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 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

specific disease

specific disease

specific disease

specific disease

group of diseases

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

Hereditary Vitreoretinopathies

X-Linked Juvenile Retinoschisis
--Named for peripheral changes, but present in only 50% of cases
--Foveal schisis present in…100%--Looks like CME, but is dry
--Split is in…RNFL--Abnormal cells: Müller cells--ERG: 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
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Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Goldmann-Favre Syndrome

Norrie Disease

Knobloch Syndrome

Goldmann-Favre Syndrome

Norrie Disease

Familial Exudative Vitreoretinopathy

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome
### Hereditary Vitreoretinopathies

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<td>X-linked, bilateral congenital blindness, associated with MR, deafness, yellow RD appearing within weeks of birth, lenses &amp; Ks opacify with time, ends in phthisis by age 10</td>
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#### Goldmann-Favre Syndrome
- Named for the disease of Frederick Goldmann and François Favre
- Characterized by progressive retinal atrophy, coloboma, cataracts, and mental retardation
- Associated with mutations in the RGC1 gene

#### Norrie Disease
- X-linked, bilateral congenital blindness
- Associated with microphthalmia, mental retardation, hearing loss, and other congenital anomalies
- Yellowish RD appears within weeks of birth
- Lenses and Ks opacify with time
- Ends in phthisis by age 10

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- Named for peripheral changes, but present in only 50% of cases
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#### Hereditary Hyaloideoretinopathies with Optically Empty Vitreous
- Presents with nyctalopia
- Is also known as enhanced S-cone dz/syndrome

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Hereditary Vitreoretinopathies

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

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  --Lenses & Ks opacify with time--Ends in...phthisis by age 10

**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

**Knobloch Syndrome**

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

**Goldmann-Favre Syndrome**

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**Familial Exudative Vitreoretinopathy**

**Knobloch Syndrome**
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Hereditary Vitreoretinopathies

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-- Foveal schisis present in... [%]

**Goldmann-Favre Syndrome**

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**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

**Knobloch Syndrome**

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--Goldmann-Favre Syndrome

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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 (cystoid macular edema)

Goldmann-Favre Syndrome

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--Split is in... [retinal layer]

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Knobloch Syndrome
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--Split is in...RNFL
(retinal nerve fiber layer)

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.

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Familial Exudative Vitreoretinopathy

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**Knobloch Syndrome**

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**Knobloch Syndrome**

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Hereditary Vitreoretinopathies

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Hereditary Vitreoretinopathies

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- Knobloch Syndrome

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--ERG: Selective loss of...b-wave
--If severe, peripheral changes can lead to...[two problems]

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Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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Knobloch Syndrome
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Hereditary Vitreoretinopathies

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Goldmann-Favre Syndrome

Familial Exudative Vitreoretinopathy

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

Goldmann-Favre Syndrome

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

Familial Exudative Vitreoretinopathy
Hereditary Vitreoretinopathies

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**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**
Goldmann-Favre Syndrome

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

Norrie Disease

(X-linked juvenile retinoschisis)
(Retinitis pigmentosa)

X-Linked Juvenile Retinoschisis

--Named for peripheral changes, but present in only 50% of cases
--Foveal schisis present in...100%
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Knobloch Syndrome

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Hereditary Vitreoretinopathies
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--Macula looks like...XLJ, but periphery looks like...RP
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--Is also known as enhanced S-cone dz/syndrome

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  $\rightarrow$ TRD
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Hereditary Vitreoretinopathies

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**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

**Knobloch Syndrome**

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Presents with…nyctalopia
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Knobloch Syndrome
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Hereditary Vitreoretinopathies

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Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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Hereditary Vitreoretinopathies
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Hereditary Hyaloideoretinopathies with Optically Empty Vitreous
### Hereditary Vitreoretinopathies

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Vitreoretinal Dystrophies

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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What’s up with the disease/syndrome ambiguity?

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

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--Presents with nyctalopia
--Is also known as enhanced S-cone dz/syndrome

**Norrie Disease**
--X-linked recessive
--Bilateral congenital blindness
--Associated with:
  --[nonocular finding 1]
  --[nonocular finding 2]

**Familial Exudative Vitreoretinopathy**

**Knobloch Syndrome**

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**
Hereditary Vitreoretinopathies

**Goldmann-Favre Syndrome**
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--Presents with...nyctalopia
--Is also known as enhanced S-cone dz/syndrome

**Norrie Disease**
--X-linked recessive
--Bilateral congenital...blindness
--Associated with:
--MR (= mental retardation)
--Hearing loss

**Familial Exudative Vitreoretinopathy**

**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**

**Goldmann-Favre Syndrome**

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

**Norrie Disease**

--X-linked recessive
--Bilateral congenital...blindness
--Associated with:
  --MR
  --Hearing loss
--Yellowish RD appears w/in...
  [time frame relative to birth]

**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

**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—Ends in phthisis by age 10

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**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

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**Goldmann-Favre Syndrome**

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Hereditary Vitreoretinopathies

Goldmann-Favre Syndrome

- Macula looks like XLJR, but periphery looks like RP
- Vitreous is optically empty
- Presents with nyctalopia
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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
  - Ends in phthisis by age 10

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

X-Linked Juvenile Retinoschisis

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Knobloch Syndrome

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

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

Goldmann-Favre Syndrome
--Macula looks like...XLJR, but periphery looks like...RP
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Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

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**Hereditary Vitreoretinopathies**
Hereditary Vitreoretinopathies

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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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**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

---
Familial Exudative Vitreoretinopathy
--Looks like…[dz]
--Normal respiratory status
--Hallmark: Failure of…temporal retina to vascularize
--Inheritance: AD (check family)
--Peripheral neo  TRD  retinal breaks and foveal dragging
--PVD, vitreous traction

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--X-linked recessive
--Bilateral congenital…blindness
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Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome

Hereditary Vitreoretinopathies
Familial Exudative Vitreoretinopathy
--Looks like...ROP--but FT and w/ normal respiratory status
--Hallmark: Failure of...temporal retina to vascularize
--Inheritance: AD (check family)--Peripheral neo TRD
-> retinal breaks and foveal dragging
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Goldmann-Favre Syndrome

Norrie Disease

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

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

### X-Linked Juvenile Retinoschisis

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- Yellowish RD appears w/in... weeks of birth
- Lenses & Ks opacify with time
- Endstage: Phthisis by age 10

### Familial Exudative Vitreoretinopathy

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

### Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

### Knobloch Syndrome
**Hereditary Vitreoretinopathies**

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- Endstage: Phthisis by age 10

**Familial Exudative Vitreoretinopathy**
- Looks like ROP but FT and w/ normal respiratory status
- Hallmark: Failure of temporal retina to vascularize

**Knobloch Syndrome**

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

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- Endstage: Phthisis by age 10

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

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**Knobloch Syndrome**

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

**Familial Exudative Vitreoretinopathy**
--Looks like ROP--but FT and w/ normal respiratory status
--Hallmark: Failure of...temporal retina to vascularize
--Inheritance:
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
--Endstage: 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
--Looks like...ROP--but FT and w/ normal respiratory status
--Hallmark: Failure of...temporal retina to vascularize
--Inheritance: AD, AR, X-linked

Knobloch Syndrome

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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

Hereditary Vitreoretinopathies
Hereditary Vitreoretinopathies

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--Foveal schisis present in 100%
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--Split is in RNFL
--Abnormal cells: Müller cells
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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 nystagmus
--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
--Endstage: Phthisis by age 10

**Familial Exudative Vitreoretinopathy**
--Looks like ROP--but FT and w/ normal respiratory status
--Hallmark: Failure of temporal retina to vascularize
--Inheritance: AD, AR, X-linked
--Peripheral neo TRD

**Knobloch Syndrome**

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous
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
--Endstage: 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
--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

Knobloch Syndrome

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous
Norrie Disease
--X-linked recessive
--Bilateral congenital blindness
--Associated with:
  --MR
  --Hearing loss
  --Yellowish RD appears within weeks of birth
--Endstage: Phthisis by age 10

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

Knobloch Syndrome
--Posterior vitreous detachment

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Familial Exudative Vitreoretinopathy
--Looks like ROP--but FT and with 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

X-Linked Juvenile Retinoschisis
--Named for peripheral changes, but present in only 50% of cases
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--Looks like CME, but is dry dz
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--Abnormal cells: Müller cells
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--If severe, peripheral changes can lead to RD, vitreous heme
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.

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
- Endstage: Phthisis by age 10

Familial Exudative Vitreoretinopathy

- Looks like...ROP--but FT and w/ normal respiratory status
- Hallmark: Failure of...temporal retina to vascularize
- Inheritance: AD, AR, X-linked
- Peripheral neo → TND → retinal breaks and foveal dragging
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Knobloch Syndrome

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous
Hereditary Vitreoretinopathies

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- Inheritance: AD, AR, X-linked
- Peripheral neo → TRD → retinal breaks and foveal dragging
- PVD, vitreous traction present

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

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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

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

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

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

Knobloch Syndrome
--Classic triad:
  --Occipital encephalocele
  --High myopia
  --Predisposition to retinal detachment

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

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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

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

Knobloch Syndrome
--Classic triad: Occipital encephalocele + high myopia + predisposition to Retinal detachment

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 kids head with the hair pushed out of the way to reveal the scalp, go with Knobloch syndrome)

Familial Exudative Vitreoretinopathy
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Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Knobloch Syndrome
Hereditary Vitreoretinopathies

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- Associated with: MR, Hearing loss
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Knobloch Syndrome
--Classic triad: Occipital encephalocele + high myopia + predisposition to retinal detachment
--RPE is atrophic

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

Hereditary Vitreoretinopathies
**Hereditary Vitreoretinopathies**

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

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

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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

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Hereditary Vitreoretinopathies
**Hereditary Vitreoretinopathies**

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**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

**Hereditary Hyaloideoretinopathies** get their own slides...

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

**Goldmann-Favre Syndrome**

**Norrie Disease**

**X-Linked Juvenile Retinoschisis**

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

**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 Hyaloideoretinopathies with Optically Empty Vitreous

--All have vitreous...veils
--All are associated with:
    --[refractive status]
Hereditary Hyaloideoretinopathies

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 Hyaloideoretinopathies

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

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

--All have vitreous...veils
--All are associated with:
  --Myopia
  --Cataracts
  --Glaucoma
  --[abn retinal finding]
Hereditary Vitreoretinopathies

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

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

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

### What is unusual about lattice in these conditions?
Hereditary Vitreoretinopathies

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

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

*What is unusual about lattice in these conditions?*
It has a **RADIAL** orientation, i.e., the lattice points toward the optic nerve, as opposed to the circumferential orientation characteristic of typical lattice
A basic distinction between two sorts of hereditary hyaloideoretinopathies yada yada yada

**Hereditary Hyaloideoretinopathies with Optically Empty Vitreous**

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

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

Ocular disease only

Associated with systemic disease
Hereditary Vitreoretinopathies

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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  --Myopia
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Ocular disease only

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Hereditary Vitreoretinopathies

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  --Myopia
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  --Glaucoma
  --Lattice degeneration

Ocular disease only

Associated with systemic disease

Wagner's disease
Hereditary Vitreoretinopathies

---

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

--- All have vitreous...veils
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--- Myopia
--- Cataracts
--- Glaucoma
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Ocular disease only

Associated with systemic disease

Wagner's disease

?
Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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Ocular disease only

Wagner's disease

Associated with systemic disease

Stickler syndrome
Hereditary Vitreoretinopathies

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    - Lattice degeneration

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?
Pierre Robin sequence (PRS)

Ocular disease only
- Wagner’s disease

Associated with systemic disease
- Stickler syndrome
Hereditary Vitreoretinopathies

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

How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence?

Ocular disease only
- Wagner’s disease

Associated with systemic disease
- Stickler syndrome
Hereditary Vitreoretinopathies

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

**Pierre Robin sequence** (PRS)

_How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence?_

PEA-err roe-BAHN

Ocular disease only

- Wagner’s disease

Associated with systemic disease

- Stickler syndrome
Hereditary Vitreoretinopathies

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

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

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

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

Micrognathia → glossoptosis → cleft palate → feeding difficulties

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

The affected joints are enlarged and hypermobile
Hereditary Vitreoretinopathies

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

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

Ocular disease only
- Wagner’s disease

Associated with systemic disease
- Stickler syndrome
Hereditary Vitreoretinopathies

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

Ocular disease only
- Wagner’s disease

Associated with systemic disease
- Stickler syndrome
Hereditary Vitreoretinopathies

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

Pierre Robin sequence (PRS)

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

It means that a single developmental malformation initiates a ‘domino effect’ which leads to other malformations, which in turn lead to significant functional issues

*In PRS, what is the ‘single developmental malformation’ that triggers the sequence?*

Micrognathia

---

Ocular disease only

- Wagner’s disease

Associated with systemic disease

- **Stickler syndrome**
Hereditary Vitreoretinopathies

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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

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**

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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?*

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*In PRS, what is the ‘single developmental malformation’ that triggers the sequence?*

Micrognathia

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

Micrognathia → glossoptosis → cleft palate → feeding difficulties

Ocular disease only

Associated with systemic disease

Wagner’s disease

Stickler syndrome
**Hereditary Vitreoretinopathies**

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

*And what is the ‘sequence,’ ie, the subsequent malformations and functional issues?*  
Micrognathia $\rightarrow$ glossoptosis $\rightarrow$ cleft palate $\rightarrow$ feeding difficulties

---

Ocular disease only  
- Wagner's disease

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

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

**Micrognathia**

- glossoptosis
- cleft palate
- feeding difficulties

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

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

Ocular disease only

- Wagner’s disease

Associated with systemic disease

- *Stickler syndrome*
Hereditary Vitreoretinopathies

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

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

**In PRS, what is the ‘single developmental malformation’ that triggers the sequence?**
Micrognathia

And what is the ‘sequence,’ i.e., the additional malformations that arise from this single developmental malformation?

- Micrognathia ➔ glossoptosis ➔ cleft palate ➔ feeding difficulties

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

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

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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 disorders/syndrome that arise from that single issue?

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

Ocular disease only
- Wagner’s disease

Associated with systemic disease
- Stickler syndrome
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Ocular disease only

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

Micrognathia → glossoptosis → cleft palate → feeding difficulties

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

The affected joints are **tight vs loose**.

---

Ocular disease only

- Wagner’s disease

Associated with systemic disease

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Hereditary Vitreoretinopathies

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**Ocular disease only**

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Hereditary Vitreoretinopathies

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And what are the subsequent malformations and functional issues?
Micrognathia ➔ glossoptosis ➔ cleft palate ➔ feeding difficulties

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

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

Ocular disease only
  Wagner’s disease

Associated with systemic disease
  Stickler syndrome, aka...
Hereditary Vitreoretinopathies

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

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It means that a single developmental malformation initiates a 'domino effect' which leads to other malformations and functional issues. In PRS, what is the 'single developmental malformation' that triggers the sequence?
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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

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

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

Ocular disease only

Wagner’s disease

Associated with systemic disease

Stickler syndrome

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

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

Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

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

Ocular disease only

Wagner’s disease

Associated with systemic disease

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

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

(Stickler syndrome)