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
-- Bilateral congenital blindness
-- Associated with:
  -- MR
  -- Hearing loss
-- Yellowish RD appears within weeks of birth
-- Lenses & Ks opacify with time
-- Ends in phthisis by age 10

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
-- Split is in RNFL
-- Abnormal cells: Müller cells
-- ERG: Diminution of the b-wave
-- If severe, peripheral changes can lead to RD, vitreous heme

Familial Exudative Vitreoretinopathy
-- Looks like ROP (but FT and normal respiratory status)
-- Hallmark: Failure of temporal retina to vascularize
-- Inheritance: AD (check family)
-- Peripheral neo → TRD → retinal breaks and foveal dragging
-- PVD, vitreous traction

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

specific disease

group of diseases

specific disease

specific disease
Norrie Disease
-- X-linked
-- Bilateral congenital blindness
-- Associated with:
  -- MR
  -- [nonocular finding] ring loss
  -- Yellowish RD appears within weeks of birth
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Hereditary Vitreoretinopathies

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Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

Goldmann-Favre Syndrome

Norrie Disease

Familial Exudative Vitreoretinopathy
Norrie Disease
--X-linked
--Bilateral congenital blindness
--Associated with:
--MR
--[nonocular finding] ring loss
--Yellowish RD appears within weeks of birth
--Lenses & Ks opacify with time
--Ends in phthisis by age 10

Hereditary Vitreoretinopathies

Familial Exudative Vitreoretinopathy
--Looks like ROP (but FT and normal respiratory status)
--Hallmark: Failure of temporal retina to vascularize
--Inheritance: AD (check family)
--Peripheral neo  TRD  retinal breaks and foveal dragging
--PVD, vitreous traction

X-Linked Juvenile Retinoschisis
--Named for peripheral changes, but present in only % of cases

Goldmann-Favre Syndrome

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

Knobloch 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
--- Bilateral congenital blindness
--- Associated with:
   - MR
   - [nonocular finding 2]
   - Ring loss
--- Yellowish RD appears within weeks of birth
--- Lenses & Ks opacify with time
--- Ends in... phthisis by age 10

Hereditary Vitreoretinopathies

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Familial Exudative Vitreoretinopathy
--- Looks like ROP (but FT and normal respiratory status)
--- Hallmark: Failure of... temporal retina to vascularize
--- Inheritance: AD (check family)
--- Peripheral neo... TRD... retinal breaks and foveal dragging
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Hereditary Hyaloideoretinopathies with Optically Empty Vitreous
--- Knobloch 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
Hereditary Vitreoretinopathies

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**

**Norrie Disease**

**Familial Exudative Vitreoretinopathy**

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

**Knobloch Syndrome**

Norrie Disease

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

Hereditary Vitreoretinopathies

X-Linked Juvenile Retinoschisis

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

--Looks like CME, but is dry
--Split is in…RNFL
--Abnormal cells: Müller cells
--ERG: Diminution of the…b-wave
--If severe, peripheral changes can lead to…RD, vitreous heme

Familial Exudative Vitreoretinopathy

--Looks like… ROP (but FT and normal respiratory status)
--Hallmark: Failure of…temporal retina to vascularize
--Inheritance: AD (check family)
--Peripheral neoTRDretinal breaks and foveal dragging
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Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

Goldmann-Favre Syndrome

Knobloch Syndrome

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Familial Exudative Vitreoretinopathy

**Hereditary Hyaloideoretinopathies**

Goldmann-Favre Syndrome

Norrie Disease

Familial Exudative Vitreoretinopathy

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome
Hereditary Vitreoretinopathies

X-Linked Juvenile Retinoschisis
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Goldmann-Favre Syndrome

Norrie Disease
--Bilateral congenital blindness
--Associated with: MR, Hearing loss
--Yellowish RD appears within weeks of birth
--Lenses & Ks opacify with time
--Ends in phthisis by age 10

Familial Exudative Vitreoretinopathy
--Looks like ROP (but FT and normal respiratory status)
--Hallmark: Failure of temporal retina to vascularize
--Inheritance: AD (check family)
--Peripheral neo \( \rightarrow \) TRD \( \rightarrow \) retinal breaks and foveal dragging
--PVD, vitreous traction

Knobloch Syndrome

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Goldmann-Favre Syndrome
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)

Familial Exudative Vitreoretinopathy

Norrie Disease
--Bilateral congenital blindness
--Associated with:
  --MR
  --Hearing loss
--Yellowish RD appears within weeks of birth
--Lenses & Ks opacify with time
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Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

Goldmann-Favre Syndrome

Knobloch Syndrome
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 [retinal layer]

**Norrie Disease**
--X-linked
--Bilateral congenital blindness
--Associated with:
  --MR
  --Hearing loss
--Yellowish RD appears within weeks of birth
--Lenses & KS opacify with time
--Ends in phthisis by age 10

**Familial Exudative Vitreoretinopathy**
--Looks like ROP (but FT and normal respiratory status)
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**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

**Knobloch Syndrome**
Hereditary Vitreoretinopathies

**Norrie Disease**
- X-linked
- Bilateral congenital blindness
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**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

**Familial Exudative Vitreoretinopathy**
- Looks like...ROP (but FT and normal respiratory status)
- Hallmark: Failure of...temporal retina to vascularize
- Inheritance: AD (check family)
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**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

**Knobloch Syndrome**
Hereditary Vitreoretinopathies

X-linked juvenile retinoschisis: RNFL schisis
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.
--Split is in RNFL.

Goldmann-Favre Syndrome

Norrie Disease
--X-linked
--Bilateral congenital blindness
--Yellowish RD appears within weeks of birth.
--Lenses & Ks opacify with time.
--Ends in phthisis by age 10.

Familial Exudative Vitreoretinopathy
--Looks like ROP, but FT and normal respiratory status.
--Hallmark: Failure of temporal retina to vascularize.
--Inheritance: AD (check family).
--Peripheral neo → TRD → retinal breaks and foveal dragging.
--PVD, vitreous traction.

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

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.
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:

**Norrie Disease**
--X-linked
--Bilateral congenital blindness
--Associated with:
  --MR
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**Goldmann-Favre Syndrome**

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**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

**Knobloch Syndrome**

Goldmann-Favre Syndrome

Norrie Disease

Familial Exudative Vitreoretinopathy

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome
Norrie Disease
--X-linked
--Bilateral congenital blindness
--Associated with:
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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
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Knobloch Syndrome
Goldmann-Favre Syndrome
Norrie Disease
Familial Exudative Vitreoretinopathy
Knobloch Syndrome
Hereditary Hyaloideoretinopathies with Optically Empty Vitreous
Hereditary Vitreoretinopathies

**Norrie Disease**
- X-linked
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- Associated with:
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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: Müller cells
- ERG: Diminution of the...[a-wave vs b-wave vs both]
  
**Familial Exudative Vitreoretinopathy**
- Looks like...ROP (but FT and normal respiratory status)
- Hallmark: Failure of...temporal retina to vascularize
- Inheritance: AD (check family)
- Peripheral neo...TRD...retinal breaks and foveal dragging
- PVD, vitreous traction

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

**Knobloch Syndrome**
Hereditary Vitreoretinopathies

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- ERG: Diminution of the b-wave

**Goldmann-Favre Syndrome**

**Norrie Disease**
- Bilateral congenital blindness
- Associated with: MR, Hearing loss
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- PVD, vitreous traction

**Knobloch Syndrome**

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**
- Macula looks like XLJR, but periphery looks like RP
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Hereditary Vitreoretinopathies

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

Goldmann-Favre Syndrome

Familial Exudative Vitreoretinopathy
-- Looks like ROP (but FT and normal respiratory status)
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Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

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
Hereditary Vitreoretinopathies

X-Linked Juvenile Retinoschisis
- Named for peripheral changes,
  but present in only 50% of cases
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- Looks like CME, but is dry
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  -- ERG: Diminution of the...b-wave
- If severe, peripheral changes can lead to...RD, vitreous heme

Norrie Disease
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- Bilateral congenital blindness
- Associated with:
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  -- MR
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- Yellowish RD appears w/in...weeks of birth
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- Ends in...phthisis by age 10

Familial Exudative Vitreoretinopathy
- Looks like... ROP (but FT and normal respiratory status)
- Hallmark: Failure of...temporal retina to vascularize
- Inheritance: AD (check family)
- Peripheral neo...TRD...retinal breaks and foveal dragging
- PVD, vitreous traction

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

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
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*
Norrie Disease

X-linked

Bilateral congenital blindness

Associated with:

MR

Hearing loss

Yellowish RD appears within weeks of birth

Lenses & Ks opacify with time

Ends in phthisis by age 10

X-Linked Juvenile Retinoschisis

Named for peripheral changes, but present in only 50% of cases

Foveal schisis present in 100%

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ERG: Diminution of the b-wave

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

Familial Exudative Vitreoretinopathy

Looks like ROP (but FT and normal respiratory status)

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Inheritance: AD (check family)

Peripheral neo -> TRD -> retinal breaks and foveal dragging

PVD, vitreous traction

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

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

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

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.
Norrie Disease

-- X-linked
-- Bilateral congenital blindness
-- Associated with:
  -- MR
  -- Hearing loss
-- Yellowish RD appears within weeks of birth
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Hereditary Vitreoretinopathies

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-- ERG: Diminution of the b-wave
-- If severe, peripheral changes can lead to RD, vitreous heme

Familial Exudative Vitreoretinopathy

-- Looks like ROP (but FT and normal respiratory status)
-- Hallmark: Failure of temporal retina to vascularize
-- Inheritance: AD (check family)
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-- PVD, vitreous traction

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

Goldmann-Favre Syndrome

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Such an ERG is said to demonstrate a ‘negative’ or ‘electronegative’ waveform
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Knobloch Syndrome

Goldmann-Favre Syndrome

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Familial Exudative Vitreoretinopathy

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous
Hereditary Vitreoretinopathies

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Familial Exudative Vitreoretinopathy

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Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

Goldmann-Favre Syndrome

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 Knobloch syndrome.

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

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

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.
Hereditary Vitreoretinopathies

X-Linked Juvenile Retinoschisis

Norrie Disease

Goldmann-Favre Syndrome

Familial Exudative Vitreoretinopathy

Knobloch Syndrome

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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
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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

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-- Looks like ROP (but FT and normal respiratory status)
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-- Inheritance: AD (check family)
-- Peripheral neophthisis → TRD → retinal breaks and foveal dragging
-- PVD, vitreous traction

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

What does CSNB stand for in this context?

CSNB stands for Congenital Stationary Night Blindness.

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.

What does CSNB stand for in this context?
CSNB stands for 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.
Norrie Disease
-- X-linked
-- Bilateral congenital blindness
-- Associated with:
  -- MR
  -- [nonocular finding] ring loss
  -- Yellowish RD appears within weeks of birth
  -- Lenses & Ks opacify with time
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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… RD, vitreous heme

Familial Exudative Vitreoretinopathy
-- Looks like… ROP (but FT and normal respiratory status)
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Hereditary Hyaloideoretinopathies with Optically Empty Vitreous
Knobloch Syndrome

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

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…

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
Norrie Disease
--X-linked
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--Associated with:
--MR
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Hereditary Vitreoretinopathies
X-Linked Juvenile Retinoschisis
--Named for peripheral changes, but present in only 50% of cases
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Hereditary Hyaloideoretinopathies with Optically Empty Vitreous
Knobloch Syndrome

Goldmann-Favre Syndrome

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

In a nutshell, what’s CSNB?

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

In a nutshell, what’s CSNB?

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Congenital stationary night blindness

In a nutshell, what’s CSNB?

What does CSNB stand for in this context?
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**Norrie Disease**

- X-linked
- Bilateral congenital blindness
- Associated with:
  - MR
  - Hearing loss
- Yellowish RD appears within weeks of birth
- Lenses & Ks opacify with time
- Ends in phthisis by age 10

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**Familial Exudative Vitreoretinopathy**

- Looks like ROP (but FT and normal respiratory status)
- Hallmark: Failure of temporal retina to vascularize
- Inheritance: AD (check family)
- Peripheral neo → TRD → retinal breaks and foveal dragging
- PVD, vitreous traction

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**Goldmann-Favre Syndrome**

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**
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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 ERG pathognomonic for XLJR?
No, several conditions can cause it. That said, the only other one likely to present in childhood is CSNB.

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The BCSC Retina book calls Goldmann-Favre a “diffuse photoreceptor dystrophy.” In what ways are photoreceptors affected?
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--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
--Bilateral congenital...blindness
--Associated with:
  --MR
  --Hearing loss
--Yellowish RD appears w/in...weeks of birth
--Lenses & Ks opacify with time
--Ends in...phthisis by age 10

Familial Exudative Vitreoretinopathy

--Looks like...ROP (but FT and normal respiratory status)
--Hallmark: Failure of...temporal retina to vascularize
--Inheritance: AD (check family)
--Peripheral neo
  \[\text{TRD}\] \[\rightarrow\] retinal breaks and foveal dragging
--PVD, vitreous traction

Knobloch Syndrome

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

--Rods: Non-functioning
  --Red/green cones: Reduced in number
  --Blue cones: Increased in number

What are the ERG findings?

--Rod response: Undetectable
--Red/green cone response: Attenuated
Hereditary Vitreoretinopathies

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**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

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--Split is in RNFL
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--Blue cones: Enhanced (hence the name of the syndrome)
Hereditary Vitreoretinopathies

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Knobloch Syndrome

Hereditary Hyaloideoretinopathies
with Optically Empty Vitreous

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Hereditary Hyaloideoretinopathies
What's up with the disease/syndrome ambiguity?
Hereditary Vitreoretinopathies

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Vitreoretinal Dystrophies

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

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 Vitreoretinopathies

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Knobloch Syndrome

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Knobloch Syndrome

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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Knobloch Syndrome
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**Norrie Disease**
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--Associated with:
  --[nonocular finding 1]
  --[nonocular finding 2]

**Knobloch Syndrome**

**Familial Exudative Vitreoretinopathy**

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**
Norrie Disease
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Knobloch Syndrome

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Hereditary Vitreoretinopathies
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Hereditary Vitreoretinopathies

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

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- X-linked
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- Associated with:
  - MR
  - [nonocular finding] ring loss
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Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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Knobloch Syndrome

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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Hereditary Vitreoretinopathies

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**Hereditary Vitreoretinopathies**

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**Knobloch Syndrome**
Hereditary Vitreoretinopathies

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--Looks like...[dz]

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Hereditary Vitreoretinopathies

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**Knobloch Syndrome**

**Goldmann-Favre Syndrome**

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**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

**Knobloch Syndrome**
## Hereditary Vitreoretinopathies

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### Familial Exudative Vitreoretinopathy

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- Hallmark: Failure of temporal retina to vascularize

### Knobloch Syndrome

### Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

### 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

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FEVR: Unvascularized temporal retina
A 41-year-old man with an unremarkable medical history presented for evaluation of a raised, pigmented peripheral retinal lesion. The patient was asymptomatic. VA was 20/40 OD and 20/25 OS. DFE OD showed a dragged macula (double-headed arrow) with temporal fibrovascular tissue. OS had a normal fovea and infratemporal retinoschisis.
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retinal breaks and foveal dragging

Knobloch Syndrome

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome
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FEVR: Foveal/disc dragging
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--May be equivalent to the congenital night-blinding dz enhanced S-cone syndrome

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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 Norrie Disease – Pseudoglioma (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: location, and CNS developmental prob
  + refractive prob
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Do Knobloch pts always have a full-blown occipital encephalocele?
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No, there is a spectrum of severity--some kids ‘only’ have a funky occipital scalp.
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Hereditary Vitreoretinopathies

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The hyaloideoretinopathies get their own slides...

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Hereditary Hyaloideoretinopathies
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

Goldmann-Favre Syndrome

Norrie Disease

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Hereditary Hyaloideoretinopathies

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

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

Hereditary Vitreoretinopathies
X-Linked Juvenile Retinoschisis
--Named for peripheral change, but present in only 50%
--Foveal schisis present in 100%
--Looks like CME, but is dry
--Split is in...RNFL
--Abnormal cells: Müller cells
--ERG: Diminution of the...b-wave
--If severe, peripheral changes can lead to...RD, vitreous heme

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous
The hyaloideoretinopathies get their own slides...

Goldmann-Favre Syndrome

Norrie Disease

Hereditary Hyaloideoretinopathies
Familial Exudative Vitreoretinopathy
--Looks like...ROP--but FT and w/ normal respiratory status
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Goldmann-Favre Syndrome

Norrie Disease

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The hyaloideoretinopathies get their own slides...

Hereditary Hyaloideoretinopathies
Familial Exudative Vitreoretinopathy
--Looks like...ROP--but FT and w/ normal respiratory status
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Hereditary Hyaloideoretinopathies with Optically Empty Vitreous
The hyaloideoretinopathies get their own slides...
Hereditary Hyaloideoretinopathies

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

--All have vitreous...[finding]
Hereditary Hyaloideoretinopathies

Hereditary Vitreoretinopathies with Optically Empty Vitreous

--All have vitreous...veils
Hereditary Vitreoretinopathies

Vitreous veils
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 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 Hyaloideoretinopathies

--All have vitreous...veils
--All are associated with:
   --Myopia
   --Cataracts
   --Glaucoma
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 Retinopathies

--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 radial orientation, i.e., the lattice points toward the optic nerve, as opposed to the circumferential orientation characteristic of typical lattice
Radially-oriented 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
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

One way of divvying up the hereditary hyaloideoretinopathies
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

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

Ocular disease only

Wagner’s disease

Associated with systemic disease
Hereditary Hyaloideoretinopathies

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 Hyaloideoretinopathies with Optically Empty Vitreous

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

Hereditary Vitreoretinopathies

Ocular disease only
  Wagner’s disease

Associated with systemic disease
  Stickler syndrome
Hereditary Hyaloideoretinopathies

All have vitreous veils

All are associated with:
- Myopia
- Glaucoma
- Cataracts
- Lattice degeneration

Hereditary Vitreoretinopathies

Ocular disease only
- Wagner’s disease

Associated with systemic disease
- Stickler syndrome

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

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

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

The affected joints are enlarged and hypermobile
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 craniofacial malformations, which one?

Pierre Robin sequence (PRS)

How do you pronounce Pierre Robin in this context?

Ocular disease only

- Wagner’s disease

Associated with systemic disease

- Stickler syndrome
Hereditary Hyaloideoretinopathies

---

Hereditary Vitreoretinopathies

- Ocular disease only
  - Wagner’s disease

- Associated with systemic disease
  - Stickler syndrome

---

Stickler syndrome is strongly associated with craniofacial malformations - which one?

**Pierre Robin sequence**

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

PEA-err roe-BAHN

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

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

The affected joints are enlarged and hypermobile.
Stickler syndrome is strongly associated with a craniofacial malformation--which one?

Pierre Robin sequence (PRS)

Two categories of craniofacial syndrome

Ocular disease only

Wagner’s disease

Associated with systemic disease

Stickler syndrome

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

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

- 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**

**Two categories of craniofacial 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

In which group does Pierre Robin sequence belong?

- Ocular disease only
- Wagner’s disease
- Associated with systemic disease
- Stickler syndrome

Craniosynostoses

- Micrognathia
- Glossoptosis
- Cleft palate
- Feeding difficulties

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

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

The affected joints are enlarged and hypermobile

Craniosynostoses

Not craniosynostoses

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
  - Pierre Robin sequence

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)

Ocular disease only
- Wagner’s disease

Associated with systemic disease
- Stickler syndrome

What are the other three ‘not craniosynostosis’ craniofacial malformations?

Craniosynostoses

Not craniosynostoses
--?
--?
--Pierre Robin sequence
--?

Two categories of craniofacial syndrome
Hereditary Vitreoretinopathies

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous -- All have vitreous veils -- All are associated with:
- Myopia
- Glaucoma
- Cataracts
- Lattice degeneration

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

**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**

Two categories of craniofacial syndrome

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

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

Craniosynostoses Not craniosynostoses

Two categories of craniofacial syndrome

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

What are the other three ‘not craniosynostosis’ craniofacial malformations?
Hereditary Vitreoretinopathies

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous
--All have vitreous veils
--All are associated with:
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- Cataracts
- Lattice degeneration

Hereditary Vitreoretinopathies
Ocular disease only
- Wagner’s disease

Associated with systemic disease
- Stickler syndrome

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

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?
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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?
The affected joints are enlarged and hypermobile.

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

Not craniosynostoses

What are the four craniosynostoses?
Hereditary Vitreoretinopathies

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

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)

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Ocular disease only
- Wagner’s disease

Associated with systemic disease
- Stickler syndrome
Hereditary Hyaloideoretinopathies

---

**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 Hyaloideoretinopathies

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

Hereditary Vitreoretinopathies

Ocular disease only
- Wagner’s disease

Associated with systemic disease
- Stickler syndrome

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
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*

*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**

*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**

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’, i.e., the subsequent malformations and functional issues? Micrognathia → glossoptosis → cleft palate → feeding difficulties

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

What does glossoptosis refer to? The position of the tongue being too posterior.

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

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

What does glossoptosis refer to? The position of the organ being too
Hereditary Vitreoretinopathies

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous—All have vitreous veils
—All are associated with:
  —Myopia
  —Glaucoma
  —Cataracts
  —Lattice degeneration

Hereditary Vitreoretinopathies

Ocular disease only
—Wagner’s disease

Associated with systemic disease
—Stickler syndrome

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

What does glossoptosis refer to?
The position of the tongue being too posterior
Hereditary Vitreoretinopathies

Pierre Robin sequence: Glossoptosis
**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

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

---

**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

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

---

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

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

The affected joints are enlarged, and hypermobile

Ocular disease only

Wagner’s disease

Associated with systemic disease

Stickler syndrome
Hereditary Vitreoretinopathies

Stickler syndrome: hypermobile joints
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.

InPRS, what is the single developmental malformation?

Micrognathia

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

Micrognathia → glossoptosis → cleft palate → feeding difficulties

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

What is the non-eponymous name for Stickler syndrome?

'Ocular disease only

Wagner’s disease

Associated with systemic disease

Stickler syndrome, aka...
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 affect which leads to other malformations.

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

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

**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.

Ocular disease only

- Wagner’s disease

Associated with systemic disease

- **Stickler syndrome, aka...**
  ‘Hereditary arthro-ophthalmopathy, Marfanoid variety’
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

Wagner’s disease?

Stickler syndrome?

*Of these two conditions, only one carries a very high risk of retinal detachment. Which one?*
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? 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 Hyaloideoretinopathies with Optically Empty Vitreous

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

Ocular disease only

Wagner’s disease
Jansen’s disease

Associated with systemic disease

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

Are Stickler pts at increased risk of RD?

(Stickler syndrome)
Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

--All have vitreous...veils
--All are associated with:
  --Myopia
  --Cataracts
  --Glucoma
  --Lattice degeneration

Ocular disease only

- Wagner’s disease
- Jansen’s disease

Associated with systemic disease

- ‘Hereditary arthro-ophthalmopathy with stiff joints’ (Weill-Marchesani-like variety)
- ‘Hereditary arthro-ophthalmopathy, Marfanoid variety’ (Stickler syndrome)

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