These three are all secondary to abnormal closure of the embryonic optic fissure.
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- **Optic Nerve Coloboma**
  - 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:
    - Funnel-shaped
    - 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
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Optic Pit/Hole
-- Associated with
   serous RD
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Morning-Glory Disc
-- DFE reveals:
   1) Funnel-shaped...
   2) Number of vessels crossing the rim seems abnormally...
   -- 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

Megalo-...
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
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Optic Pit/Hole

-- Associated with serous RD in adulthood

Morning-Glory Disc

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These three are all secondary to abnormal closure of the embryonic optic fissure
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…
2) Can be…
3) Part of the…

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…
-- Tissue is…

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

Myelinated…

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

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**
Developmental Abnormalities of the Optic Nerve Head

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

- Myelinated RNFL
- Megalopapilla
- Optic Nerve Coloboma
- Optic Pit/Hole
- Morning-Glory Disc

Optic Nerve Coloboma

Megalopapilla

Myelinated RNFL

Optic Nerve…

Morning-Glory Disc

Optic Pit/Hole

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
-- 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
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-- 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
Developmental Abnormalities of the Optic Nerve Head

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

Optic Nerve Coloboma

Megalopapilla

Optic Pit/Hole

Myelinated RNFL

Morning-Glory Disc

Optic Nerve Hypoplasia

Two-words Syndrome

Tilted-Disc 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
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Optic Nerve Coloboma
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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

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

2. Optic Pit/Hole

3. Morning-Glory Disc

- Megalopapilla
- Myelinated RNFL
- Optic Nerve Hypoplasia
- Tilted-Disc Syndrome
Optic Nerve Coloboma
1) May resemble… **deep cupping**
2)
3)

Optic Pit/Hole

Morning-Glory Disc

Optic Nerve Hypoplasia

Myelinated RNFL

Megalopapilla

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 OD. Note the well-demarcated inferior excavation with thinning of the inferior neuroretinal rim and preservation of the superior rim. ONH OS is normal.

Optic nerve coloboma
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

---

Tilted-Disc Syndrome
1) Superior pole appears… **elevated**
2) Associated with… **situs inversus** of retinal vessels
3) Fundus abnormality produces… **myopic astigmatism**
4) VF testing reveals… **bitemporal hemianopia** that doesn’t… with refract
Optic Nerve Coloboma
1) May resemble **deep cupping**
2) Can be **bilateral, asymmetric**
3) Part of the **CHARGE association**

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

---

Morning-Glory Disc
1) Funnel-shaped
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3) Tissue is contractile, so cup seems to open and close (like its namesake, the morning-glory flower)
4) VA usually 20/200, but can be 20/20->NLP
5) 1/3 develop serous RD

---

Optic Pit/Hole
1) Associated with serous RD in adulthood

---

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

---

Tilted-Disc Syndrome
1) Superior pole appears elevated, inferior recessed
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Myelinated RNFL
-- Myelin normally starts at LGN, ends at lamina cribrosa
-- Can be patchy, discontinuous
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---

Megalopapilla

---

**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

Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome
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

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

Optic Pit/Hole

Morning-Glory Disc

What is the CHARGE association?

Coloboma
H
A
R
G
E

Megalopapilla

Hypoplasia

Tilted-Disc Syndrome

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

What is the CHARGE association?

Coloboma
H
A
R
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Optic Pit/Hole

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What is the CHARGE association?

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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…
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  - 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

Megalopapilla

Hypoplasia

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head

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

What is the CHARGE association?
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Optic Nerve Coloboma
- 1) May resemble…deep cupping
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Optic Pit/Hole

Morning-Glory Disc

Megalopapilla

Hypoplasia

Tilted-Disc Syndrome
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.

Coloboma
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. 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
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)

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

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 Coloboma

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

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 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

Morning-Glory Disc
--DFE reveals:
1) A funnel-shaped… [classic term]
2) Number of vessels crossing the rim seems abnormally…

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
--Abnormally low number of axons
--DFE: Small pale disc with…double ring sign
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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

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…respect the vertical and may…resolve with refraction

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

---

Optic Nerve Coloboma

---

Optic Pit/Hole

---

Morning-Glory Disc

---

Myelinated RNFL

---

Optic Nerve Hypoplasia

---

Megalopapilla

---

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

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

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

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

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) 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

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

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

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

Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

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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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

In addition to their number, what else is unusual about the vessels at the ONH?

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

Myelinated RNFL
--Myelin normally starts at LGN, ends at lamina cribrosa
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In addition to their number, what else is unusual about the vessels at the ONH?
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

- **Megalopapilla**

- **Myelinated RNFL**

- **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...

In addition to their number, what else is unusual about the vessels at the ONH? They all emanate from the rim of the disc.
Developmental Abnormalities of the Optic Nerve Head

Morning-glory disc: Lotsa vessels, emanating from the rim
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
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
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
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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...[#/#], but can be...[range]

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) Can be bilateral, asymmetric
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Optic Pit/Hole
--Associated with serous RD in adulthood

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

Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

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

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

Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

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
- May resemble deep cupping
- Can be bilateral, asymmetric
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Optic Pit/Hole
- Associated with serous RD in adulthood

Morning-Glory Disc
- DFE reveals:
  1) A funnel-shaped excavation
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Optic Nerve Hypoplasia

Tilted-Disc Syndrome
- Superior pole appears elevated, inferior recessed
- Associated with situs inversus of retinal vessels
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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

Is morning-glory disc usually unilateral, or bilateral?

Is there a gender predilection?
Yes, there is a ♀ preponderance

It has two associations of particular note—what are they?
--PHACE syndrome
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--VA usually 20/20, but can be 20/20<->NLP
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Is morning-glory disc usually unilateral, or bilateral?
Unilateral

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

Developmental Abnormalities of the Optic Nerve Head

Megalopapilla

RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

Morning-Glory Disc

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

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

Optic Nerve Hypoplasia

---Abnormally low number of axons
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--Myelin normally starts at LGN, ends at lamina cribrosa
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Yes, there is a ♀ preponderance

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--PHACE syndrome
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What does PHACE stand for?
P = Posterior fossa malformations
H = Hemangiomas
A = Arterial lesions
C = Cardiac anomalies
E = Eye anomalies

Where is the hemangioma located?
The face, +/- scalp
Is it large, or small?
Large

Megalopapilla
--Abnormally large diameter of disc and cup
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Optic Nerve Hypoplasia
--Abnormally low number of axons
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PHACE syndrome: Large plaque-like hemangioma of the face and scalp
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An occlusive vascular condition primarily affecting the terminal internal carotids and proximal anterior and middle cerebral arteries

Is it common, or rare?
Quite rare

Is there a racial predilection?
Yes, it is more common in Asians, especially Japanese and Korean individuals

Is there a gender predilection?
Yes, there is a modest ♀ preponderance

Is there an age predilection?
It manifests most commonly in childhood (with a second, smaller peak in the fourth decade)
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Normal angiogram

Angiogram in moyamoya
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What is moyamoya disease (MMD)? An occlusive vascular condition primarily affecting the terminal internal carotids and proximal anterior and middle cerebral arteries

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Is it common, or rare? Quite rare

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Optic Pit/Hole
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These three are all secondary to abnormal closure of the embryonic optic fissure

--Is morning-glory disc usually unilateral, or bilateral?
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If an Asian child—in the real world or on the OKAP/Boards—has a morning-glory disc and neurological issues, get the angiography—it’s moyamoya!

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‘Occlusive cerebrovascular condition in young Asian women’ should bring to mind another condition as well. What is it?

Takayasu’s arteritis

In a nutshell, what is Takayasu’s arteritis?
An occlusive vasculitis that affects larger vessels

Is it common, or rare?
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Is there an age predilection?
Yes, most cases present in early adulthood

Asians

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Is there a gender predilection?
Yes, there is a ♀ preponderance

It has two associations of particular note—what are they?
--PHACE syndrome
--Moyamoya disease

What is moyamoya disease (MMD)?
An occlusive vascular condition primarily affecting the terminal internal carotids and proximal anterior and middle cerebral arteries

Is it common, or rare?
Quite rare

Is there a racial predilection?
Yes, it is more common in Asians

Is there a gender predilection?
Yes, there is a modest female preponderance

Is there an age predilection?
It manifests most commonly in the fourth decade

‘Occlusive cerebrovascular condition in young Asian women’ should bring to mind another condition as well. What is it?
Takayasu's arteritis

In a nutshell, what is Takayasu's arteritis?
An occlusive vasculitis that affects large vessels

Is it common, or rare?
Rare

Asians
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

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

Developmental Abnormalities of the Optic Nerve Head

Morning-Glory Disc

Unilateral

Is there a gender predilection?
Yes, there is a ♀ preponderance

It has two associations of particular note—what are they?
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Is it common, or rare?

Is there an age predilection?
Developmental Abnormalities of the Optic Nerve Head

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In a nutshell, what is Takayasu’s arteritis?
- An occlusive vasculitis that affects large vessels

Is it common, or rare?
- Rare

Is there an age predilection?
- Yes, most cases present in early adulthood
Morning-Glory Disc

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

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For more on Takayasu's arteritis, see slide set R23

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What is Takayasu's arteritis? An occlusive vasculitis that affects large vessels

In a nutshell, what is Takayasu's arteritis? Rare

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Developmental Abnormalities of the Optic Nerve Head

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Optic Nerve Coloboma
1) May resemble…deep cupping
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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 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

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Developmental Abnormalities of the Optic Nerve Head

An 8-year-old with VA 20/20 OU, IOP 12 OU, VF and RNFL normal OU

Megalopapilla
Developmental Abnormalities of the Optic Nerve Head

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

Megalopapilla
1) Abnormally large diameter of...disc and cup
2) VF testing may reveal an...[specific VF finding]

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Developmental Abnormalities of the Optic Nerve Head

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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?

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

- **Periventricular leukomalacia**
  - Causative event: CNS ischemia in the perinatal period
  - How CNS ischemia leads to an enlarged cup: Via retrograde trans-synaptic degeneration of ganglion cell axons

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

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

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Developmental Abnormalities of the Optic Nerve Head

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

Tilted-Disc Syndrome
1) Superior pole appears...elevated
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What is the causative event, and when does it occur?
CNS ischemia in the perinatal period

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CNS ischemia in the perinatal period

How does CNS ischemia lead to an enlarged cup?

Megalopapilla
1) Abnormally large diameter of...disc and cup
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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?
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How does CNS ischemia lead to an enlarged cup?
Via retrograde trans-synaptic degeneration of ganglion cell axons

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1) Superior pole appears... elevated
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Developmental Abnormalities of the Optic Nerve Head

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

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

**Optic Nerve Hypoplasia**

**Tilted-Disc Syndrome**
1) Superior pole appears...elevated
2) Inferior...recessed
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**Megalopapilla**
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**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 respect the vertical and may resolve with refraction

And it (almost) goes without saying... What condition must be considered in any individual with an enlarged cup? **Glaucoma**
Developmental Abnormalities of the Optic Nerve Head

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**Megalopapilla**
1) Abnormally large diameter of...disc and cup
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**Myelinated RNFL**
1) Myelin normally ends at the...[location]
2) 3)

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

**Tilted-Disc Syndrome**
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Optic Nerve Coloboma
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Myelinated RNFL
1) Myelin normally ends at the...lamina cribrosa
2)
3)

Megalopapilla
1) Abnormally large diameter of...disc and cup
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Optic Nerve Hypoplasia

Tilted-Disc Syndrome

These three are all secondary to abnormal closure of the embryonic optic fissure
Optic Nerve Hypoplasia
--Abnormally low number of axons
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Where does myelination normally begin?

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
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Megalopapilla
1) Abnormally large diameter of disc and cup
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1) May resemble deep cupping
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Optic Pit/Hole
--Associated with serous RD in adulthood

Morning-Glory Disc
--DFE reveals:
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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

Where does myelination normally begin?
At the lateral geniculate nucleus

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

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(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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--Abnormally low number of axons
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Optic Nerve Hypoplasia
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2)
3)
4)
5)

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

Optic nerve hypoplasia

Hypoplastic optic nerve heads
Developmental Abnormalities of the Optic Nerve Head

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

Hypoplastic optic nerve heads

Optic nerve hypoplasia
Developmental Abnormalities of the Optic Nerve Head

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2) DFE: Small pale disc with...[2-word desc.] sign
3) VF defects...invariably present
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A hypoplastic nerve with double-ring sign can easily be mistaken for what?

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Developmental Abnormalities of the Optic Nerve Head

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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)

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

Optic nerve hypoplasia: *Double-ring sign*
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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Optic Nerve Hypoplasia
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3) VA...[range]
4) [range]

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Developmental Abnormalities of the Optic Nerve Head

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

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20/20<->NLP
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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
5) VF defects...invariably present

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

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 Coloboma
1) May resemble...deep cupping
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These three are all secondary to abnormal closure of the embryonic optic fissure
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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What is the classic structural pituitary abnormality associated with ON hypoplasia?

Optic Nerve Coloboma
1) May resemble deep cupping
2) Can be bilateral, asymmetric
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Optic Pit/Hole
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1) Superior pole appears elevated, inferior... recessed
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**Myelinated RNFL**
1) Myelin normally ends at the... lamina cribrosa
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--VA usually 20/200, but can be 20/20–NLP
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--Obtain MRI brain (will unveil a variety of CNS abnormalities are associated with it): anterior pituitary hypoplasia coupled with an ectopic posterior pituitary
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Note the posterior ectopic bright spot at the upper infundibulum (long arrow) and hypoplastic anterior pituitary (short arrow)
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What pituitary-related deficiencies may be present?
Anything up to and including panhypopituitarism
If an infant/child has ON hypoplasia and...short stature, think: Growth-hormone deficiency
If an infant/child has ON hypoplasia and...neonatal jaundice, think: Hypothyroidism
If an infant/child has ON hypoplasia and...hypoglycemia/seizures, think: Panhypopituitarism
If an infant/child has ON hypoplasia and...diabetes insipidus, think: Hypocortisolism
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These three are all secondary to abnormal closure of the embryonic optic fissure

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--Obtain MRI brain—A variety of CNS abnormalities are associated with it (will unpack this in the upcoming 4 D’s section)
--Get endocrine consult—multiple hormonal deficiencies are associated with this—mainly pituitary-related
What steps should you at least consider taking?

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

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

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1) May resemble deep cupping
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If an infant/child has ON hypoplasia and...
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...neonatal jaundice, think: Hypothyroidism
...neonatal jaundice...

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1) Abnormally low number of axons
2) DFE: Small pale disc with double ring sign
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Developmental Abnormalities of the Optic Nerve Head

**Tilted-Disc Syndrome**
1) Superior pole appears elevated, inferior recessed.
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**Myelinated RNFL**
1) Myelin normally ends at the lamina cribrosa.
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You see a child with optic nerve hypoplasia. What steps should you at least consider taking?

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These are probably low-yield factoids in isolation. But what isn’t low-yield is making the mental note ‘If a neonate or young child presents with ON hypoplasia along with anything remotely hormone-related, expedite neuroimaging and Peds Endo referral!’

...short stature, think: Growth-hormone deficiency
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Developmental Abnormalities of the Optic Nerve Head

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

Optic Nerve Coloboma
1) May resemble...deep cupping
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As for the four Ds of optic nerve hypoplasia:
What are the **4 D’s** of optic nerve hypoplasia?

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---

---

**Hints forthcoming**…
What are the 4 D’s of optic nerve hypoplasia?

1. Drink (heavy EtOH consumption during pregnancy)
2. Diabetes
3. Drugs (especially Dilantin or other seizure meds)
4. De Morsier syndrome

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
Developmental Abnormalities of the Optic Nerve Head

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

- **Drink** (ie, heavy EtOH consumption during pregnancy)
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Concern mom’s life while she is pregnant with the child who will have ON hypoplasia

A congenital condition with significant CNS findings

(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)

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

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

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

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What triad constitutes de Morsier syndrome?
What are the **4 D’s** of optic nerve hypoplasia?

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

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---**De Morsier syndrome**

What triad constitutes de Morsier syndrome? --two words

--hypoplasia (duh)
What are the 4 D’s of optic nerve hypoplasia?

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-- Optic nerve hypoplasia (duh)

-- Absence of the

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Developmental Abnormalities of the Optic Nerve Head

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--Absence of the septum pellucidum

--?
Developmental Abnormalities of the Optic Nerve Head

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

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

--Diabetes

--Drugs (especially Dilantin or other seizure meds)

--De Morsier syndrome

What triad constitutes de Morsier syndrome?
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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 noneponymous name of 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 septum pellucidum

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What is the noneponymous name of de Morsier syndrome?

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

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

--- **Diabetes**

--- **Drugs** (especially **Dilantin** or other seizure meds)

--- **De Morsier syndrome**

**What triad constitutes de Morsier syndrome?**

--- **Optic** nerve hypoplasia (duh)
--- Absence of the **septum** pellucidum
--- Agenesis of the corpus callosum

**What is the noneponymous name of de Morsier syndrome?**

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

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

-- Diabetes

-- Drugs (especially Dilantin or other seizure meds)

--- De Morsier syndrome

What triad constitutes de Morsier syndrome?

-- Optic nerve hypoplasia (duh)
-- Absence of the septum pellucidum
-- Agenesis of the corpus callosum

What is the noneponymous name of de Morsier syndrome?

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

- **D**rink (i.e., heavy EtOH consumption)
- **D**iabetes
- **D**rugs (especially Dilantin or other seizure meds)
- **D**e Morsier syndrome

Note: The listed triad is from the BCSC Peds book.

What triad constitutes de Morsier syndrome?
- Optic nerve hypoplasia (duh)
- Absence of the septum pellucidum
- Agenesis of the corpus callosum

*Note: The listed triad is from the BCSC Peds book. Per the Neuro book, the triad includes hypothalamic-pituitary axis dysfunction, not corpus callosum agenesis (although it states that “the corpus callosum may be thinned or absent”). EyeWiki states the term de Morsier syndrome is “used to describe the association between ONH and the absence of septum pellucidum, deficiency of pituitary hormones and agenesis of corpus callosum.” Which is correct? I dunno. Caveat emptor.*
What are the 4 D’s of optic nerve hypoplasia?

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--- De Morsier syndrome

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---De Morsier syndrome---

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Developmental Abnormalities of the Optic Nerve Head

Of the four, which is the most common cause of optic nerve hypoplasia?

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

--Diabetes

--Drugs (especially Dilantin or other seizure meds)

--De Morsier syndrome
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Of the four, which is the most common cause of optic nerve hypoplasia?

Diabetes

*Maternal diabetes is notorious for causing a specific pattern of optic nerve hypoplasia—what is it?*
What are the 4 D’s of optic nerve hypoplasia?

- Drink (ie, heavy EtOH consumption during pregnancy)
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Of the four, which is the most common cause of optic nerve hypoplasia? Diabetes

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Superior segmental optic nerve hypoplasia (SSONH)
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What is the appearance of the nerve head in SSONH?

--De Morsier syndrome
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What is the appearance of the nerve head in SSONH?

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

De Morsier syndrome
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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Does DM-induced SSONH tend to be unilateral, or bilateral?

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*What pattern of VF loss is associated with SSONH?*
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
What are the 4 D's of optic nerve hypoplasia?

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---

*Does gestational diabetes place a fetus at risk for SSONH?*  
Tough call. I could find no mention of this in the BCSC, Focal Points, or EyeWiki. However, a different (and quite prominent) Academy source explicitly states gestational DM is **not** a risk factor.

*Gestational Diabetes?*

Maternal diabetes nerve hypoplasia

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De Morsier syndrome
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...

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

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Developmental Abnormalities of the Optic Nerve Head

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3)
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Developmental Abnormalities of the Optic Nerve Head

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Tilted-Disc Syndrome aka...Fuch's Coloboma
1) Superior pole appears...elevated, inferior...recessed
2)
3)
4)
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

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Tilted-Disc Syndrome aka Fuch’s Coloboma
1) Superior pole appears elevated, inferior recessed
2) Associated with [two words] of retinal vessels
3)
4)

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Developmental Abnormalities of the Optic Nerve Head

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aka Fuch's Coloboma
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Huh? I thought situs inversus meant all the organs were on the wrong side of the body, or something. What does it mean in this context?
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It means the... nasal v temp... vessels run... direction for a short interval before heading off in the right direction
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These three are all secondary to abnormal closure of the embryonic optic fissure

Huh? I thought situs inversus meant all the organs were on the wrong side of the body, or something. What does it mean in this context?

It means the temporal vessels run nasally for a short interval before heading off in the right direction
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.

Normal OD posterior pole for comparison

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

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  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

Optic Nerve Coloboma

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

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**
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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
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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…[specific VF finding] that doesn’t respect the vertical, and may resolve with…[simple clinical maneuver]
Developmental Abnormalities of the Optic Nerve Head

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Tilted-Disc Syndrome aka...Fuch’s Coloboma
1) Superior pole appears...elevated, inferior...recessed
2) Ritus inversus of retinal vessels produces...myopic astigmatism that doesn’t...bitemporal loss
3) May resolve...refraction

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

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

bitemporal loss
Developmental Abnormalities of the Optic Nerve Head

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

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

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

Tilted-disc syndrome: Superior bitemporal VF loss
Developmental Abnormalities of the Optic Nerve Head

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**Tilted-Disc Syndrome** aka...Fuch's Coloboma
1) Superior pole appears...elevated
2) Inferonasal ONH is longer than other regions.
3) Inferonasal retina is more myopic than others.
4) VF testing reveals...bitemporal loss
5) May resolve with...refraction

---

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

- Usually...20/20, but can be...
- 20/20<->NLP
- 1/3 develop...serous RD
Developmental Abnormalities of the Optic Nerve Head

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Optic Nerve Hypoplasia
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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
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3) Fundus abnormality produces myopic astigmatism
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aka... Fuch’s Coloboma
1) Superior pole appears... elevated, inferior... recessed
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Developmental Abnormalities of the Optic Nerve Head

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Developmental Abnormalities of the Optic Nerve Head

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

Megalopapilla
1) Abnormally large diameter of disc and cup
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Myelinated RNFL
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1) Superior pole appears elevated, inferior recessed
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4) VF testing reveals bitemporal loss that doesn't respect the vertical, and may resolve with refraction

When you hear 'superior bitemporal VF defect' two words come to mind, and they ain't Fuchs coloboma—they are _________.

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 which localizes in the superotemporal VF. And because Fuchs coloboma is almost always a bilateral condition, the VF loss is too.

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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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Optic Nerve Coloboma
When you hear ‘superior bitemporal VF defect’ two words come to mind, and they ain’t Fuchs coloboma—they are pituitary tumor.

Megalopapilla
1) Abnormally large diameter of... disc and cup
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When you hear ‘superior bitemporal VF defect’ two words come to mind, and they ain’t Fuchs coloboma—they are pituitary tumor.

OK, fair. But is there a way to tell from the VF itself? Indeed there is—a bitemporal VF defect secondary to a pituitary tumor will always respect the vertical midline, whereas one secondary to Fuchs coloboma will not. Put another way: A pituitary tumor, but not a Fuchs coloboma, is expected to produce bitemporal hemianopic VF loss.
Developmental Abnormalities of the Optic Nerve Head

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Optic Nerve Coloboma
--Asymmetric and asymmetric

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 which localizes in the superotemporal VF. And because Fuchs coloboma is almost always a bilateral condition, the VF loss is too.

When you hear 'superior bitemporal VF defect' two words come to mind, and they ain’t Fuchs coloboma—they are pituitary tumor. How can you tell whether a bitemporal VF cut results from a pituitary tumor as opposed to Fuchs coloboma?

Optic Nerve Coloboma

Optic Nerve Hypoplasia

Megalopapilla

Myelinated RNFL

Optic Nerve Coloboma

Optic Nerve Hypoplasia

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Megalopapilla

Myelinated RNFL

Tilted-Disc Syndrome
aka Fuch’s Coloboma
1) Superior pole appears elevated, inferior recessed
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When the ONH is tilted, the inferonasal region becomes more myopic and the VF testing reveals an enlarged blind spot.

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(but not hemianopic)
Developmental Abnormalities of the Optic Nerve Head

Tilted-disc syndrome: Superior bitemporal VF loss
(Note that the VF loss does not respect the vertical midline)
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Finally: Fuchs coloboma is associated with a rare inherited retinal condition. What is it?

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Fuchs coloboma is associated with a rare inherited retinal condition. What is it?

CSNB
What does CSNB stand for in this context?
Congenital stationary night blindness
In a nutshell, what's CSNB?
A congenital condition in which a dearth of functioning rods leads to nyctalopia, nystagmus, and variably decreased VA
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--- rods vs cones

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4) VF testing reveals bitemporal loss that doesn’t respect the vertical, and may resolve with refraction

--- rods vs cones

--- abb.

**CSNB**

What does CSNB stand for in this context?

Congenital stationary night blindness

In a nutshell, what’s CSNB?

A congenital condition in which a dearth of functioning rods leads to nystagmus, nyctalopia, and variably decreased VA

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**Finally: Fuchs coloboma is associated with a rare inherited retinal condition. What is it?**

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**Tilted-Disc Syndrome aka Fuch’s Coloboma**

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--- abb.
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

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

Tilted-Disc Syndrome aka Fuch’s Coloboma