Blame the most recent (at the time of this writing) version of the *Retina* book--in one chapter the condition is referred to as 'enhanced S-cone syndrome,' whereas in another the same condition is 'enhanced S-cone **disease**.' (It even has separate entries in the Index.)

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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Norrie Disease

--inheritance

Familial Exudative Vitreoretinopathy

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Norrie Disease

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--Bilateral congenital…[*VA*]

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--Associated with:
--[nonocular finding 1]
--[nonocular finding 2]

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--Associated with:
--MR (= *mental retardation*)
--Hearing loss

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--Yellowish RD appears w/in…
  [time frame relative to birth]

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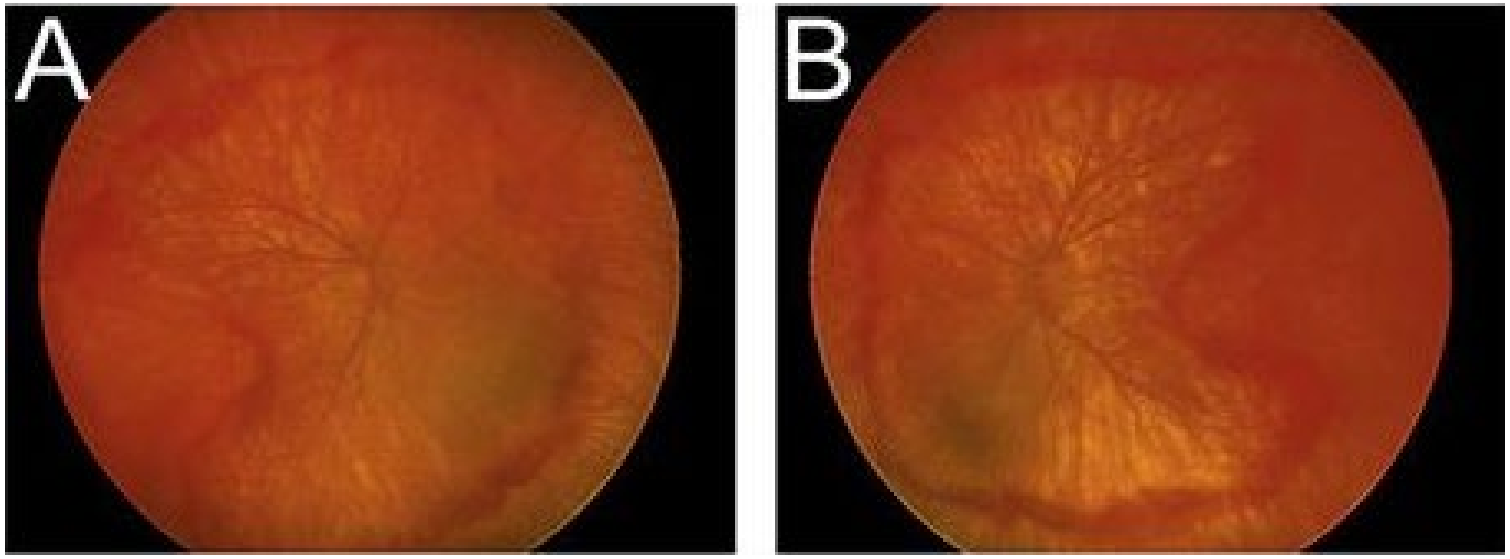

Fundus photograph of 34-week-old infant with Norrie Disease. **A** and **B**. Pretreatment images of the right and left eye demonstrating incomplete retinal vasculogenesis with neovascularization and hemorrhage, and incomplete foveal vascularization.

Norrie disease: Yellowish RDs

Hereditary Vitreoretinopathies

The yellowish (sometimes described as gray-yellowish) retinal mess has been mistaken for a retinal tumor. For this reason, it is sometimes referred to as a ***pseudoglioma***. (This fact will become more relevant in a couple of slides.)

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--Lenses & Ks…[*same change*]

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--Looks like…[*dz*]

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Familial Exudative Vitreoretinopathy

--Looks like…ROP--but FT and w/ normal respiratory status

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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Familial Exudative Vitreoretinopathy

--Looks like…ROP--but FT and w/ normal respiratory status
--Hallmark: Failure of…*[normal prenatal event]*

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

Hereditary Vitreoretinopathies

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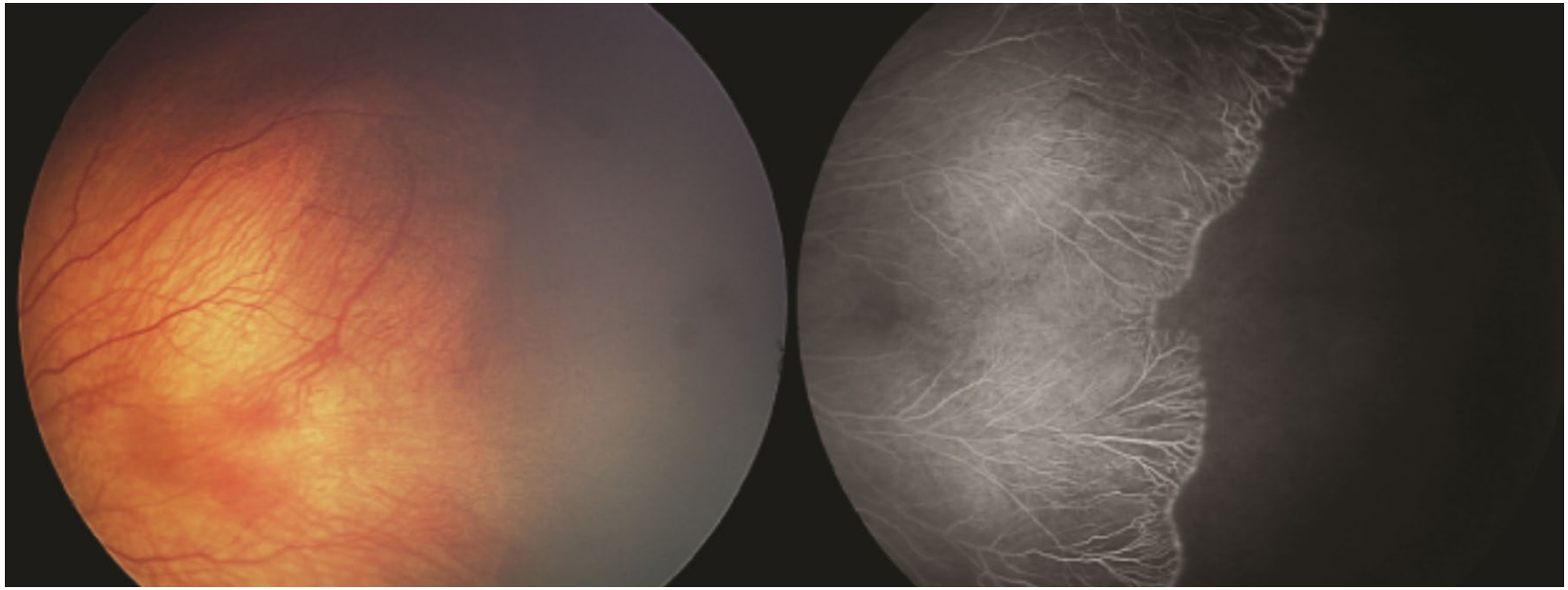
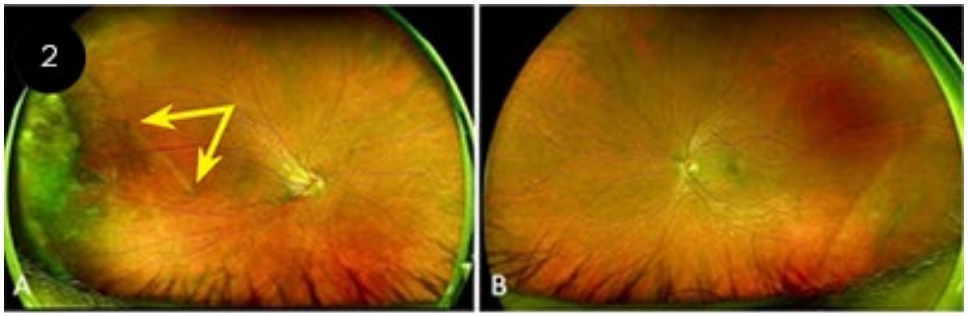


FEVR: Unvascularized temporal retina

Hereditary Vitreoretinopathies



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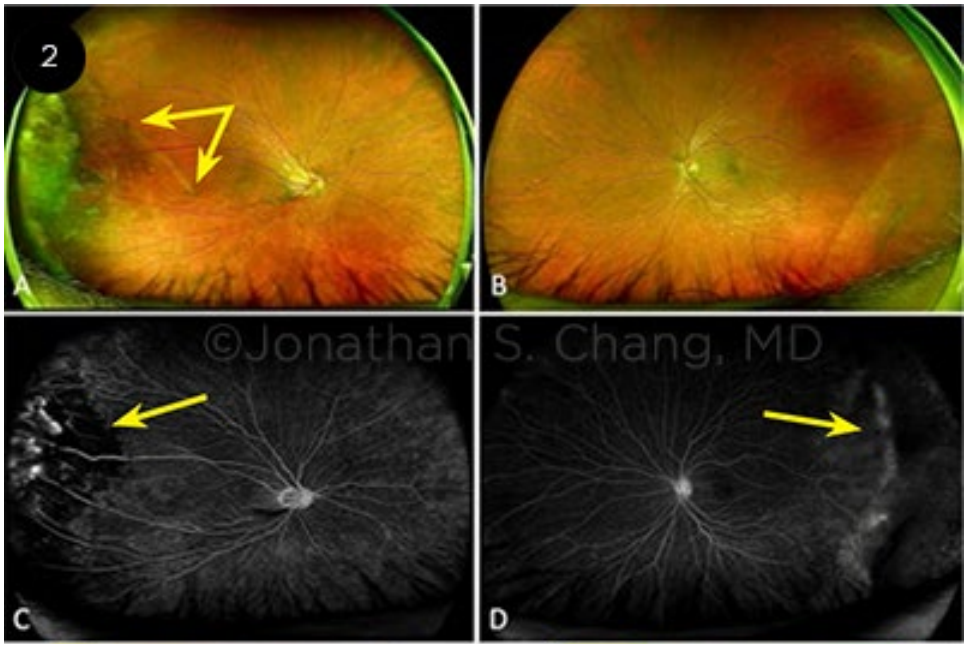

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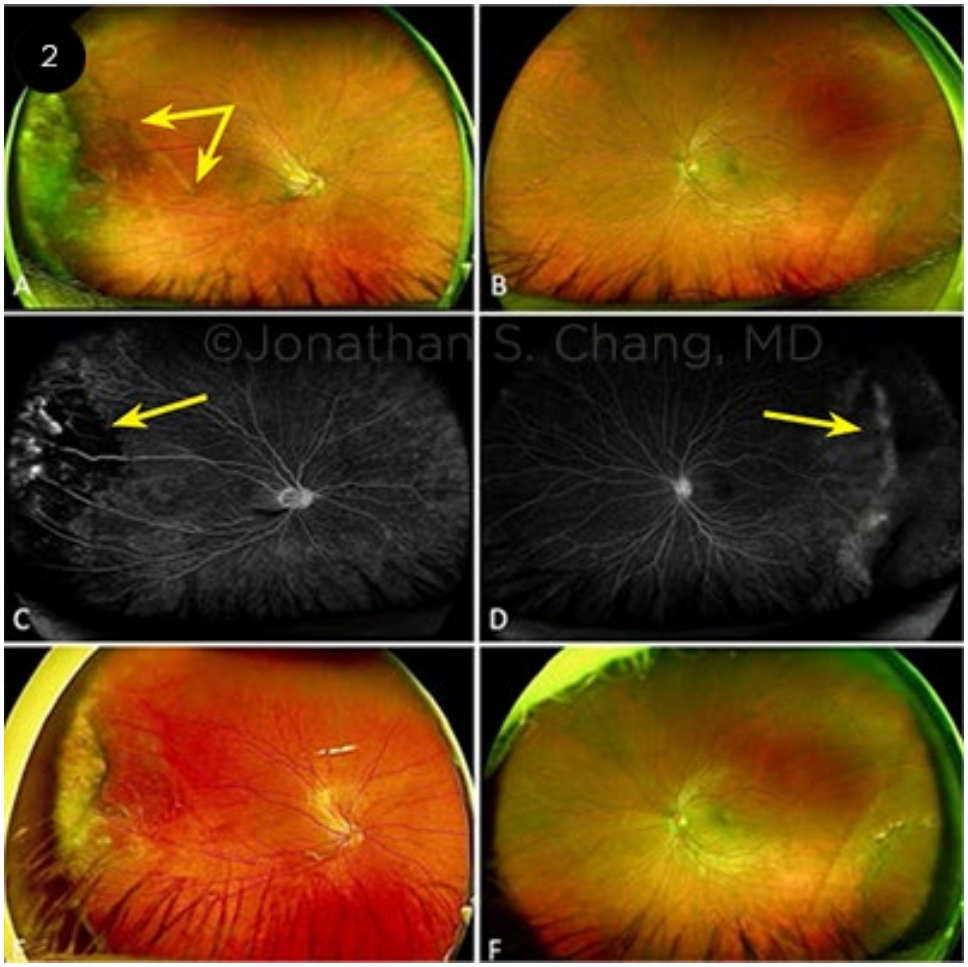
Hereditary Vitreoretinopathies



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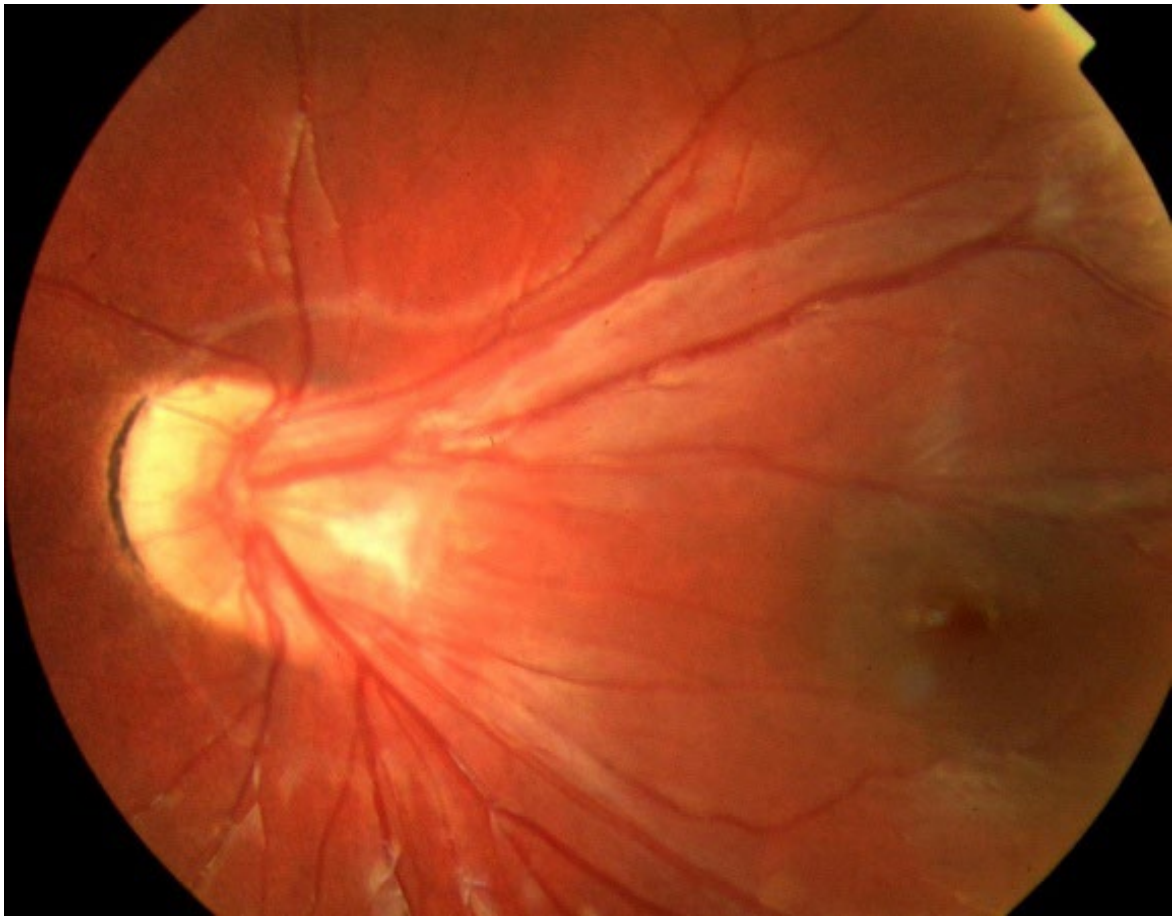

FEVR: Foveal/disc dragging

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Goldmann-Favre

X-Li… Reti…

--Name… but p…
--Fove…
--Looks…
--Split …
--Abno…
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Norrie disease and the ***X-linked version of FEVR*** have been traced to defects in a signaling protein called ***norrin***. The gene responsible is the ***N****orrie* ***D****isease* – ***P****seudoglioma (NDP) gene.* Multiple dz-causing mutations of this gene have been identified. In addition to Norrie's and *X*-linked FEVR, the NDP gene has been implicated in the pathogenesis of *some* (not all) cases of several other retinal conditions, including:
--Persistent hyperplastic primary vitreous (PHPV)
--Retinopathy of prematurity (ROP)
--Coats disease
The precise role played by the NDP gene in PHPV, ROP and Coats disease has yet to be elucidated.

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--Classic triad: predisposition to location, and CNS developmental prob + refractive prob + retinal prob

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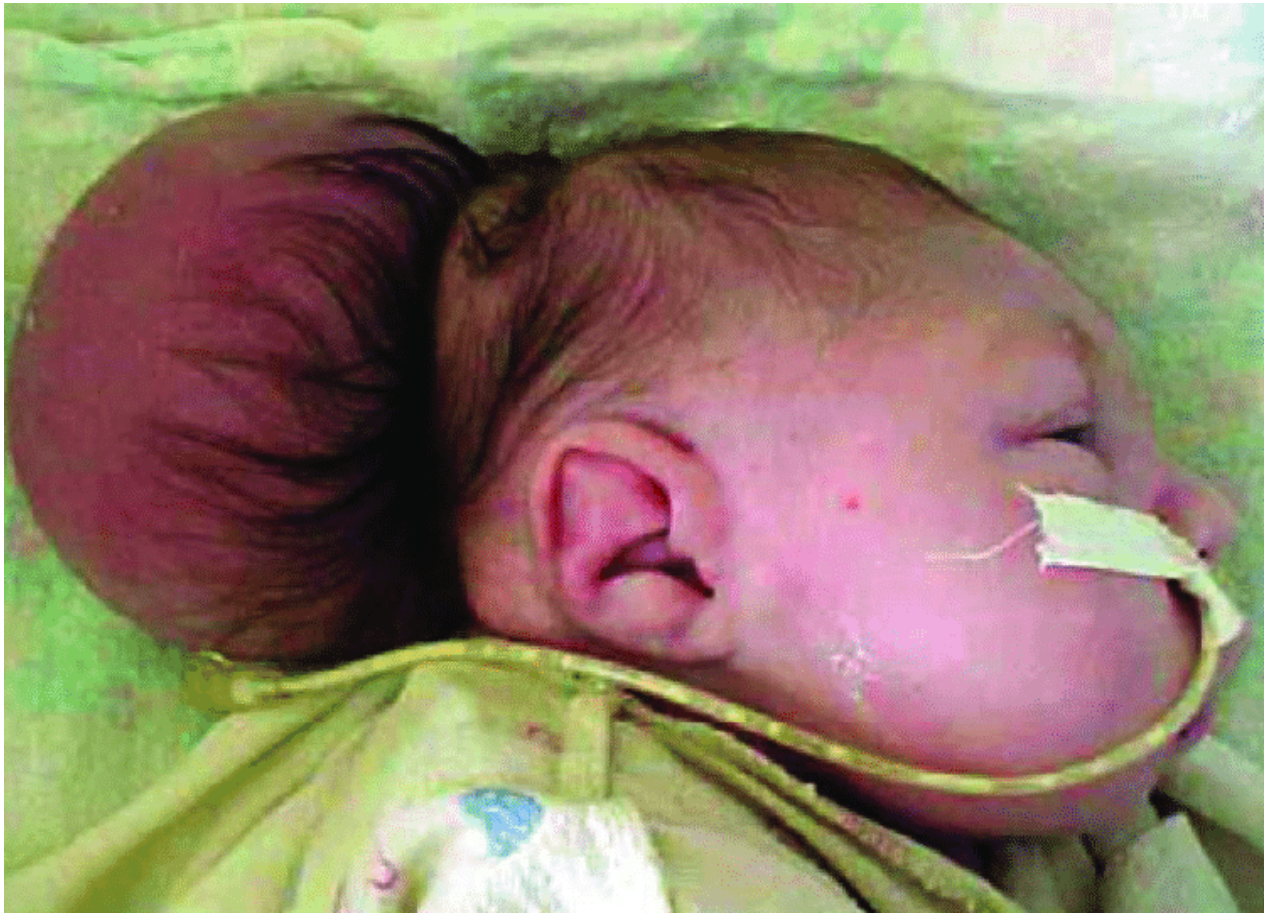
--Classic triad: Occipital encephalocele + high myopia + predisposition to retinal detachment

Hereditary Vitreoretinopathies



Knobloch syndrome: Occipital encephalocele

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Hereditary Hyaloideoretin… with Optically Empty V…

Do Knobloch pts always have a full-blown occipital encephalocele?

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Do Knobloch pts always have a full-blown occipital encephalocele?
No, there is a spectrum of severity--some kids 'only' have a funky occipital scalp.

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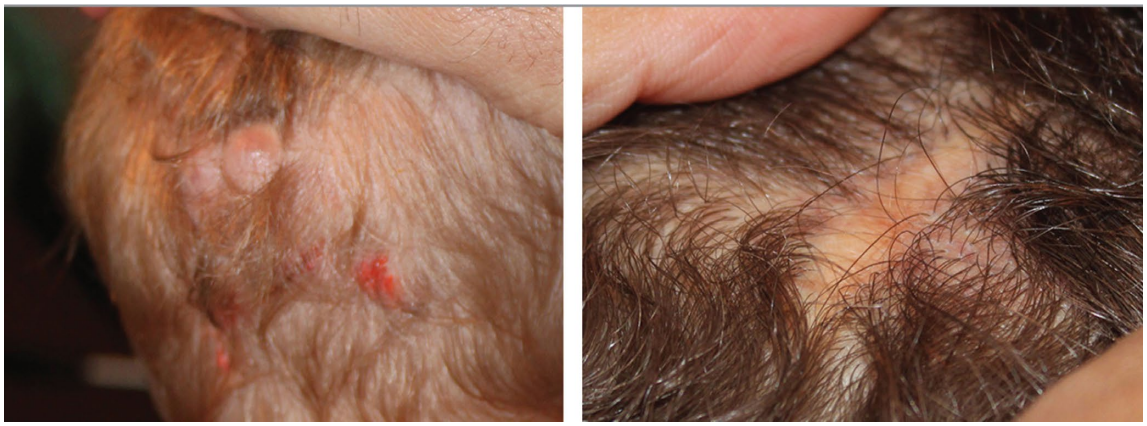
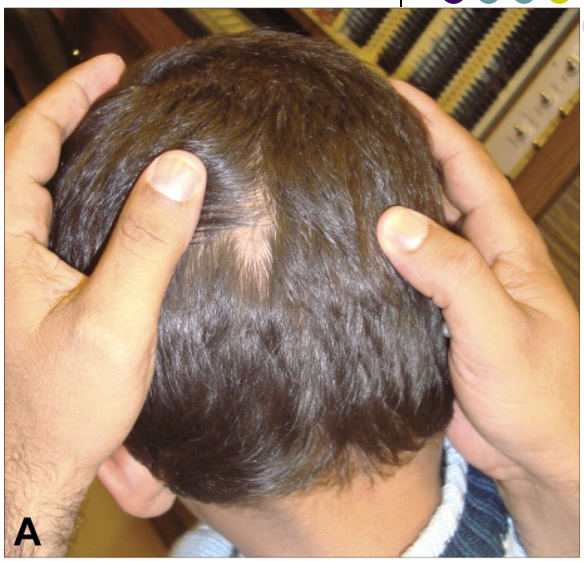
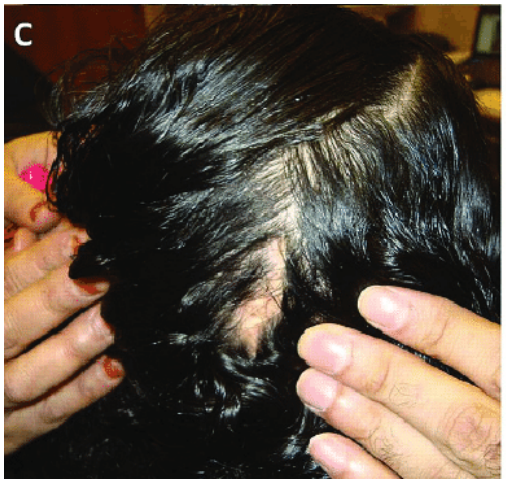
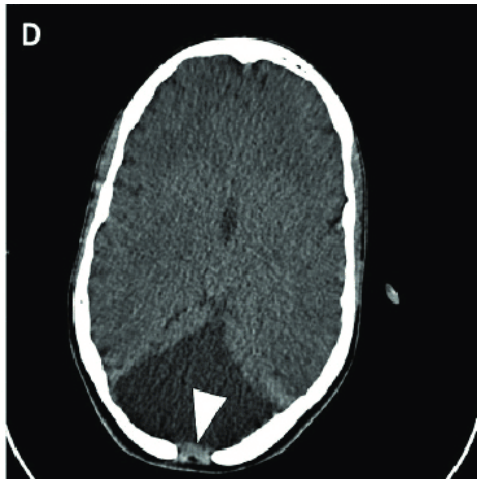
Do Knobloch pts always have a full-blown occipital encephalocele? No, there is a spectrum of severity--some kids 'only' have a funky occipital scalp. **Protip**: If shown a photo of the back of a kid's head, with the hair pushed out of the way to reveal the scalp, go with Knobloch syndrome.

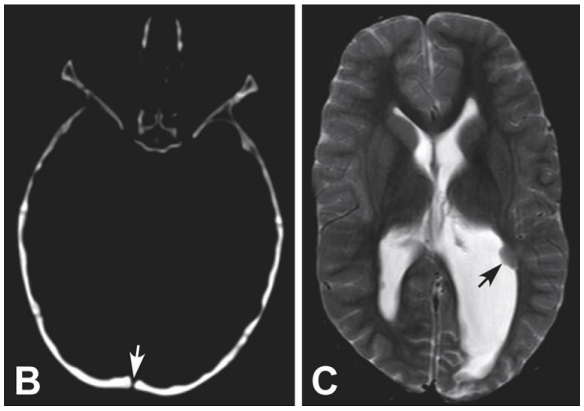
Hereditary Vitreoretinopathies











Knobloch syndrome: Funky occipital scalp

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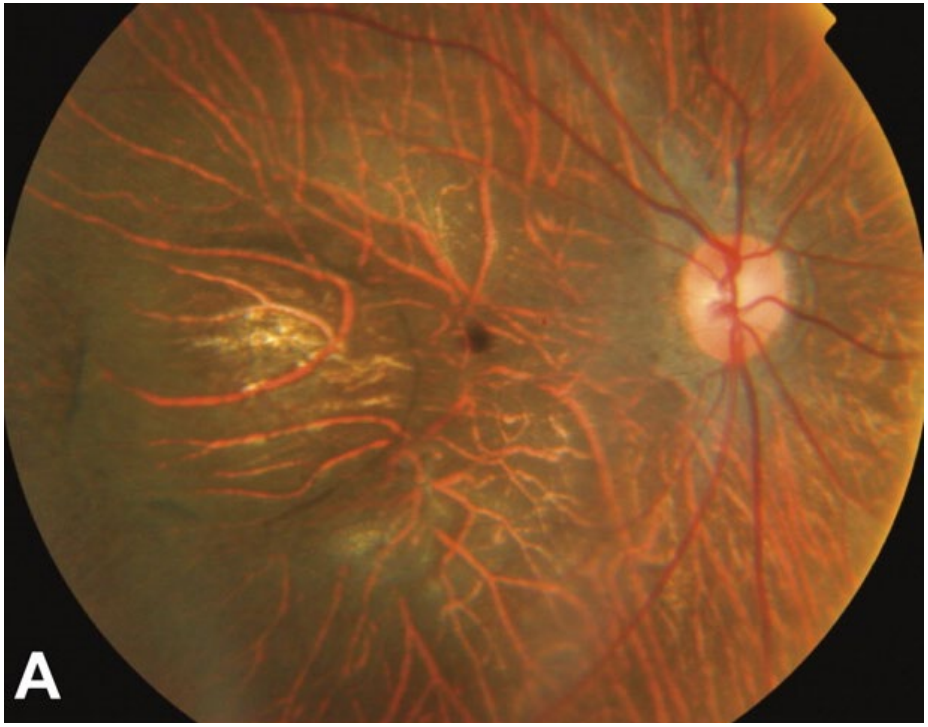
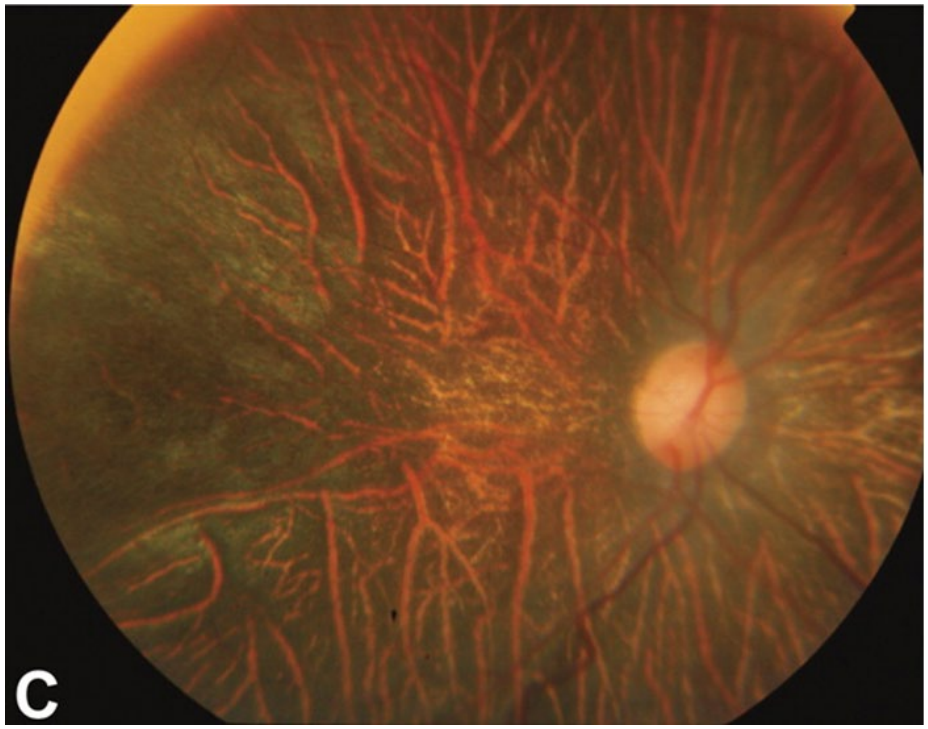
--RPE is atrophic

Hereditary Vitreoretinopathies





Knobloch syndrome: Atrophic RPE

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--Classic triad: Occipital encephalocele + high myopia + predisposition to retinal detachment
--RPE is atrophic
--Irides are structural issue

Hereditary Vitreoretinopathies

X-Linked Juvenile Retinoschisis

--Named for peripheral changes, but present in only 50% of cases
--Foveal schisis present in…100%
--Looks like CME, but is **dry** dz
--Split is in…RNFL
--Abnormal cells: Müller cells
--ERG: Diminution of the…*b*-wave
--If severe, peripheral changes can lead to…RD, vitreous heme

Goldmann-Favre Syndrome

--Macula looks like…XLJR, but periphery looks like…RP
--Vitreous is…optically empty
Presents with…nyctalopia
--Is also known as *enhanced S-cone dz/syndrome*

Norrie Disease

--X-linked recessive
--Bilateral congenital…blindness
--Associated with:
--MR
--Hearing loss
--Yellowish RD appears w/in… weeks of birth
--Lenses & Ks opacify with time
--End stage: Phthisis by age 10

Familial Exudative Vitreoretinopathy

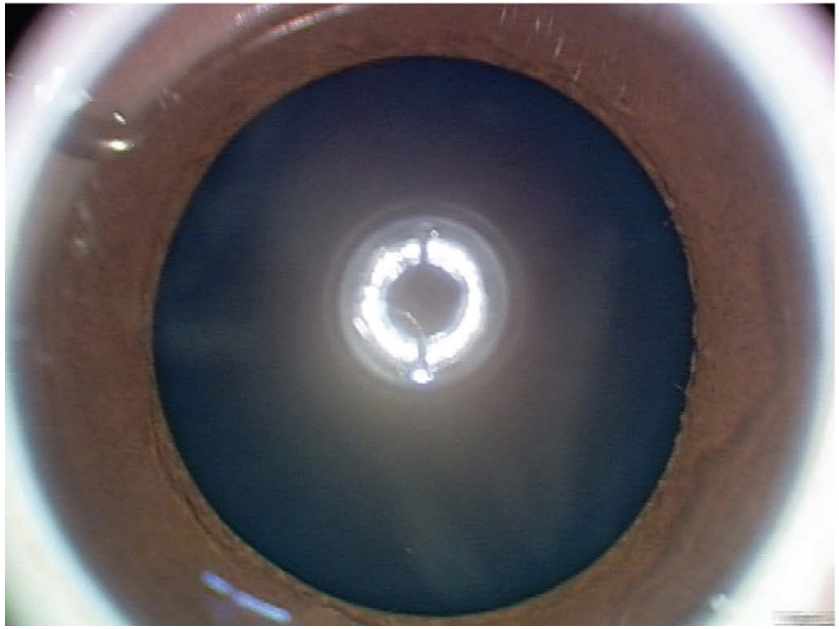
--Looks like…ROP--but FT and w/ normal respiratory status
--Hallmark: Failure of…temporal retina to vascularize
--Inheritance: AD, AR, X-linked
--Peripheral neo → TRD → retinal breaks and foveal dragging
--PVD, vitreous traction present

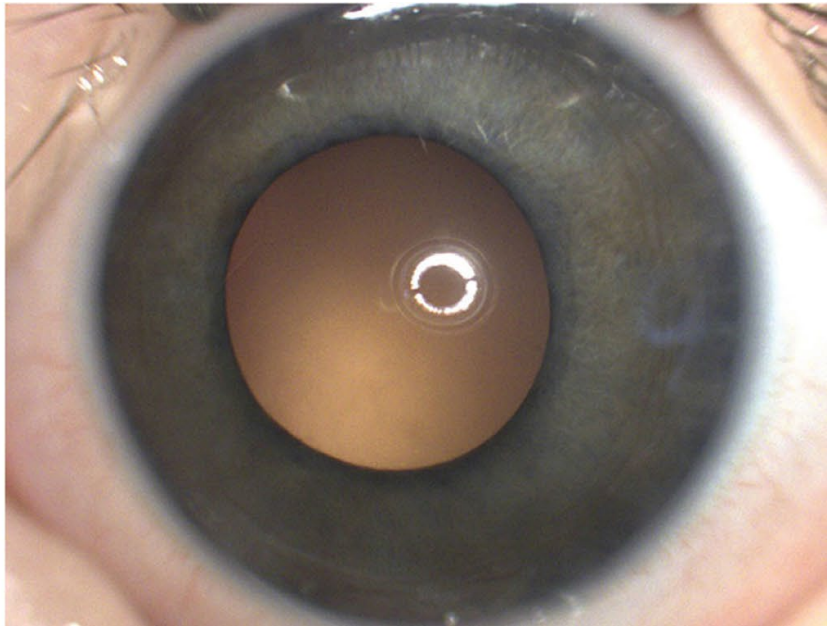
Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

--Classic triad: Occipital encephalocele + high myopia + predisposition to retinal detachment
--RPE is atrophic
--Irides are cryptless

Hereditary Vitreoretinopathies





Knobloch syndrome: Cryptless irides

Hereditary Vitreoretinopathies

Goldmann-Favre Syndrome

--Macula looks like…XLJR,
but periphery looks like…RP

Norrie Disease

--X-linked recessive
--Bilateral congenital…blindness
--Associated with:
--MR
loss
RD appears w/in…
birth
Ks opacify with time
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X-Linked Juvenile Retinoschisis

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--Abnormal cells: Müller c
--ERG: Diminution of the…
--If severe, peripheral changes can lead to…RD, vitreous heme

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

The hyaloideoretinopathies get their own slides…

Exudative …etinopathy

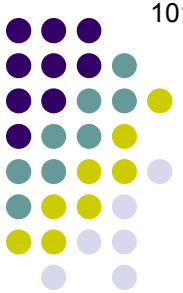
--Looks like…ROP--but FT and w/ normal respiratory status
--Hallmark: Failure of…temporal retina to vascularize
--Inheritance: AD, AR, X-linked
--Peripheral neo → TRD → retinal breaks and foveal dragging
--PVD, vitreous traction present

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

--Classic triad: Occipital encephalocele + high myopia + predisposition to retinal detachment
--RPE is atrophic
--Irides are cryptless

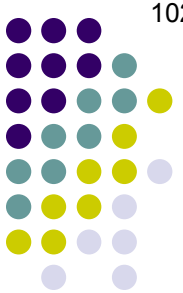
Hereditary Vitreoretinopathies



Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

--All have vitreous…*[finding]*

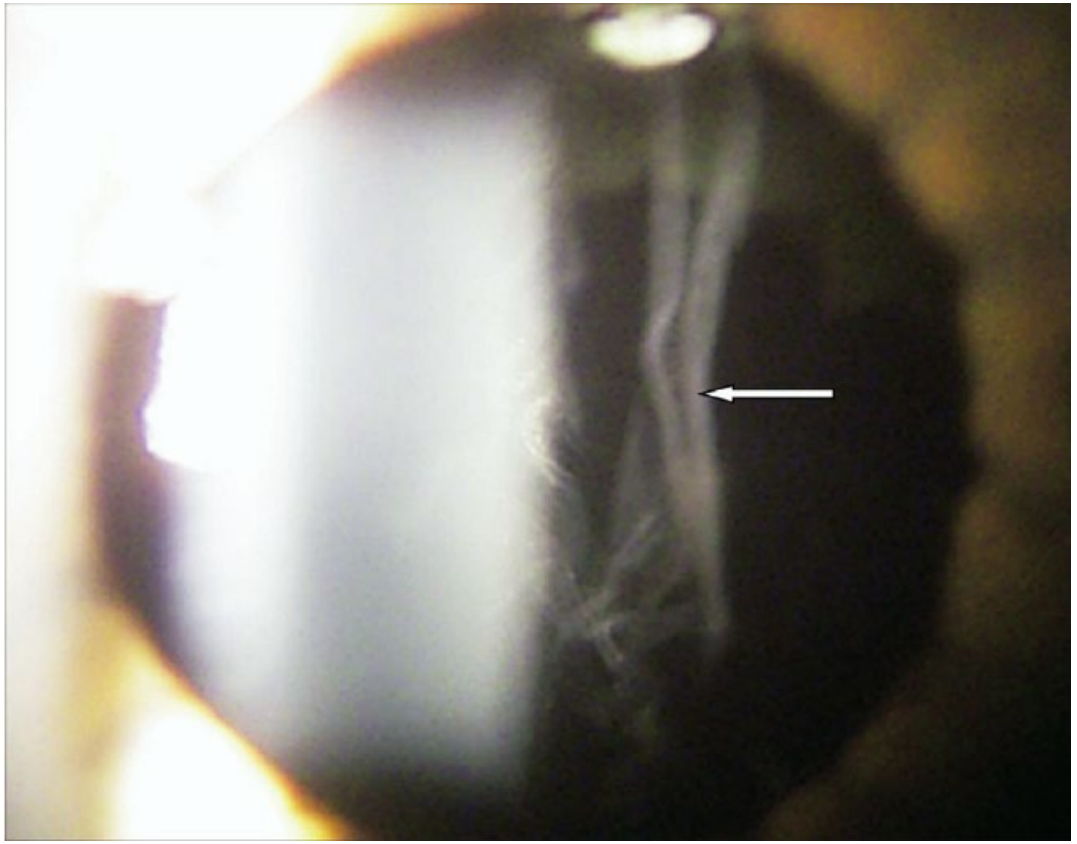
Hereditary Vitreoretinopathies



Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

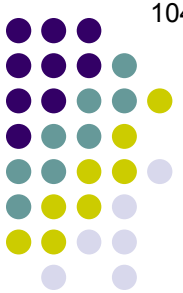
--All have vitreous…veils

Hereditary Vitreoretinopathies



Vitreous veils

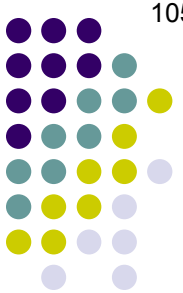
Hereditary Vitreoretinopathies



Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

--All have vitreous…veils
--All are associated with:
--[refractive status]

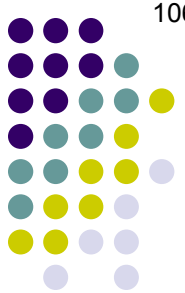
Hereditary Vitreoretinopathies



Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

--All have vitreous…veils
--All are associated with:
  --**Myopia**

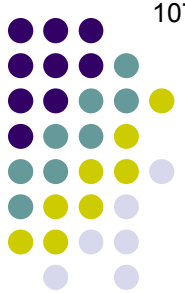
Hereditary Vitreoretinopathies



Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

--All have vitreous…veils
--All are associated with:
  --**Myopia** *--[blinding dz]*

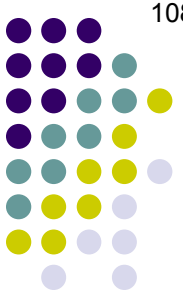
Hereditary Vitreoretinopathies



Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

--All have vitreous…veils
--All are associated with:
--**Myopia** --**Glaucoma**

Hereditary Vitreoretinopathies



Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

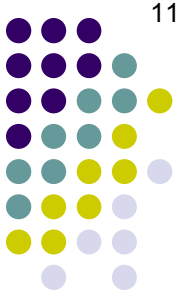
--All have vitreous…veils
--All are associated with:
--**Myopia** --**Glaucoma**
--[common eye prob]

Hereditary Vitreoretinopathies

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

--All have vitreous…veils
--All are associated with:
--**Myopia** --**Glaucoma**
--**Cataracts**

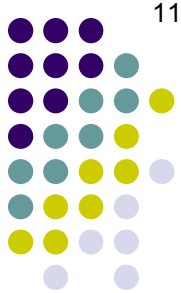
Hereditary Vitreoretinopathies



Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

--All have vitreous…veils
--All are associated with:
--**Myopia**
--**Cataracts**
--**Glaucoma**
--[abn retinal finding]

Hereditary Vitreoretinopathies



Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

--All have vitreous…veils
--All are associated with:
- --**Myopia**
- --**Cataracts**
- --**Glaucoma**
- --**Lattice degeneration**

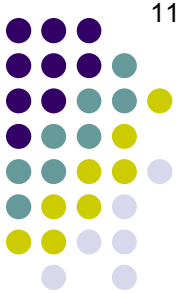
Hereditary Vitreoretinopathies

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

--All have vitreous…veils
--All are associated with:
--**Myopia**
--**Cataracts**
--**Glaucoma**
--**Lattice degeneration**

What is unusual about lattice in these conditions?

Hereditary Vitreoretinopathies

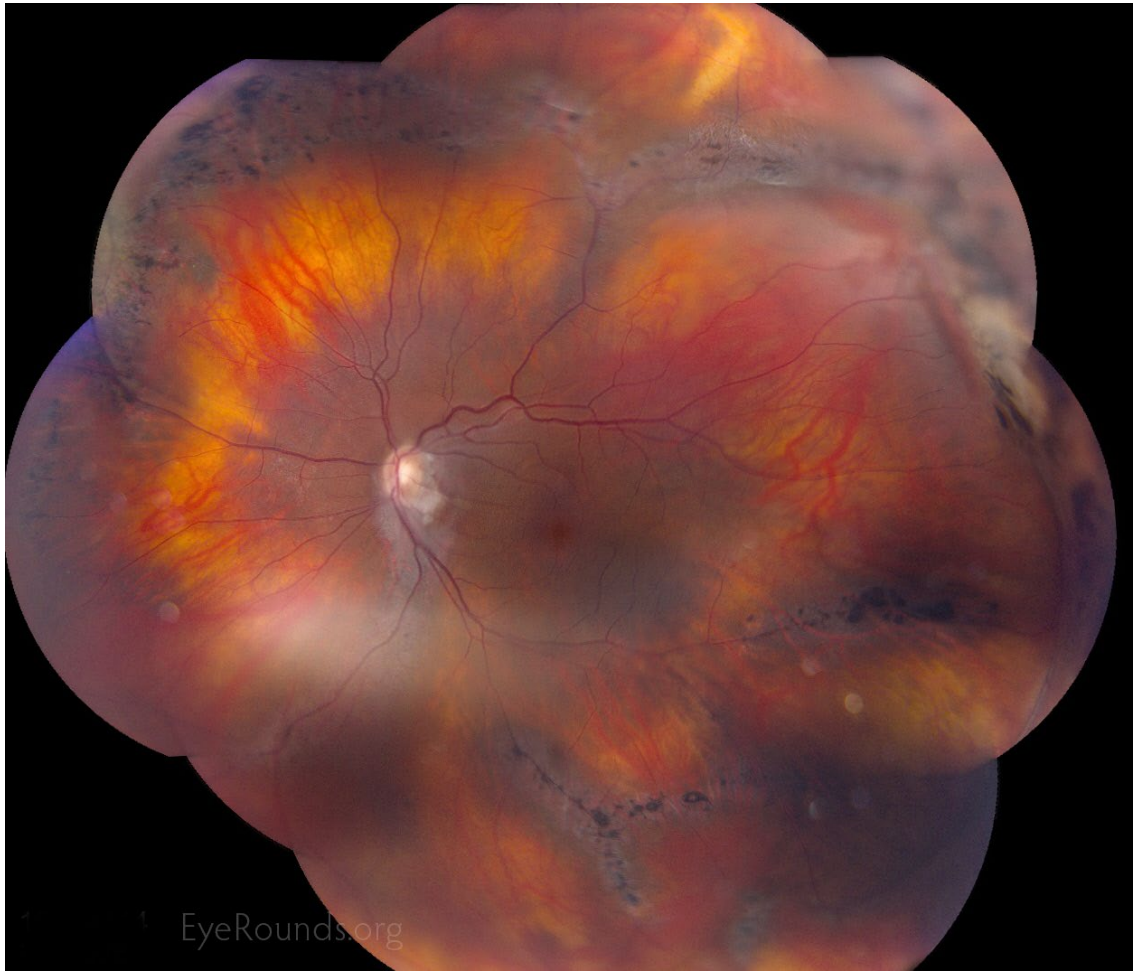


Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

--All have vitreous…veils
--All are associated with:
--**Myopia**
--**Cataracts**
--**Glaucoma**
--**Lattice degeneration**

What is unusual about lattice in these conditions?
It has a **R**
A
D
I
A
L orientation, i.e., the lattice points toward the optic nerve, as opposed to the circumferential orientation characteristic of typical lattice

Hereditary Vitreoretinopathies



Radially-oriented lattice degeneration

Hereditary Vitreoretinopathies

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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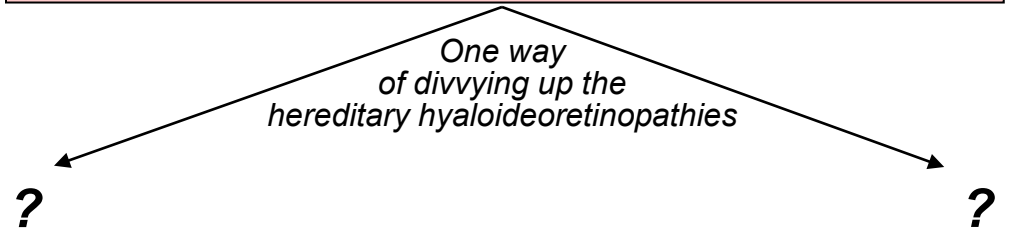
- --**Glaucoma**
- --**Lattice degeneration**



Hereditary Vitreoretinopathies

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

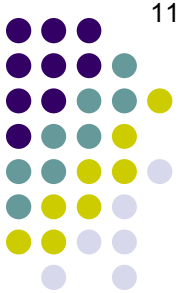
--All have vitreous…veils
--All are associated with:
- --**Myopia**
- --**Cataracts**
- --**Glaucoma**
- --**Lattice degeneration**

One way of divvying up the hereditary hyaloideoretinopathies

Ocular disease only

Associated with systemic disease

Hereditary Vitreoretinopathies



Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

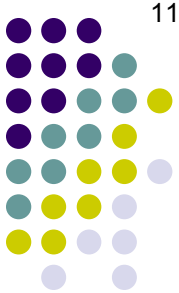
--All have vitreous…veils
--All are associated with:
- --**Myopia**
- --**Cataracts**
- --**Glaucoma**
- --**Lattice degeneration**

Ocular disease only

?

Associated with systemic disease

Hereditary Vitreoretinopathies



Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

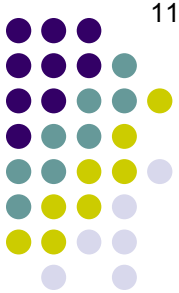
--All have vitreous…veils
--All are associated with:
- --**Myopia**
- --**Cataracts**
- --**Glaucoma**
- --**Lattice degeneration**

Ocular disease only

- Wagner's disease

Associated with systemic disease

Hereditary Vitreoretinopathies



Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

--All have vitreous…veils
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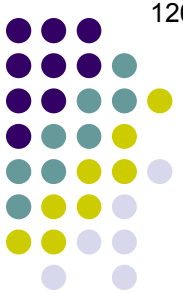
- --**Myopia**
- --**Cataracts**
- --**Glaucoma**
- --**Lattice degeneration**

Ocular disease only
- Wagner's disease

Associated with systemic disease
- ***?***

Hereditary Vitreoretinopathies



Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

--All have vitreous…veils
--All are associated with:
- **--Myopia**
- **--Cataracts**
- **--Glaucoma**
- **--Lattice degeneration**

Ocular disease only
- Wagner's disease

Associated with systemic disease
- Stickler syndrome

Hereditary Vitreoretinopathies

Stickler syndrome is strongly associated with a craniofacial malformation--which one?

Ocular disease only

- Wagner's disease

Associated with systemic disease

- **Stickler syndrome**

Hereditary Vitreoretinopathies

Stickler syndrome is strongly associated with a craniofacial malformation--which one?
Pierre Robin sequence (PRS)

Ocular disease only

- Wagner's disease

Associated with systemic disease

- **Stickler syndrome**

Hereditary Vitreoretinopathies

Stickler syndrome is strongly associated with a specific malformation, which one?

Pierre Robin sequence (P

How do you pronounce Pierre Robin *in this context?*

Ocular disease only

- Wagner's disease

Associated with systemic disease

- **Stickler syndrome**

Hereditary Vitreoretinopathies

Stickler syndrome is strongly associated with a pierre robin sequence which one?

Pierre Robin sequence (P

How do you pronounce Pierre Robin *in this context?*
PEA-err roe-BAHN

Ocular disease only

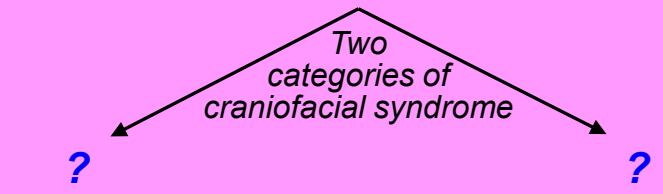
- Wagner's disease

Associated with systemic disease

- **Stickler syndrome**

Hereditary Vitreoretinopathies

Stickler syndrome is strongly associated with a ***craniofacial malformation****--which one?*
Pierre Robin sequence (PRS)



The Peds *book divides the craniofacial malformations into two groups—what are they?*

Ocular disease only

- Wagner's disease

Associated with systemic disease

- **Stickler syndrome**

Hereditary Vitreoretinopathies

Stickler syndrome is strongly associated with a ***craniofacial malformation****--which one?*

Pierre Robin sequence (PRS)

Two categories of craniofacial syndrome

Craniosynostoses

Not craniosynostoses

The Peds *book divides the craniofacial malformations into two groups—what are they?*

Ocular disease only

- Wagner's disease

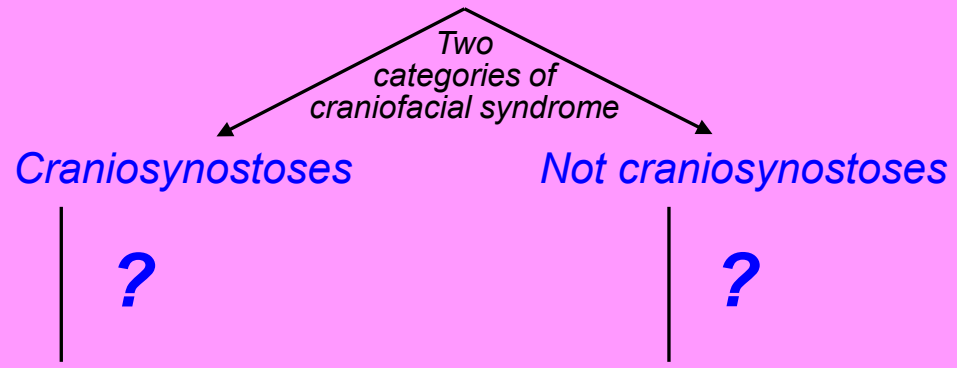
Associated with systemic disease

- **Stickler syndrome**

Hereditary Vitreoretinopathies

*Stickler syndrome is strongly associated with a **craniofacial malformation**--which one?*

Pierre Robin sequence (PRS)



In which group does Pierre Robin sequence belong?

Ocular disease only

- Wagner's disease

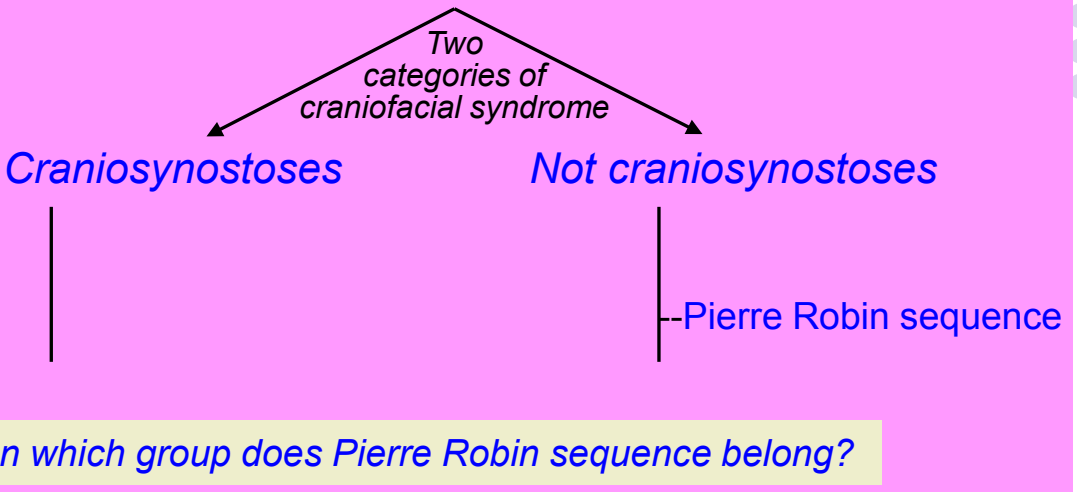
Associated with systemic disease

- **Stickler syndrome**

Hereditary Vitreoretinopathies

*Stickler syndrome is strongly associated with a **craniofacial malformation**--which one?*

Pierre Robin sequence (PRS)



In which group does Pierre Robin sequence belong?

Ocular disease only

- Wagner's disease

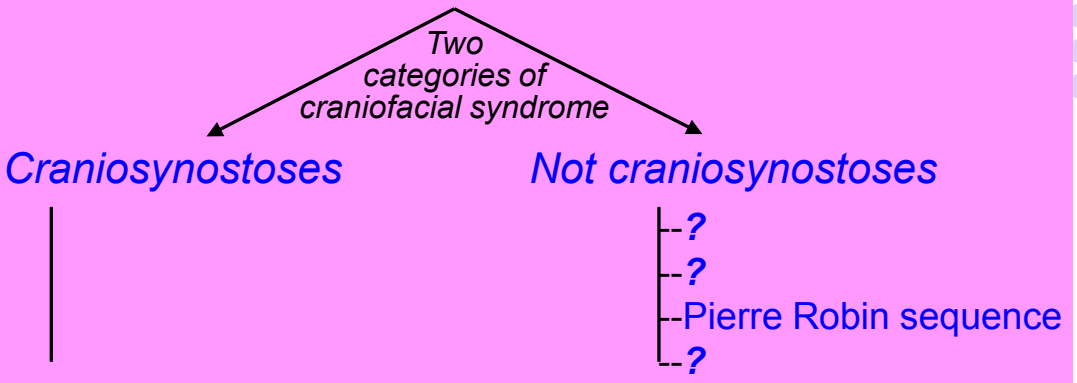
Associated with systemic disease

- **Stickler syndrome**

Hereditary Vitreoretinopathies

*Stickler syndrome is strongly associated with a **craniofacial malformation**--which one?*

Pierre Robin sequence (PRS)



What are the other three 'not craniosynostosis' craniofacial malformations?

Ocular disease only

- Wagner's disease

Associated with systemic disease

- **Stickler syndrome**

Hereditary Vitreoretinopathies

*Stickler syndrome is strongly associated with a **craniofacial malformation**--which one?*

Pierre Robin sequence (PRS)

Two categories of craniofacial syndrome

Craniosynostoses

Not craniosynostoses

- Goldenhar
- Treacher Collins
- Pierre Robin sequence
- Fetal alcohol syndrome

What are the other three 'not craniosynostosis' craniofacial malformations?

Ocular disease only

- Wagner's disease

Associated with systemic disease

- **Stickler syndrome**

Hereditary Vitreoretinopathies

*Stickler syndrome is strongly associated with a **craniofacial malformation**--which one?*

Pierre Robin sequence (PRS)

Two categories of craniofacial syndrome

Craniosynostoses

- *--?*
- *--?*
- *--?*
- *--?*

Not craniosynostoses

- --Goldenhar
- --Treacher Collins
- --Pierre Robin sequence
- --Fetal alcohol syndrome

What are the four craniosynostoses?

Ocular disease only

- Wagner's disease

Associated with systemic disease

- **Stickler syndrome**

Hereditary Vitreoretinopathies

*Stickler syndrome is strongly associated with a **craniofacial malformation**--which one?*

Pierre Robin sequence (PRS)

Two categories of craniofacial syndrome

Craniosynostoses

--Crouzon
--Apert
--Pfeiffer
--Saethre-Chotzen

Not craniosynostoses

--Goldenhar
--Treacher Collins
--Pierre Robin sequence
--Fetal alcohol syndrome

What are the four craniosynostoses?

Ocular disease only

└ Wagner's disease

Associated with systemic disease

└ **Stickler syndrome**

Hereditary Vitreoretinopathies

Stickler syndrome is strongly associated with a craniofacial malformation--which one?
Pierre Robin sequence (PRS)

With regard to congenital anomalies, what is meant by the term sequence*?*

Ocular disease only

- Wagner's disease

Associated with systemic disease

- **Stickler syndrome**

Hereditary Vitreoretinopathies

Stickler syndrome is strongly associated with a craniofacial malformation--which one?
Pierre Robin sequence (PRS)

With regard to congenital anomalies, what is meant by the term sequence*?*
It means that a single developmental malformation initiates a 'domino effect' which leads to other malformations, which in turn lead to significant functional issues

Ocular disease only

- Wagner's disease

Associated with systemic disease

- **Stickler syndrome**

Hereditary Vitreoretinopathies

Stickler syndrome is strongly associated with a craniofacial malformation--which one?
Pierre Robin sequence (PRS)

With regard to congenital anomalies, what is meant by the term sequence*?*
It means that a single developmental malformation initiates a 'domino effect' which leads to other malformations, which in turn lead to significant functional issues

In PRS, what is the 'single developmental malformation' that triggers the sequence?

Ocular disease only

- Wagner's disease

Associated with systemic disease

- **Stickler syndrome**

Hereditary Vitreoretinopathies

Stickler syndrome is strongly associated with a craniofacial malformation--which one?
Pierre Robin sequence (PRS)

With regard to congenital anomalies, what is meant by the term sequence*?*
It means that a single developmental malformation initiates a 'domino effect' which leads to other malformations, which in turn lead to significant functional issues

In PRS, what is the 'single developmental malformation' that triggers the sequence?
Micrognathia

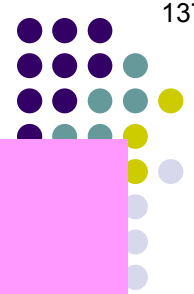
Ocular disease only

- Wagner's disease

Associated with systemic disease

- **Stickler syndrome**

Hereditary Vitreoretinopathies



Stickler syndrome is strongly associated with a craniofacial malformation--which one?
Pierre Robin sequence (PRS)

With regard to congenital anomalies, what is meant by the term sequence*?*
It means that a single developmental malformation initiates a 'domino effect' which leads to other malformations, which in turn lead to significant functional issues

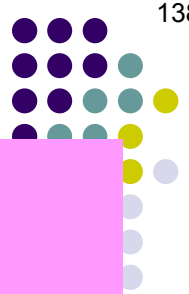
In PRS, what is the [illegible] malformation? What triggers the sequence?
Micrognathia

What does micrognathia *mean?*

Ocular disease only
- Wagner's disease

Associated with systemic disease
- **Stickler syndrome**

Hereditary Vitreoretinopathies



Stickler syndrome is strongly associated with a craniofacial malformation--which one?
Pierre Robin sequence (PRS)

With regard to congenital anomalies, what is meant by the term sequence*?*
It means that a single developmental malformation initiates a 'domino effect' which leads to other malformations, which in turn lead to significant functional issues

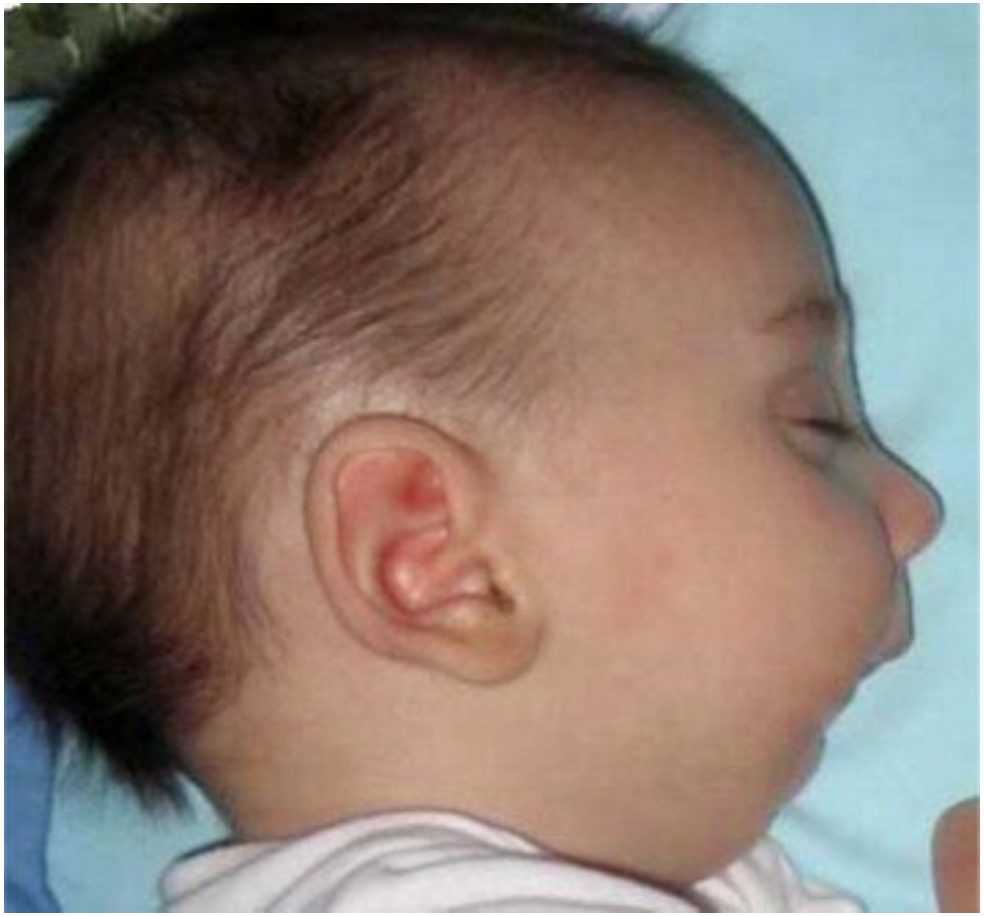
In PRS, what is the [illegible] that triggers the sequence?
Micrognathia

What does micrognathia *mean?*
It means 'severe hypoplasia of the mandible'

Ocular disease only
- Wagner's disease

Associated with systemic disease
- **Stickler syndrome**

Hereditary Vitreoretinopathies



Pierre Robin sequence: Micrognathia

Hereditary Vitreoretinopathies

Stickler syndrome is strongly associated with a craniofacial malformation--which one?
Pierre Robin sequence (PRS)

With regard to congenital anomalies, what is meant by the term sequence*?*
It means that a single developmental malformation initiates a 'domino effect' which leads to other malformations, which in turn lead to significant functional issues

In PRS, what is the 'single developmental malformation' that triggers the sequence?
Micrognathia

And what is the 'sequence,' ie, the subsequent malformations and functional issues?

Ocular disease only

- Wagner's disease

Associated with systemic disease

- **Stickler syndrome**

Hereditary Vitreoretinopathies

Stickler syndrome is strongly associated with a craniofacial malformation--which one?
Pierre Robin sequence (PRS)

With regard to congenital anomalies, what is meant by the term sequence*?*
It means that a single developmental malformation initiates a 'domino effect' which leads to other malformations, which in turn lead to significant functional issues

In PRS, what is the 'single developmental malformation' that triggers the sequence?
Micrognathia

And what is the 'sequence,' ie, the subsequent malformations and functional issues?
Micrognathia→glossoptosis→cleft palate→feeding difficulties

Ocular disease only

- Wagner's disease

Associated with systemic disease

- **Stickler syndrome**

Hereditary Vitreoretinopathies

Stickler syndrome is strongly associated with a craniofacial malformation--which one?
Pierre Robin sequence (PRS)

With regard to congenital anomalies, what is meant by the term sequence*?*
It means that a single developmental malformation initiates a 'domino effect' which leads to other malformations, which in turn lead to significant functional issues

In PRS, what is the 'single developmental malformation' that triggers the sequence?
Micrognathia

And what is the 'sequence,' ie, [illegible] *issues?*
Micrognathia → **glossoptosis** →

What does glossoptosis *refer to?*

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The position of the tongue being too posterior

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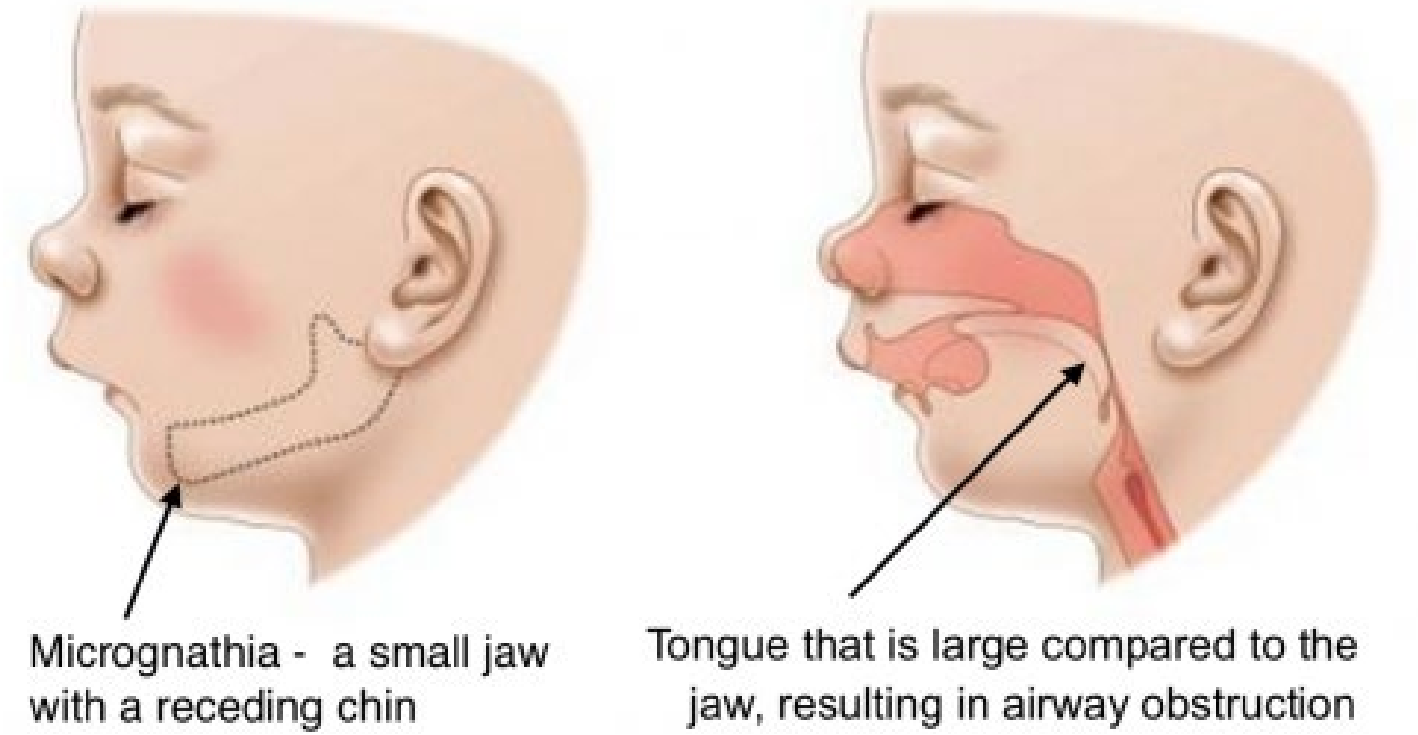
Hereditary Vitreoretinopathies



Micrognathia - a small jaw with a receding chin

Tongue that is large compared to the jaw, resulting in airway obstruction

Pierre Robin sequence: Glossoptosis

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And what is the 'sequence,' ie, the subsequent malformations and functional issues?
Micrognathia→glossoptosis→cleft palate→feeding difficulties

Stickler syndrome is also associated with arthropathy. How does this manifest?

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Stickler syndrome is also associated with arthropathy. How does this manifest?
The affected joints are size, and tight vs loose

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The affected joints are enlarged , and hypermobile

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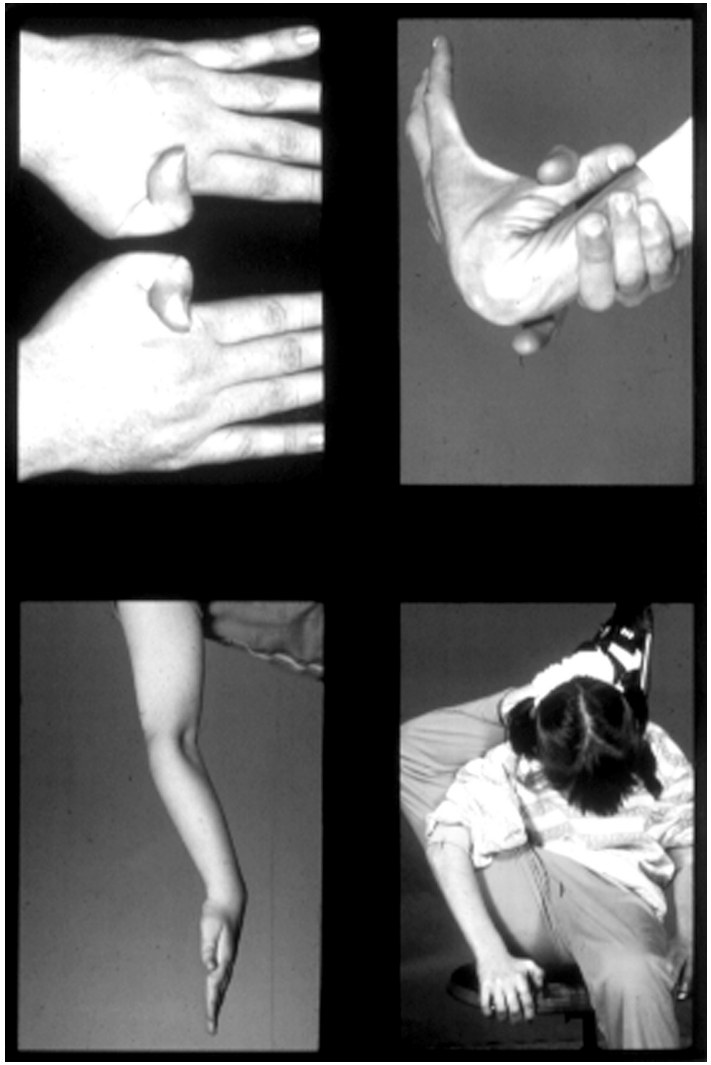
Hereditary Vitreoretinopathies



Stickler syndrome: hypermobile joints

Hereditary Vitreoretinopathies

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What is the non-eponymous name for Stickler syndrome?

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What is the non-eponymous name for Stickler syndrome?
'Hereditary arthro-ophthalmopathy, **Marfanoid** variety.' Note that this term may have fallen out of favor (it appeared in my *Retina* book back in the day, but not in the most recent edition.) I'm mentioning it as a way to 'make stick' the nature of the arthropathy associated with Stickler syndrome.

Stickler syndrome is also associated with arthropathy. How does this manifest?
The affected joints are enlarged , and hypermobile

Ocular disease only

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- **Stickler syndrome,** *aka…*
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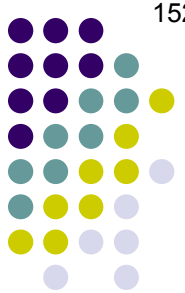
Hereditary Vitreoretinopathies



Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

--All have vitreous…veils
--All are associated with:
- --**Myopia**
- --**Cataracts**
- --**Glaucoma**
- --**Lattice degeneration**

Ocular disease only

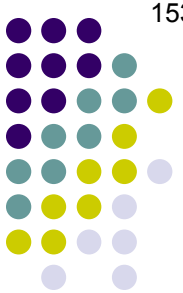
Wagner's disease?

Associated with systemic disease

Stickler syndrome?

Of these two conditions, only one carries a very high risk of retinal detachment. Which one?

Hereditary Vitreoretinopathies



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Ocular disease only

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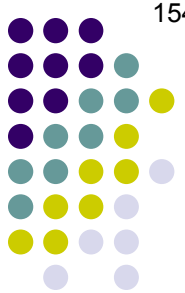
Associated with systemic disease

- **Stickler syndrome**

Of these two conditions, only one carries a very high risk of retinal detachment. Which one?

Stickler syndrome. BTW, this fact (the high RD risk associated with Stickler) is emphasized by the *BCSC* books--may be worth your time to commit it to memory.

Hereditary Vitreoretinopathies



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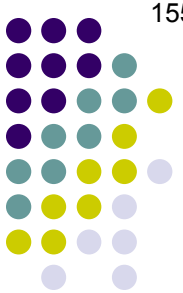
- Jansen's disease

Associated with systemic disease

- 'Hereditary arthro-ophthalmopathy with stiff joints' (Weill-Marchesani-like variety)
- ro-ophthalmopathy;
- ty (**Stickler syndrome**)

Are Stickler pts at increased risk of RD?

Hereditary Vitreoretinopathies



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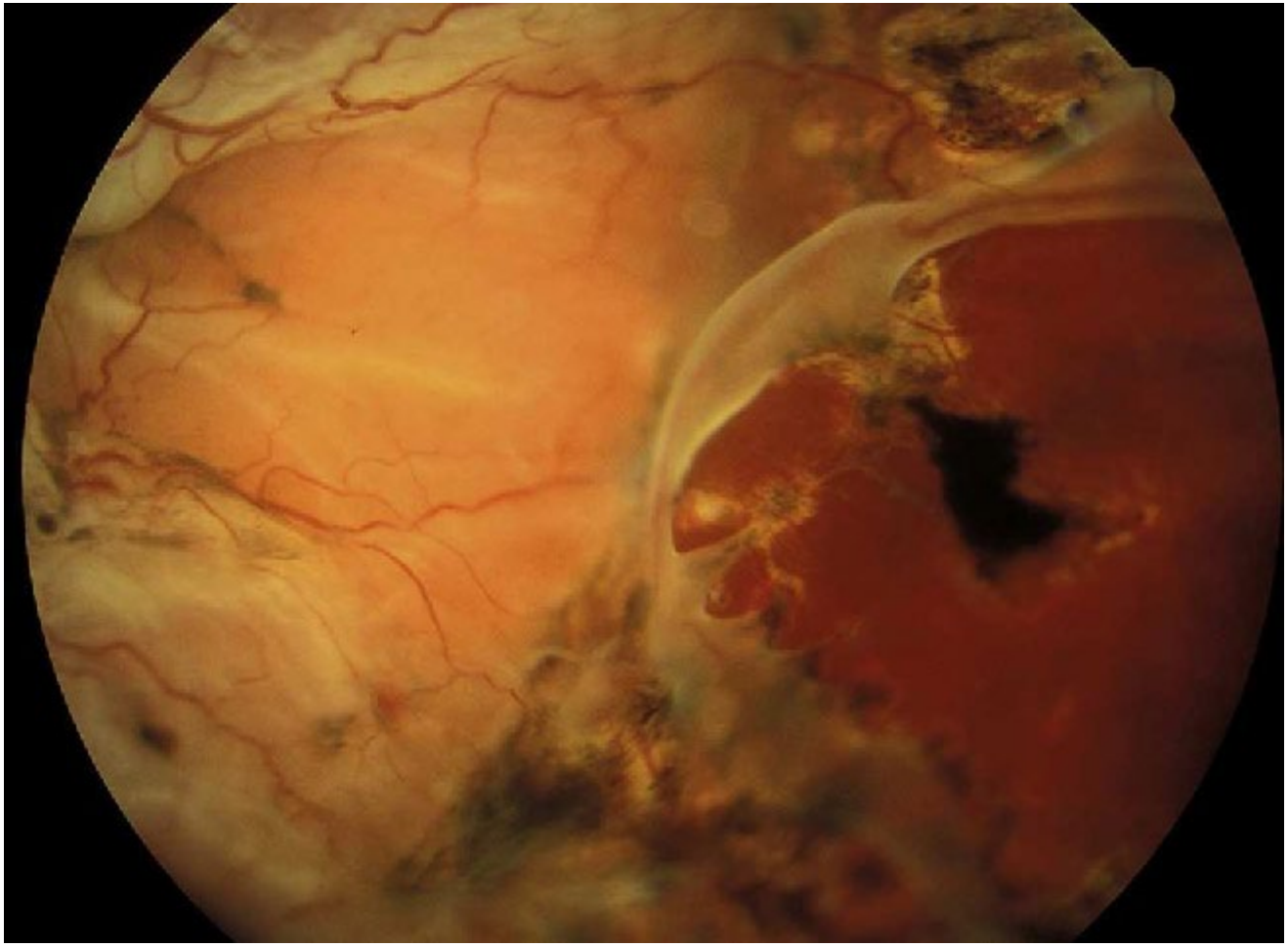
Associated with systemic disease

- 'Hereditary arthro-ophthalmopathy with stiff joints' (Weill-Marchesani-like variety)

Are Stickler pts at increased risk of RD?
Very much so. Even worse, their RDs are associated with large multiple breaks, rendering repair difficult

(**Stickler syndrome**)

Hereditary Vitreoretinopathies



Stickler syndrome: RD