These three are all secondary to abnormal closure of the embryonic optic fissure

Optic Nerve…

Optic…

[Two-words] Disc
These three are all secondary to abnormal closure of the embryonic optic fissure

Optic Nerve Coloboma

Optic Pit/Hole

Morning-Glory Disc

Developmental Abnormalities of the Optic Nerve Head
Developmental Abnormalities of the Optic Nerve Head

Optic Nerve Coloboma

Optic Pit/Hole

Morning-Glory Disc

These three are all secondary to abnormal closure of the embryonic optic fissure

Megalo-…

Developmental Abnormalities of the Optic Nerve Head

1) May resemble deep cupping

2) Can be bilateral, asymmetric

3) Part of the CHARGE association

Optic Pit/Hole -- Associated with serous RD in adulthood

Morning-Glory Disc -- DFE reveals:

1) Funnel-shaped

2) Number of vessels crossing the rim seems abnormally high

-- Tissue is contractile, so cup seems to open and close (like its namesake, the morning-glory flower)

-- VA usually 20/200, but can be 20/20- NLP

-- 1/3 develop serous RD
Developmental Abnormalities of the Optic Nerve Head

Megalopapilla

-- Abnormally large diameter of disc and cup
-- VF testing may reveal an enlarged blind spot

Optic Nerve Coloboma

1) May resemble deep cupping
2) Can be bilateral, asymmetric
3) Part of the CHARGE association

Optic Pit/Hole

-- Associated with serous RD in adulthood

Morning-Glory Disc

-- DFE reveals:
  1) Funnel-shaped
  2) Number of vessels crossing the rim seems abnormally high
-- Tissue is contractile, so cup seems to open and close (like its namesake, the morning-glory flower)
-- VA usually 20/20, but can be 20/20->NLP
-- 1/3 develop serous RD

These three are all secondary to abnormal closure of the embryonic optic fissure
Developmental Abnormalities of the Optic Nerve Head

These three are all secondary to abnormal closure of the embryonic optic fissure

- Megalopapilla
- Optic Nerve Coloboma
- Optic Pit/Hole
- Morning-Glory Disc
Developmental Abnormalities of the Optic Nerve Head

- Myelinated RNFL
  - Myelin normally starts at LGN, ends at lamina cribrosa
  - Can be patchy, discontinuous
  - Corresponding VF has absolute scotoma

- Megalopapilla
  - Abnormally large diameter of disc and cup
  - VF testing may reveal an enlarged blind spot

- Optic Nerve Coloboma
- Optic Pit/Hole
- Morning-Glory Disc

These three are all secondary to abnormal closure of the embryonic optic fissure
Developmental Abnormalities of the Optic Nerve Head

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- Optic Nerve Coloboma
  - 1) May resemble deep cupping
  - 2) Can be bilateral, asymmetric
  - 3) Part of the CHARGE association

- Optic Pit/Hole
  - Associated with serous RD in adulthood

- Morning-Glory Disc
  - DFE reveals:
    1) Funnel-shaped
    2) Number of vessels crossing the rim seems abnormally high
  - Tissue is contractile, so cup seems to open and close (like its namesake, the morning-glory flower)
  - VA usually 20/20, but can be 20/20->NLP
  - 1/3 develop serous RD

- Optic Nerve Coloboma
  - Abnormally low number of axons
  - DFE: Small pale disc with double ring sign
  - VA 20/20->NLP
  - VF defects invariably present
  - Remember the 4 D’s (more on this shortly)

These three are all secondary to abnormal closure of the embryonic optic fissure
Myelinated RNFL
-- Myelin normally starts at LGN, ends at lamina cribrosa
-- Can be patchy, discontinuous
-- Corresponding VF has absolute scotoma

Megalopapilla
-- Abnormally large diameter of disc and cup
-- VF testing may reveal an enlarged blind spot

Optic Nerve Hypoplasia
-- Abnormally low number of axons
-- DFE: Small pale disc with double ring sign
-- VA: 20/20 -> NLP
-- VF defects invariably present
-- Remember the 4 D's (more on this shortly)

Optic Nerve Coloboma
1) May resemble deep cupping
2) Can be bilateral, asymmetric
3) Part of the CHARGE association

Optic Pit/Hole
-- Associated with serous RD in adulthood

Morning-Glory Disc
-- DFE reveals:
  1) Funnel-shaped
  2) Number of vessels crossing the rim seems abnormally high
-- Tissue is contractile, so cup seems to open and close (like its namesake, the morning-glory flower)
-- VA usually 20/200, but can be 20/20 -> NLP
-- 1/3 develop serous RD

Developmental Abnormalities of the Optic Nerve Head

These three are all secondary to abnormal closure of the embryonic optic fissure
Developmental Abnormalities of the Optic Nerve Head

These three are all secondary to abnormal closure of the embryonic optic fissure

Optic Nerve Coloboma

Optic Pit/Hole

Morning-Glory Disc

Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

[Two-words] Syndrome

These three are all secondary to abnormal closure of the embryonic optic fissure
Developmental Abnormalities of the Optic Nerve Head

- Myelinated RNFL
  - Myelin normally starts at LGN, ends at lamina cribrosa
  - Can be patchy, discontinuous
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  - Abnormally large diameter of disc and cup
  - VF testing may reveal an enlarged blind spot

- Optic Nerve Hypoplasia
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  - May resemble deep cupping
  - Can be bilateral, asymmetric
  - Part of the CHARGE association

- Optic Pit/Hole
  - Associated with serous RD in adulthood

- Morning-Glory Disc
  - DFE reveals:
    1) Funnel-shaped
    2) Number of vessels crossing the rim seems abnormally high
  - Tissue is contractile, so cup seems to open and close (like its namesake, the morning-glory flower)
  - VA usually 20/200, but can be 20/20->NLP
  - 1/3 develop serous RD

- Tilted-Disc Syndrome
  1) Superior pole appears elevated, inferior recessed
  2) Associated with situs inversus of retinal vessels
  3) Fundus abnormality produces myopic astigmatism
  4) VF testing reveals bitemporal hemianopia that doesn't respect the vertical and may resolve with refraction

These three are all secondary to abnormal closure of the embryonic optic fissure.
Developmental Abnormalities of the Optic Nerve Head

These three are all secondary to abnormal closure of the embryonic optic fissure

Optic Nerve Coloboma
1) May resemble… *common acquired appearance*
2) 
3)

Optic Pit/Hole

Morning-Glory Disc

Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome
Developmental Abnormalities of the Optic Nerve Head

Optic Nerve Coloboma
1) May resemble...deep cupping
2)
3)

Optic Pit/Hole

Morning-Glory Disc

These three are all secondary to abnormal closure of the embryonic optic fissure

Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

These three are all secondary to abnormal closure of the embryonic optic fissure

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Optic nerve coloboma OD. Note the well-demarcated inferior excavation with thinning of the inferior neuroretinal rim and preservation of the superior rim. ONH OS is normal.
Developmental Abnormalities of the Optic Nerve Head

Optic nerve coloboma: More examples
Developmental Abnormalities of the Optic Nerve Head

Optic Nerve Coloboma
1) May resemble... **deep cupping**
2) Can be... [*laterality*]
3)

Optic Pit/Hole

Morning-Glory Disc

These three are all secondary to abnormal closure of the embryonic optic fissure

Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

1/3 develop... serous RD
Developmental Abnormalities of the Optic Nerve Head

Optic Nerve Coloboma
1) May resemble...**deep cupping**
2) Can be...**bilateral, asymmetric**
3)

Optic Pit/Hole

Morning-Glory Disc

These three are all secondary to abnormal closure of the embryonic optic fissure

Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

- Myelin normally starts at LGN, ends at lamina cribrosa
- Can be patchy, discontinuous
- Corresponding VF has absolute scotoma

- Abnormally large diameter of disc and cup
- VF testing may reveal an enlarged blind spot

- Abnormally low number of axons
- DFE: Small pale disc with double ring sign
- VA 20/20<>NLP
- VF defects invariably present
- Remember the 4 D’s (more on this shortly)

- Associated with serous RD in adulthood
- DFE reveals:
  1) Funnel-shaped...
  2) Number of vessels crossing the rim seems abnormally high
- Tissue is contractile, so cup seems to open and close (like its namesake, the morning-glory flower)
- VA usually 20/200, but can be 20/20<>NLP
- 1/3 develop serous RD
Developmental Abnormalities of the Optic Nerve Head

These three are all secondary to abnormal closure of the embryonic optic fissure.

Optic Nerve Coloboma
1) May resemble...deep cupping
2) Can be...bilateral, asymmetric
3) Part of the...association

Optic Pit/Hole

Morning-Glory Disc

- Myelinated RNFL
  - Myelin normally starts at LGN, ends at lamina cribrosa
  - Can be patchy, discontinuous
  - Corresponding VF has absolute scotoma

- Meegalopapilla

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- Megalopapilla

- Megalopapilla

- Megalopapilla

Optic Nerve Hypoplasia
- Abnormally low number of axons
- DFE: Small pale disc with double ring sign
- VA: 20/20->NLP
- VF defects invariably present
- Remember the 4 D's (more on this shortly)

Optic Pit/Hole
- Associated with serous RD in adulthood

Morning-Glory Disc
- DFE reveals:
  1) Funnel-shaped...
  2) Number of vessels crossing the rim seems abnormally high
- Tissue is contractile, so cup seems to...open and close (like its namesake, the morning-glory flower)
- VA usually...20/200, but can be...
  20/20->NLP
- 1/3 develop...serous RD

Tilted-Disc Syndrome
1) Superior pole appears...elevated
2) Inferior...recessed
3) Fundus abnormality produces...myopic astigmatism
4) VF testing reveals...bitemporal hemianopia that doesn't...

Tilted-Disc Syndrome
1) Superior pole appears...elevated
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These three are all secondary to abnormal closure of the embryonic optic fissure

Optic Nerve Coloboma
1) May resemble...deep cupping
2) Can be...bilateral, asymmetric
3) Part of the...CHARGE association

Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Morning-Glory Disc

Tilted-Disc Syndrome
Developmental Abnormalities of the Optic Nerve Head

- Myelinated RNFL
  - Myelin normally starts at LGN, ends at lamina cribrosa
  - Can be patchy, discontinuous
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- Megalopapilla
  - Abnormally large diameter of disc and cup
  - VF testing may reveal an enlarged blind spot

- Optic Nerve Hypoplasia
  - Abnormally low number of axons
  - DFE: Small pale disc with double ring sign
  - VA 20/20->NLP
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  - Remember the 4 D’s (more on this shortly)

- Optic Pit/Hole
  - Associated with serous RD in adulthood

- Morning-Glory Disc
  - DFE reveals:
    1) Funnel-shaped…
    2) Number of vessels crossing the rim seems abnormally…
    3) Tissue is… contractile, so cup seems to… open and close (like its namesake, the morning-glory flower)
  - VA usually… 20/200, but can be… 20/20->NLP
  - 1/3 develop… serous RD

These three are all secondary to abnormal closure of the embryonic optic fissure

What is the CHARGE association?

- Coloboma
- Heart abnormalities
- Choanal atresia
- Retardation
- Genitourinary abnormalities
- Ear abnormalities

Optic Nerve Coloboma
1) May resemble…deep cupping
2) Can be…bilateral, asymmetric
3) Part of the…CHARGE association

Optic Pit/Hole

Morning-Glory Disc

Tilted-Disc Syndrome
Developmental Abnormalities of the Optic Nerve Head

- Myelinated RNFL:
  - Myelin normally starts at LGN, ends at lamina cribrosa.
  - Can be patchy, discontinuous.
  - Corresponding VF has absolute scotoma.

- Megalopapilla:
  - Abnormally large diameter of disc and cup.
  - VF testing may reveal an enlarged blind spot.

- Optic Nerve Hypoplasia:
  - Abnormally low number of axons.
  - DFE: Small pale disc with double ring sign.
  - VA: 20/20 <-> NLP.
  - VF defects invariably present.
  - Remember the 4 D's (more on this shortly).

- Optic Pit/Hole:
  - Associated with serous RD in adulthood.

- Morning-Glory Disc:
  - DFE reveals:
    1) Funnel-shaped.
    2) Number of vessels crossing the rim seems abnormally high.
  - Tissue is contractile, so cup seems to open and close (like its namesake, the morning-glory flower).
  - VA usually 20/200, but can be 20/20 <-> NLP.
  - 1/3 develop serous RD.

- These three are all secondary to abnormal closure of the embryonic optic fissure.

- Optic Nerve Coloboma:
  1) May resemble deep cupping.
  2) Can be bilateral, asymmetric.
  3) Part of the CHARGE association.

- What is the CHARGE association?
  - Coloboma
  - Heart abnormalities
  - Choanal Atresia
  - Retardation
  - Genitourinary abnormalities
  - Ear abnormalities

- Optic Pit/Hole

- Tilted-Disc Syndrome
  - Superior pole appears elevated, inferior recessed.
  - Associated with situs inversus of retinal vessels.
  - Fundus abnormality produces myopic astigmatism.
  - VF testing reveals bitemporal hemianopia that doesn't respect the vertical and may resolve with refraction.

- Morning-Glory Disc

- Tilted-Disc Syndrome

- Hypoplasia
Developmental Abnormalities of the Optic Nerve Head

A very subtle coloboma OS (note the ‘tongue’ of relative pallor of the RPE and choroid just below and slightly nasal to the nerve) in a child with the CHARGE association.
A very subtle coloboma OS (note the ‘tongue’ of relative pallor of the RPE and choroid just below and slightly nasal to the nerve) in a child with the CHARGE association. Although functionally insignificant, this sign has as much diagnostic importance as a marked coloboma.
Developmental Abnormalities of the Optic Nerve Head

These three are all secondary to abnormal closure of the embryonic optic fissure

**Optic Nerve Coloboma**
1) May resemble... **deep cupping**
2) Can be... **bilateral, asymmetric**
3) Part of the... **CHARGE association**

**Optic Pit/Hole**
--Associated with... **[retinal condition, and age]**

**Morning-Glory Disc**

**Megalopapilla**

**Myelinated RNFL**

**Optic Nerve Hypoplasia**

**Tilted-Disc Syndrome**
1) Superior pole appears... **elevated**
2) Inferior... **recessed**
3) Fundus abnormality produces... **myopic astigmat**
4) VF testing reveals... **bitemporal hemianopia** that doesn't... **resolve with refraction**
Developmental Abnormalities of the Optic Nerve Head

These three are all secondary to abnormal closure of the embryonic optic fissure

Optic Nerve Coloboma
1) May resemble... deep cupping
2) Can be... bilateral, asymmetric
3) Part of the... CHARGE association

Optic Pit/Hole
--Associated with... serous RD in adulthood

Morning-Glory Disc

Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

These are all secondary to abnormal closure of the embryonic optic fissure.
Optic nerve pits are colobomatous defects in the optic nerve, most common inferotemporally. Most optic nerve pits are asymptomatic, but they can occasionally cause serous macular detachments as seen in this fundus photograph. The OCT scan shows extensive subretinal fluid extending from the optic nerve.

Optic nerve pit with serous RD
Developmental Abnormalities of the Optic Nerve Head

These three are all secondary to abnormal closure of the embryonic optic fissure

Optic Nerve Coloboma
1) May resemble...deep cupping
2) Can be...bilateral, asymmetric
3) Part of the...CHARGE association

Optic Pit/Hole
--Associated with...serous RD in adulthood

Morning-Glory Disc
--DFE reveals:
  1) A funnel-shaped... [classic term]
  2)

Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

Optic Nerve Coloboma
1) May resemble...deep cupping
2) Can be...bilateral, asymmetric
3) Part of the...CHARGE association

Optic Pit/Hole
--Associated with...serous RD in adulthood

Morning-Glory Disc
--DFE reveals:
  1) A funnel-shaped... [classic term]
  2)
Developmental Abnormalities of the Optic Nerve Head

*These three are all secondary to abnormal closure of the embryonic optic fissure*

**Optic Nerve Coloboma**
1) May resemble *deep cupping*
2) Can be *bilateral, asymmetric*
3) Part of the *CHARGE association*

**Optic Pit/Hole**
--Associated with *serous RD in adulthood*

**Morning-Glory Disc**
--DFE reveals:
1) A funnel-shaped *excavation*
2) *Megalopapilla*

**Optic Nerve Hypoplasia**
--Abnormally *low number of axons*
--DFE: Small pale disc with *double ring sign*
--VA *20/20<->NLP*
--VF defects *invariably present*
--Remember the *4 D's*

**Megalopapilla**

**Myelinated RNFL**
--Myelin normally starts at LGN, ends at lamina cribrosa
--Can be patchy, discontinuous--Corresponding VF has absolute scotoma

**Tilted-Disc Syndrome**
1) Superior pole appears *elevated*, inferior *recessed*
2) Associated with *situs inversus* of retinal vessels
3) Fundus abnormality produces *myopic astigmatism*
4) VF testing reveals *bitemporal hemianopia* that *doesn't respect the vertical* and *may resolve with refraction*
Developmental Abnormalities of the Optic Nerve Head

These three are all secondary to abnormal closure of the embryonic optic fissure

**Optic Nerve Coloboma**
1) May resemble **deep cupping**
2) Can be **bilateral, asymmetric**
3) Part of the **CHARGE association**

**Optic Pit/Hole**
--Associated with **serous RD in adulthood**

**Morning-Glory Disc**
--DFE reveals:
  1) A funnel-shaped **excavation**
  2) Number of vessels crossing the rim seems abnormally... *high v low*

**Megalopapilla**

**Myelinated RNFL**
--Myelin normally starts at LGN, ends at lamina cribrosa
--Can be patchy, discontinuous--Corresponding VF has absolute scotoma

**Optic Nerve Hypoplasia**
--Abnormally low number of axons
--DFE: Small pale disc with double ring sign
--VA 20/20<->NLP
--VF defects invariably present
--Remember the **4 D's** (more on this shortly)

**Tilted-Disc Syndrome**
1) Superior pole appears **elevated**, inferior **recessed**
2) Associated with **situs inversus** of retinal vessels
3) Fundus abnormality produces **myopic astigmatism**
4) VF testing reveals **bitemporal hemianopia** that doesn't...
Developmental Abnormalities of the Optic Nerve Head

Optic Pit/Hole
--Associated with...serous RD in adulthood

Optic Nerve Coloboma
1) May resemble...deep cupping
2) Can be...bilateral, asymmetric
3) Part of the...CHARGE association

Optic Nerve Hypoplasia
--Abnormally low number of axons
--DFE: Small pale disc with double ring sign
--VA 20/20<->NLP
--VF defects invariably present
--Remember the 4 D's (more on this shortly)

Morning-Glory Disc
--DFE reveals:
  1) A funnel-shaped...excavation
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Myelinated RNFL
--Myelin normally starts at LGN, ends at lamina cribrosa
--Can be patchy, discontinuous--Corresponding VF has absolute scotoma

Megalopapilla
--Abnormally large diameter of disc and cup
--VF testing may reveal an enlarged blind spot

Tilted-Disc Syndrome
1) Superior pole appears...elevated
2) Inferior...recessed
3) Fundus abnormality produces...myopic astigmat
4) VF testing reveals...bitemporal hemianopia that doesn't respect the vertical and may resolve with refraction

These three are all secondary to abnormal closure of the embryonic optic fissure
Developmental Abnormalities of the Optic Nerve Head

Morning-glory disc
Developmental Abnormalities of the Optic Nerve Head

These three are all secondary to abnormal closure of the embryonic optic fissure:

- **Optic Nerve Coloboma**
  1. May resemble **deep cupping**
  2. Can be **bilateral, asymmetric**
  3. Part of the **CHARGE association**

- **Optic Pit/Hole**
  --Associated with **serous RD in adulthood**

- **Morning-Glory Disc**
  --DFE reveals:
    1. A funnel-shaped **excavation**
    2. Number of vessels crossing the rim seems abnormally **high**
  --Tissue is... **descriptor**, so cup seems to... **type of change** (like its namesake, the morning-glory flower)

- **Megalopapilla**

- **Myelinated RNFL**

- **Optic Nerve Hypoplasia**

- **Tilted-Disc Syndrome**
  1. Superior pole appears **elevated**, inferior **recessed**
  2. Associated with **situs inversus** of retinal vessels
  3. Fundus abnormality produces **myopic astigmatism**
  4. VF testing reveals **bitemporal hemianopia** that doesn’t... **resolve with refraction**
Developmental Abnormalities of the Optic Nerve Head

Optic Pit/Hole
--Associated with...serous RD in adulthood

These three are all secondary to abnormal closure of the embryonic optic fissure

Optic Nerve Coloboma
1) May resemble...deep cupping
2) Can be...bilateral, asymmetric
3) Part of the...CHARGE association

Optic Pit/Hole
--Associated with...serous RD in adulthood

Morning-Glory Disc
--DFE reveals:
1) A funnel-shaped...excavation
2) Number of vessels crossing the rim seems abnormally...high
--Tissue is...contractile, so cup seems to...open and close (like its namesake, the morning-glory flower)

Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome
Optic Pit/Hole
--Associated with... serous RD in adulthood

Optic Nerve Coloboma
1) May resemble... deep cupping
2) Can be... bilateral, asymmetric
3) Part of the... CHARGE association

Optic Nerve Hypoplasia
--Abnormally low number of axons
--DFE: Small pale disc with double ring sign
--VA 20/20<->NLP
--VF defects invariably present
--Remember the 4 D's (more on this shortly)

Morning-Glory Disc
--DFE reveals:
  1) A funnel-shaped... excavation
  2) Number of vessels crossing the rim seems abnormally... high
--Tissue is... contractile, so cup seems to... open and close (like its namesake, the morning-glory flower)
--VA usually... [range], but can be...

Megalopapilla

Myelinated RNFL
--Myelin normally starts at LGN, ends at lamina cribrosa
--Can be patchy, discontinuous--Corresponding VF has absolute scotoma

Tilted-Disc Syndrome
1) Superior pole appears... elevated, inferior... recessed
2) Associated with... situs inversus of retinal vessels
3) Fundus abnormality produces... myopic astigmatism
4) VF testing reveals... bitemporal hemianopia that doesn't respect the vertical and may resolve with refraction

These three are all secondary to abnormal closure of the embryonic optic fissure
**Developmental Abnormalities of the Optic Nerve Head**

- **Optic Pit/Hole**
  - Associated with **serous RD in adulthood**

- **Optic Nerve Coloboma**
  - 1) May resemble **deep cupping**
  - 2) Can be **bilateral, asymmetric**
  - 3) Part of the **CHARGE association**

- **Morning-Glory Disc**
  - DFE reveals:
    1) A funnel-shaped **excavation**
    2) Number of vessels crossing the rim seems abnormally **high**
  - Tissue is **contractile**, so cup seems to **open and close** (like its namesake, the morning-glory flower)
  - VA usually **20/200**, but can be **20/20<->NLP**

- **Tilted-Disc Syndrome**
  - Superior pole appears **elevated**, inferior **recessed**
  - Associated with **situs inversus** of retinal vessels
  - Fundus abnormality produces **myopic astigmatism**
  - VF testing reveals **bitemporal hemianopia** that doesn’t respect the vertical and may resolve with refraction

- **Megalopapilla**

- **Myelinated RNFL**
  - Myelin normally starts at LGN, ends at lamina cribrosa
  - Can be patchy, discontinuous
  - Corresponding VF has absolute scotoma

- **Optic Nerve Hypoplasia**
  - Abnormally low number of axons
  - DFE: Small pale disc with **double ring sign**
  - VA **20/20<->NLP**
  - VF defects invariably present
  - Remember the **4 D’s** (more on this shortly)

These three are all secondary to abnormal closure of the embryonic optic fissure
Developmental Abnormalities of the Optic Nerve Head

Optic Nerve Coloboma
1) May resemble...deep cupping
2) Can be...bilateral, asymmetric
3) Part of the...CHARGE association

Optic Pit/Hole
--Associated with...serous RD in adulthood

Morning-Glory Disc
--DFE reveals:
  1) A funnel-shaped...excavation
  2) Number of vessels crossing the rim seems abnormally...high
--Tissue is...contractile, so cup seems to...open and close (like its namesake, the morning-glory flower)
--VA usually...20/200, but can be...
  20/20<->NLP
--1/3 develop...[retinal condition]

Optic Nerve Hypoplasia

Megalopapilla

Myelinated RNFL

Tilted-Disc Syndrome

These three are all secondary to abnormal closure of the embryonic optic fissure
Developmental Abnormalities of the Optic Nerve Head

Optic Nerve Coloboma
1) May resemble... **deep cupping**
2) Can be... **bilateral, asymmetric**
3) Part of the... **CHARGE association**

Optic Pit/Hole
--Associated with... **serous RD in adulthood**

Morning-Glory Disc
--DFE reveals:
1) A funnel-shaped... **excavation**
2) Number of vessels crossing the rim seems abnormally... **high**
--Tissue is... **contractile**, so cup seems to... **open and close** (like its namesake, the morning-glory flower)
--VA usually... **20/200**, but can be...
**20/20<->NLP**
--1/3 develop... **serous RD**

These three are all secondary to abnormal closure of the embryonic optic fissure

Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome
Developmental Abnormalities of the Optic Nerve Head

Morning-Glory Disc
--DFE reveals:
  1) A funnel-shaped... excavation
  2) Number of vessels crossing the rim seems abnormally... high
--Tissue is... contractile, so cup seems to... open and close (like its namesake, the morning-glory flower)
--VA usually... 20/200, but can be... 20/20<->NLP
--1/3 develop... serous RD

Optic Nerve Coloboma
  1) May resemble... deep cupping
  2) Can be... bilateral, asymmetric
  3) Part of the... CHARGE association

Optic Pit/Hole
--Associated with... serous RD in adulthood

Optic Nerve Hypoplasia
--Abnormally low number of axons
--DFE: Small pale disc with... double ring sign
--VA 20/20<->NLP
--VF defects invariably present
--Remember the 4 D's (more on this shortly)

Megalopapilla
  1) Abnormally large diameter of... [? and ?] up
  2) VF testing may reveal an... enlarged blind spot

Myelinated RNFL
--Myelin normally starts at LGN, ends at lamina cribrosa
--Can be patchy, discontinuous--Corresponding VF has absolute scotoma

Tilted-Disc Syndrome
  1) Superior pole appears... elevated
  2) Associated with... situs inversus of retinal vessels
  3) Fundus abnormality produces... myopic astigmatism
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Developmental Abnormalities of the Optic Nerve Head

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--DFE reveals:
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An 8-year-old white male with visual acuity of 20/20 OU, IOP 12 OU, visual fields and RNFL normal OU

Megalopapilla
Developmental Abnormalities of the Optic Nerve Head

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2) VF testing may reveal an... [specific VF finding]

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These three are all secondary to abnormal closure of the embryonic optic fissure
Developmental Abnormalities of the Optic Nerve Head

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**An abnormally large cup in a preemie with cerebral palsy is suggestive of what condition?**
Developmental Abnormalities of the Optic Nerve Head

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An abnormally large cup in a preemie with cerebral palsy is suggestive of what condition?
Periventricular leukomalacia

These three are all secondary to abnormal closure of the embryonic optic fissure

An abnormally large cup in a preemie with cerebral palsy is suggestive of what condition?
Periventricular leukomalacia

CNS ischemia in the perinatal period

Via retrograde trans-synaptic degeneration of ganglion cell axons

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Developmental Abnormalities of the Optic Nerve Head

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An abnormally large cup in a preemie with cerebral palsy is suggestive of what condition?
Periventricular leukomalacia

What is the causative event, and when does it occur?

Tilted-Disc Syndrome
1) Superior pole appears...elevated
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Developmental Abnormalities of the Optic Nerve Head

These three are all secondary to abnormal closure of the embryonic optic fissure

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What is the causative event, and when does it occur?
CNS ischemia in the perinatal period

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Developmental Abnormalities of the Optic Nerve Head

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**An abnormally large cup in a preemie with cerebral palsy is suggestive of what condition?**
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**What is the causative event, and when does it occur?**
CNS ischemia in the perinatal period

**How does CNS ischemia lead to an enlarged cup?**

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CNS ischemia in the perinatal period

How does CNS ischemia lead to an enlarged cup?
Via retrograde trans-synaptic degeneration of ganglion cell axons

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Tilted-Disc Syndrome
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Megalopapilla
1) Abnormally large diameter of disc and cup
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Optic Nerve Hypoplasia

Tilted-Disc Syndrome

And it (almost) goes without saying... What condition must be considered in any individual with an enlarged cup?

These three are all secondary to abnormal closure of the embryonic optic fissure
Developmental Abnormalities of the Optic Nerve Head

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Glaucoma
Developmental Abnormalities of the Optic Nerve Head

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- **Where does myelination normally begin?**
  - At the lateral geniculate nucleus

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**Where does myelination normally begin?**
At the lateral geniculate nucleus

(Note: Because we’re talking about the axons of retinal ganglion cells, it’s probably better to say that myelination begins at the lamina cribrosa and ends at the LGN.)
Developmental Abnormalities of the Optic Nerve Head

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--Remember the 4 D’s (more on this shortly)

Tilted-Disc Syndrome
1) Superior pole appears…elevated, inferior…recessed
2) Associated with…situs inversus of retinal vessels
3) Fundus abnormality produces…myopic astigmatism
4) VF testing reveals…bitemporal hemianopia that doesn’t respect the vertical and may…resolve with refraction
Developmental Abnormalities of the Optic Nerve Head

Myelinated RNFL
Developmental Abnormalities of the Optic Nerve Head

These three are all secondary to abnormal closure of the embryonic optic fissure

Optic Nerve Coloboma
1) May resemble deep cupping
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**Optic Nerve Hypoplasia**
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2)
3)
4)
5)

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Developmental Abnormalities of the Optic Nerve Head

Hypoplastic optic nerve heads

Magnified image of the optic discs above. Black arrows: optic nerve edge. Blue arrows: scleral canal edge

Optic nerve hypoplasia
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Optic Nerve Hypoplasia
A hypoplastic nerve with double-ring sign can easily be mistaken for what?
Developmental Abnormalities of the Optic Nerve Head

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Remember the...4 D's
(more on this shortly)

These three are all secondary to abnormal closure of the embryonic optic fissure

A hypoplastic nerve with double-ring sign can easily be mistaken for what?
A normal sized optic nerve head and cup (the outer edge of the ring is interpreted as the edge of the optic rim)
Developmental Abnormalities of the Optic Nerve Head

Stereo image of a very hypoplastic nerve. At first glance this looks like a big pale nerve. But look closely—there’s just a little stump of optic nerve (it’s where the vessels emanate) surrounded by a pale ring (double-ring sign).

Stereo instructions: If you’re emmetropic, throw on some +3s, lean in, and bring into focus the image in the middle. If you’re myopic, take off your specs and do the same. (If you’re a hyperope, good luck.)

Optic nerve hypoplasia: Double-ring sign
Developmental Abnormalities of the Optic Nerve Head

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2) DFE: Small pale disc with double ring sign
3) VA [range]
4)
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*These three are all secondary to abnormal closure of the embryonic optic fissure*
### Developmental Abnormalities of the Optic Nerve Head

These three are all secondary to abnormal closure of the embryonic optic fissure

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5) Is the VF loss associated with optic nerve hypoplasia progressive, or nonprogressive?

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- Is the VF loss associated with optic nerve hypoplasia progressive, or nonprogressive? Nonprogressive
Developmental Abnormalities of the Optic Nerve Head

a) Optic nerve hypoplasia OD; normal ONH OS. b) Automated perimetry demonstrating VF loss OD. c) Three-year VF index demonstrating no progressive loss (the slight trend toward *improvement* is due to a learning effect.)

Optic nerve hypoplasia and VF loss
Developmental Abnormalities of the Optic Nerve Head

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You see a child with optic nerve hypoplasia. What should you do for her?

Get MRI brain (various CNS abnormalities are associated with optic-nerve hypoplasia), and refer for endocrine evaluation (multiple endocrine deficiencies are associated, especially pituitary-related)
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3) VA...20/15-NLP
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5) Remember the 4 D's (more on this shortly)

You see a child with optic nerve hypoplasia. What should you do for her? Get MRI brain (various CNS abnormalities are associated with optic-nerve hypoplasia), and refer for endocrine evaluation (multiple endocrine deficiencies are associated, especially pituitary-related)
Developmental Abnormalities of the Optic Nerve Head

**Tilted-Disc Syndrome**
1) Superior pole appears elevated, inferior recessed
2) Associated with situs inversus of retinal vessels
3) Fundus abnormality produces myopic astigmatism
4) VF testing reveals bitemporal hemianopia that doesn’t respect the vertical and may resolve with refract

**Morning-Glory Disc**
--DFE reveals:
1) A funnel-shaped excavation
2) Number of vessels crossing the rim seems abnormally high
--Tissue is contractile, so cup seems to open and close (like its namesake, the morning-glory flower)
--VA usually 20/200, but can be 20/20<->NLP
--1/3 develop serous RD

**Optic Nerve Coloboma**
1) May resemble deep cupping
2) Can be bilateral, asymmetric
3) Part of the CHARGE association

**Optic Pit/Hole**
--Associated with serous RD in adulthood

**Megalopapilla**
1) Abnormally large diameter of disc and cup
2) VF testing may reveal an enlarged blind spot

**Myelinated RNFL**
1) Myelin normally ends at the lamina cribrosa
2) Can be patchy and discontinuous
3) Corresponding VF has an absolute scotoma

**Optic Nerve Hypoplasia**
1) Abnormally low number of axons
2) DFE: Small pale disc with double ring sign
3) VA 20/15<->NLP
4) VF defects invariably present
5) Remember the 4 D’s

**Tilted-Disc Syndrome aka**
By what eponymous name is tilted-disc syndrome known?
Developmental Abnormalities of the Optic Nerve Head

- **Tilted-Disc Syndrome**
  1) Superior pole appears elevated, inferior recessed
  2) Associated with situs inversus of retinal vessels
  3) Fundus abnormality produces myopic astigmatism
  4) VF testing reveals bitemporal hemianopia that doesn’t respect the vertical and may resolve with refraction

- **Morning-Glory Disc**
  --DFE reveals:
  1) A funnel-shaped excavation
  2) Number of vessels crossing the rim seems abnormally high
  --Tissue is contractile, so cup seems to open and close (like its namesake, the morning-glory flower)
  --VA usually 20/200, but can be 20/20<->NLP
  --1/3 develop serous RD

- **Optic Nerve Coloboma**
  1) May resemble deep cupping
  2) Can be bilateral, asymmetric
  3) Part of the CHARGE association

- **Optic Pit/Hole**
  --Associated with serous RD in adulthood

- **Megalopapilla**
  1) Abnormally large diameter of disc and cup
  2) VF testing may reveal an enlarged blind spot

- **Myelinated RNFL**
  1) Myelin normally ends at the lamina cribrosa
  2) Can be patchy and discontinuous
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- **Optic Nerve Hypoplasia**
  1) Abnormally low number of axons
  2) DFE: Small pale disc with double ring sign
  3) VA 20/15<->NLP
  4) VF defects invariably present
  5) Remember the 4 D’s

- **Tilted-Disc Syndrome aka Fuch’s Coloboma**
  By what eponymous name is tilted-disc syndrome known?
  Fuch’s coloboma
Developmental Abnormalities of the Optic Nerve Head

Tilted-Disc Syndrome aka... Fuch’s Coloboma
1) Superior pole appears... elevated, inferior... recessed
2) VF testing reveals... bitemporal hemianopia that doesn't respect the vertical and may resolve with refraction

Optic Nerve Coloboma
1) May resemble... deep cupping
2) Can be... bilateral, asymmetric
3) Part of the... CHARGE association

Optic Pit/Hole
--Associated with... serous RD in adulthood

Morning-Glory Disc
--DFE reveals:
  1) A funnel-shaped... excavation
  2) Number of vessels crossing the rim seems abnormally... high
--Tissue is... contractile, so cup seems to... open and close (like its namesake, the morning-glory flower)
--VA usually... 20/200, but can be...
  20/20<->NLP
--1/3 develop... serous RD

Megalopapilla
1) Abnormally large diameter of... disc and cup
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Myelinated RNFL
1) Myelin normally ends at the... lamina cribrosa
2) Can be... patchy and discontinuous
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1) Abnormally low number of... axons
2) DFE: Small pale disc with... double ring sign
3) VA... 20/15<->NLP
4) VF defects... invariably present
5) Remember the... 4 D's

These three are all secondary to abnormal closure of the embryonic optic fissure

Megalopapilla
1) Abnormally large diameter of... disc and cup
2) VF testing may reveal an... enlarged blind spot
Developmental Abnormalities of the Optic Nerve Head

Morning-Glory Disc
--DFE reveals:
1) A funnel-shaped...excavation
2) Number of vessels crossing the rim seems abnormally...high
--Tissue is...contractile, so cup seems to...open and close (like its namesake, the morning-glory flower)
--VA usually...20/200, but can be...20/20<->NLP
--1/3 develop...serous RD

Optic Nerve Coloboma
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2) Can be...bilateral, asymmetric
3) Part of the...CHARGE association

Optic Pit/Hole
--Associated with...serous RD in adulthood

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2) DFE: Small pale disc with...double ring sign
3) VA...20/15<->NLP
4) VF defects...invariably present
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Tilted-Disc Syndrome aka...Fuch's Coloboma
1) Superior pole appears...elevated, inferior...recessed
2)
3)
4)

Megalopapilla
1) Abnormally large diameter of...disc and cup
2) VF testing may reveal an...enlarged blind spot

Myelinated RNFL
1) Myelin normally ends at the...lamina cribrosa
2) Can be...patchy and discontinuous
3) Corresponding VF has an...absolute scotoma

These three are all secondary to abnormal closure of the embryonic optic fissure
Developmental Abnormalities of the Optic Nerve Head

Tilted-disc syndrome. If you use your imagination, you can see that the superior pole is elevated relative to the inferior.
Developmental Abnormalities of the Optic Nerve Head

Morning-Glory Disc
--DFE reveals:
  1) A funnel-shaped excavation
  2) Number of vessels crossing the rim seems abnormally high
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Optic Nerve Hypoplasia
1) Abnormally low number of axons
2) DFE: Small pale disc with double ring sign
3) VA 20/15<->NLP
4) VF defects invariably present
5) Remember the 4 D's

Tilted-Disc Syndrome aka Fuch's Coloboma
1) Superior pole appears elevated, inferior recessed
2) Associated with [two words] of retinal vessels
3)
4)
Developmental Abnormalities of the Optic Nerve Head

Morning-Glory Disc
--DFE reveals:
1) A funnel-shaped excavation
2) Number of vessels crossing the rim seems abnormally high
--Tissue is contractile, so cup seems to open and close (like its namesake, the morning-glory flower)
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Tilted-Disc Syndrome aka Fuch’s Coloboma
1) Superior pole appears elevated, inferior recessed
2) Associated with situs inversus of retinal vessels
3)
4)
Developmental Abnormalities of the Optic Nerve Head

Situs inversus OD. Note how the temporal vessels exiting the ONH briefly run nasally prior to heading temporally

Situs inversus of the retinal vessels

Normal OD posterior pole for comparison
Developmental Abnormalities of the Optic Nerve Head

Morning-Glory Disc
--DFE reveals:
1) A funnel-shaped...excavation
2) Number of vessels crossing the rim seems abnormally...high
--Tissue is...contractile, so cup seems to...open and close (like its namesake, the morning-glory flower)
--VA usually...20/200, but can be...
20/20<->NLP
--1/3 develop...serous RD

Optic Nerve Coloboma
1) May resemble...deep cupping
2) Can be...bilateral, asymmetric
3) Part of the...CHARGE association

Optic Pit/Hole
--Associated with...serous RD in adulthood

Megalopapilla
1) Abnormally large diameter of...disc and cup
2) VF testing may reveal an...enlarged blind spot

Myelinated RNFL
1) Myelin normally ends at the...lamina cribrosa
2) Can be...patchy and discontinuous
3) Corresponding VF has an...absolute scotoma

Optic Nerve Hypoplasia
1) Abnormally low number of...axons
2) DFE: Small pale disc with...double ring sign
3) VA...20/15<->NLP
4) VF defects...invariably present
5) Remember the...4 D's

Tilted-Disc Syndrome aka...Fuch's Coloboma
1) Superior pole appears...elevated, inferior...recessed
2) Associated with...situs inversus of retinal vessels
3) Fundus abnormality produces...[refractive error]
4)
Developmental Abnormalities of the Optic Nerve Head

Morning-Glory Disc
--DFE reveals:
  1) A funnel-shaped...excavation
  2) Number of vessels crossing the rim seems abnormally...high
  --Tissue is...contractile, so cup seems to...open and close (like its namesake, the morning-glory flower)
  --VA usually...20/200, but can be...20/20<->NLP
  --1/3 develop...serous RD

Optic Nerve Coloboma
1) May resemble...deep cupping
2) Can be...bilateral, asymmetric
3) Part of the...CHARGE association

Optic Pit/Hole
--Associated with...serous RD in adulthood

Megalopapilla
1) Abnormally large diameter of...disc and cup
2) VF testing may reveal an...enlarged blind spot

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1) Myelin normally ends at the...lamina cribrosa
2) Can be...patchy and discontinuous
3) Corresponding VF has an...absolute scotoma

Optic Nerve Hypoplasia
1) Abnormally low number of...axons
2) DFE: Small pale disc with...double ring sign
3) VA...20/15<->NLP
4) VF defects...invariably present
5) Remember the...4 D's

Tilted-Disc Syndrome aka...Fuch's Coloboma
1) Superior pole appears...elevated, inferior...recessed
2) Associated with...situs inversus of retinal vessels
3) Fundus abnormality produces...myopic astigmatism
4)
Developmental Abnormalities of the Optic Nerve Head

*These three are all secondary to abnormal closure of the embryonic optic fissure*

**Optic Nerve Coloboma**
1) May resemble... *deep cupping*
2) Can be... *bilateral, asymmetric*
3) Part of the... *CHARGE association*

**Optic Pit/Hole**
--Associated with... *serous RD in adulthood*

**Morning-Glory Disc**
--DFE reveals:
1) A funnel-shaped... *excavation*
2) Number of vessels crossing the rim seems abnormally... *high*
--Tissue is... *contractile*, so cup seems to... *open and close* (like its namesake, the morning-glory flower)
--VA usually... *20/200*, but can be...
   20/20<->NLP
--1/3 develop... *serous RD*

**Megalopapilla**
1) Abnormally large diameter of... *disc and cup*
2) VF testing may reveal an... *enlarged blind spot*

**Myelinated RNFL**
1) Myelin normally ends at the... *lamina cribrosa*
2) Can be... *patchy and discontinuous*
3) Corresponding VF has an... *absolute* scotoma

**Optic Nerve Hypoplasia**
1) Abnormally low number of... *axons*
2) DFE: Small pale disc with... *double ring* sign
3) VA... *20/15<->NLP*
4) VF defects... *invariably present*
5) Remember the... *4 D’s*

**Tilted-Disc Syndrome aka... Fuch’s Coloboma**
1) Superior pole appears... *elevated*, inferior... *recessed*
2) Associated with... *situs inversus* of retinal vessels
3) Fundus abnormality produces... *myopic astigmatism*
4) VF testing reveals... *[specific VF finding]* that doesn’t respect the vertical, and may resolve with... *[simple clinical maneuver]*
Developmental Abnormalities of the Optic Nerve Head

**Morning-Glory Disc**
--DFE reveals:
1) A funnel-shaped... **excavation**
2) Number of vessels crossing the rim seems abnormally... **high**
--Tissue is... **contractile**, so cup seems to... **open and close** (like its namesake, the morning-glory flower)
--VA usually... **20/200**, but can be... **20/20<->NLP**
--1/3 develop... **serous RD**

**Megalopapilla**
1) Abnormally large diameter of... **disc and cup**
2) VF testing may reveal an... **enlarged blind spot**

**Optic Nerve Coloboma**
1) May resemble... **deep cupping**
2) Can be... **bilateral, asymmetric**
3) Part of the... **CHARGE association**

**Optic Pit/Hole**
--Associated with... **serous RD in adulthood**

**Myelinated RNFL**
1) Myelin normally ends at the... **lamina cribrosa**
2) Can be... **patchy and discontinuous**
3) Corresponding VF has an... **absolute** scotoma

**Optic Nerve Hypoplasia**
1) Abnormally low number of... **axons**
2) DFE: Small pale disc with... **double ring** sign
3) VA... **20/15<->NLP**
4) VF defects... **invariably present**
5) Remember the... **4 D’s**

**Tilted-Disc Syndrome aka... Fuch’s Coloboma**
1) Superior pole appears... **elevated**, inferior... **recessed**
2) Associated with... **situs inversus** of retinal vessels
3) Fundus abnormality produces... **myopic astigmatism**
4) VF testing reveals... **bitemporal hemianopia** that doesn’t respect the vertical, and may resolve with... **refraction**

**These three are all secondary to abnormal closure of the embryonic optic fissure**
Developmental Abnormalities of the Optic Nerve Head

Optic Nerve Coloboma
1) May resemble...deep cupping
2) Can be...bilateral, asymmetric
3) Part of the...CHARGE association

Optic Pit/Hole
--Associated with...serous RD in adulthood

Morning-Glory Disc
--DFE reveals:
1) A funnel-shaped...excavation
2) Number of vessels crossing the rim seems abnormally...high
--Tissue is...contractile, so cup seems to...open and close (like its namesake, the morning-glory flower)
--VA usually...20/200, but can be...20/20<->NLP
--1/3 develop...serous RD

Megalopapilla
1) Abnormally large diameter of...disc and cup
2) VF testing may reveal an...enlarged blind spot

Myelinated RNFL
1) Myelin normally ends at the...lamina cribrosa
2) Can be...patchy and discontinuous
3) Corresponding VF has an...absolute scotoma

Optic Nerve Hypoplasia
1) Abnormally low number of...axons
2) DFE: Small pale disc with...double ring sign
3) VA...20/15<->NLP
4) VF defects...invariably present
5) Remember the...4 D's

Tilted-Disc Syndrome aka...Fuch's Coloboma
1) Superior pole appears...elevated, inferior...recessed
2) Situs inversus of retinal vessels
3) Myopic astigmatism due to...recessed...hyperopia produces...myopic astigmatism
4) Defects...bitemporal hemianopia that doesn't
5) May resolve with...refraction

Does the VF defect tend to involve the superior field, or the inferior?
Developmental Abnormalities of the Optic Nerve Head

Morning-Glory Disc
--DFE reveals:
1) A funnel-shaped…excavation
2) Number of vessels crossing the rim seems abnormally…high
--Tissue is…contractile, so cup seems to…open and close (like its namesake, the morning-glory flower)
--VA usually…20/200, but can be…20/20<->NLP
--1/3 develop…serous RD

Optic Pit/Hole
--Associated with…serous RD in adulthood

Optic Nerve Coloboma
1) May resemble…deep cupping
2) Can be…bilateral, asymmetric
3) Part of the…CHARGE association

Optic Nerve Hypoplasia
1) Abnormally low number of…axons
2) DFE: Small pale disc with…double ring sign
3) VA…20/15<->NLP
4) VF defects…invariably present
5) Remember the…4 D’s

Megalopapilla
1) Abnormally large diameter of…disc and cup
2) VF testing may reveal an…enlarged blind spot

Myelinated RNFL
1) Myelin normally ends at the…lamina cribrosa
2) Can be…patchy and discontinuous
3) Corresponding VF has an…absolute scotoma

Tilted-Disc Syndrome aka…Fuch’s Coloboma
1) Superior pole appears…elevated, inferior…recessed
2) Situs inversus of retinal vessels
3) Myopic astigmatism
4) VF defects…bitemporal hemianopia

Does the VF defect tend to involve the superior field, or the inferior?
Superior
Developmental Abnormalities of the Optic Nerve Head

Tilted-disc syndrome: Classic superior bitemporal VF loss not respecting the midline (note also the situs inversus of the retinal vasculature)
Developmental Abnormalities of the Optic Nerve Head

These three are all secondary to abnormal closure of the embryonic optic fissure

Optic Nerve Coloboma
1) May resemble...deep cupping
2) Can be...bilateral, asymmetric
3) Part of...CHARGE association

Megalopapilla
1) Abnormally large diameter of...disc and cup
2) VF testing may reveal an...enlarged blind spot

Myelinated RNFL
1) Myelin normally ends at the...lamina cribrosa
2) Can be...patchy and discontinuous

Morning-Glory Disc
--DFE reveals:
1) A funnel-shaped...excavation
2) Number of vessels crossing the rim seems abnormally...high
--Tissue is...contractile, so cup seems...open and close...like its namesake, the morning-glory flower)
--VA usually...20/200, but can be...20/20<->NLP
--1/3 develop...serous RD

Optic Nerve Hypoplasia
1) Abnormally low number of...axons
2) DFE: Small pale disc with...double ring sign
3) VA...20/15<->NLP
4) VF defects...invariably present
5) Remember the...4 D's

Tilted-Disc Syndrome
aka...Fuch's Coloboma
1) Superior pole appears...elevated, inferior...recessed
2) Associated with...situs inversus of retinal vessels
3) Fundus abnormality produces...myopic astigmatism
4) VF testing reveals...bitemporal hemianopia that doesn't...and may resolve with...refraction

How on earth does tilting of the discs lead to a superior bitemporal VF defect?

Does the VF defect tend to involve the superior field, or the inferior?
Superior

Mittus inversus of retinal vessels
Developmental Abnormalities of the Optic Nerve Head

These three are all secondary to abnormal closure of the embryonic optic fissure:

- **Morning-Glory Disc**
  1) A funnel-shaped excavation
  2) Number of vessels crossing the rim seems abnormally high
  -- Tissue is contractile, so cup seems to open and close (like its namesake, the morning-glory flower)
  -- VA usually 20/200, but can be 20/20<->NLP
  -- 1/3 develop serous RD

- **Optic Nerve Coloboma**
  1) May resemble deep cupping
  2) Can be bilateral, asymmetric
  3) Part of the CHARGE association

- **Megalopapilla**
  1) Abnormally large diameter of disc and cup
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  1) Abnormally low number of axons
  2) DFE: Small pale disc with double ring sign
  3) VA 20/15<->NLP
  4) VF defects invariably present
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- **Tilted-Disc Syndrome** aka Fuch's Coloboma
  1) Superior pole appears elevated, inferior recessed
  2) Associated with situs inversus of retinal vessels
  3) Fundus abnormality produces myopic astigmatism

How on earth does tilting of the discs lead to a superior bitemporal VF defect? It's actually pretty simple.

The recession of the inferonasal ONH is quite dramatic—almost staphyloma-ish. Because of this, the ‘axial length’ (AL) of the inferonasal peripapillary retina is longer than that of other retinal regions. The excess AL of this portion of the retina renders it more myopic than the rest, and thus the refractive correction used during the performance of the VF test—derived from the foveal refraction, with its unaffected AL—is not myopic enough for the inferonasal retina.

Does the VF defect tend to involve the superior field, or the inferior?

Superior
Developmental Abnormalities of the Optic Nerve Head

Morning-Glory Disc
--DFE reveals:
1) A funnel-shaped excavation
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--VA usually...20/200, but can be...20/20<->NLP
--1/3 develop...serous RD

Optic Nerve Coloboma
1) May resemble...deep cupping
2) Can be...bilateral, asymmetric
3) Part of the...CHARGE association

Megalopapilla
1) Abnormally large diameter of...disc and cup
2) VF testing may reveal an...enlarged blind spot

Optic Nerve Hypoplasia
1) Abnormally low number of...axons
2) DFE: Small pale disc with...double ring sign
3) VA...20/15<->NLP
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1) Abnormally large diameter of...disc and cup
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Myelinated RNFL
1) Myelin normally ends at the...lamina cribrosa
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Tilted-Disc Syndrome
aka...Fuch's Coloboma
1) Superior pole appears...elevated, inferior...recessed
2) Associated with...situs inversus of retinal vessels
3) Fundus abnormality produces...myopic astigmatism
4) VF testing reveals...bitemporal hemianopia

How on earth does tilting of the discs lead to a superior bitemporal VF defect?
It's actually pretty simple.
The recession of the inferonasal ONH is quite dramatic—almost staphyloma-ish. Because of this, the 'axial length' (AL) of the inferonasal peripapillary retina is longer than that of other retinal regions. The excess AL of this portion of the retina renders it more myopic than the rest, and thus the refractive correction used during the performance of the VF test—derived from the foveal refraction, with its unaffected AL—is not myopic enough for the inferonasal retina. The subsequent uncorrected myopia of the inferonasal retina produces a refractive scotoma in the superotemporal VF.

Does the VF defect tend to involve the superior field, or the inferior?
Superior

bitemporal hemianopia
Developmental Abnormalities of the Optic Nerve Head

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--Tissue is contractile, so cup seems to open and close (like its namesake, the morning-glory flower)
--VA usually 20/200, but can be 20/20-►NLP
--1/3 develop serous RD

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2) Can be bilateral, asymmetric
3) Part of the CHARGE association

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aka Fuch's Coloboma
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How on earth does tilting of the discs lead to a superior bitemporal VF defect?
It's actually pretty simple. The recession of the inferonasal ONH is quite dramatic—almost staphyloma-ish. Because of this, the 'axial length' (AL) of the inferonasal peripapillary retina is longer than that of other retinal regions. The excess AL of this portion of the retina renders it more myopic than the rest, and thus the refractive correction used during the performance of the VF test—derived from the foveal refraction, with its unaffected AL—is not myopic enough for the inferonasal retina. The subsequent uncorrected myopia of the inferonasal retina produces a refractive scotoma in the superotemporal VF.

This implies that the VF defect will resolve if the 'proper' refractive correction is employed. Does it?

Does the VF defect tend to involve the superior field, or the inferior?
Superior
How on earth does tilting of the discs lead to a superior bitemporal VF defect?

It’s actually pretty simple. The recession of the inferonasal ONH is quite dramatic—almost staphyloma-ish. Because of this, the ‘axial length’ (AL) of the inferonasal peripapillary retina is longer than that of other retinal regions. The excess AL of this portion of the retina renders it more myopic than the rest, and thus the refractive correction used during the performance of the VF test—derived from the foveal refraction, with its unaffected AL—is not myopic enough for the inferonasal retina. The subsequent uncorrected myopia of the inferonasal retina produces a refractive scotoma in the superotemporal VF.

This implies that the VF defect will resolve if the ‘proper’ refractive correction is employed. Does it? Indeed it does.

Does the VF defect tend to involve the superior field, or the inferior?

Superior

bitemporal hemianopia
Developmental Abnormalities of the Optic Nerve Head

**Optic Nerve Coloboma**
1. May resemble... **deep cupping**
2. Can be... **bilateral, asymmetric**
3. Part of the... **CHARGE association**

**Optic Pit/Hole**
--Associated with... **serous RD in adulthood**

**Morning-Glory Disc**
--DFE reveals:
1. A funnel-shaped... **excavation**
2. Number of vessels crossing the rim seems abnormally... **high**
--Tissue is... **contractile**, so cup seems to... **open and close** (like its namesake, the morning-glory flower)
--VA usually... **20/200**, but can be...
20/20<->NLP
--1/3 develop... **serous RD**

**Megalopapilla**
1. Abnormally large diameter of... **disc and cup**
2. VF testing may reveal an... **enlarged blind spot**

**Myelinated RNFL**
1. Myelin normally ends at the... **lamina cribrosa**
2. Can be... **patchy and discontinuous**
3. Corresponding VF has an... **absolute scotoma**

**Optic Nerve Hypoplasia**
1. Abnormally low number of... **axons**
2. DFE: Small pale disc with... **double ring sign**
3. VA... **20/15<->NLP**
4. VF defects... **invariably present**
5. Remember the... **4 D's**

**Tilted-Disc Syndrome**
1. Superior pole appears... **elevated**
2. Inferior... **recessed**
3. Associated with... **situs inversus** of retinal vessels
4. Fundus abnormality produces... **myopic astigmatism**
5. VF testing reveals... **bitemporal hemianopia** that doesn't respect the vertical, and may resolve with... **refraction**

As for the four Ds of optic nerve hypoplasia:

---

These three are all secondary to abnormal closure of the embryonic optic fissure.
What are the 4 D’s of optic nerve hypoplasia?

--D

--D

Hints forthcoming…

--D

--D
What are the **4 D’s** of optic nerve hypoplasia?

Concern mom’s life while she is pregnant w/ the child who will have ON hypoplasia

*Hints forthcoming…*

A congenital condition with significant CNS findings
What are the **4 D’s** of optic nerve hypoplasia?

- **Drink** (ie, heavy EtOH consumption during pregnancy)
- **Diabetes**
- **Drugs** (especially *D*ilantin or other seizure meds)
- **De Morsier syndrome**

(5 D’s if you count this one)

(should be lower-case, but it looked funny)
What are the 4 D’s of optic nerve hypoplasia?

--Drink (ie, heavy EtOH consumption during pregnancy)

In other words, optic-nerve hypoplasia is part of the

--Diabetes

--Drugs (especially Dilantin or other seizure meds)

--De Morsier syndrome
What are the 4 D’s of optic nerve hypoplasia?

--*Drink* (ie, heavy EtOH consumption during pregnancy)
In other words, optic-nerve hypoplasia is part of the *fetal alcohol syndrome*

--*Diabetes*

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--*De Morsier syndrome*
What are the 4 D’s of optic nerve hypoplasia?

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What triad constitutes de Morsier syndrome?

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--Absence of the

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--Optic nerve hypoplasia (duh)
--Absence of the septum pellucidum
--Agenesis of the corpus callosum
Developmental Abnormalities of the Optic Nerve Head

(A), MRI brain showing prominent cerebrospinal fluid spaces around optic nerve suggestive of optic nerve hypoplasia (yellow triangles). (B), absence of septum pellucidum (white triangle). (C), absence of septum pellucidum (white triangle)

de Morsier syndrome
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What is the nonponymous name of de Morsier syndrome?

Septo-optic dysplasia
Developmental Abnormalities of the Optic Nerve Head

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--Absence of the septum pellucidum

(Agenesis of the corpus callosum)

--Pituitary dwarfism

Note: The listed triad is from the BCSC Peds book. Per the Neuro book, the triad includes pituitary dwarfism, not corpus callosum agenesis (although it states that “the corpus callosum may be thinned or absent”)

Developmental Abnormalities of the Optic Nerve Head
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Of the four, which is the most common cause of optic nerve hypoplasia?

Maternal diabetes is notorious for causing a specific pattern of optic nerve hypoplasia--what is it?

Superior segmental optic nerve hypoplasia (SSONH)
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Pretty much what you would expect based on the name--a normal-appearing nerve save for a thin superior rim, with associated thinning of the superior nerve fiber layer

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Developmental Abnormalities of the Optic Nerve Head

(Don’t be fooled by the superior ‘double ring sign’!)

Superior segmental ON hypoplasia
Developmental Abnormalities of the Optic Nerve Head

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Bitemporal inferior loss that doesn’t respect the vertical midline

De Morsier syndrome
Developmental Abnormalities of the Optic Nerve Head

Superior segmental ON hypoplasia: Inferior VF defects