Hereditary Vitreoretinopathies

- 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, nonocular finding, ring loss
  - Yellowish RD appears w/in 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%
  - Looks like CME, but is dry
  - Split is in RNFL
  - Abnormal cells: Müller cells
  - ERG: Selective loss of b-wave
  - If severe, peripheral changes can lead to RD, vitreous heme

- Norrie Disease
  - X-linked
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  - Associated with MR, Hearing loss
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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

- Hereditary Hyaloideoretinopathies
  - With Optically Empty Vitreous
    - Specific disease
    - Specific disease
    - Specific disease
    - Specific disease
    - Specific disease
    - Group of diseases

- Knobloch Syndrome
  - Specific disease
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

Goldmann-Favre Syndrome

X-Linked Juvenile Retinoschisis

Familial Exudative Vitreoretinopathy

Knobloch Syndrome

Hereditary Vitreoretinopathies

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Goldmann-Favre Syndrome

Norrie Disease

Familial Exudative Vitreoretinopathy
Hereditary Vitreoretinopathies

**X-Linked Juvenile Retinoschisis**

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

**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...TRD...retinal breaks and foveal dragging

--PVD, vitreous traction

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

**Knobloch Syndrome**

Goldmann-Favre Syndrome

Familial Exudative Vitreoretinopathy

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

Norrie Disease
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

Goldmann-Favre Syndrome

Norrie Disease

Familial Exudative Vitreoretinopathy

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

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
Hereditary Vitreoretinopathies

X-linked juvenile retinoschisis: Peripheral retinoschisis
Norrie Disease
--X-linked
--Bilateral congenital blindness
--Associated with:
  --MR
  --[nonocular finding] ring 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...[%]

Goldmann-Favre Syndrome

Norrie Disease

Familial Exudative Vitreoretinopathy

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

Goldmann-Favre Syndrome

Knobloch Syndrome

Knobloch Syndrome

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Goldmann-Favre Syndrome
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

X-Linked Juvenile Retinoschisis
-- Named for peripheral changes, but present in only 50% of cases
-- Foveal schisis present in...100%

Goldmann-Favre Syndrome

Norrie Disease

Familial Exudative Vitreoretinopathy

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

Goldmann-Favre Syndrome

Knobloch Syndrome

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous
Hereditary Vitreoretinopathies

X-linked juvenile retinoschisis: Foveal cysts
X-linked juvenile retinoschisis: Foveal cysts
Norrie Disease
--X-linked--Bilateral congenital blindness--Associated with:
--MR--[nonocular finding ring 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...100%--Looks like CME, but is dry dz
(CME = cystoid macular edema)

Norrie Disease

Goldmann-Favre Syndrome

Familial Exudative Vitreoretinopathy

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

Goldmann-Favre Syndrome

Knobloch Syndrome

Norrie Disease

Familial Exudative Vitreoretinopathy

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous
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

**Familial Exudative Vitreoretinopathy**
--Looks like ROP (but FT and normal respiratory status)
--Hallmark: Failure of temporal retina to vascularize
--Inheritance: AD (check family)
--Peripheral neovascularization leads to TRD, vitreous traction

**Knobloch Syndrome**

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

**Goldmann-Favre 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
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

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
  (RNFL = retinal nerve fiber layer)

Goldmann-Favre Syndrome

Familial Exudative Vitreoretinopathy

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

Goldmann-Favre Syndrome

Norrie Disease

Familial Exudative Vitreoretinopathy

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome
Hereditary Vitreoretinopathies

X-linked juvenile retinoschisis: RNFL schisis
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

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

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.

Goldmann-Favre Syndrome

Norrie Disease

Knobloch Syndrome

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous
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

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:

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

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

Goldmann-Favre Syndrome

Norrie Disease

Hereditary Hyaloideoretinopathies

Goldmann-Favre Syndrome

Norrie Disease

Familial Exudative Vitreoretinopathy

Knobloch 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

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

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**

**X-Linked Juvenile Retinoschisis**
- Named for peripheral changes, but present in only 50% of cases
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- If severe, peripheral changes can lead to RD, vitreous heme

**Goldmann-Favre 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  TRD  retinal breaks and foveal dragging
- PVD, vitreous traction

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

**Knobloch Syndrome**

**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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--Split is in RNFL
--Abnormal cells: Müller cells
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**Goldmann-Favre Syndrome**

**Norrie Disease**

--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 → TRD → retinal breaks and foveal dragging
--PVD, vitreous traction

**Knobloch Syndrome**

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

**Goldmann-Favre Syndrome**

**Norrie Disease**

--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: ERG

Normal

Patient

Absent b-wave

a-wave

b-wave
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%
--Looks like CME, but is dry dz
--Split is in…RNFL
--Abnormal cells: Müller cells
--ERG: Selective loss of…b-wave
--If severe, peripheral changes can lead to…[two problems]

Goldmann-Favre Syndrome

Familial Exudative Vitreoretinopathy
--Looks like…RO (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
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

X-Linked Juvenile Retinoschisis
--Named for peripheral changes, but present in only 50% of cases
--Foveal schisis present in... 100%
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--Split is in... RNFL
--Abnormal cells: Müller cells
--ERG: Selective loss of... b-wave
--If severe, peripheral changes can lead to... RD, vitreous heme

Goldmann-Favre Syndrome

Norrie Disease

Familial Exudative Vitreoretinopathy

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome
Hereditary Vitreoretinopathies

Goldmann-Favre Syndrome
--Macula looks like... but periphery looks like...

Norrie Disease
--X-linked
--Bilateral congenital blindness
--Associated with:
  --MR
  --[nonocular finding 2] ring loss
  --Yellowish RD appears w/in... 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%
--Looks like CME, but is dry dz
--Split is in... RNFL
--Abnormal cells: Müller cells
--ERG: Selective loss of... b-wave
--If severe, peripheral changes can lead to... RD, vitreous heme

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

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... peripheral changes can lead to... RD, vitreous heme
Hereditary Vitreoretinopathies

**Goldmann-Favre Syndrome**
--Macula looks like...XLR, but periphery looks like...RP

**Norrie Disease**
--X-linked
--Bilateral congenital blindness
--Associated with:
  --MR
  --[nonocular finding] ring 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)
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--PVD, vitreous traction

**X-Linked Juvenile Retinoschisis**
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--If severe, peripheral changes can lead to...RD, vitreous heme

**Knobloch Syndrome**

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**
(XLJR = X-linked juvenile retinoschisis)
(RP = retinitis pigmentosa)
Hereditary Vitreoretinopathies

Goldmann-Favre syndrome
**Hereditary Vitreoretinopathies**

**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
  --[nonocular finding 2] ring 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
\[\rightarrow\] TRD
\[\rightarrow\] retinal breaks and foveal dragging
--PVD, vitreous traction

**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: Selective loss of...b-wave
--If severe, peripheral changes can lead to...RD, vitreous heme

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

**Knobloch Syndrome**
Hereditary Vitreoretinopathies

**Norrie Disease**
- X-linked
- Bilateral congenital blindness
- Associated with: MR, Hearing loss
- Yellowish RD appears within weeks of birth
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**X-Linked Juvenile Retinoschisis**
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- Split is in RNFL
- Abnormal cells: Müller cells
- ERG: Selective loss of 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

**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**
- Goldmann-Favre Syndrome
- Norrie Disease
**Hereditary Vitreoretinopathies**

**Goldmann-Favre Syndrome**
--Macula looks like... XLJR, but periphery looks like... RP
--Vitreous is... optically empty
Presents with... nyctalopia

**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

**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: Selective loss of... 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**

**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...RNFL
--Abnormal cells: Müller cells
--ERG: Selective loss of...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

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

Norrie Disease
--X-linked
--Bilateral congenital blindness
--Associated with:
  --MR
  --[nonocular finding 2] ring loss
  --Yellowish RD appears w/in...weeks of birth
  --Lenses & Ks opacify with time--Ends in...phthisis by age 10
Hereditary Vitreoretinopathies

**Goldmann-Favre Syndrome**
--Macula looks like...XLJR, but periphery looks like...RP
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--Presents with...nyctalopia
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**Norrie Disease**
--X-linked
--Bilateral congenital blindness
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--Yellowish RD appears within weeks of birth
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**X-Linked Juvenile Retinoschisis**
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--Looks like CME, but is dry dz
--Split is in...RNFL
--Abnormal cells: Müller cells
--ERG: Selective loss of...b-wave
--If severe, peripheral changes can lead to...RD, vitreous heme

**Knobloch Syndrome**

**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
  \[\text{TRD} \rightarrow \text{retinal breaks and foveal dragging}\]
--PVD, vitreous traction

**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
- Bilateral congenital... blindness
- Associated with:
  - MR
  - [nonocular finding] ring loss
  - Yellowish RD appears w/in... weeks of birth
  - Lenses & Ks opacify with time
  - Ends in... ptysis 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... RD, vitreous heme

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome
Hereditary Vitreoretinopathies

**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
  --[nonocular finding] ring 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→ TRD→ 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:

**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 cell
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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...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 w/in... weeks of birth
--Lenses & Ks opacify with time--Ends in...phthisis by age 10

Knobloch Syndrome
The BCSC Retina book calls Goldmann-Favre a “diffuse photoreceptor dystrophy.” In what ways are photoreceptors affected?
--Rods: Non-functioning

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
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Hereditary Vitreoretinopathies

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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--X-linked
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Vitreoretinal Dystrophies

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What’s up with the disease/syndrome ambiguity?

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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
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- PVD, vitreous traction

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.)
Norrie Disease
--X-linked recessive
--Bilateral congenital blindness
--Associated with:
  --MR
  --Hearing loss
--Yellowish RD appears within weeks of birth
--Lenses & Ks...[same change]
  (K = cornea)

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

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Knobloch Syndrome

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Familial Exudative Vitreoretinopathy
--Looks like...ROP (but FT and normal respiratory status)
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Hereditary Vitreoretinopathies

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Knobloch Syndrome
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Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

Hereditary Vitreoretinopathies

Hereditary Hyaloideoretinopathies
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Knobloch Syndrome

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Knobloch Syndrome

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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Hereditary Vitreoretinopathies
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**Knobloch Syndrome**

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**Goldmann-Favre Syndrome**

**Knobloch Syndrome**
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 (2A, double-headed arrow) with temporal fibrovascular tissue. OS (2B) had a normal fovea and infratemporal retinoschisis. FA OD demonstrated a V-shaped area of avascular/limited perfusion in the temporal retina (2C, arrow), with mild leakage. FA OS showed mild nonperfusion in the far temporal periphery (2D, arrow). After the patient received laser photocoagulation therapy to right eye, he had no further complications, no increase in fibrosis, and no new areas of neovascularization (2E, 2F).
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Knobloch Syndrome

Hereditary Vitreoretinopathies
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### Knobloch Syndrome

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Knobloch Syndrome
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FEVR: Foveal/disc dragging
**Hereditary Vitreoretinopathies**

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(PVD = posterior vitreous detachment)
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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**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

**Knobloch Syndrome**

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

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Knobloch Syndrome
--Classic triad: Occipital encephalocele + high myopia + predisposition to retinal detachment
Knobloch syndrome: Occipital encephalocele
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Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch pts always have a full-blown occipital encephalocele?
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**Hereditary Hyaloideoretinopathies**

**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: Selective loss of b-wave
--If severe, peripheral changes can lead to RD, vitreous heme

**Norrie Disease**
--X-linked recessive
--Bilateral congenital blindness
--Associated with:
  --MR
  --Hearing loss
--Yellowish RD appears within weeks of birth
--Lenses & Ks opacify with time
--End stage: Phthisis by age 10

**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

**Familial Exudative Vitreoretinopathy**
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Hereditary Vitreoretinopathies

Norrie Disease
--X-linked recessive
--Bilateral congenital blindness
--Associated with:
  --MR
  --[nonocular finding 2] ring loss
--Yellowish RD appears w/in… weeks of birth
--Lenses & Ks opacify with time--End stage: Phthisis by age 10

Goldmann-Favre Syndrome
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Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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

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.
Knobloch syndrome: Funky occipital scalp
Hereditary Vitreoretinopathies

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--Peripheral neo → TRD → retinal breaks and foveal dragging
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**Knobloch Syndrome**
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--RPE is atrophic

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

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Hereditary Vitreoretinopathies

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Knobloch syndrome: Atrophic RPE
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Knobloch Syndrome
--Classic triad: Occipital encephalocele + high myopia + predisposition to retinal detachment
--RPE is atrophic
--Irids are structural issue
Hereditary Vitreoretinopathies

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

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

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Goldmann-Favre Syndrome
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Hereditary Hyaloideoretinopathies with Optically Empty Vitreous
The hyaloideoretinopathies get their own slides...

Goldmann-Favre
Syndrome

Norrie Disease

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous
--All have vitreous [finding]
--All are associated with:
--myopia
--glaucoma
--cataracts
--lattice degeneration

The hyaloideoretinopathies get their own slides...

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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Hereditary Vitreoretinopathies

Hereditary Hyaloideoretinopathies
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The hyaloideoretinopathies get their own slides...
Hereditary Hyaloideoretinopathies

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

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Hereditary Vitreoretinopathies

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Hereditary Vitreoretinopathies

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What is unusual about lattice in these conditions?
Hereditary Hyaloideoretinopathies

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---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.
Hereditary Vitreoretinopathies

Radially-oriented lattice degeneration
Hereditary Vitreoretinopathies

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One way of divvying up the hereditary hyaloideoretinopathies

? ?
Hereditary Vitreoretinopathies

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Ocular disease only
Associated with systemic disease

One way of divvying up the hereditary hyaloideoretinopathies
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Ocular disease only

Wagner's disease

Associated with systemic disease
Hereditary Vitreoretinopathies

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Hereditary Vitreoretinopathies

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)

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**Hereditary Hyaloideoretinopathies**

With Optically Empty Vitreous

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**Hereditary Vitreoretinopathies**

Ocular disease only

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Stickler syndrome is strongly associated with a craniofacial malformation—

**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

How do you pronounce Pierre Robin in this context?

**PEA-err roe-BAHN**
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)

Craniosynostoses

Not craniosynostoses

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Ocular disease only

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**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

Stickler syndrome

Associated with systemic disease

Wagner’s disease

Craniosynostoses

Not craniosynostoses

In which group does Pierre Robin sequence belong?
Hereditary Vitreoretinopathies

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Two categories of craniofacial syndrome

Craniosynostoses Not craniosynostoses

---?

---?

---Pierre Robin sequence

---?

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

Ocular disease only

Wagner’s disease

Associated with systemic disease

Stickler syndrome
Hereditary Vitreoretinopathies

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--Goldenhar
--Treacher Collins
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What are the four craniosynostoses?
Hereditary Vitreoretinopathies

---

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

Pierre Robin sequence (PRS)

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

---

**Ocular disease only**

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**Associated with systemic disease**

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**Two categories of craniofacial syndrome**

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**What are the four craniosynostoses?**

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Hereditary Vitreoretinopathies

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- Wagner’s disease

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Pierre Robin sequence: Micrognathia
Hereditary Vitreoretinopathies

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*InPRS, 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

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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

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**glossoptosis**

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- glossoptosis → micrognathia → cleft palate
- cleft palate → feeding difficulties

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

Pierre Robin sequence: Glossoptosis

Micrognathia - a small jaw with a receding chin
Tongue that is large compared to the jaw, resulting in airway obstruction
Hereditary Vitreoretinopathies

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

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Ocular disease only

![Diagram showing ocular disease only and associated with systemic disease]

- Wagner’s disease
- **Stickler syndrome**

Associated with systemic disease
Hereditary Vitreoretinopathies

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Associated with systemic disease

Stickler syndrome
Hereditary Vitreoretinopathies

Stickler syndrome: hypermobile joints
Hereditary Vitreoretinopathies

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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 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.
Hereditary Vitreoretinopathies

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The affected joints are enlarged, and hypermobile

Ocular disease only

- Wagner's disease

Associated with systemic disease

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

**What is the non-eponymous name for Stickler syndrome?**

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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

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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

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

**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)
- 'Hereditary arthro-ophthalmopathy, Marfanoid variety' (Stickler syndrome)

Are Stickler pts at increased risk of RD?
Hereditary Vitreoretinopathies

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

--All have vitreous…veils
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**Associated with systemic disease**

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

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*Are Stickler pts at increased risk of RD?*

Very much so. Even worse, their RDs are associated with large multiple breaks, rendering repair difficult.
Hereditary Vitreoretinopathies

Stickler syndrome: RD