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

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

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
specific disease

Knobloch Syndrome
Hereditary Vitreoretinopathies

X-Linked Juvenile Retinoschisis

Goldmann-Favre Syndrome

Norrie Disease

Familial Exudative Vitreoretinopathy

Knobloch Syndrome

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous
Hereditary Vitreoretinopathies

**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 % of cases

**Goldmann-Favre 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 \( \rightarrow \) TRD \( \rightarrow \) 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**
Norrie Disease
--X-linked--Bilateral congenital blindness--Associated with:
--MR--Hearing loss--Yellowish RD appears w/in 21 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
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 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%

**Goldmann-Favre Syndrome**

**Familial Exudative Vitreoretinopathy**

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

**Knobloch Syndrome**

**Hereditary Vitreoretinopathies**
X-linked juvenile retinoschisis: Foveal cysts
X-linked juvenile retinoschisis: Foveal cysts
Norrie Disease
--X-linked--Bilateral congenital blindness--Associated with:
--MR--Hearing loss--Yellowish RD appears w/in 3-4 weeks of birth--Lenses & Ks opacify with time--Ends in amblyopia, 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)

Goldmann-Favre Syndrome

Norrie Disease

Familial Exudative Vitreoretinopathy

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome
Hereditary Vitreoretinopathies

**X-Linked Juvenile Retinoschisis**

--Named for peripheral changes, but present in only 50% of cases
--Foveal schisis present in...100%
--Looks like CME, but is dry dz
--Split is in...[retinal layer]

**Knobloch Syndrome**

**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
--Peripheral neo TRD
--PVD, vitreous traction

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

**Knobloch Syndrome**

**Goldmann-Favre Syndrome**

--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]
Hereditary Vitreoretinopathies

**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
  
  \(\text{RNFL} = \text{retinal nerve fiber layer}\)

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

**Knobloch Syndrome**

**Goldmann-Favre 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→retinal breaks and foveal dragging
- PVD, vitreous traction
Hereditary Vitreoretinopathies

X-linked juvenile retinoschisis: RNFL schisis
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
--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

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

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch 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%
--Looks like CME, but is dry dz
--Split is in...RNFL
--Abnormal cells:

Goldmann-Favre Syndrome

Norrie Disease

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
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...100%
--Looks like CME, but is dry dz
--Split is in...RNFL
--Abnormal cells: Müller cells

Goldmann-Favre Syndrome

Familial Exudative Vitreoretinopathy

Knobloch Syndrome

Knobloch Syndrome

Goldmann-Favre Syndrome

Norrie Disease

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous
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 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
  -- Peripheral 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
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: Müller cells
--ERG: Selective loss of b-wave

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: 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

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

**Goldmann-Favre Syndrome**

**Knobloch Syndrome**

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

**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]
Hereditary Vitreoretinopathies

**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 RD, vitreous heme

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

**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 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...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...one dz
but periphery looks like...diff dz

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
--Macula looks like...XLJR, but periphery looks like...RP
(XLJR = X-linked juvenile retinoschisis)
(RP = retinitis pigmentosa)

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

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
--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 Vitreoretinopathies

Goldmann-Favre syndrome
Goldmann-Favre Syndrome
--Macula looks like XLJR, but periphery looks like RP
--Vitreous is optically empty

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

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous
Norrie Disease
--X-linked
--Bilateral congenital blindness
--Associated with:
  --MR
  --Hearing loss
  --[nonocular finding] ring 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 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 neophthisis, TRD, retinal breaks and foveal dragging
--PVD, vitreous traction

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

**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

**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 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

**Knobloch Syndrome**

**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...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

**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

**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**
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
--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

Knobloch Syndrome
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 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
--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

Familial Exudative Vitreoretinopathy

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

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

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

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

Knobloch Syndrome

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

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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

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

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

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

Knobloch Syndrome

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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-Troud

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous
Hereditary Vitreoretinopathies

Goldmann-Favre Syndrome
--Macula looks like...XLJR, but periphery looks like...RP
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--Bilateral congenital blindness
--Associated with: MR, Hearing loss
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--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

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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
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- 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

Norrie Disease
- X-linked recessive
- Bilateral congenital...blindness
- Associated with:
  - MR
  - Hearing loss
  - Yellowish RD appears w/in...weeks of birth

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
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--Lenses & Ks [same change]
  (K = cornea)
--Ends in phthisis by age 10

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

Goldmann-Favre Syndrome
--Macula looks like XLJR, but periphery looks like RP
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Hereditary Vitreoretinopathies

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--Looks like…[dz]

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(ROP = Retinopathy of prematurity)
(FT = Full term)

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HEREDITARY VITREORETINOPATHIES

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).
Hereditary Vitreoretinopathies

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  bad thing 1
  and
  bad thing 2
(neo = short for 'neovascularization')
(TRD = tractional retinal detachment)
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FEVR: Foveal/disc dragging
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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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--Classic triad: predisposition to location, and CNS developmental prob + refractive prob + retinal prob

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

Knobloch syndrome: Funky occipital scalp
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
--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

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

**Knobloch Syndrome**

--Classic triad: Occipital encephalocele + high myopia + predisposition to retinal detachment
--RPE is...
Norrie Disease
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Familial Exudative Vitreoretinopathy
--Looks like...ROP--but FT and w/ normal respiratory status
--Hallmark: Failure of...temporal retina to vascularize
--Inheritance: AD, AR, X-linked
--Peripheral neo → TRD → retinal breaks and foveal dragging
--PVD, vitreous traction present

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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

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--Irides are structural issue

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Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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

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Hereditary Vitreoretinopathies
Hereditary Vitreoretinopathies

Knobloch syndrome: Cryptless irides
Hereditary Vitreoretinopathies

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The hyaloideoretinopathies get their own slides...

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**Hereditary Hyaloideoretinopathies**
--All have vitreous...
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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 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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  --Cataracts
Hereditary Vitreoretinopathies

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

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  --Glaucoma
  --[abn retinal finding]
Hereditary Vitreoretinopathies

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Hereditary Vitreoretinopathies

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What is unusual about lattice in these conditions?
Hereditary Vitreoretinopathies

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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

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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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
Hereditary Hyaloideoretinopathies

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  --Cataracts
  --Glaucma
  --Lattice degeneration

Ocular disease only
  Wagner's disease

Associated with systemic disease
Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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Associated with systemic disease

  Stickler syndrome
Hereditary Vitreoretinopathies

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

Ocular disease only
- Wagner's disease

Associated with systemic disease
- Stickler syndrome
Hereditary Vitreoretinopathies

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

Ocular disease only
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Hereditary Vitreoretinopathies

Stickler syndrome is strongly associated with systemic disease. Which one?

Pierre Robin sequence (PRS)

Ocular disease only

Wagner’s disease

Associated with systemic disease

Stickler syndrome

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,’ 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.

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

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Pierre Robin sequence (PRS)

Two categories of craniofacial syndrome

Ocular disease only
- Wagner’s disease

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

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Craniosynostoses

Not craniosynostoses

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Stickler syndrome
Hereditary Hyaloideoretinopathies

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**

Stickler syndrome is associated with [systemic disease](#).

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Two categories of craniofacial syndrome

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Hereditary Vitreoretinopathies

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Ocular disease only
  
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Associated with systemic disease
  
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Two categories of craniofacial syndrome

Craniosynostoses

Not craniosynostoses
  
  - Wagner’s disease
  
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What are the other three ‘not craniosynostosis’ craniofacial malformations?
Hereditary Vitreoretinopathies

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

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**Associated with systemic disease**
- **Stickler syndrome**

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Two categories of craniofacial syndrome

- Craniosynostoses
- Not craniosynostoses

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

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

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Stickler syndrome is strongly associated with a craniofacial malformation—who one? Pierre Robin sequence (PRS)
Hereditary Hyaloideoretinopathies

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Two categories of craniofacial syndrome

Craniosynostoses

Not craniosynostoses

What are the four craniosynostoses?

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

Craniosynostoses

Not craniosynostoses

-? -? -? -?

Ocular disease only

Associated with systemic disease

Stickler syndrome

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

Craniosynostoses Not craniosynostoses

-? -? -? -?

Ocular disease only

Associated with systemic disease

Stickler syndrome

- Wagner's disease

What are the four craniosynostoses?

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

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Ocular disease only

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Stickler syndrome

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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)

---

**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

Micrognathia: The affected joints are enlarged and hypermobile.
Hereditary Vitreoretinopathies

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

Wagner’s disease

Associated with systemic disease

*Stickler syndrome*
Hereditary Vitreoretinopathies

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Ocular disease only
- Wagner’s disease

Associated with systemic disease
- Stickler syndrome
Hereditary Vitreoretinopathies

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Micrognathia

What does micrognathia mean?

- Micrognathia

Ocular disease only
- Wagner’s disease

Associated with systemic disease
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Hereditary Vitreoretinopathies

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

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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

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Ocular disease only

- Wagner’s disease

Associated with systemic disease

- Stickler syndrome
**Hereditary Vitreoretinopathies**

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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

Hereditary Hyaloideoretinopathies

--All have vitreous...veils

--All are associated with:

- Myopia
- Glaucoma
- Cataracts
- Lattice degeneration

Hereditary Vitreoretinopathies

--Ocular disease only

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Micrognathia

And what is the ‘sequence’, ie, the single developmental malformation 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
Hereditary Vitreoretinopathies

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Pierre Robin sequence (PRS)

With regard to congenital anomalies, what is meant by the term sequence?
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In PRS, what is the ‘single developmental malformation’ that triggers the sequence?
Micrognathia

And what is the ‘sequence’, i.e., the malformations that result from the initial 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

Ocular disease only
Wagner’s disease

Associated with systemic disease
Stickler syndrome
Hereditary Vitreoretinopathies

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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 chain reaction of additional malformations and functional issues?

| Micrognathia | glossoptosis | The position of the tongue being too posterior |

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

The affected joints are enlarged and hypermobile

What does glossoptosis refer to?

Ocular disease only  
Wagner’s disease

Associated with systemic disease  
Stickler syndrome
Hereditary Vitreoretinopathies

Pierre Robin sequence: Glossoptosis
Hereditary Vitreoretinopathies

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Micrognathia → glossoptosis → cleft palate → feeding difficulties

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

Ocular disease only

Associated with systemic disease

- Wagner’s disease

- Stickler syndrome
Hereditary Vitreoretinopathies

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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 **size**, and **tight vs loose**

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Ocular disease only

- Wagner’s disease

Associated with systemic disease

- **Stickler syndrome**
Hereditary Vitreoretinopathies

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**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

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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.

*What is the non-eponymous name for Stickler 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

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)

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Micrognathia

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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, 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
  --Glaucma
  --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

Are Stickler pts at increased risk of RD?

Associated with systemic disease

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

‘Hereditary arthro-ophthalmopathy, Marfanoid variety’ (Stickler syndrome)
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)

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 arthro-ophthalmopathy, Marfanoid variety’ (Stickler syndrome)
Hereditary Vitreoretinopathies

Stickler syndrome: RD