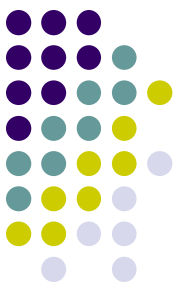


Developmental Abnormalities of the Optic Nerve Head



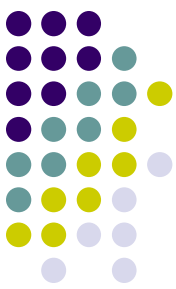
These three are all secondary to abnormal closure of the embryonic optic fissure

Optic Nerve...

Optic...

[Two-words] Disc

Developmental Abnormalities of the Optic Nerve Head



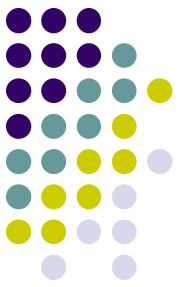
These three are all secondary to abnormal closure of the embryonic optic fissure

Optic Nerve Coloboma

Optic Pit/Hole

Morning-Glory Disc

Developmental Abnormalities of the Optic Nerve Head



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

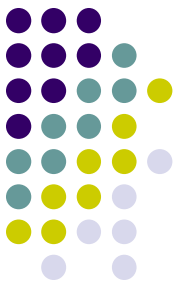
Optic Nerve Coloboma

Optic Pit/Hole

Morning-Glory Disc

Megalo-...

Developmental Abnormalities of the Optic Nerve Head



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

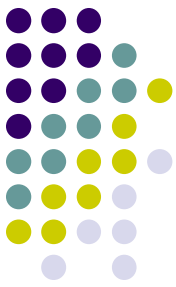
Optic Nerve Coloboma

Optic Pit/Hole

Morning-Glory Disc

Megalopapilla

Developmental Abnormalities of the Optic Nerve Head



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

Optic Nerve Coloboma

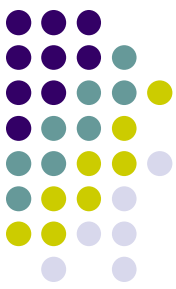
Optic Pit/Hole

Morning-Glory Disc

Megalopapilla

Myelinated...

Developmental Abnormalities of the Optic Nerve Head



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

Optic Nerve Coloboma

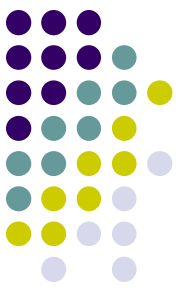
Optic Pit/Hole

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

Optic Nerve Coloboma

Optic Pit/Hole

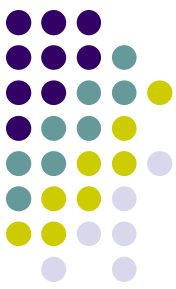
Morning-Glory Disc

Megalopapilla

Myelinated RNFL

Optic Nerve...

Developmental Abnormalities of the Optic Nerve Head



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

Optic Nerve Coloboma

Optic Pit/Hole

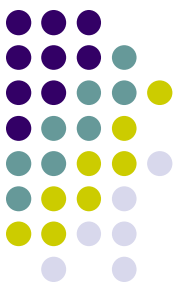
Morning-Glory Disc

Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Developmental Abnormalities of the Optic Nerve Head



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

Optic Nerve Coloboma

Optic Pit/Hole

Morning-Glory Disc

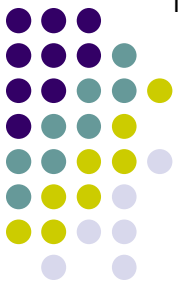
Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

[Two-words] Syndrome

Developmental Abnormalities of the Optic Nerve Head



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

Optic Nerve Coloboma

Optic Pit/Hole

Morning-Glory Disc

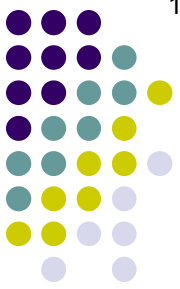
Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

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... *[common acquired appearance]*
- 2)
- 3)

Optic Pit/Hole

Morning-Glory Disc

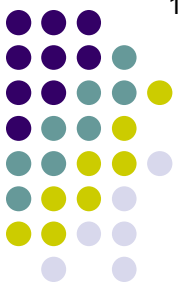
Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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

Optic Nerve Coloboma

- 1) May resemble...**deep cupping**
- 2)
- 3)

Optic Pit/Hole

Morning-Glory Disc

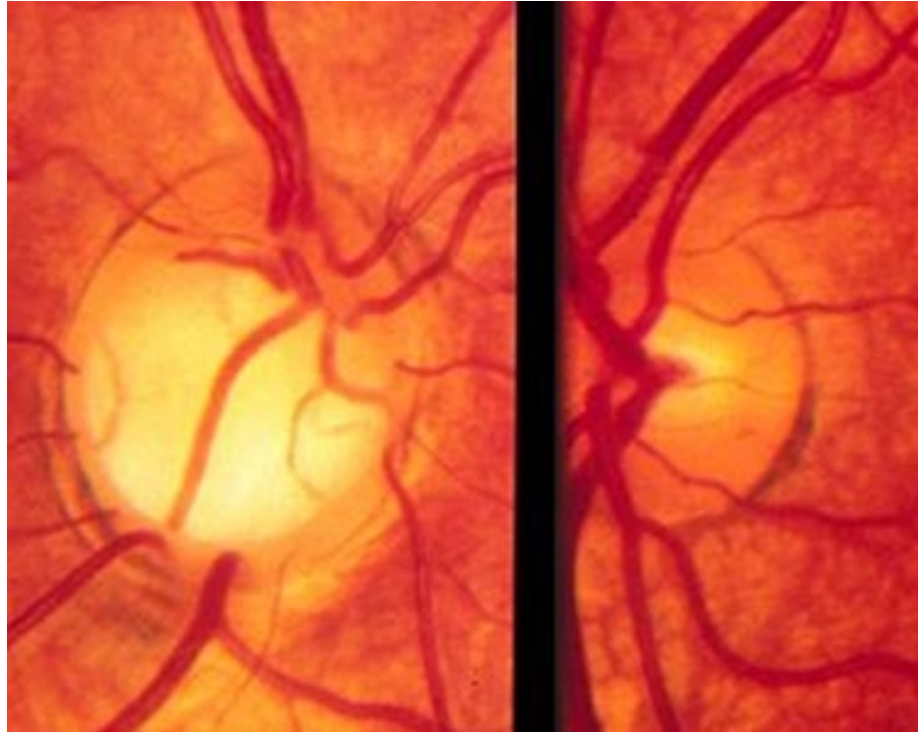
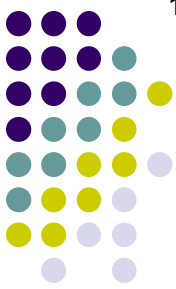
Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

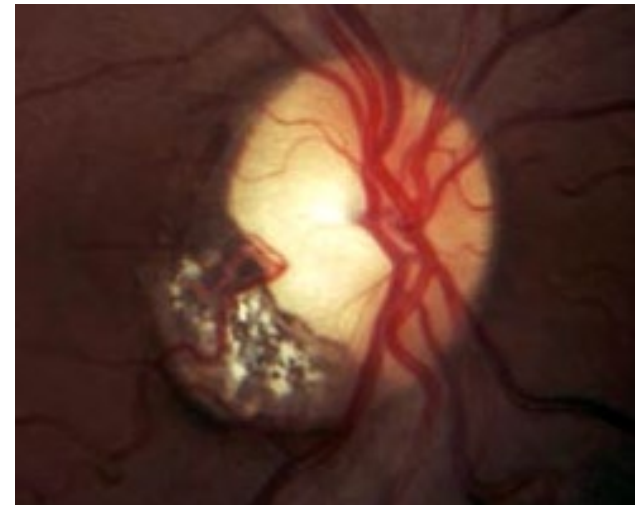
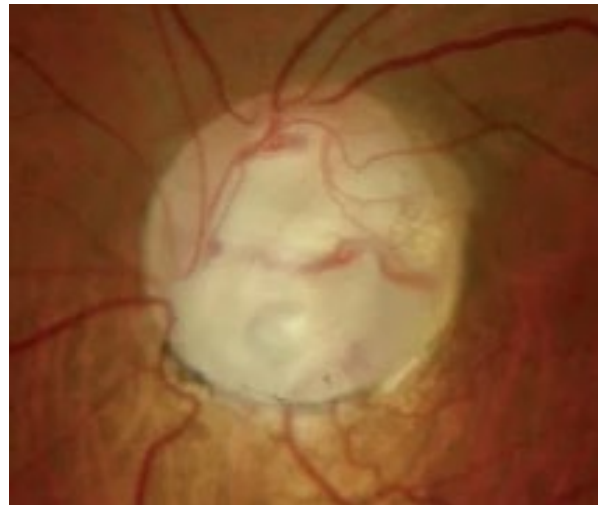
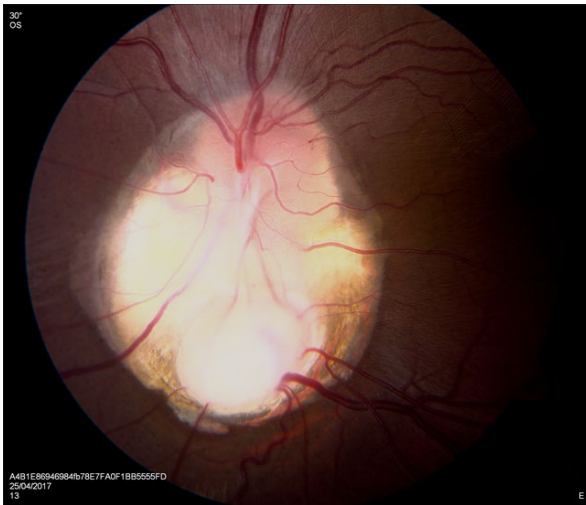
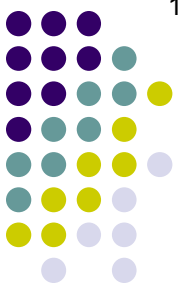
Developmental Abnormalities of the Optic Nerve Head



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

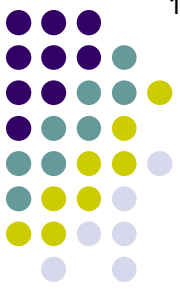
Optic nerve coloboma

Developmental Abnormalities of the Optic Nerve Head



Optic nerve coloboma: More examples

Developmental Abnormalities of the Optic Nerve Head



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



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)

Optic Pit/Hole

Morning-Glory Disc

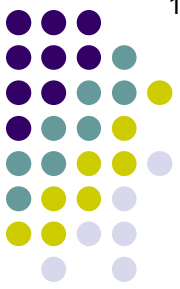
Megalopapilla

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...**[battle cry?] association**

Optic Pit/Hole

Morning-Glory Disc

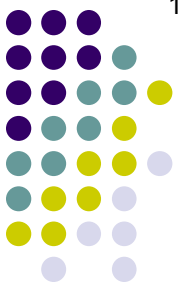
Megalopapilla

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

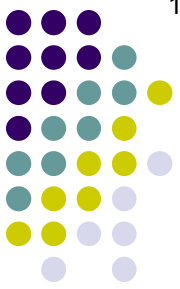
Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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

What is the CHARGE association?

Coloboma

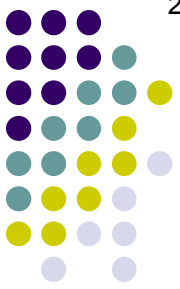
**H
A
R
G
E**

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

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

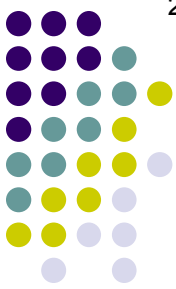
What is the CHARGE association?

Coloboma
Hear abnormalities
Choanal **A**tresia
Retardation
Genitourinary abnormalities
Ear abnormalities

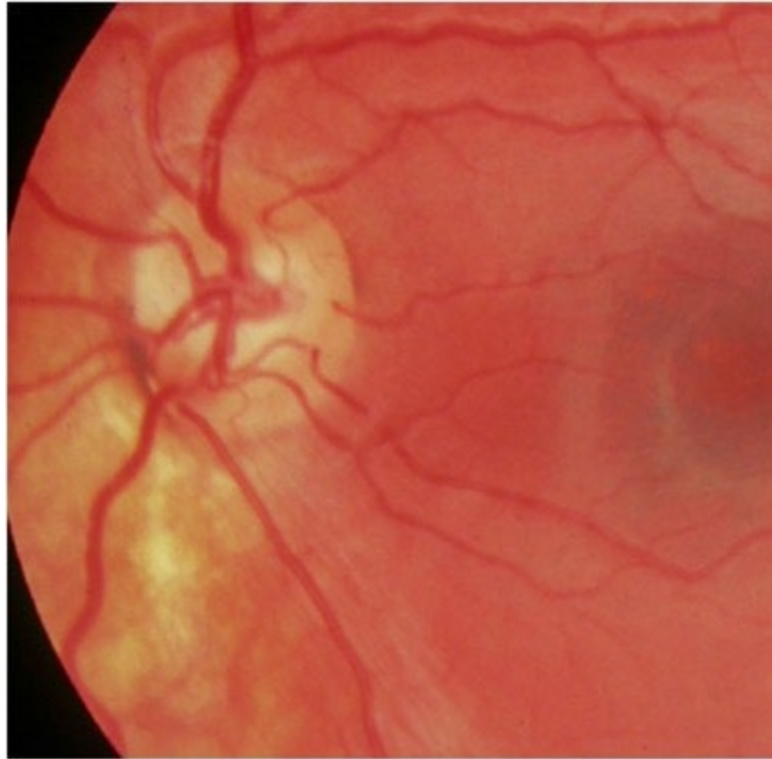
Hypoplasia

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



21



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

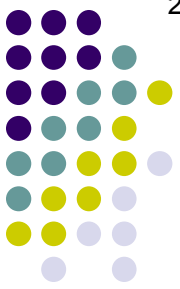
Developmental Abnormalities of the Optic Nerve Head



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.*

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

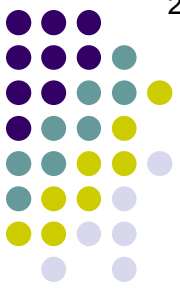
Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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

Optic Nerve Coloboma

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

Optic Pit/Hole

--Associated with...**serous RD in adulthood**

Morning-Glory Disc

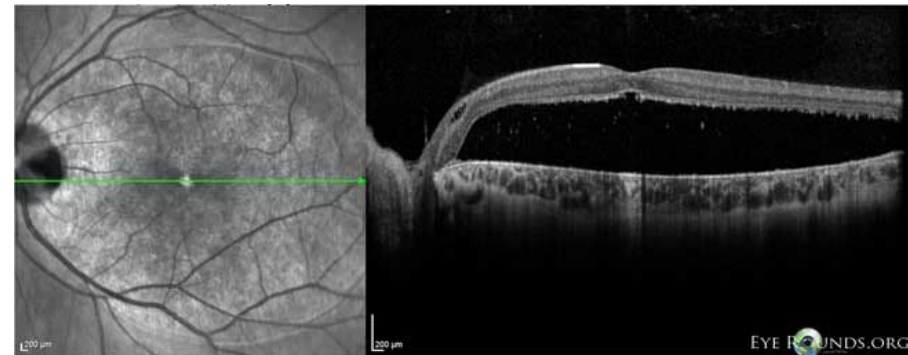
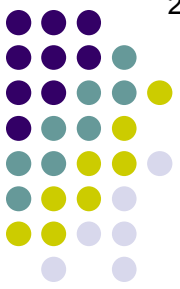
Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

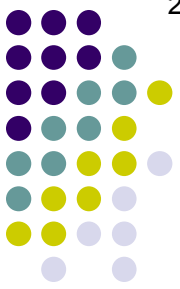
Developmental Abnormalities of the Optic Nerve Head



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



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

Optic Nerve Coloboma

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

Optic Pit/Hole

--Associated with... **serous RD in adulthood**

Morning-Glory Disc

--DFE reveals:

- 1) A funnel-shaped... *[classic term]*
- 2)

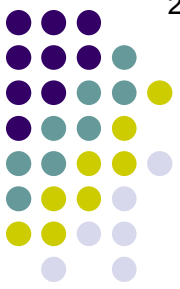
Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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

Optic Nerve Coloboma

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

Optic Pit/Hole

--Associated with...**serous RD in adulthood**

Morning-Glory Disc

--DFE reveals:

- 1) A funnel-shaped...**excavation**
- 2)

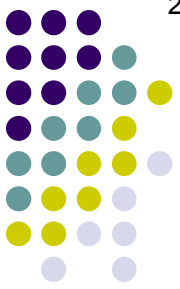
Megalopapilla

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]*

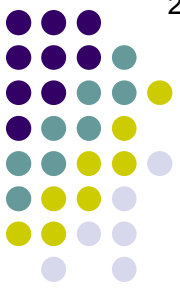
Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

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**

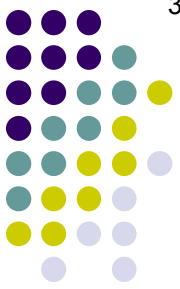
Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

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**

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

Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

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**

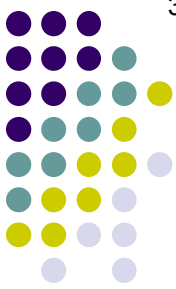
In addition to their number, what else is unusual about the vessels at the ONH? They all emanate from the rim of the disc

Megalopapilla

Myelinated RNFL

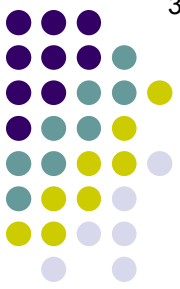
Optic Nerve Hypoplasia

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)

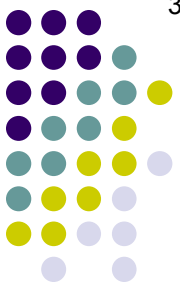
Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

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)

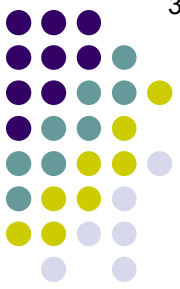
Megalopapilla

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]**

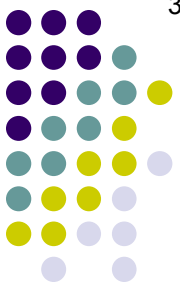
Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

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**

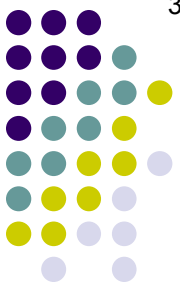
Megalopapilla

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...**20/200**, but can be...**20/20<->NLP**

--1/3 develop... *[retinal condition]*

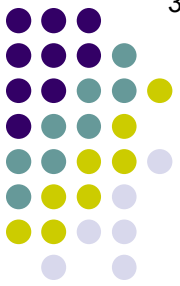
Megalopapilla

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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**

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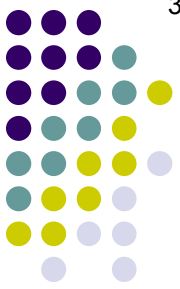
Megalopapilla

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

Is morning-glory disc usually unilateral, or bilateral?

Megalopapilla

RNFL

Optic Nerve Hypoplasia

Morning-Glory Disc

--DFE reveals:

- 1) A funnel-shaped...**excavation**
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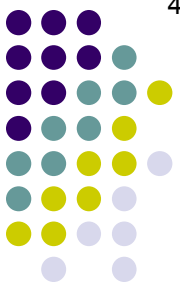
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Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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Unilateral

Megalopapilla

RNFL

Optic Nerve Hypoplasia

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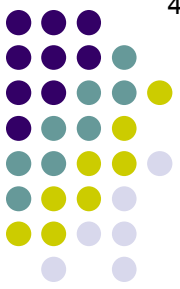
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Developmental Abnormalities of the Optic Nerve Head



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Megalopapilla

RNFL

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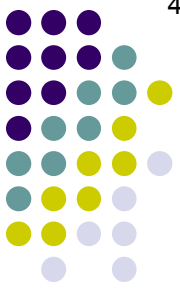
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Is there a gender predilection?

Yes, there is a ♀ preponderance

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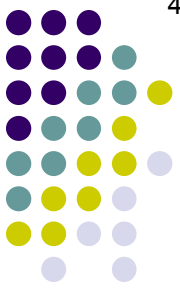
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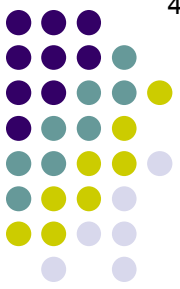
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It has two associations of particular note—what are they?

--?

--?

Megalopapilla

RNFL

Optic Nerve Hypoplasia

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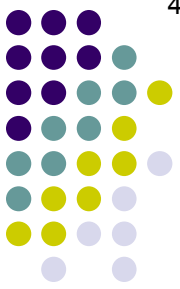
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-- **acronym** syndrome

--?

Megalopapilla

RNFL

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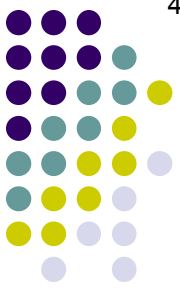
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RNFL

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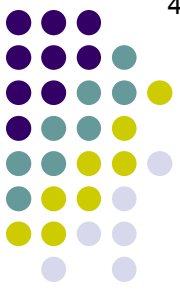
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--disease

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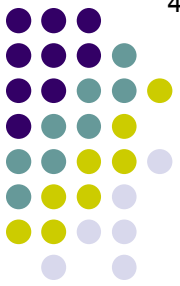
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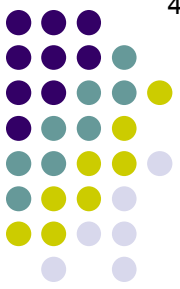
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P
H
A
C
E

Morning-Glory

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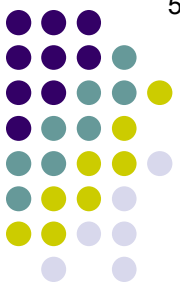
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Posterior fossa malformations

Hemangiomas

Arterial lesions

Cardiac anomalies

Eye anomalies

Morning-Glory

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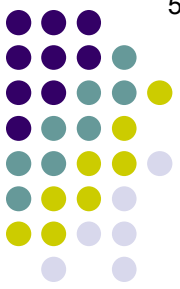
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It has two associations

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--Moyamoya disease

What does PHACE stand for?

Posterior fossa anomalies

Hemangiomas

Arterial lesions

Cardiac anomalies

Eye anomalies

Where is the hemangioma located?

Megalopapilla

RNFL

Alasia

Morning-Glory

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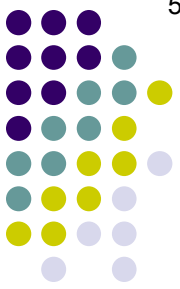
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Arterial lesions

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Eye anomalies

Where is the hemangioma located?

The face, +/- scalp

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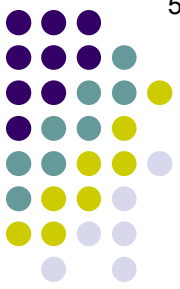
Megalopapilla

RNFL

Glaucoma

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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Unilateral

Is there a gender predilection?

Yes, there is a ♀ preponderance

It has two associations

--**PHACE syndrome**

--Moyamoya disease

What does PHACE stand for?

Posterior fossa

Hemangiomas

Arterial lesions

Cardiac anomalies

Eye anomalies

Where is the hemangioma located?

The face, +/- scalp

Is it large, or small?

Megalopapilla

RNFL

Alasia

Morning-Glory

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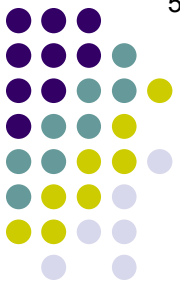
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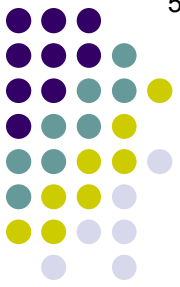
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Arterial lesions

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Eye anomalies

Where is the hemangioma located?

The face, +/- scalp

Is it large, or small?

Large

What is the classic term for its shape?

Morning-Glory

--DFE reveals

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2) Number of vessels crossing the disc seems abnormally...**high**

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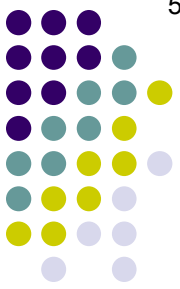
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Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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It has two associations

--**PHACE syndrome**

--Moyamoya disease

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Posterior fossa

Hemangiomas

Arterial lesions

Cardiac anomalies

Eye anomalies

Where is the hemangioma located?

The face, +/- scalp

Is it large, or small?

Large

What is the classic term for its shape?

'Plaquelike'

Megalopapilla

RNFL

Alasia

Morning-Glory

--DFE reveals

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2) Number of vessels crossing the disc seems abnormally...**high**

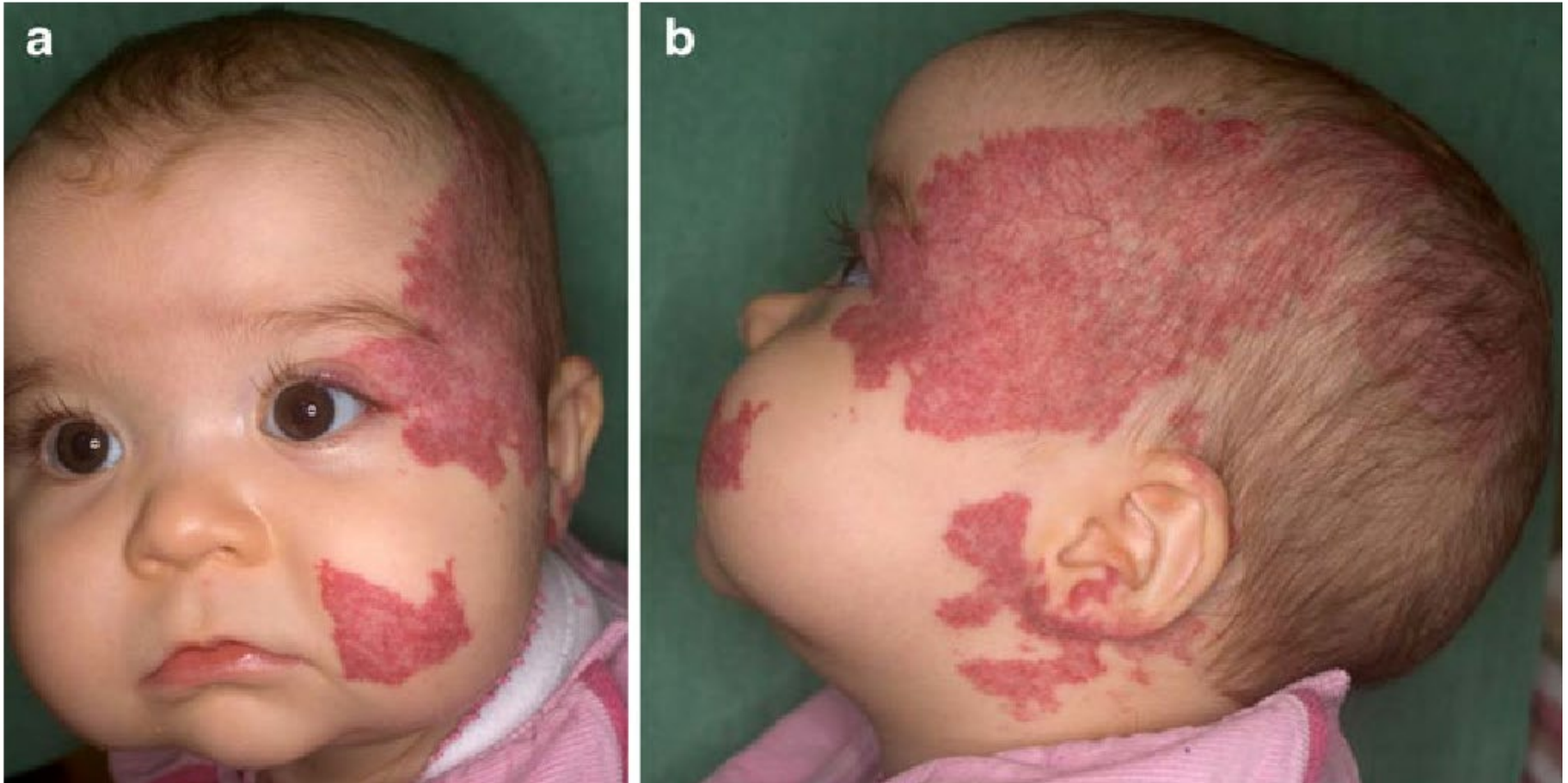
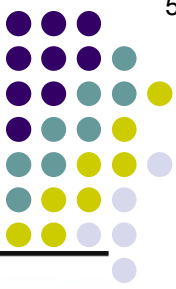
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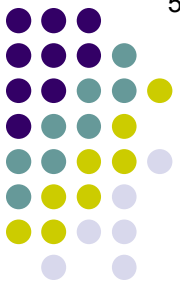
Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



PHACE syndrome: Large plaque-like hemangioma of the face and scalp

Developmental Abnormalities of the Optic Nerve Head



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

Is morning-glory disc usually unilateral, or bilateral?

Unilateral

Megalopapilla

Is there a gender predilection?

Yes, there is a ♀

What is moyamoya disease (MMD)?

It has two associations

--PHACE syndrome

--**Moyamoya disease**

Morning-glory disc

--DFE reveals

1) A funnel-shaped disc

2) Numerous small vessels

seems to be normal

--Tissue is normal

to...open

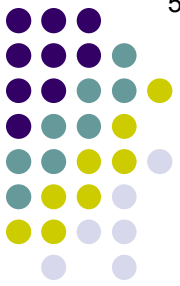
the morning-glory disc

--VA usually normal

20/20 or better

--1/3 develop...serous RD

Developmental Abnormalities of the Optic Nerve Head



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Unilateral

Megalopapilla

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What is moyamoya disease (MMD)?

An vascular condition primarily affecting the terminal and proximal arteries

two arteries

two arteries

It has two associations

--PHACE syndrome

--**Moyamoya disease**

Morning

--DFE re

1) A fun

2) Num

seems

--Tissue i

to...ope

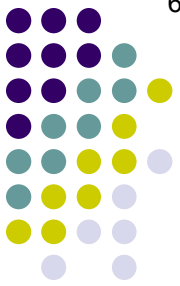
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--VA usua

20/20<

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Developmental Abnormalities of the Optic Nerve Head



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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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Morning

--DFE reveals

1) A fundus

2) Number of

seems

--Tissue

to...open

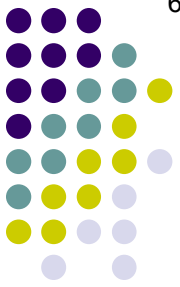
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--VA usually

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--PHACE syndrome

--Moyamoya

What's the story behind the name moyamoya?

Morning-glory disc

--DFE

1) A fundus

2) Num

seems

--Tissue

to...ope

the mor

--VA usual

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--**Moyamoya**

What's the story behind the name moyamoya?

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.

Mor

--DFE

1) A fun

2) Num

seems

--Tissue

to...ope

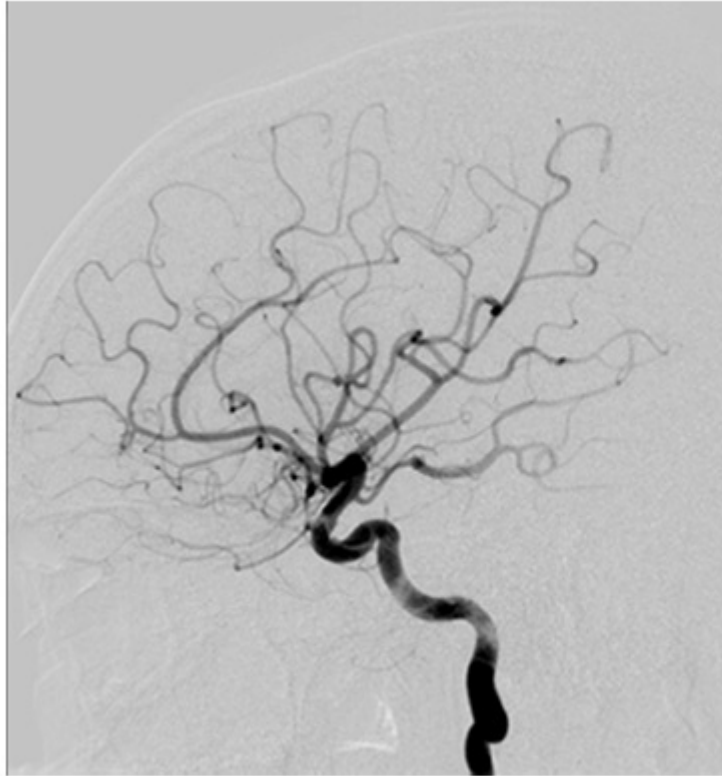
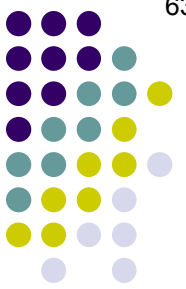
the mor

--VA usual

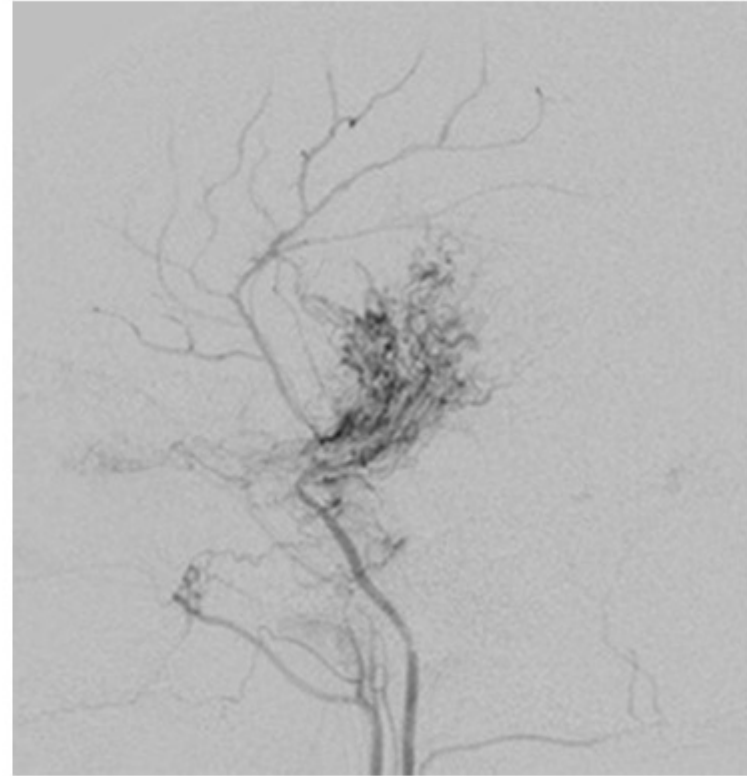
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Developmental Abnormalities of the Optic Nerve Head

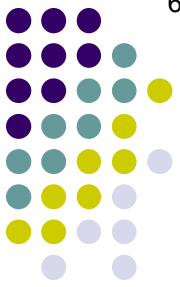


Normal angiogram



Angiogram in moyamoya

Developmental Abnormalities of the Optic Nerve Head



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An occlusive vascular condition primarily affecting the terminal internal carotids and proximal anterior and middle cerebral arteries

What's the story behind the name moyamoya?

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.

Mor

--DFB

1) A fun

2) Num

seems

--Tissue

to...ope

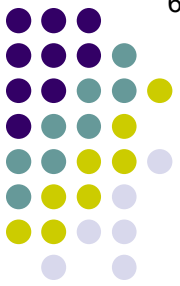
the mor

--VA usua

20/20<

--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

Is morning-glory disc usually unilateral, or bilateral?

Unilateral

Megalopapilla

Is there a gender predilection?

Yes, there is a ♀

What is moyamoya disease (MMD)?

An occlusive vascular condition primarily affecting the terminal internal carotids and proximal anterior and middle cerebral arteries

It has two associated syndromes

--PHACE syndrome

--**Moyamoya disease**

Is it common, or rare?

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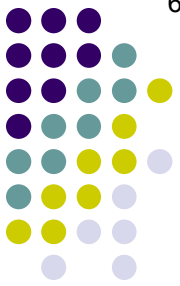
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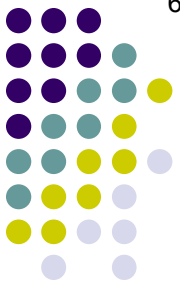
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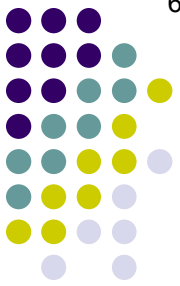
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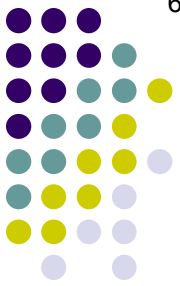
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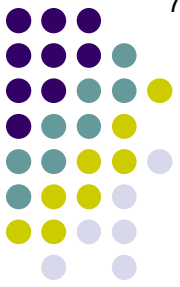
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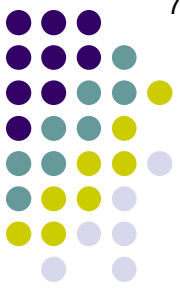
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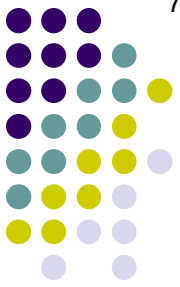
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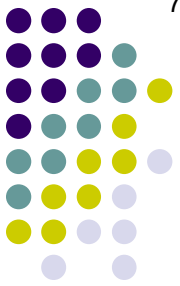
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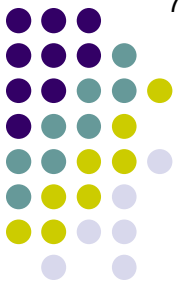
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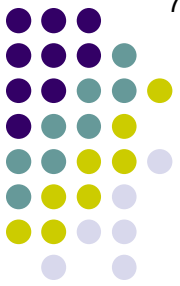
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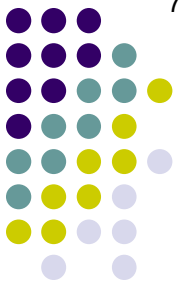
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Is there an age predilection?

It manifests most commonly in the 20/20-30 decade (with a second, smaller peak in the 50-60 decade)

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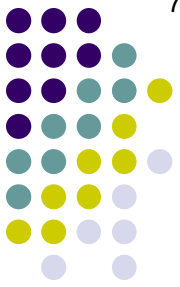
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It manifests most commonly in childhood (with a second, smaller peak in the fourth decade)

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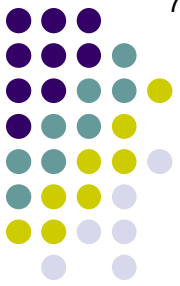
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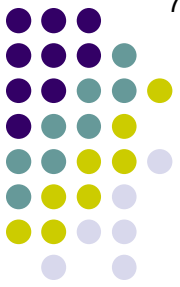
How does moyamoya present in childhood?

It manifests most commonly in **childhood** (with a second, smaller peak in the

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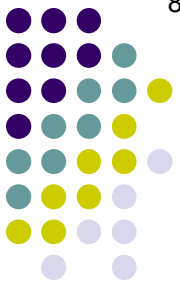
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With TIAs

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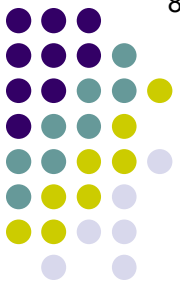
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With TIAs

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!**

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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?

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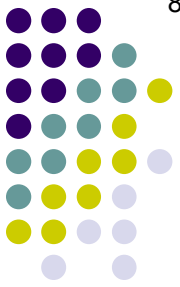
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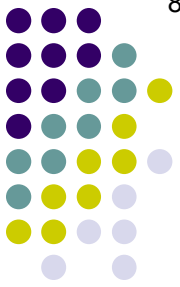
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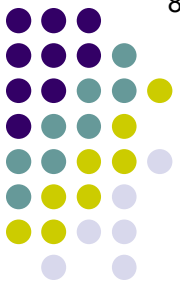
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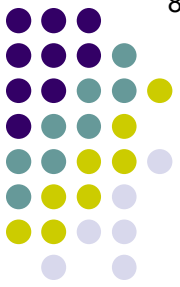
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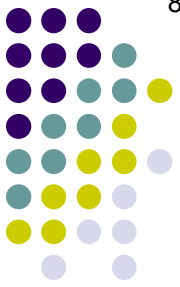
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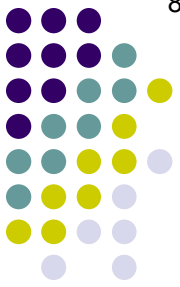
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Megalopapilla

Is there a gender predilection?

Yes, there is a ♀

What is moyamoya disease (MMD)?

An occlusive vascular condition primarily affecting the terminal internal carotids and proximal anterior and middle cerebral arteries

It has two associated conditions

--PHACE syndrome

--Moyamoya disease

Is it common, or rare?

Quite rare

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

Takayasu's arteritis

Morning-glory disc

--DFE reveals

1) A funnel-shaped

2) Numerous small

seems to be

--Tissue is

to...open

the more

--VA usually

20/20 or

--1/3 develop

Is there a racial predilection?

Yes, it is more common in **Asians**

Is there a gender predilection?

Yes, there is a modest **female**

Is there an age predilection?

It manifests most commonly in the fourth decade)

In a nutshell, what is Takayasu's arteritis?

An occlusive vasculitis that affects large vessels

Is it common, or rare?

Developmental Abnormalities of the Optic Nerve Head



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

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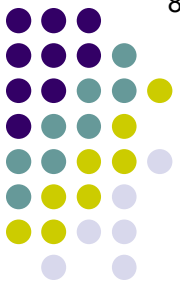
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Developmental Abnormalities of the Optic Nerve Head



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--DFE reveals

1) A funnel-shaped disc

2) Numerous small vessels

--Tissue is normal

--to...open

--the morning-glory disc

--VA usually normal

--20/20 vision

--1/3 develop...serous RD

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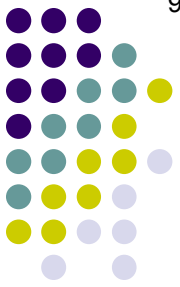
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Developmental Abnormalities of the Optic Nerve Head



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Megalopapilla

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Is it common, or rare?

Rare

Is there an age predilection?

Yes, most cases present in early adulthood

Morning-glory disc

--DFE reveals

1) A funnel-shaped disc

2) Numerous small vessels

seems to be normal

--Tissue is normal

to...open

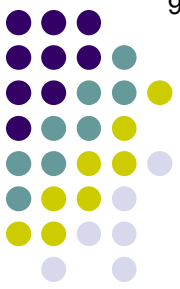
the morning-glory disc

--VA usually normal

20/20 or better

--1/3 develop...serous RD

Developmental Abnormalities of the Optic Nerve Head



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

--PHACE syndrome Quite rare

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women' should bring to mind another condition as well.

What is it?

Takayasu's arteritis

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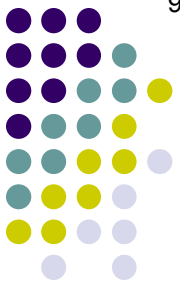
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Developmental Abnormalities of the Optic Nerve Head



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

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- 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... **[? and ?] up**
- 2) VF testing may reveal an... **enlarged blind spot**

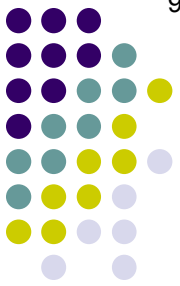
Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

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Developmental Abnormalities of the Optic Nerve Head



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Megalopapilla

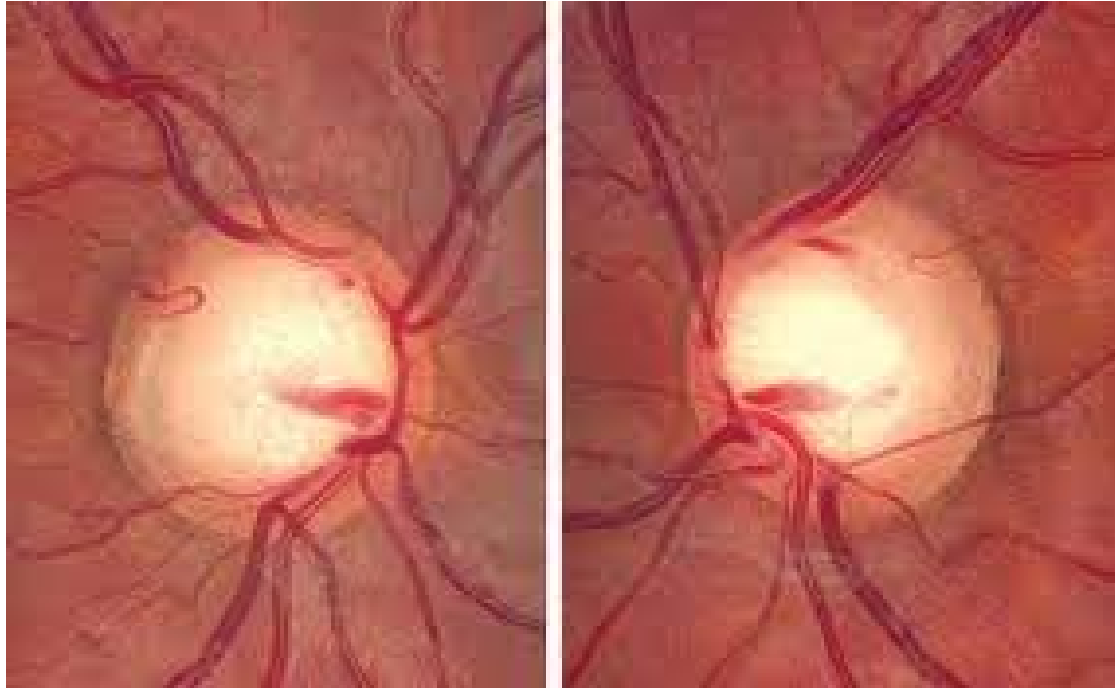
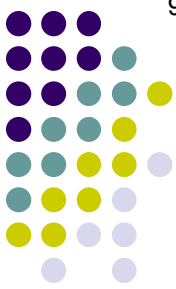
- 1) Abnormally large diameter of...**disc and cup**
- 2) **VF testing may reveal an...enlarged blind spot**

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

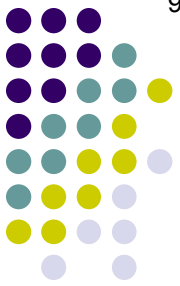
Developmental Abnormalities of the Optic Nerve Head



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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Megalopapilla

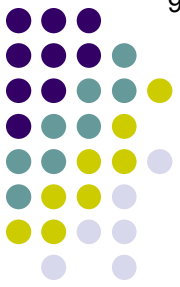
- 1) Abnormally large diameter of...**disc and cup**
- 2) VF testing may reveal an... *[specific VF finding]*

Myelinated RNFL

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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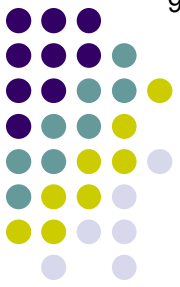
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Myelinated RNFL

Optic Nerve Hypoplasia

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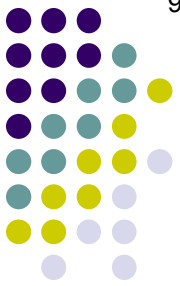
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An abnormally large cup in a preemie with cerebral palsy is suggestive of what condition?

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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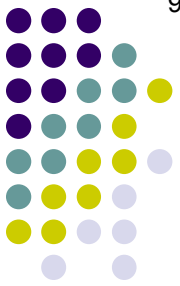
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Periventricular leukomalacia

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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Megalopapilla

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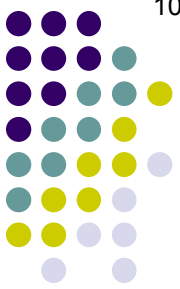
Periventricular leukomalacia

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

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head

100



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

Optic Nerve Coloboma

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Optic Pit/Hole

--Associated with...**serous RD in adulthood**

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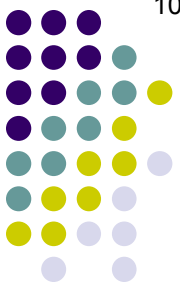
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An abnormally large cup in a preemie with cerebral palsy is suggestive of what condition?
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What is the causative event, and when does it occur?
CNS ischemia in the perinatal period

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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Optic Pit/Hole

--Associated with...**serous RD in adulthood**

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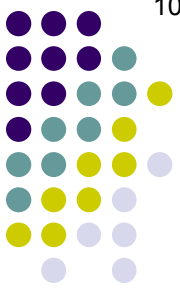
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CNS ischemia in the perinatal period

How does CNS ischemia lead to an enlarged cup?

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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

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Megalopapilla

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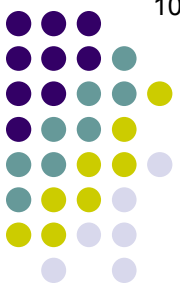
CNS ischemia in the perinatal period

How does CNS ischemia lead to an enlarged cup?

Via retrograde trans-synaptic degeneration of ganglion cell axons

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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Optic Nerve Coloboma

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Optic Pit/Hole

--Associated with...**serous RD in adulthood**

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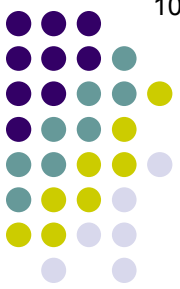
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And it (almost) goes without saying...What condition must be considered in any individual with an enlarged cup?

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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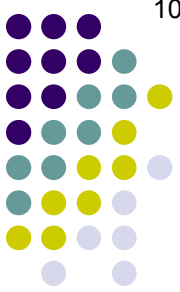
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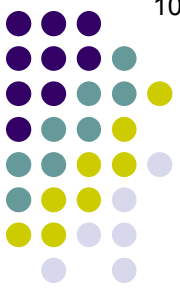
Myelinated RNFL

- 1) Myelin normally ends at the... **[location]**
- 2)
- 3)

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

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Megalopapilla

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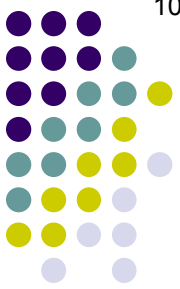
Myelinated RNFL

- 1) Myelin normally ends at the...**lamina cribrosa**
- 2)
- 3)

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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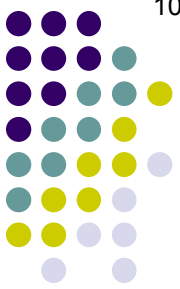
Myelinated RNFL

- 1) Myelin normally ends at the...**lamina cribrosa**
- 2)
- 3)

Where does myelination normally begin?

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...**20/200**, but can be...**20/20<->NLP**

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

Megalopapilla

- 1) Abnormally large diameter of...**disc and cup**
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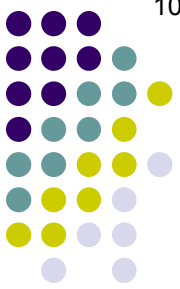
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- 2)
- 3)

Where does myelination normally begin?
At the lateral geniculate nucleus

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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Myelinated RNFL

- 1) Myelin normally ends at the...**lamina cribrosa**
- 2)
- 3)

Where does myelination normally begin?
At the lateral geniculate nucleus

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

Developmental Abnormalities of the Optic Nerve Head



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

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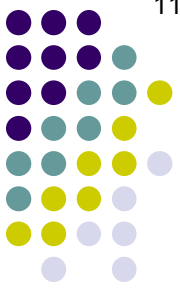
Myelinated RNFL

- 1) Myelin normally ends at the...**lamina cribrosa**
- 2) Can be...**[how confluent/continuous?]**
- 3)

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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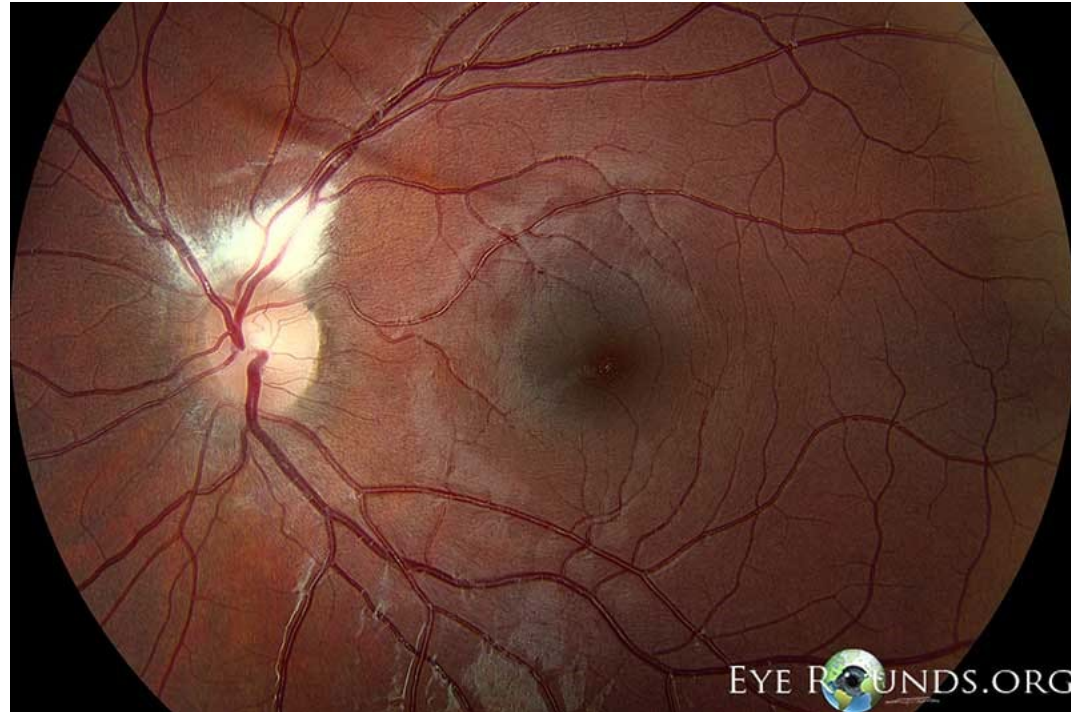
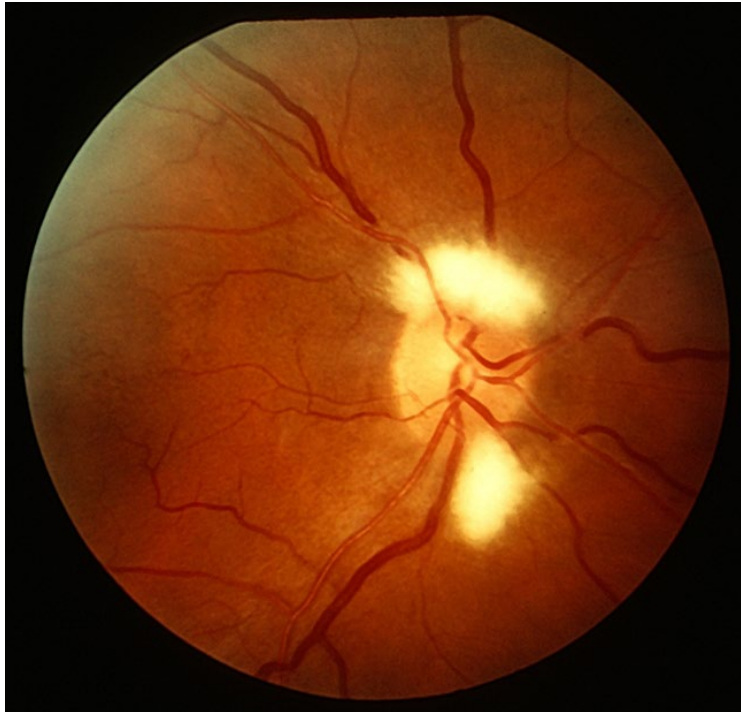
Myelinated RNFL

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Optic Nerve Hypoplasia

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



Myelinated RNFL

Developmental Abnormalities of the Optic Nerve Head



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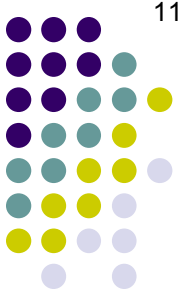
Myelinated RNFL

- 1) Myelin normally ends at the...**lamina cribrosa**
- 2) Can be...**patchy and discontinuous**
- 3) Corresponding VF has an...**[absolute v relative] scotoma**

Optic Nerve Hypoplasia

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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Myelinated RNFL

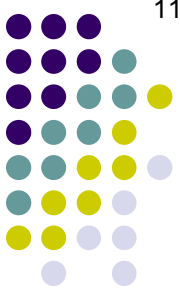
- 1) Myelin normally ends at the...**lamina cribrosa**
- 2) Can be...**patchy and discontinuous**
- 3) Corresponding VF has an...**absolute** scotoma

Optic Nerve Hypoplasia

- 1) Abnormally low number of... *[structure?]*
- 2)
- 3)
- 4)
- 5)

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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

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Myelinated RNFL

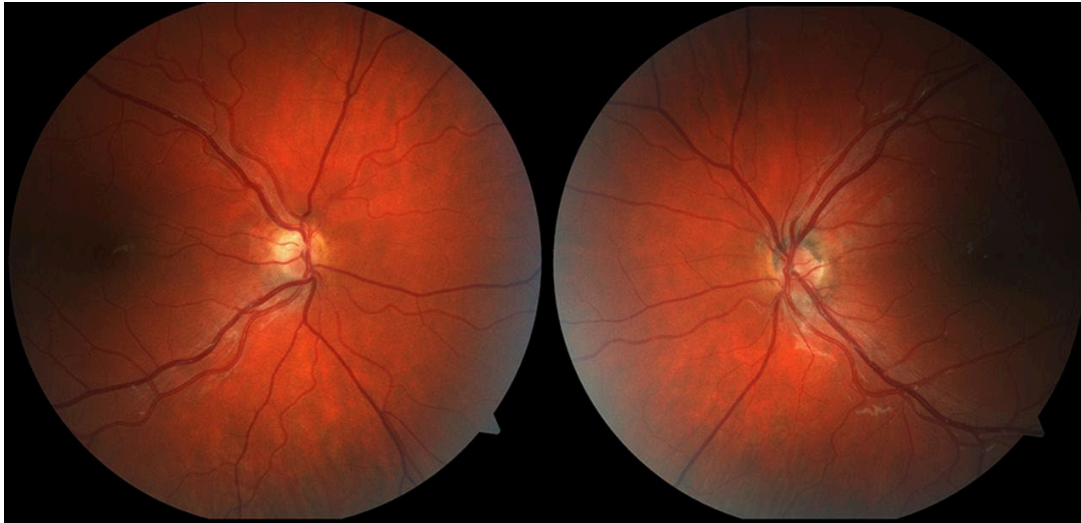
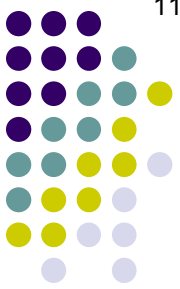
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Optic Nerve Hypoplasia

- 1) Abnormally low number of...**axons**
- 2)
- 3)
- 4)
- 5)

Tilted-Disc Syndrome

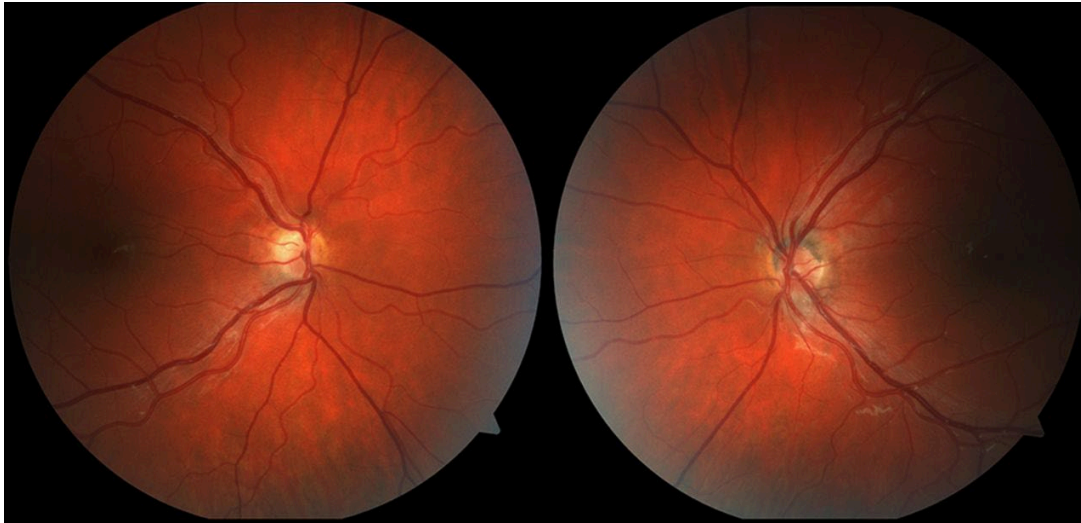
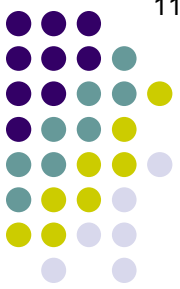
Developmental Abnormalities of the Optic Nerve Head



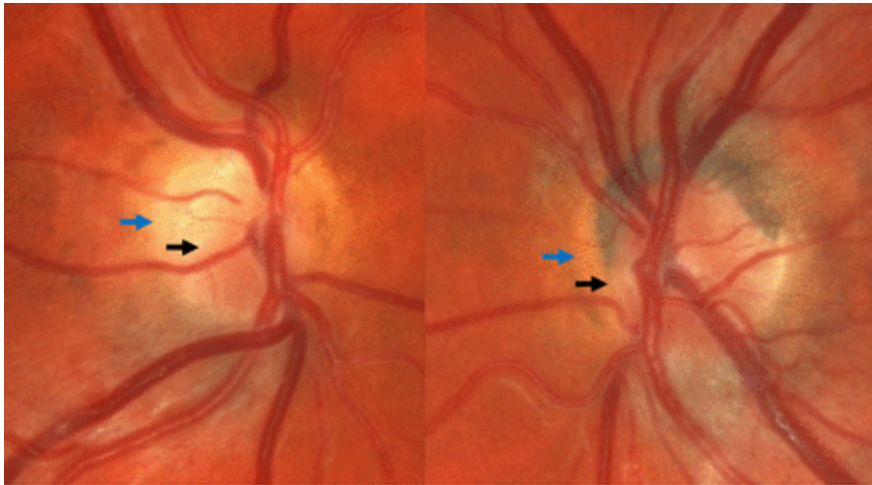
Hypoplastic optic nerve heads

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

Developmental Abnormalities of the Optic Nerve Head



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

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Megalopapilla

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Myelinated RNFL

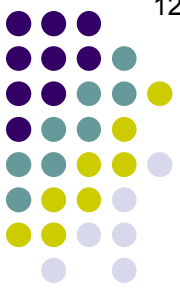
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- 3) Corresponding VF has an...**absolute** scotoma

Optic Nerve Hypoplasia

- 1) Abnormally low number of...**axons**
- 2) DFE: Small pale disc with...**[2-word desc.]** sign
- 3)
- 4)
- 5)

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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

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Myelinated RNFL

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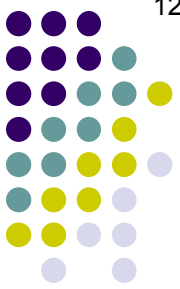
Optic Nerve Hypoplasia

- 1) Abnormally low number of...**axons**
- 2) DFE: Small pale disc with...**double ring** sign
- 3)
- 4)
- 5)

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head

121



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

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--VA usually...**20/200**, but can be...**20/20** → **NLP**

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

Megalopapilla

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Myelinated RNFL

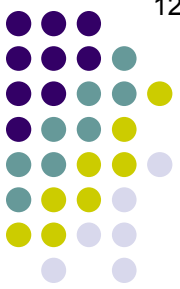
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- 2) Can be...**patchy and discontinuous**
- 3) Corresponding VF has an...**absolute scotoma**

Optic Nerve Hypoplasia

- 1) Abnormally low number of...**axons**
- 2) DFE: Small pale disc with...**double ring sign**
- 3)

A hypoplastic nerve with double-ring sign can easily be mistaken for what?

Developmental Abnormalities of the Optic Nerve Head



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

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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...**lamina cribrosa**
- 2) Can be...**patchy** and **discontinuous**
- 3) Corresponding VF has an...**absolute scotoma**

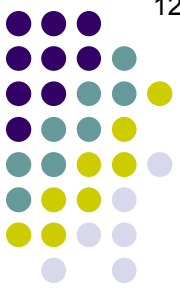
Optic Nerve Hypoplasia

- 1) Abnormally low number of...**axons**
- 2) DFE: Small pale disc with...**double ring** sign
- 3)

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)

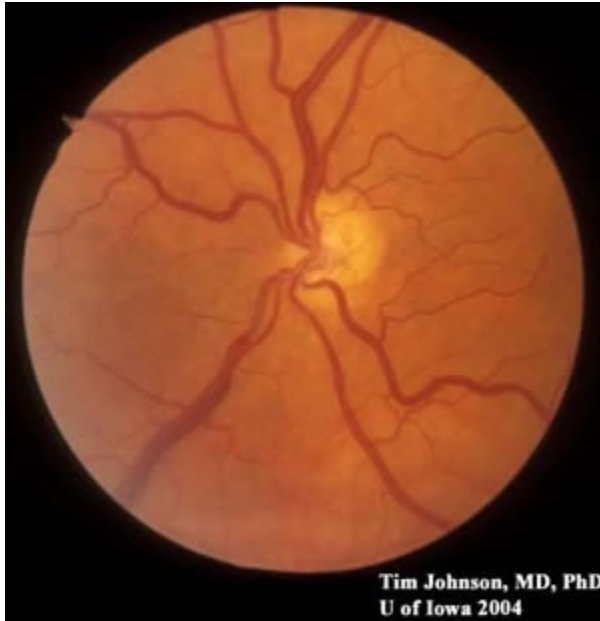
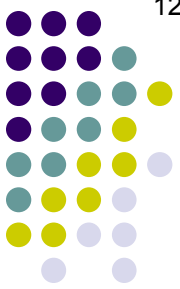
Developmental Abnormalities of the Optic Nerve Head



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*

Developmental Abnormalities of the Optic Nerve Head

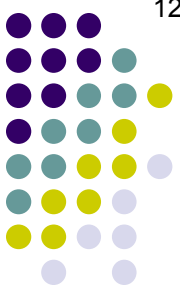


Stereo image of a very hypoplastic nerve. At first glance this looks like a big pale nerve. But look closely—there's just a little stump of optic nerve (it's where the vessels emanate) surrounded by a pale ring (double-ring sign).

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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--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

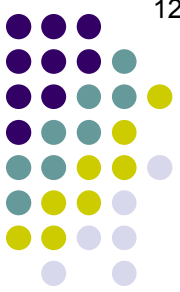
- 1) Myelin normally ends at the...**lamina cribrosa**
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Optic Nerve Hypoplasia

- 1) Abnormally low number of...**axons**
- 2) DFE: Small pale disc with...**double ring** sign
- 3) VA...**[range]**
- 4)
- 5)

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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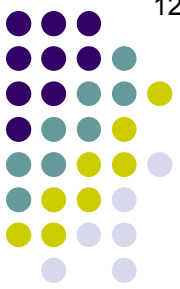
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Optic Nerve Hypoplasia

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- 3) VA...**20/15<->NLP**
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Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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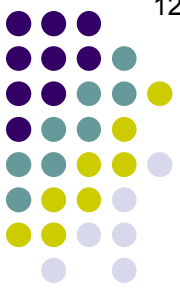
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Optic Nerve Hypoplasia

- 1) Abnormally low number of...**axons**
- 2) DFE: Small pale disc with...**double ring** sign
- 3) VA...**20/15<->NLP**
- 4) VF defects... *[how likely?]*
- 5)

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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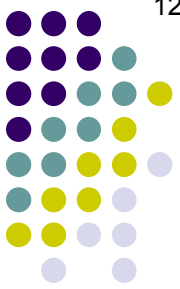
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- 2) Can be...**patchy and discontinuous**
- 3) Corresponding VF has an...**absolute** scotoma

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)

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...**20/200**, but can be...**20/20<->NLP**
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Megalopapilla

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

Myelinated RNFL

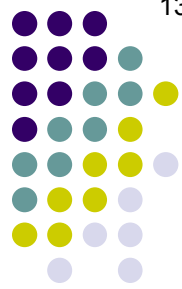
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Is the VF loss associated with optic nerve hypoplasia progressive, or nonprogressive?

Developmental Abnormalities of the Optic Nerve Head



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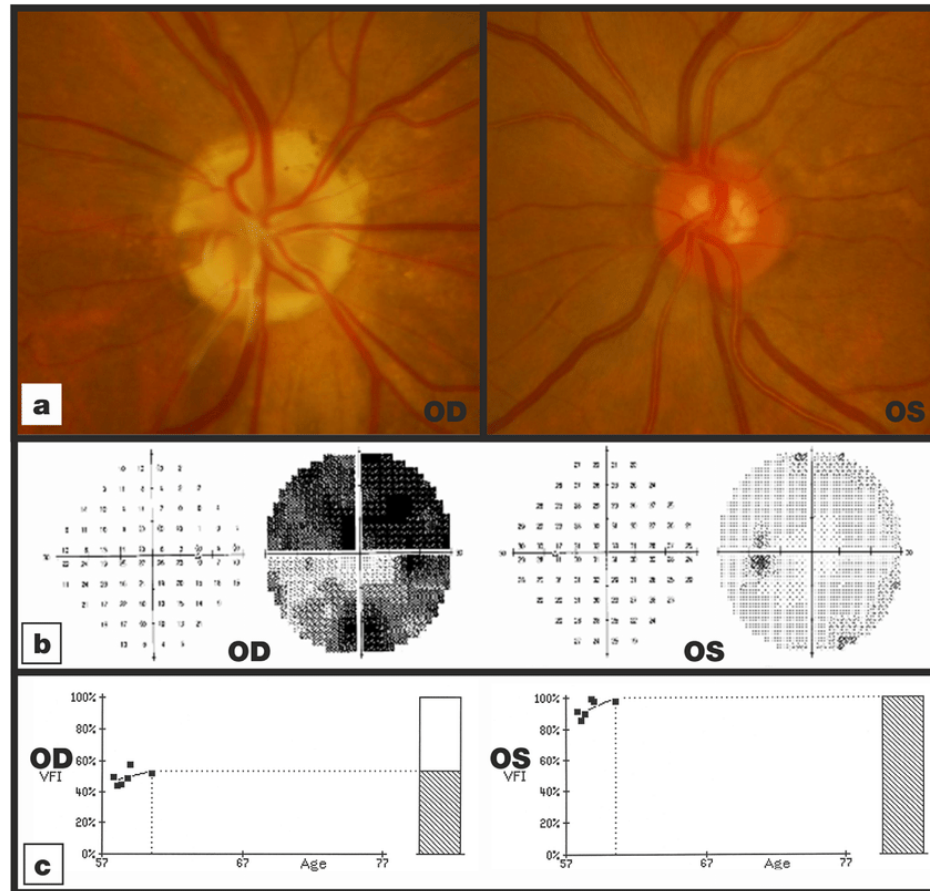
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Is the VF loss associated with optic nerve hypoplasia progressive, or nonprogressive?
Nonprogressive

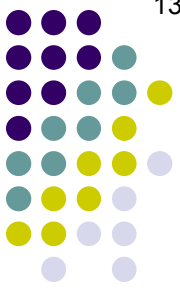
Developmental Abnormalities of the Optic Nerve Head



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

Optic nerve hypoplasia and VF loss

Developmental Abnormalities of the Optic Nerve Head



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

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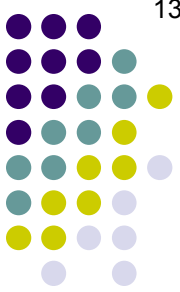
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Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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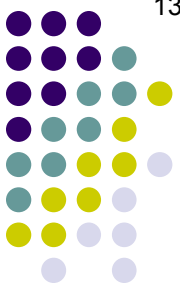
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Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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

Megalopapilla

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

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

Optic Nerve Co

- 1) May resemble... --
- 2) Can be...**bilater**
- 3) Part of the...**CH**

Optic F

--Associated with...**serous RD**
in adulthood

Morning-Glory Disc

--DFE reveals:

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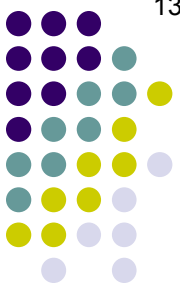
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Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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

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--Obtaining

imaging

--Getting an

service

consult

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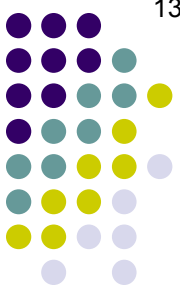
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Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



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

Megalopapilla

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

--Obtaining MRI brain

--Getting an endocrine consult

Optic Nerve Co

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Optic F

--Associated with...**serous RD** in adulthood

Morning-Glory Disc

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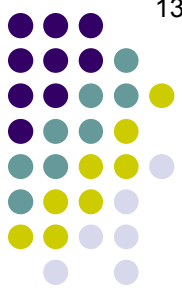
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- 5) Remember the...**4 D's** (more on this shortly)

Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head

137



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

Megalopapilla

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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**

Optic Nerve Co

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Morning-Glory Disc

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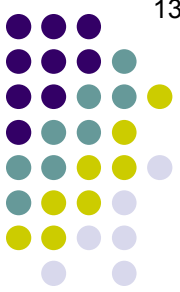
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--Getting an **endocrine consult**—multiple hormonal deficiencies are associated

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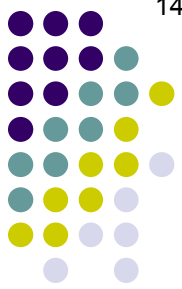
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Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve



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

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consi

--Obta

(will u

--Gett

(main

MRI brain

pituitary- related

variety of CNS abnormalities are associated with it
(including 4 D's section)

ult—multiple hormonal deficiencies are associated

Optic F

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Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve



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

Anterior pituitary hypoplasia coupled with an [redacted] posterior pituitary

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Developmental Abnormalities of the Optic Nerve



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

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How will this manifest on neuroimaging?

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Developmental Abnormalities of the Optic Nerve



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 vs dim** spot at the **upper v lower** infundibulum

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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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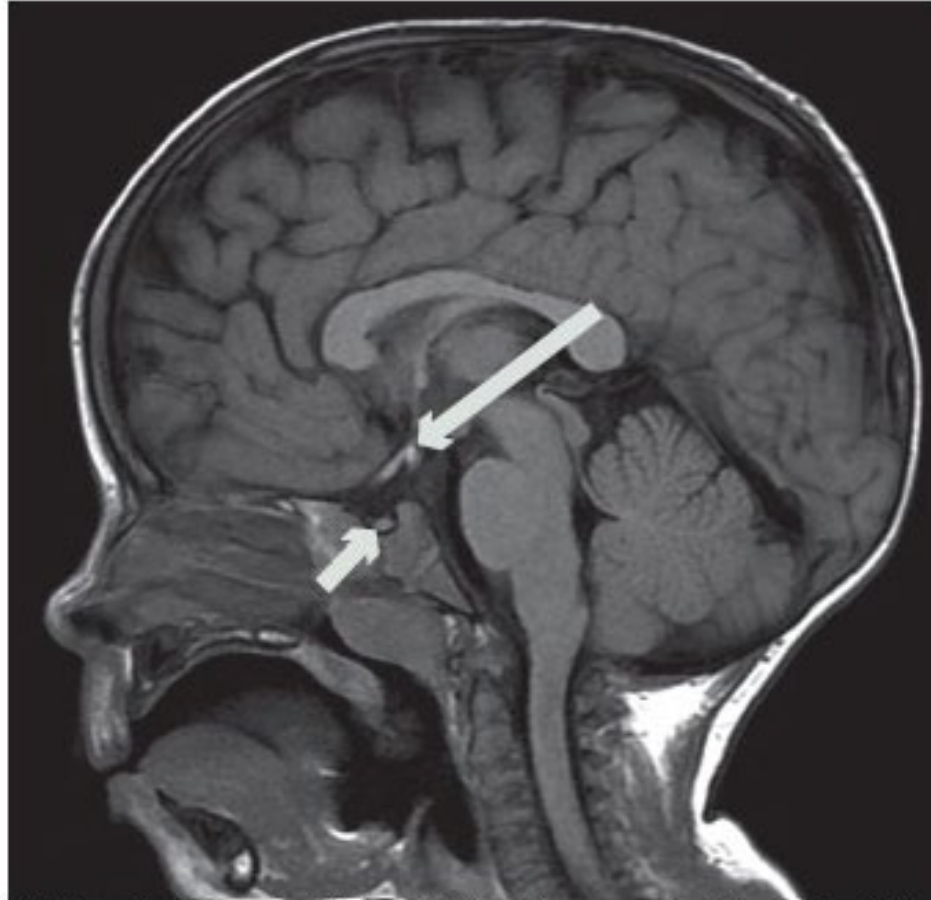
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- 2) DFE: Small pale disc with...**double ring** sign
- 3) VA...**20/15<->NLP**
- 4) VF defects...**invariably present**
- 5) Remember the...**4 D's** (more on this shortly)

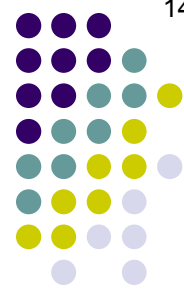
Tilted-Disc Syndrome

Developmental Abnormalities of the Optic Nerve Head



Note the posterior ectopic bright spot at the upper infundibulum (*long arrow*) and hypoplastic anterior pituitary (*short arrow*)

Developmental Abnormalities of the Optic Nerve



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- 1) May resemble...
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consi

--Obta

(will u

--Gett

(main

MRI brain

pituitary- related

variety of CNS abnormalities are associated with it (including 4 D's section)

ult—multiple hormonal deficiencies are associated

Optic F

What pituitary-related deficiencies may be present?

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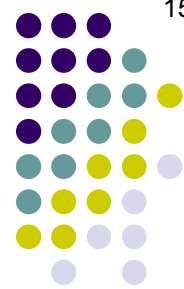
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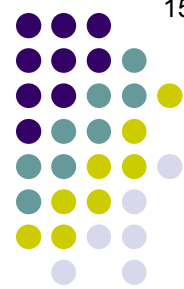
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1) Abnormally large diameter of...**disc** and **cup**

2) Abnormal contour of...**disc**. What steps should you at least

Optic Nerve Considerations

- 1) May resemble...
- 2) Can be...**bilateral**
- 3) Part of the...**CH**

consider

--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

These are probably low-yield factoids in isolation.

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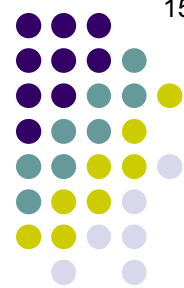
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*These are probably low-yield factoids in isolation. But what **isn't** low-yield is making the mental note 'If a neonate or young child presents with ON hypoplasia along with anything remotely hormone-related, expedite neuroimaging and Peds Endo referral!'*

...*short stature, think: Growth-hormone deficiency*
 ...*neonatal jaundice, think: Hypothyroidism*
 ...*hypoglycemia/seizures, think: Panhypopituitarism*
 ...*diabetes insipidus, think: Hypocortisolism*

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Developmental Abnormalities of the Optic Nerve Head

160



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

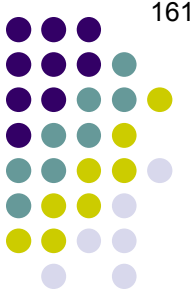
- 1) Myelin normally ends at the...**lamina cribrosa**
- 2) Can be...**patchy** and **discontinuous**
- 3) Corresponding VF has an...**absolute scotoma**

Optic Nerve Hypoplasia

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As for the four Ds of optic nerve hypoplasia:

Developmental Abnormalities of the Optic Nerve Head



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

--**D**

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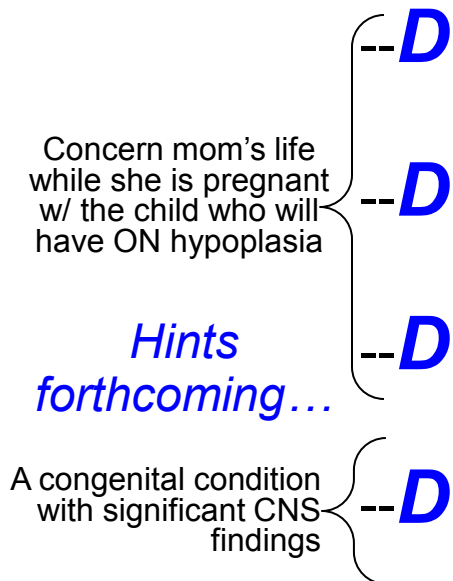
--**D**

*Hints
forthcoming...*

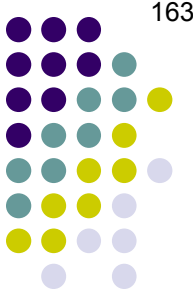
Developmental Abnormalities of the Optic Nerve Head



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



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 **D**ilantin or other seizure meds)

--**De Morsier syndrome**

(5 D's if you count this one)

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

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

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)

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

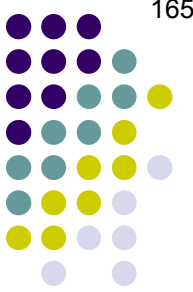
three words

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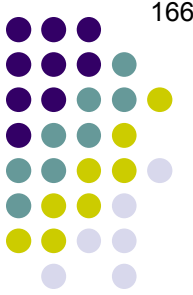
In other words, optic-nerve hypoplasia is part of the *fetal alcohol syndrome*

--**Diabetes**

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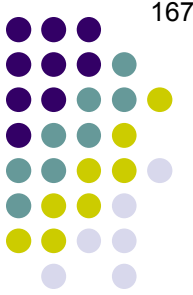
What triad constitutes de Morsier syndrome?

--?

--?

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-- two words hypoplasia (duh)

--?

--?

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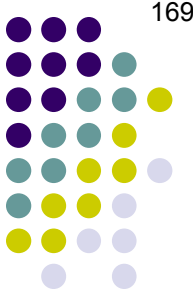
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two words

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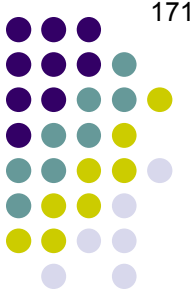
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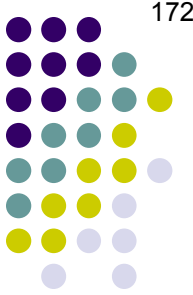
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two diff words

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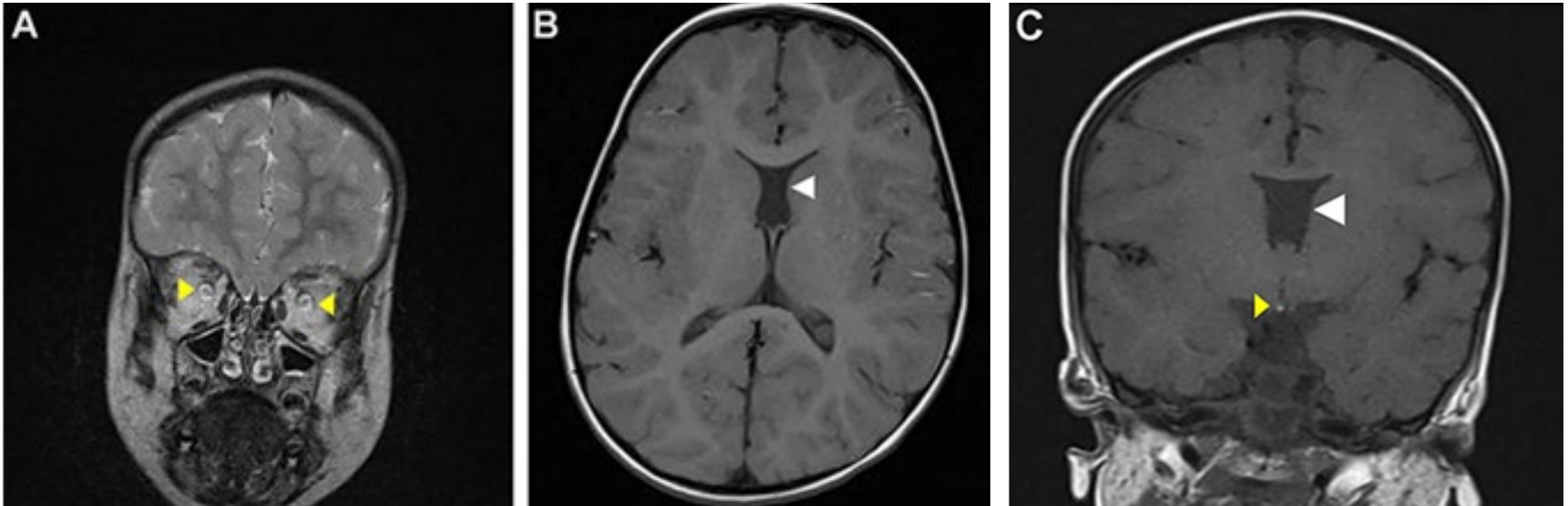
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--Absence of the septum pellucidum

--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

Developmental Abnormalities of the Optic Nerve Head



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

--**D**rink (ie, heavy EtOH consumption during pregnancy)

--**D**iabetes

--**D**rugs (especially **D**ilantin or other seizure meds)

--**D**e Morsier syndrome

What triad constitutes de Morsier syndrome?

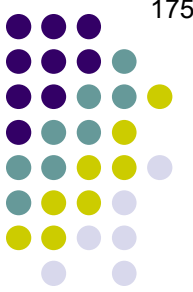
--**O**ptic nerve hypoplasia (duh)

--Absence of the **septum** pellucidum

--Agensis of the corpus callosum

What is the noneponymous name of de Morsier syndrome?

Developmental Abnormalities of the Optic Nerve Head



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What is the noneponymous name of de Morsier syndrome?
Septo-

Developmental Abnormalities of the Optic Nerve Head



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What is the noneponymous name of de Morsier syndrome?
Septo-optic

Developmental Abnormalities of the Optic Nerve Head



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Septo-optic dysplasia

Developmental Abnormalities of the Optic Nerve Head



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

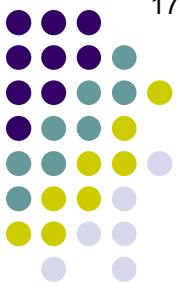
Note: The listed triad is from the BCSC *Peds* book.

--**De Morsier syndrome**

What triad constitutes de Morsier syndrome?

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Developmental Abnormalities of the Optic Nerve Head



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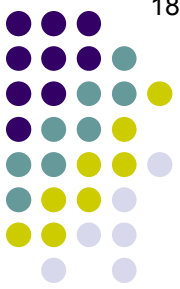
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Developmental Abnormalities of the Optic Nerve Head



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Developmental Abnormalities of the Optic Nerve Head



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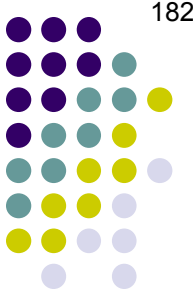
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Developmental Abnormalities of the Optic Nerve Head



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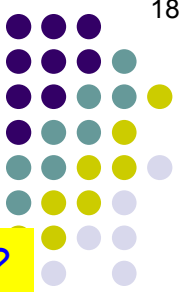
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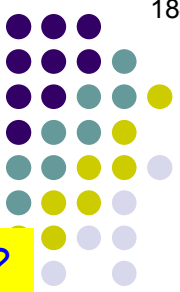
Developmental Abnormalities of the Optic Nerve Head



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

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- Diabetes**
- Drugs** (especially **D**ilantin or other seizure meds)
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Developmental Abnormalities of the Optic Nerve Head



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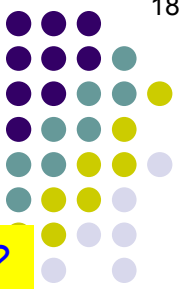
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Developmental Abnormalities of the Optic Nerve Head



Of the four, which is the most common cause of optic nerve hypoplasia?
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Maternal diabetes is notorious for causing a specific pattern of optic nerve hypoplasia—what is it?

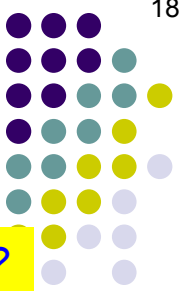
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Developmental Abnormalities of the Optic Nerve Head



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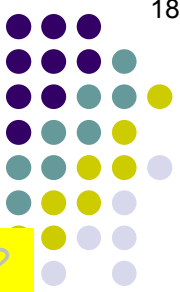
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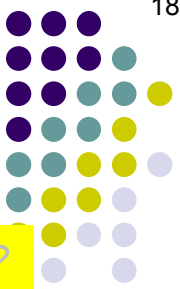
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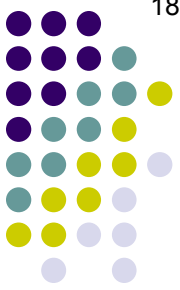
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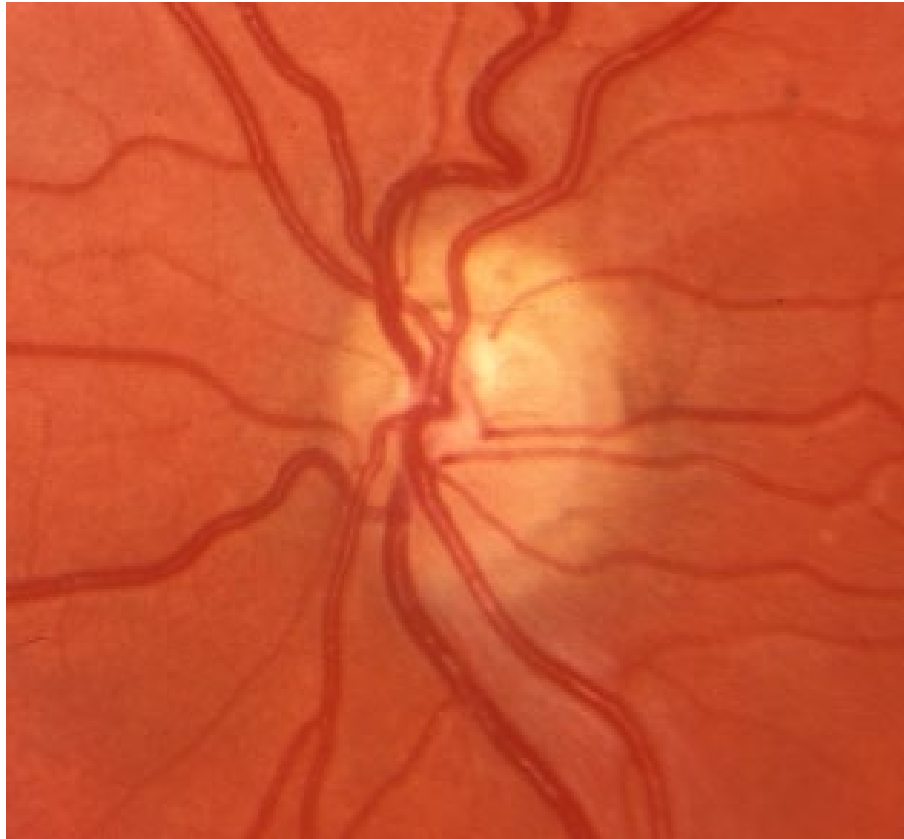
Pretty much what you would expect based on the name--a normal-appearing nerve save for a thin superior rim, with associated thinning of the superior nerve fiber layer

--*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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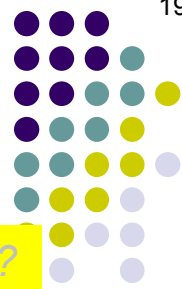
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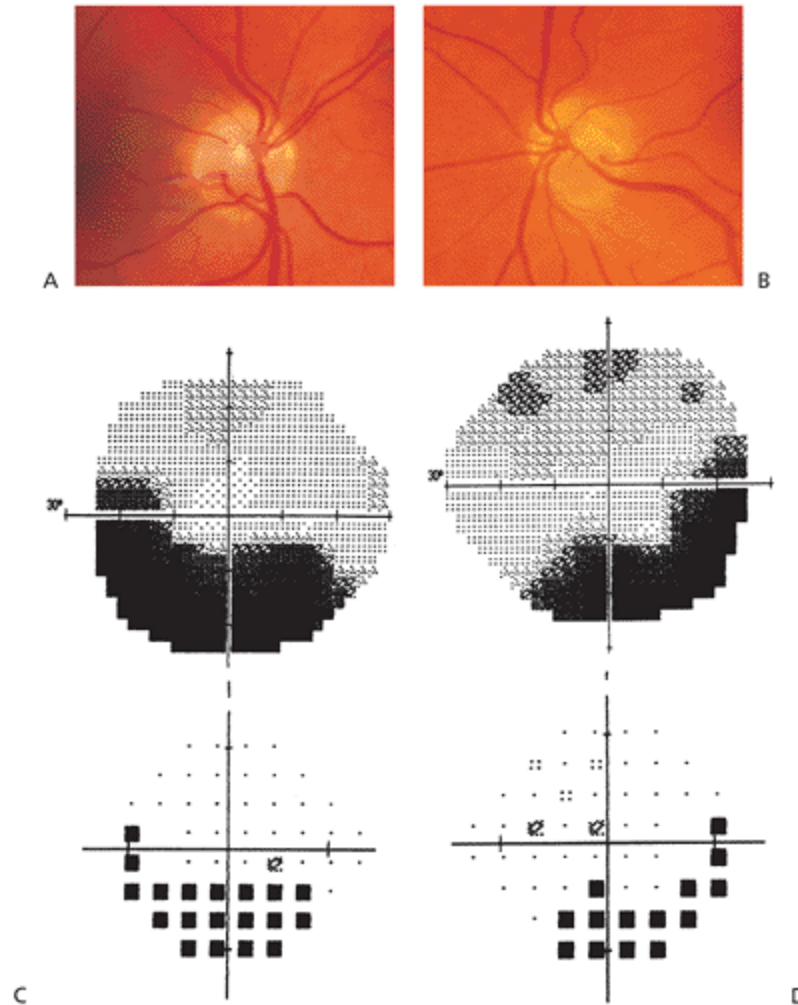
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What pattern of VF loss is associated with SSONH?

Bitemporal **inferior** loss that doesn't respect the vertical midline

--**D**e Morsier syndrome

Developmental Abnormalities of the Optic Nerve Head



Superior segmental ON hypoplasia: Inferior VF defects

Developmental Abnormalities of the Optic Nerve Head



Does **gestational** diabetes place a fetus at risk for SSONH?

Gestational **Diabetes?**

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What is the appearance of the nerve head in SSONH?

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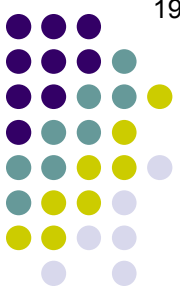
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Developmental Abnormalities of the Optic Nerve Head



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**
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Myelinated RNFL

- 1) Myelin normally ends at the...**lamina cribrosa**
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- 3) Corresponding VF has an...**absolute** scotoma

Optic Nerve Hypoplasia

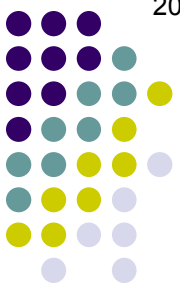
- 1) Abnormally low number of...**axons**
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- 5) Remember the...**4 D's**

Tilted-Disc Syndrome *aka...*

By what eponymous name is tilted-disc syndrome known?

Developmental Abnormalities of the Optic Nerve Head

200



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Tilted-Disc Syndrome *aka...Fuch's Coloboma*

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Developmental Abnormalities of the Optic Nerve Head

201



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Tilted-Disc Syndrome *aka...Fuch's Coloboma*

- 1) Superior pole appears...**[elevated v recessed]**, inferior...**[elevated v recessed]**
- 2)
- 3)
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Developmental Abnormalities of the Optic Nerve Head

202



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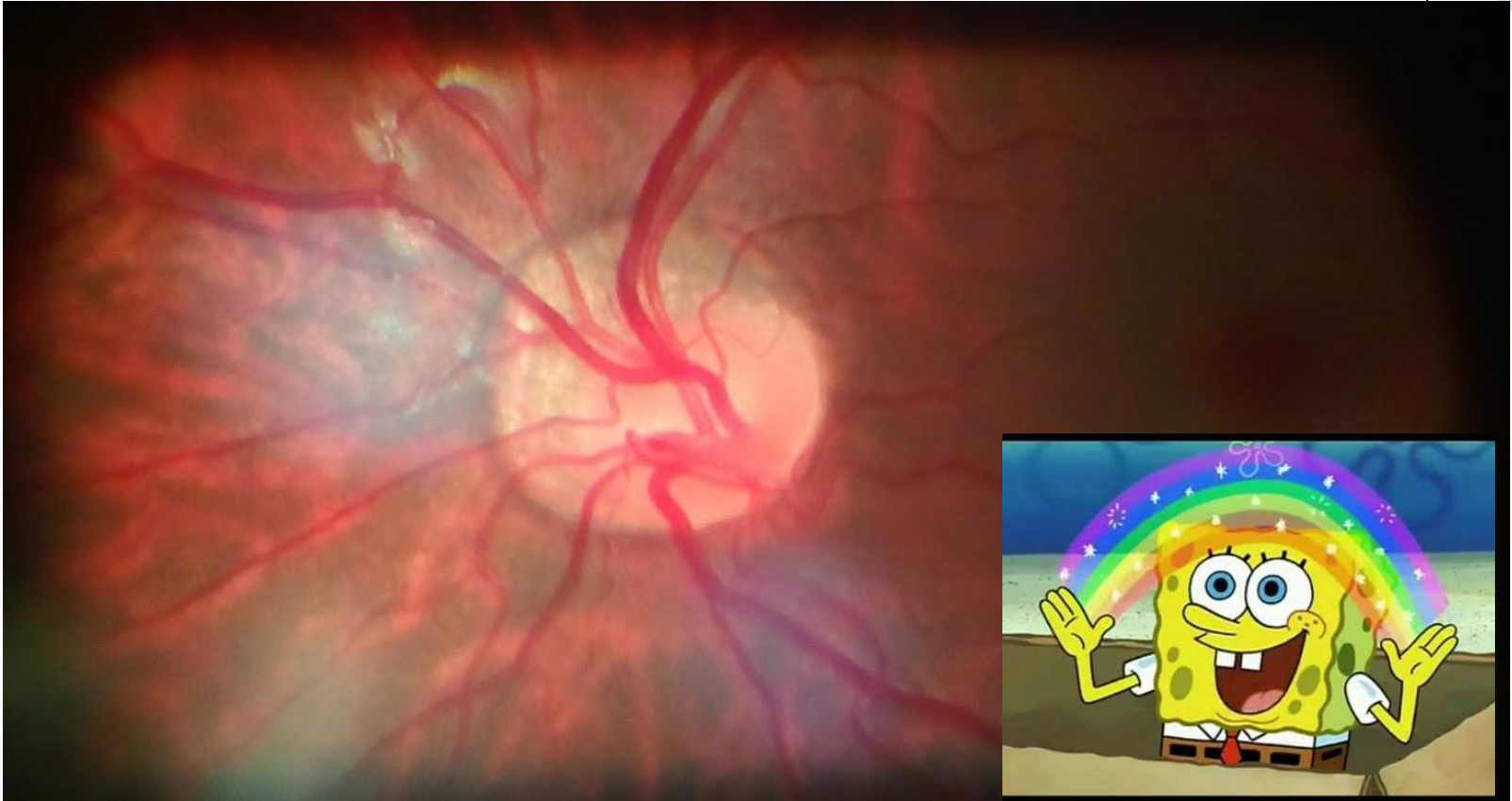
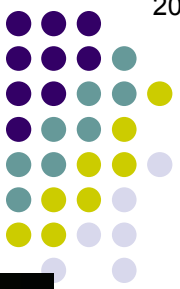
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Tilted-Disc Syndrome *aka...Fuch's Coloboma*

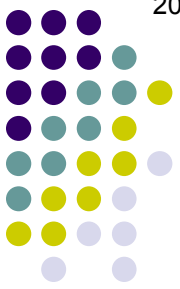
- 1) Superior pole appears...**elevated**, inferior...**recessed**
- 2)
- 3)
- 4)

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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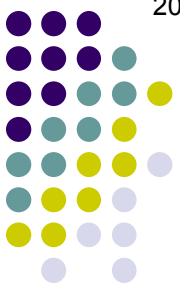
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- 1) Superior pole appears...**elevated**, inferior...**recessed**
- 2) Associated with... *[two words]* of retinal vessels
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Myelinated RNFL

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

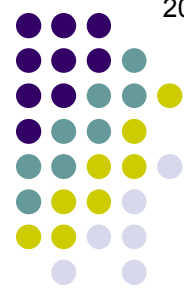
Optic Nerve Hypoplasia

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

Tilted-Disc Syndrome *aka...Fuch's Coloboma*

- 1) Superior pole appears...**elevated**, inferior...**recessed**
- 2) Associated with...**situs inversus** of retinal vessels
- 3)
- 4)

Developmental Abnormalities of the Optic Nerve Head



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

Optic Nerve Coloboma

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

Optic Pit/Hole

--Associated with...**serous RD in adulthood**

Morning-Glory Disc

--DFE reveals:

- 1) A
- 2) N
- see
- Tiss
- to...
- the morning-glory flower)
- VA usually...**20/200**, but can be...
- 20/20<->NLP**
- 1/3 develop...**serous RD**

Megalopapilla

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Optic Nerve Hypoplasia

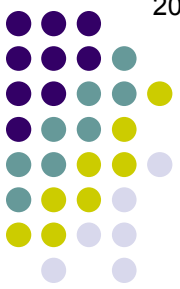
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Huh? I thought situs inversus meant all the organs were on the wrong side of the body, or something. What does it mean in this context?

situs inversus of retinal vessels

Developmental Abnormalities of the Optic Nerve Head

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

Optic Nerve Coloboma

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Optic Nerve Hypoplasia

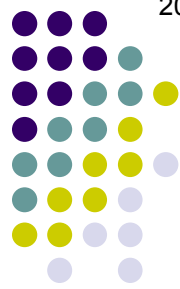
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- 4) VF defects...**invariably present**

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

2) Associated with...**situs inversus** of retinal vessels

Developmental Abnormalities of the Optic Nerve Head



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

Optic Nerve Coloboma

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

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Morning-Glory Disc

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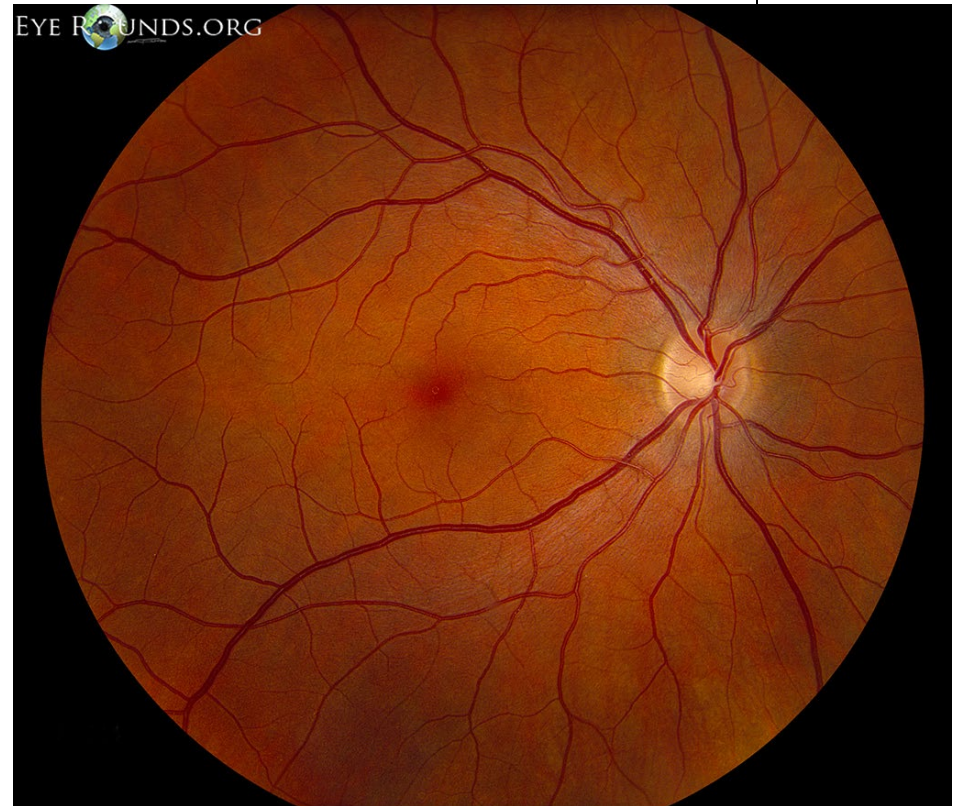
It means the temporal vessels run nasally for a short interval before heading off in the right direction

2) Associated with...**situs inversus** of retinal vessels

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

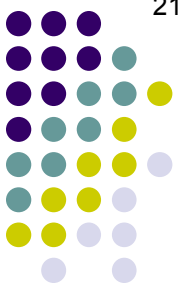


Normal OD posterior pole for comparison

Optic nerve hypoplasia: *Double-ring sign*

Developmental Abnormalities of the Optic Nerve Head

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

Optic Nerve Coloboma

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Optic Pit/Hole

--Associated with...**serous RD in adulthood**

Morning-Glory Disc

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- 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

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Optic Nerve Hypoplasia

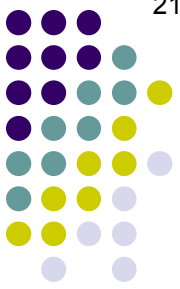
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- 5) Remember the...**4 D's**

Tilted-Disc Syndrome *aka...Fuch's Coloboma*

- 1) Superior pole appears...**elevated**, inferior...**recessed**
- 2) Associated with...**situs inversus** of retinal vessels
- 3) Fundus abnormality produces... **[refractive error]**
- 4)

Developmental Abnormalities of the Optic Nerve Head

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

Optic Nerve Coloboma

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Optic Pit/Hole

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Optic Nerve Hypoplasia

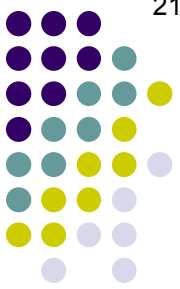
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- 1) Superior pole appears...**elevated**, inferior...**recessed**
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- 3) Fundus abnormality produces...**myopic astigmatism**
- 4)

Developmental Abnormalities of the Optic Nerve Head

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

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Optic Pit/Hole

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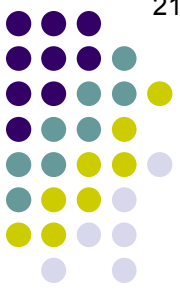
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- 2) Associated with...**situs inversus** of retinal vessels
- 3) Fundus abnormality produces...**myopic astigmatism**
- 4) VF testing reveals... *[specific VF finding]* that doesn't respect the vertical, and may resolve with... *[simple clinical maneuver]*

Developmental Abnormalities of the Optic Nerve Head



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

Optic Nerve Coloboma

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- 3) Fundus abnormality produces...**myopic astigmatism**
- 4) VF testing reveals...**bitemporal loss** 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

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Optic Pit/Hole

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

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

Megalopapilla

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Tilted-Disc Syndrome aka...Fuch's Coloboma

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ectopia bitemporalis of retinal vessels produces...**myopic astigmatism** and may resolve with...**refraction**

bitemporal loss

Developmental Abnormalities of the Optic Nerve Head



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

Optic Nerve Coloboma

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Optic Pit/Hole

--Associated with...**serous RD** in adulthood

Morning-Glory Disc

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

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Superior

Megalopapilla

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Myelinated RNFL

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- 3) Corresponding VF has an...**absolute scotoma**

Optic Nerve Hypoplasia

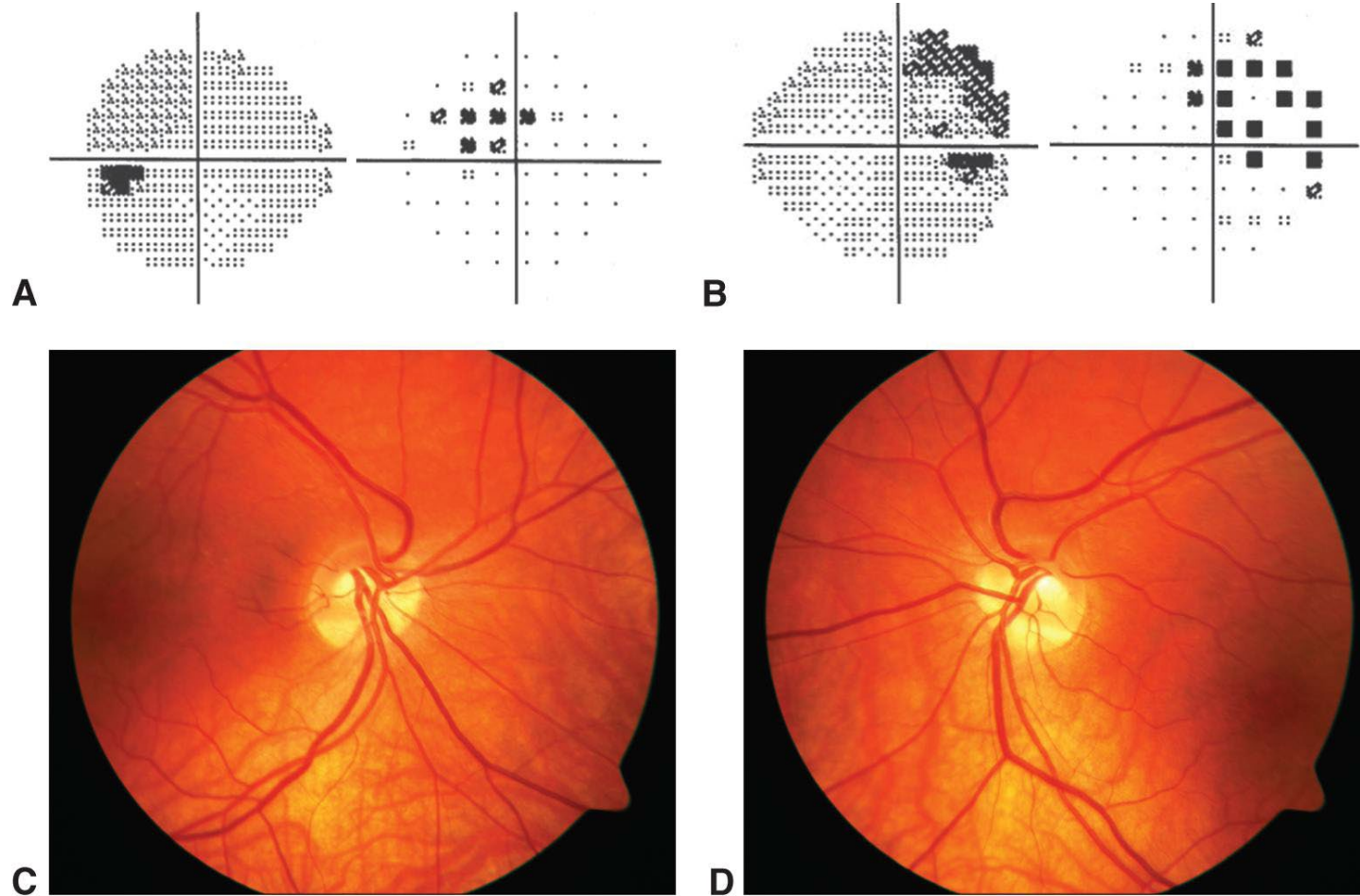
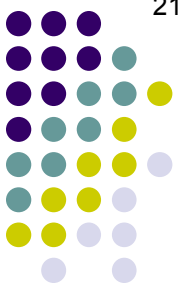
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Tilted-Disc Syndrome aka...Fuch's Coloboma

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ectopia of retinal vessels
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...**bitemporal loss** that doesn't
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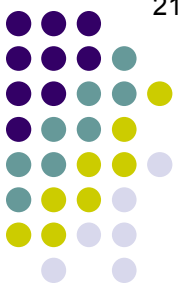
Developmental Abnormalities of the Optic Nerve Head



Tilted-disc syndrome: Superior bitemporal VF loss

Developmental Abnormalities of the Optic Nerve Head

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

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

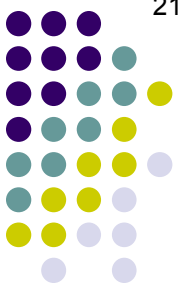
VF usually...**20/200**, but can be...
20/20 <-> **NLP**
--1/3 develop...**serous RD**

3) Fundus abnormal
4) VF testing reveals
respect the vertical

bitemporal loss that doesn't
and may resolve with...**refraction**

Developmental Abnormalities of the Optic Nerve Head

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

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

It's actually pretty simple.

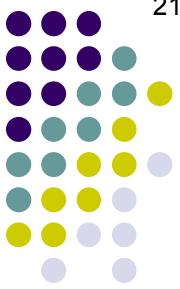
The recession of the inferonasal ONH is quite dramatic—almost staphyloma-ish. Because of this, the 'axial length' (AL) of the inferonasal peripapillary retina is longer than that of other retinal regions.

VF usually...**20/200**, but can be...
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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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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.

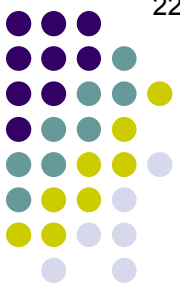
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Developmental Abnormalities of the Optic Nerve Head

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

Optic Nerve Coloboma

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Optic Pit/Hole

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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.

VF usually...**20/200**, but can be...

20/20 <-> **NLP**

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

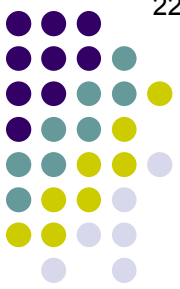
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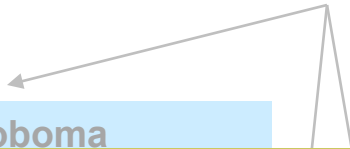
bitemporal loss that doesn't
and may resolve with...**refraction**

Developmental Abnormalities of the Optic Nerve Head

221



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



Optic Nerve Coloboma

- 1) May resemble
- 2) Can be...bilateral
- 3) Part of the...

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

Megalopapilla

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

Myelinated RNFL

Optic

--Ass

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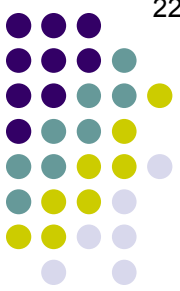
--1/3 develop...serous RD

- 3) Fundus abnormal...astigmatism
- 4) VF testing reveals...that doesn't respect the vertical...refraction

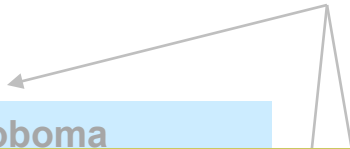
bitemporal loss

Developmental Abnormalities of the Optic Nerve Head

222



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



Megalopapilla

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- 2) Can be...**bil**
- 3) Part of the...

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

Myelinated RNFL

Opt

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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.

VF usually...**20/200**, but can be...

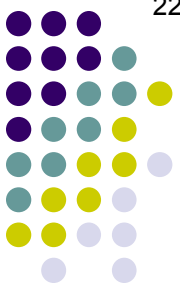
20/20<->NLP

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

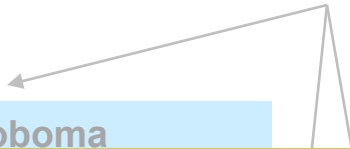
- 3) Fundus abnormal...**astigmatism**
- 4) VF testing reveals...**bitemporal loss** that doesn't respect the vertical...**refraction**

Developmental Abnormalities of the Optic Nerve Head

223



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



Optic Nerve Coloboma

- 1) May resemble
- 2) Can be...bilateral
- 3) Part of the...

*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?*

Megalopapilla

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

Myelinated RNFL

Optic

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VF usually...20/200, but can be...

20/20<->NLP

--1/3 develop...serous RD

- 3) Fundus abnormal...astigmatism
- 4) VF testing reveals...that doesn't respect the vertical...refraction

bitemporal loss

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

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

- 1) May resemble
- 2) Can be... **bilabial**
- 3) Part of the...

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.

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✓/✓ usually...20/200, but can be...

20/20<->NLP

--1/3 develop...serous RD

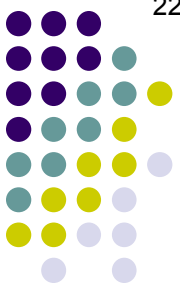
Myelinated PNEI

4) VF testing reveals **bitemporal loss** that doesn't respect the vertical meridian → ...refraction

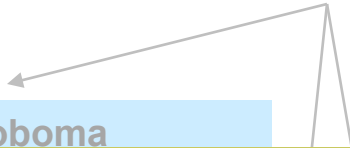
bitemporal loss

Developmental Abnormalities of the Optic Nerve Head

225



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



Optic Nerve Coloboma

- 1) May resemble
- 2) Can be...bilateral
- 3) Part of the...

Megalopapilla

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

Myelinated RNFL

*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?

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.

VF usually...20/200, but can be...

20/20<->NLP

--1/3 develop...serous RD

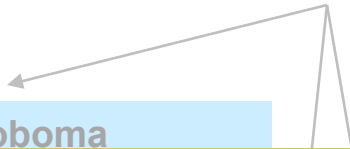
- 3) Fundus abnormal...astigmatism
- 4) VF testing reveals...that doesn't respect the vertical...refraction

bitemporal loss

Developmental Abnormalities of the Optic Nerve Head



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**

Optic Nerve Coloboma

- 1) May resemble
- 2) Can be...**bil**
- 3) Part of the...

*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 vs horizontal** midline, whereas one 2ndry to Fuchs coloboma will not.

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.

VF usually...20/200, but can be...

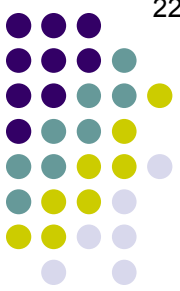
20/20<->NLP

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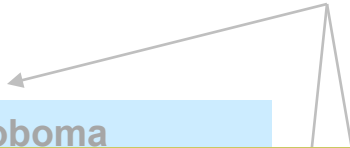
- 3) Fundus abnormal...**astigmatism**
- 4) VF testing reveals...**bitemporal loss**...that doesn't respect the vertical...**refraction**

Developmental Abnormalities of the Optic Nerve Head

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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**

Optic Nerve Coloboma

- 1) May resemble
- 2) Can be...**bil**
- 3) Part of the...

*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.

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.

VF usually...**20/200**, but can be...

20/20<->NLP

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

- 3) Fundus abnormal...**astigmatism**
- 4) VF testing reveals...**bitemporal loss** that doesn't respect the vertical...**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
- 2) Can be...bilateral
- 3) Part of the...

Megalopapilla

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

Myelinated RNFL

*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.

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.

VF usually...20/200, but can be...

20/20<->NLP

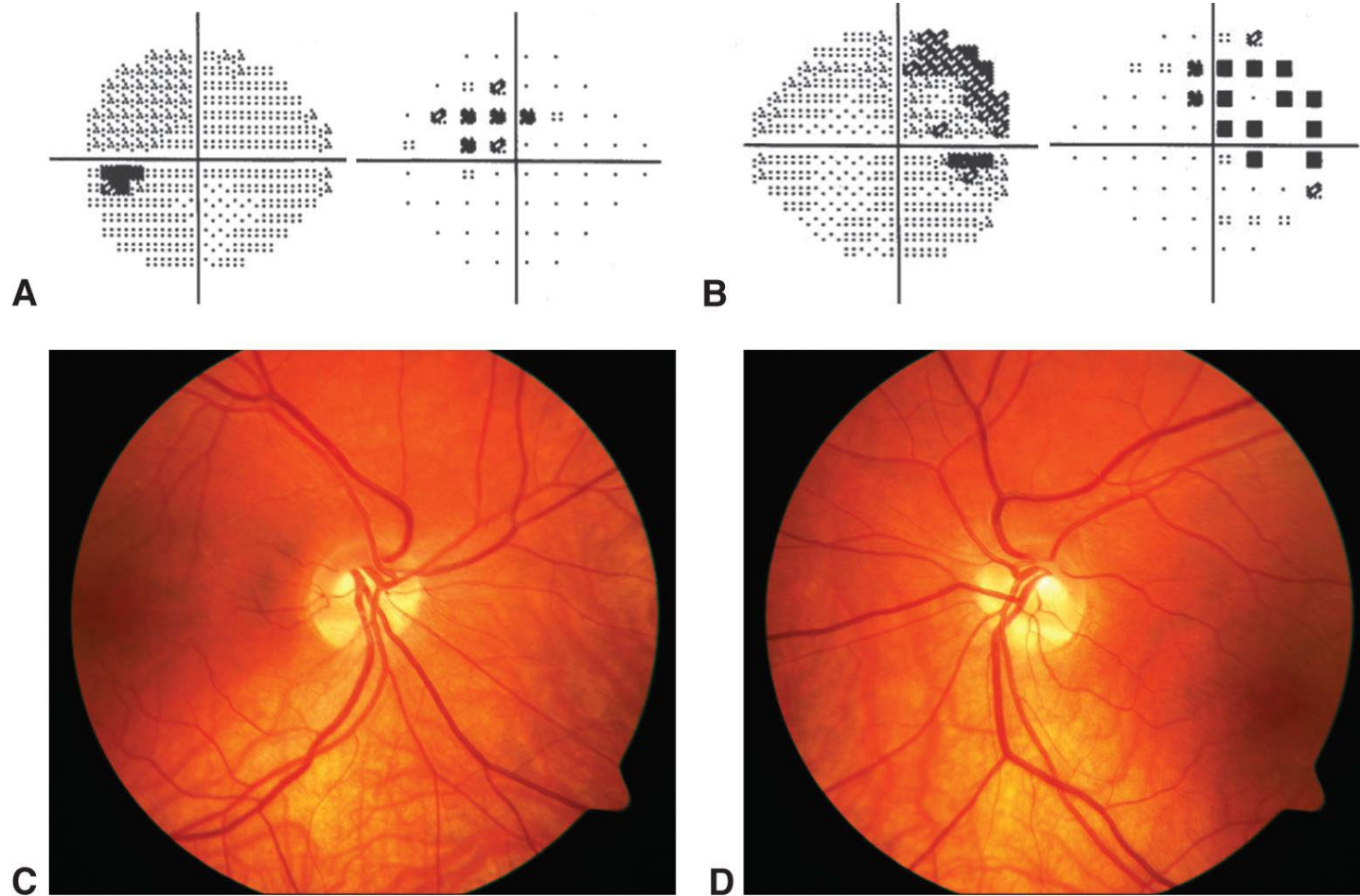
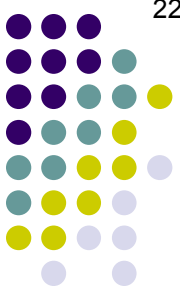
--1/3 develop...serous RD

- 3) Fundus abnormal...astigmatism
- 4) VF testing reveals...that doesn't respect the vertical...refraction

bitemporal loss

(but not hemianopic)

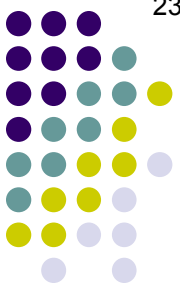
Developmental Abnormalities of the Optic Nerve Head



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

Developmental Abnormalities of the Optic Nerve Head

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

Optic Nerve Coloboma

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

Optic Pit/Hole

--Associated with...**serous RD** in adulthood

Morning-Glory Disc

--DFE reveals:

- 1) A funnel-shaped...
- 2) Number of vessels

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

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

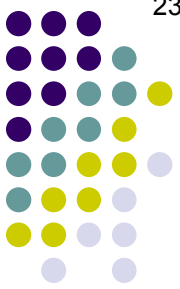
ng sign

Tilted-Disc Syndrome aka...*Fuch's Coloboma*

- 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, and may resolve with...**refraction**

Developmental Abnormalities of the Optic Nerve Head

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

Optic Nerve Coloboma

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

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Megalopapilla

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- 2) VF testing may reveal an...**enlarged blind spot**

Myelinated RNFL

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

CSNB

ng sign

Tilted-Disc Syndrome aka...*Fuch's Coloboma*

- 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, and may resolve with...**refraction**

Developmental Abnormalities of the Optic Nerve Head

232



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

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Myelinated RNFL

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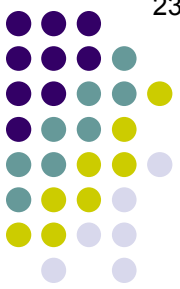
What does CSNB stand for in this context?

ng sign

Tilted-Disc Syndrome aka...*Fuch's Coloboma*

- 2) Associated with...**situs inversus** of retinal vessels
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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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--1/3 develop...**serous RD**

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- 1) Abnormally large diameter of...**disc** and **cup**
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Myelinated RNFL

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CSNB

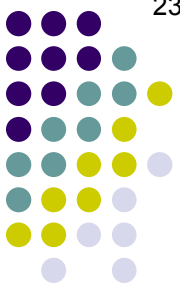
What does CSNB stand for in this context?
Congenital stationary night blindness

ing sign

Tilted-Disc Syndrome aka...Fuch's Coloboma

- 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, 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

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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**
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Myelinated RNFL

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CSNB

What does CSNB stand for in this context?
Congenital stationary night blindness

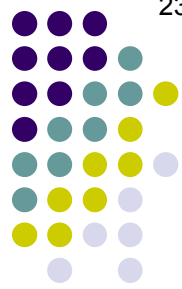
In a nutshell, what's CSNB?

Tilted-Disc Syndrome aka...Fuch's Coloboma

- 2) Associated with...**situs inversus** of retinal vessels
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- 4) VF testing reveals...**bitemporal loss** that doesn't respect the vertical, and may resolve with...**refraction**

Developmental Abnormalities of the Optic Nerve Head

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

Optic Nerve Coloboma

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

Optic Pit/Hole

--Associated with...**serous RD** in adulthood

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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**

Megalopapilla

- 1) Abnormally large diameter of...**disc** and **cup**
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Myelinated RNFL

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 leads to **rod**, **cone**, and variably decreased **abn.**

rods vs
cones

abn.

ing sign

Tilted-Disc Syndrome aka...Fuch's Coloboma

- 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, and may resolve with...**refraction**

Developmental Abnormalities of the Optic Nerve Head

236



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

Optic Nerve Coloboma

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

--Associated with...**serous RD** in adulthood

Morning-Glory Disc

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Myelinated RNFL

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

Tilted-Disc Syndrome aka...Fuch's Coloboma

- 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, and may resolve with...**refraction**