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
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

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

Developmental Abnormalities of the Optic Nerve Head

Megalo-...
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

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

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

Myelinated…
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
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

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

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-- 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
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Optic Pit/Hole
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Morning-Glory Disc
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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

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  - 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

- 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

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
Developmental Abnormalities of the Optic Nerve Head

- Myelinated RNFL
  - Myelin normally starts at LGN, ends at lamina cribrosa
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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

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

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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

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

Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

Optic Pit/Hole

Morning-Glory Disc
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

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

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

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

**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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**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 respect the vertical and may resolve with refraction

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

**Myelinated RNFL**
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- Corresponding VF has absolute scotoma

**Optic Pit/Hole**
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... **[battle cry?] association**

Optic Pit/Hole

Morning-Glory Disc

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 \textit{deep cupping}
2) Can be \textit{bilateral, asymmetric}
3) Part of the \textbf{CHARGE association}

Optic Pit/Hole

Morning-Glory Disc

Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head
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

Megalopapilla

Hypoplasia

Tilted-Disc Syndrome

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

What is the CHARGE association?

C - Coloboma
H - Hypoplasia
A - Anomalies of the ear
R - Retardation
G - Genitourinary abnormalities
E - Eye abnormalities

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

Myelinated RNFL
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Optic Nerve Hypoplasia
--Abnormally low number of axons
--DFE: Small pale disc with double ring sign
--VA 20/20<->NLP
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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

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.
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) Associated with... *situs inversus*
4) Fundus abnormality produces... *myopic astigmatism*
5) VF testing reveals... *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

Myelinated RNFL
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Optic Nerve Hypoplasia
--Abnormally low number of axons
--DFE: Small pale disc with double ring sign
--VA 20/20<->NLP
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Optic Pit/Hole
--Associated with serous RD in adulthood

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Morning-Glory Disc

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

Tilted-Disc Syndrome
1) Superior pole appears...elevated, inferior...recessed
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
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
Morning-Glory Disc
--DFE reveals:
1) A funnel-shaped... *classic term*
2)

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)

Megalopapilla

Myelinated RNFL
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Megalopapilla

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 Coloboma
1) May resemble... *deep cupping*
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3) Part of the... CHARGE association

Optic Pit/Hole
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Optic Nerve Coloboma
1) May resemble... *deep cupping*
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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)

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 respect the vertical and may... resolve with refraction
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) 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
--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)

Developmental Abnormalities of the Optic Nerve Head

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

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

Optic Nerve Hypoplasia

Tilted-Disc Syndrome
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

Optic Nerve Coloboma
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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
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

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

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...
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

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

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

Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

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

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

**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... *[descriptor]*, so cup seems to... *[type of change]* (like its namesake, the morning-glory flower)

**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*
2) Inferior... *recessed*
3) Associated with... *situs inversus of retinal vessels*
4) 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…CHASE 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
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...[###], but can be...[range]

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 respect the vertical and may resolve with refraction
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*

---

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

**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... *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
--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]

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**
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--VA usually...**20/200**, but can be...**20/20<->NLP**
--1/3 develop...**serous RD**

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

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

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**Tilted-Disc Syndrome**
1) Superior pole appears...**elevated**, inferior...**recessed**
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**Megalopapilla**

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

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**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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Is morning-glory disc usually unilateral, or bilateral?

Unilateral

Is there a gender predilection?

Yes, there is a female (♀) preponderance

It has two associations of particular note—what are they?

--PHACE syndrome
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Developmental Abnormalities of the Optic Nerve Head

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Megalopapilla

RNFL

Optic Nerve Hypoplasia

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Is morning-glory disc usually unilateral, or bilateral? Unilateral

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Morning-Glory Disc

Is the optic nerve head abnormally myelinated?
Yes, myelinated RNFL is present
--Myelin normally starts at LGN, ends at lamina cribrosa
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--Abnormally large diameter of disc and cup
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Where is the hemangioma located?
The face, +/- scalp involvement

Is it large, or small? Large

What is the classic term for its shape? 'Plaquelike'

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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? Unilateral
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It has two associations—what are they?
--PHACE syndrome
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What does PHACE stand for?
P —Posterior fossa malformations
H —Hemangiomas
A —Arterial lesions
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Where is the hemangioma located? The face, +/- scalp

Is the hemangioma large? Large
What is the classic term for its shape? 'Plaquelike'

Tilted-Disc Syndrome
1) Superior pole appears elevated, inferior recessed
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1) May resemble deep cupping
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**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

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Moyamoya disease
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

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The chronic occlusions result in the development of collaterals. On angiography, these collaterals manifest as wispy areas that have been likened to a puff of smoke. Moyamoya is the Japanese word for this 'hazy puff of smoke' appearance.
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Normal angiogram

Angiogram in moyamoya
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Tilted-Disc Syndrome
1) Superior pole appears elevated, inferior recessed
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Is morning-glory disc usually unilateral, or bilateral?

Unilateral

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

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Yes, it is more common in Asians, especially Japanese and Korean individuals
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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

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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Abnormally low number of axons

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How does moyamoya present in childhood?
It manifests most commonly in childhood

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How does moyamoya present in childhood?
With TIA's

It manifests most commonly in childhood
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How does moyamoya present in childhood?
With TIAs

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!
Developmental Abnormalities of the Optic Nerve Head

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--DFE reveals:
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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

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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

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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*

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1) May resemble... *deep cupping*
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**Megalopapilla**

1) Abnormally large diameter of... *disc and cup*
2) 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)

**Tilted-Disc Syndrome**

1) Superior pole appears... *elevated*, inferior... *recessed*

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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?

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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

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

- **Megalopapilla**
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- **Optic Pit/Hole**
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- **Morning-Glory Disc**
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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?**
  - CNS ischemia in the perinatal period

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

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--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

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?
CNS ischemia in the perinatal period

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

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
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Developmental Abnormalities of the Optic Nerve Head

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1) May resemble deep cupping
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And it (almost) goes without saying…What condition must be considered in any individual with an enlarged cup?

Optic Nerve Hypoplasia

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**Glaucoma**

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2) 3)

Where does myelination normally begin?

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Where does myelination normally begin?
At the lateral geniculate nucleus

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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.)

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Optic Nerve Hypoplasia
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2)
3)
4)
5)

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Hypoplastic optic nerve heads

Optic nerve hypoplasia
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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1) Abnormally low number of axons
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3) A hypoplastic nerve with double-ring sign can easily be mistaken for what?

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3)

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).

Optic nerve hypoplasia: *Double-ring sign*
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Is the VF loss associated with optic nerve hypoplasia progressive, or nonprogressive?

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

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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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5) Remember the…**[mnemonic mnemonic]**

Tilted-Disc Syndrome

1) Superior pole appears…**elevated**, inferior…**recessed**
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3) Fundus abnormality produces…**myopic astigmat**
4) VF testing reveals…**bitemporal hemianopia** that doesn’t…**respect the vertical** and may…**resolve with refraction**

Tilted-Disc Syndrome aka…**Fuch’s coloboma**
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You see a child with optic nerve hypoplasia. What steps should you at least consider taking?

-- Obtaining MRI brain—a variety of CNS abnormalities are associated with it (will unpack this in the upcoming 4 D’s section)
-- Getting an endocrine consult—multiple hormonal deficiencies are associated (mainly pituitary-related)
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--- MRI brain ---

pituitary-related

What steps should you at least consider if you see a child with ON hypoplasia?

--- Pituitary-related ---

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Anterior pituitary hypoplasia coupled with an...ectopic posterior pituitary

How will this manifest on neuroimaging?

As an absent/hypoplastic anterior pituitary coupled with a...bright spot at the upper infundibulum
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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
...neonatal jaundice, think: Hypothyroidism
...hypoglycemia/seizures, think: Panhypopituitarism
...diabetes insipidus, think: Hypocortisolism
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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 (more on this shortly)

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

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

You see a child with optic nerve hypoplasia. What steps should you at least consider taking?

--Obtaining MRI brain—A variety of CNS abnormalities are associated with it (will unpack this in the upcoming 4 D's section)
--Getting an endocrine consult—Multiple hormonal deficiencies are associated (mainly pituitary-related)

What is the classic structural pituitary abnormality associated with ON hypoplasia?
Anterior pituitary hypoplasia coupled with an ectopic posterior pituitary

How will this manifest on neuroimaging?
As an absent/hypoplastic anterior pituitary coupled with a bright spot at the upper infundibulum

What steps should you at least consider taking?

What pituitary-related deficiencies may be present?
Anything up to and including panhypopituitarism

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**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**

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   - 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 (more on this shortly).

**Optic Nerve Coloboma**

1. May resemble deep cupping.
2. Can be bilateral, asymmetric.
3. Part of the...CHARGE association.
4. MRI brain...pituitary-related.

**Optic Pit/Hole**

--Associated with serous RD in adulthood.

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

**Megalopapilla**

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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:

- the morning-glory flower)
- VA usually...20/200, but can be... 20/20<->NLP
- 1/3 develop...serous RD

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

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

**Optic Nerve Coarctation**
1. May resemble... (details not fully visible)
2. Can be... (details not fully visible)
3. Part of the... (details not fully visible)

**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 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 (more on this shortly).

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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... (details not fully visible)

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*You see a child with optic nerve hypoplasia. What steps should you at least consider taking?*

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--Getting an endocrine consult—multiple hormonal deficiencies are associated (mainly pituitary-related).

*What is the classic structural pituitary abnormality associated with ON hypoplasia?*

Anterior pituitary hypoplasia coupled with an ectopic posterior pituitary.

*How will this manifest on neuroimaging?*

As an absent/hypoplastic anterior pituitary coupled with a bright spot at the upper infundibulum.

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--Getting an endocrine consult—multiple hormonal deficiencies are associated (mainly pituitary-related).

*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*

---

*What is the classic structural pituitary abnormality associated with ON hypoplasia?*

Anterior pituitary hypoplasia coupled with an ectopic posterior pituitary.

*How will this manifest on neuroimaging?*

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Optic Pit/Hole

1) Associated with serous RD in adulthood.

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

Optic Nerve Hypoplasia

1) Abnormally low number of axons.

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3) VA 20/15<->NLP.

4) VF defects invariably present.

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

What is the classic structural pituitary abnormality associated with ON hypoplasia?

Anterior pituitary hypoplasia coupled with an ectopic posterior pituitary.

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As an absent/hypoplastic anterior pituitary coupled with a bright spot at the upper infundibulum.

What are some pituitary-related deficiencies that may be present?

Anything up to and including panhypopituitarism.

What pituitary-related deficiencies may be present?

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If an infant/child has ON hypoplasia and...

...short stature, think: Growth-hormone deficiency.

...neonatal jaundice, think:

the morning-glory flower.

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

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--1/3 develop...serous RD.

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Morning-Glory Disc

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...

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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.

2) Can be bilateral, asymmetric.

3) Part of the CHARGE association.
Developed 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*

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

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

**Optic Nerve Coloboma**
1) May resemble… *deep cupping*
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**Optic Nerve Hypoplasia**
1) Abnormally low number of… *axons*
2) *DFE*: Small pale disc with… *double ring sign*
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You see a child with optic nerve hypoplasia. What steps should you at least consider taking?

--Obtaining… *MRI brain*—a variety of CNS abnormalities are associated with it (will unpack this in the upcoming *4 D’s*’ section)
--Getting an endocrine consult—multiple hormonal deficiencies are associated—*mainly pituitary-related*

What is the classic *structural* pituitary abnormality associated with ON hypoplasia?
Anterior pituitary hypoplasia coupled with an… *ectopic posterior pituitary*

*How will this manifest on neuroimaging?*
As an absent/hypoplastic anterior pituitary coupled with a bright spot at the upper infundibulum

*What steps should you at least consider taking?*

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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…*neonatal jaundice, think*: Hypothyroidism

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Optic nerve hypoplasia
1) Abnormally low number of… *axons*
2) DFE: Small pale disc with… *double ring sign*
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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*

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

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Megalopapilla
1) Abnormally large diameter of… disc and cup

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Morning-Glory Disc
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**If an infant/child has ON hypoplasia and**
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Developmental Abnormalities of the Optic Nerve Head

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   --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

Optic Nerve Coloboma

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--Obtaining MRI brain—a variety of CNS abnormalities are associated with it
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What is the classic structural pituitary abnormality associated with ON hypoplasia?
Anterior pituitary hypoplasia coupled with an ectopic posterior pituitary

How will this manifest on neuroimaging?
As an absent/hypoplastic anterior pituitary coupled with a bright spot at the upper infundibulum

What steps should you at least consider taking?
--Obtaining MRI brain—a variety of CNS abnormalities are associated with it
--Getting an endocrine consult—multiple hormonal deficiencies are associated

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
...neonatal jaundice, think: Hypothyroidism
...hypoglycemia/seizures, think: Panhypopituitarism
...diabetes insipidus, think: Hypocortisolism

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. 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.

Myelinated RNFL

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

Megalopapilla

1) Abnormally large diameter of disc and cup. VF testing may reveal an enlarged blind spot. VA usually 20/200, but can be 20/20 <-> NLP. 1/3 develop serous RD.

Morning-Glory Disc

--DFE reveals:
1) A funnel-shaped excavation. 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.

--Obtaining MRI brain—a variety of CNS abnormalities are associated with it (will unpack this in the upcoming 4 D's section)
--Getting an endocrine consult—multiple hormonal deficiencies are associated with it. These are probably low-yield factoids in isolation.

Optic Nerve Coloboma

1) May resemble deep cupping. Can be bilateral, asymmetric. Part of the CHARGE association. What steps should you at least consider taking?
--Obtaining MRI brain—a variety of CNS abnormalities are associated with it
--Getting an endocrine consult—multiple hormonal deficiencies are associated

Optic Nerve Hypoplasia

1) Abnormally low number of axons. DFE: Small pale disc with double ring sign. VA 20/15 <-> NLP. VF defects invariably present. Remember the 4 D's (more on this shortly)

What is the classic structural pituitary abnormality associated with ON hypoplasia? Anterior pituitary hypoplasia coupled with an ectopic posterior pituitary.

What steps should you at least consider taking?
--Obtaining MRI brain—a variety of CNS abnormalities are associated with it (will unpack this in the upcoming 4 D’s section)
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Optic Pit/Hole

Associated with serous RD in adulthood.

Morning-Glory Disc

--DFE reveals:
1) A funnel-shaped excavation. 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.

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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

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 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 (more on this shortly)

What is the classic structural pituitary abnormality associated with ON hypoplasia?
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What steps should you at least consider taking?
--Obtaining MRI brain—a variety of CNS abnormalities are associated with it
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What are the pituitary-related deficiencies that may be present?
Anything up to and including panhypopituitarism

If an infant/child has ON hypoplasia and...
...short stature, think: Growth-hormone deficiency
...neonatal jaundice, think: Hypothyroidism
...hypoglycemia/seizures, think: Panhypopituitarism
...diabetes insipidus, think: Hypocortisolism
...9/10 brain abnormalities, think:...
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
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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

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

As for the four Ds of optic nerve hypoplasia:
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?

- Drink (heavy EtOH consumption during pregnancy)
- Diabetes
- Drugs (especially Dilantin or other seizure meds)
- 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
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

(should be lower-case, but it looked funny)

A congenital condition with significant CNS findings

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

--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**

--- **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)

--Diabetes

--Drugs (especially Dilantin or other seizure meds)

What triad constitutes de Morsier syndrome?

--De Morsier syndrome

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

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

--Diabetes

--Drugs (especially Dilantin or other seizure meds)

What triad constitutes de Morsier syndrome?

--De Morsier syndrome

--?

--?

-- two words

hypoplasia (duh)
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)

What triad constitutes de Morsier syndrome?

--Optic nerve hypoplasia (duh)

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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
--?
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

--?
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?

--Optic nerve hypoplasia (duh)

--Absence of the septum pellucidum

--Agenesis of the two diff words
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)

What triad constitutes de Morsier syndrome?

--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
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?

--- **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-**
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?

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

--Drunkenness (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

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?

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

What triad constitutes de Morsier syndrome?

--Optic nerve hypoplasia (duh)
--Absence of the septum pellucidum
--Agenesis of the corpus callosum
--?
What are the 4 D’s of optic nerve hypoplasia?

---

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”).

---

What triad constitutes de Morsier syndrome?

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De Morsier syndrome

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

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

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.”

De Morsier syndrome

- Optic nerve hypoplasia (duh)
- Absence of the septum pellucidum
- Agenesis of the corpus callosum
- Hypothalamic-pituitary axis dysfunction
What are the 4 D’s of optic nerve hypoplasia?

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?

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

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

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?
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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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?

Superior segmental optic nerve hypoplasia (SSONH)
What are the 4 D's of optic nerve hypoplasia?

- Drink (ie, heavy EtOH consumption during pregnancy)
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- Drugs (especially Dilantin or other seizure meds)
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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?

Superior segmental optic nerve hypoplasia (SSONH)

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"

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

Bilateral

What pattern of VF loss is associated with SSONH?

Bitemporal inferior loss that doesn't respect the vertical midline
What are the 4 D's of optic nerve hypoplasia?

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--De Morsier syndrome
Developmental Abnormalities of the Optic Nerve Head

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

Superior segmental ON hypoplasia
What are the 4 D's of optic nerve hypoplasia?

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- Drugs (especially Dilantin or other seizure meds)
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Bilateral

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?

- Drink (i.e., heavy EtOH consumption during pregnancy)
- Diabetes
- Drugs (especially Dilantin or other seizure meds)
- De Morsier syndrome

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

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Gestational Diabetes?

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.

Gestational diabetes? Of the four, would you say this is the most common cause of optic nerve hypoplasia? Diabetes? Yes and no. Let me explain.

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

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

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

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?

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**De Morsier syndrome**
Developmental Abnormalities of the Optic Nerve Head

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--De Morsier syndrome

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. Further, this source goes on to say that SSONH risk is associated with insulin-dependent status of the mother. This assertion is supported by two Focal Points publications, but is not mentioned in the BCSC. Caveat emptor.
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
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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

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

By what eponymous name is tilted-disc syndrome known?
Developmental Abnormalities of the Optic Nerve Head

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Tilted-Disc Syndrome aka… Fuch’s Coloboma
By what eponymous name is tilted-disc syndrome known?
Fuch’s coloboma

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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) **[elevated]**
3) **[elevated]**
4) **[recessed]**
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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.
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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? It means the... nasal v temp vessels run... direction for a short interval before heading off in the right direction

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Morning-Glory Disc --DFE reveals:
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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

These three are all secondary to abnormal closure of the embryonic optic fissure
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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Tilted-Disc Syndrome aka...Fuch's Coloboma
1) Superior pole appears...elevated, inferior...recessed
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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]

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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
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Tilted-Disc Syndrome aka...Fuch's Coloboma
1) Superior pole appears...elevated, inferior...recessed
2) Axial images of the vitreous...vitreous...avitreous...recessed
3) Inferior pole...imated...scissors...scissors...recessed
4) VF testing reveals...bitemporal loss
5) Remember the...4 D's

Optic Pit/Hole
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Optic Nerve Coloboma
1) May resemble...deep cupping
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1) Abnormally large diameter of...disc and cup
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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 loss that doesn't respect the vertical midline, and may resolve with refraction

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?

For starters, by examining the pt. The ONHs in a Fuchs coloboma pt will be highly tilted and manifest situs inversus, whereas the ONHs of a pituitary-tumor pt will be unremarkable or edematous.

OK, fair. But is there a way to tell from the VF itself?

Indeed there is—a bitemporal VF defect 2ndry to a pituitary tumor will always respect the vertical midline, whereas one 2ndry 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

Tilted-disc syndrome: Superior bitemporal VF loss
(Note that the VF loss does not respect the vertical midline)
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--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

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

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
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Finally: 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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