

Phakomatoses are a **massive** topic. (Don't believe me? Take a glance at the number of slides in this set.) Try to get through the whole set once a month or so. There's a TLDR at the end, so when it's crunch time (ie, the last few weeks before the OKAPs, WQE or Boards), just flip through the TLDR a few times every day to keep it fresh. *You got this!*

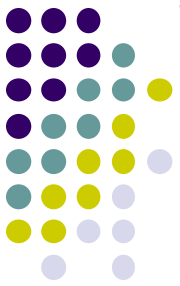
Before you begin: There's a natural break around slide 282; I placed a *break time!* slide at that location.



Phakomatoses are known also as what sort of syndrome?

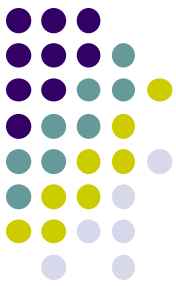


Phakomatoses are known also as what sort of syndrome?
Neuro-oculocutaneous syndromes



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In general terms, how do phakomatoses present?



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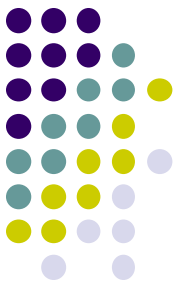
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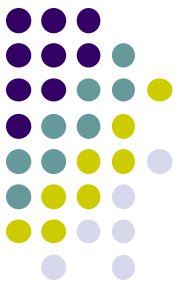
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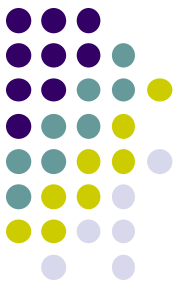


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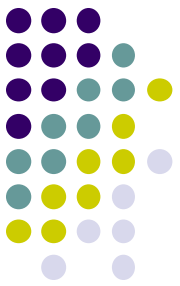


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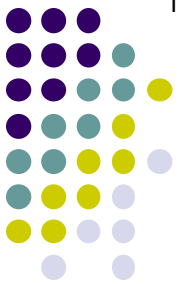
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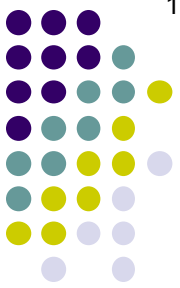
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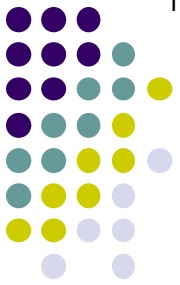
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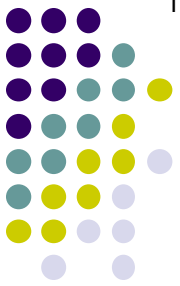
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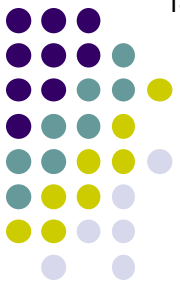
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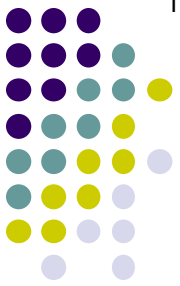
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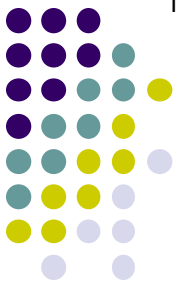
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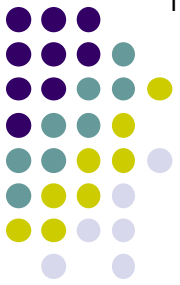
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That a lesion is a hamartoma or choristoma indicates what about its status vis a vis malignancy?
That it is **benign**



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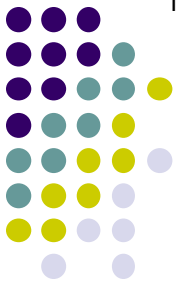
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Is there a single, universally accepted definition of the term phakomatosis?



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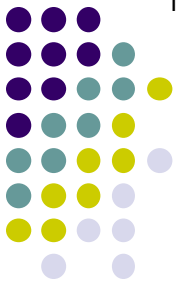
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
Is there a single, universally accepted definition of the term phakomatosis?

Unfortunately not, and for this reason, the conditions so labelled will vary from source to source



Q

A phakomatosis by any other name...by what other name is each syndrome known?

 Abbreviations used henceforth

NF1 • Neurofibromatosis type 1:

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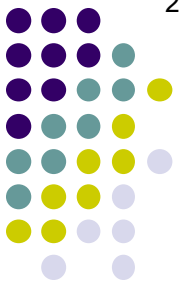
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Start here with the other name for NF1

 Abbreviations used henceforth



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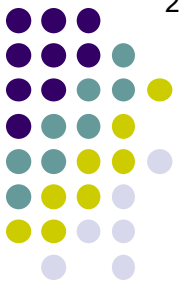
NF1 • Neurofibromatosis type 1: von Recklinghausen syndrome



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


Abbreviations used henceforth



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NF1 • Neurofibromatosis type 1: von Recklinghausen syndrome

TS • Tuberous sclerosis:

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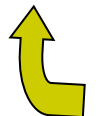
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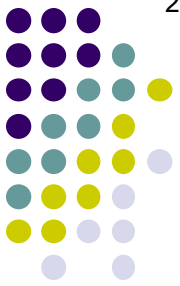
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Next for TS

 Abbreviations used henceforth



A

A phakomatosis by any other name...by what other name is each syndrome known?



Abbreviations used henceforth

NF1 • Neurofibromatosis type 1: von Recklinghausen syndrome

TS • Tuberous sclerosis: Bournville disease



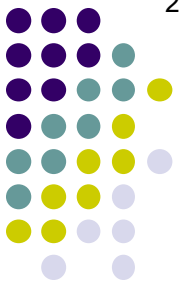
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


Abbreviations used henceforth

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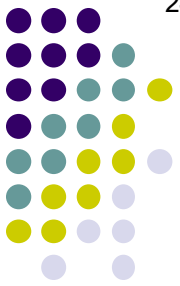


Etc

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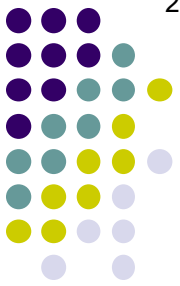
- NF1 • Neurofibromatosis type 1: von Recklinghausen syndrome
- TS • Tuberous sclerosis: Bournville disease
- SWS • Sturge-Weber syndrome: Encephalotrigeminal angiomatosis



Other names you might encounter for SWS:
Encephalofacial angiomatosis
Cerebrofacial angiomatosis



Abbreviations used henceforth



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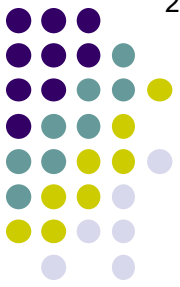
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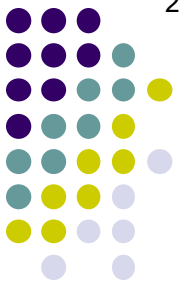
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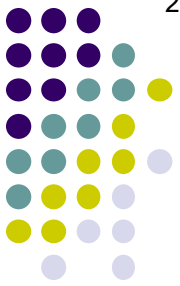
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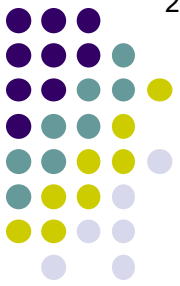


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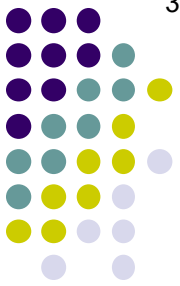
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- NF2 ● Neurofibromatosis type 2:



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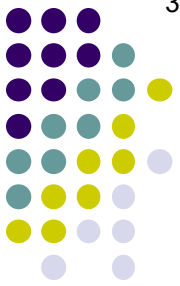


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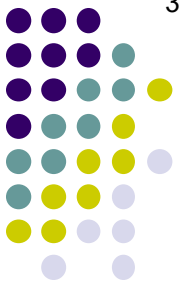
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MISME is an acronym. What does it stand for?

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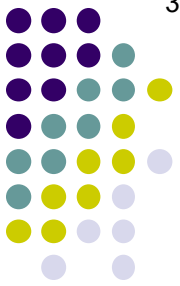
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- M**ultiple
- I**nherited
- S**chwannomas,
- M**eningiomas (and)
- E**pendymomas



Q

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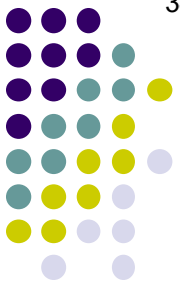
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- NF2 • Neurofibromatosis type 2: MISME syndrome
- RA • Racemose angioma:

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


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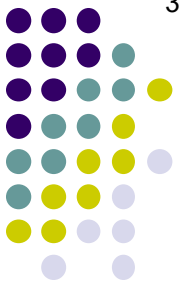
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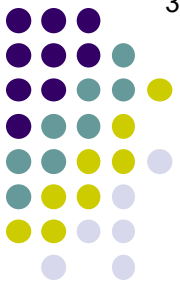


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- AT ● Ataxia-telangiectasia:



Abbreviations used henceforth



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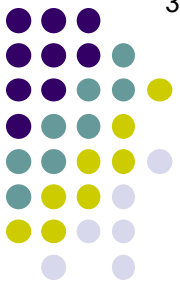


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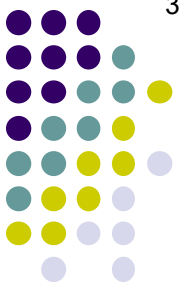
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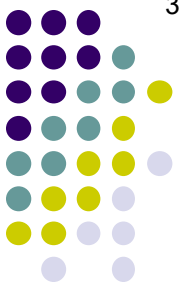
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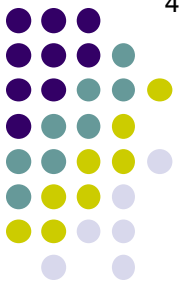
- *Phakomatoses: Inheritance patterns*
 - These four are **AD...**
 - ?
 - ?
 - ?
 - ?



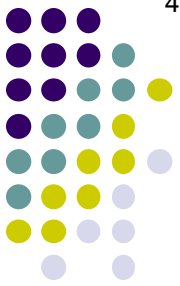
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 - NF1
 - von Hippel-Lindau
 - Tuberous sclerosis



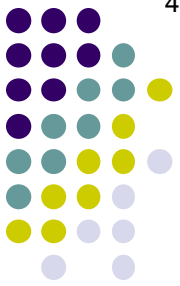
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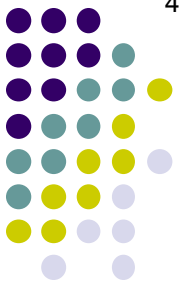
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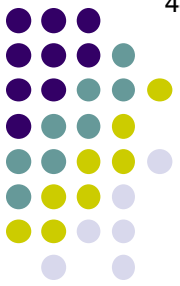
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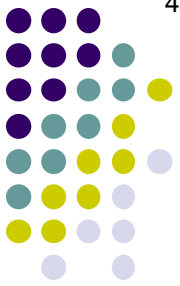
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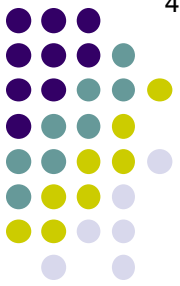


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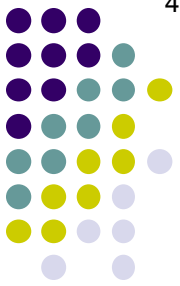
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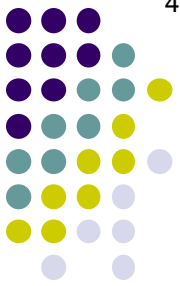
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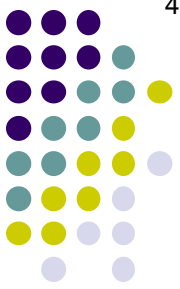
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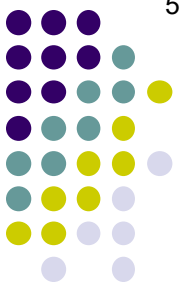
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*Hold the phone! To say that 'almost' all pts are female means that **some** IP pts are male. If IP is X-linked dominant and lethal in hemizygous individuals, how could there be **any** male pts?*

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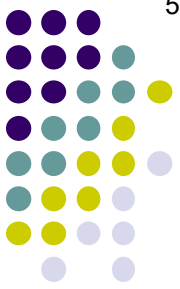
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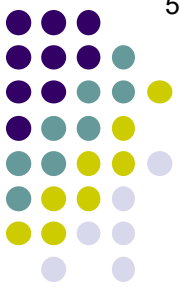
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CNS issue #1

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--Absence or dysgenesis of the

CNS issue #2 (two words)

--?

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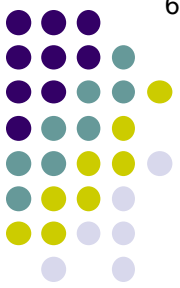
--Absence

--Choriore

**Aicardi syndrome has its own slide-set (FELT20)—
see it for more on this condition**

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Of course, even the inherited conditions can occur sporadically. For each, what percent of cases are sporadic?

● Phakomatoses

- These four are *AD*...

% Sporadic

NF2	?
● NF1	?
● von Hippel-Lindau	?
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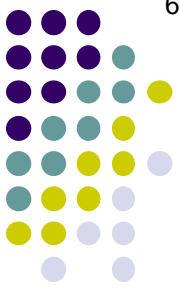
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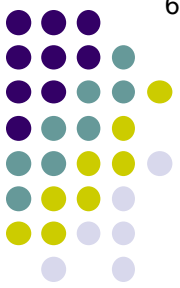
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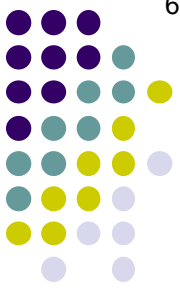
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Why is the sporadic-occurrence rate of A-T essentially zero?



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Why is the sporadic-occurrence rate of A-T essentially zero?

Because it is AR, it can occur sporadically only if someone heterozygous for it happens to suffer a mutation of the **other** copy of the responsible gene—an unlikely event.

Phakomatoses



NF1

--

central vs peripheral

 NF

Phakomatoses



NF1

--*Peripheral* NF

Phakomatoses



NF1

--*Peripheral* NF

--Most lesions due to abnormal one cell type or diff cell type cells

Phakomatoses



NF1

--*Peripheral* NF

--Most lesions due to abnormal **melanocytes** or **neuroglial** cells

Phakomatoses



NF1

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How are these cell lines related embryologically?

Phakomatoses



NF1

--Peripheral NF

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melanocytes or **neuroglial cells**

How are these cell lines related embryologically?
Both derive from **neural-crest cells**

Briefly, what's the backstory on neural crest cells—what are they, how do they develop?

Phakomatoses



NF1

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NCCs are a subtype of **embryo cell type** cells

Phakomatoses



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Phakomatoses



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NCCs are a subtype of neuroectodermal cells. Early v Late in embryogenesis, some of the neuroectodermal cells located along the dorsal v ventral aspect of the structure (two words) are induced to transition into NCCs.

Phakomatoses



NF1

--Peripheral NF

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Phakomatoses



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Phakomatoses



NF1

--Peripheral NF

--Most lesions due to abnormal

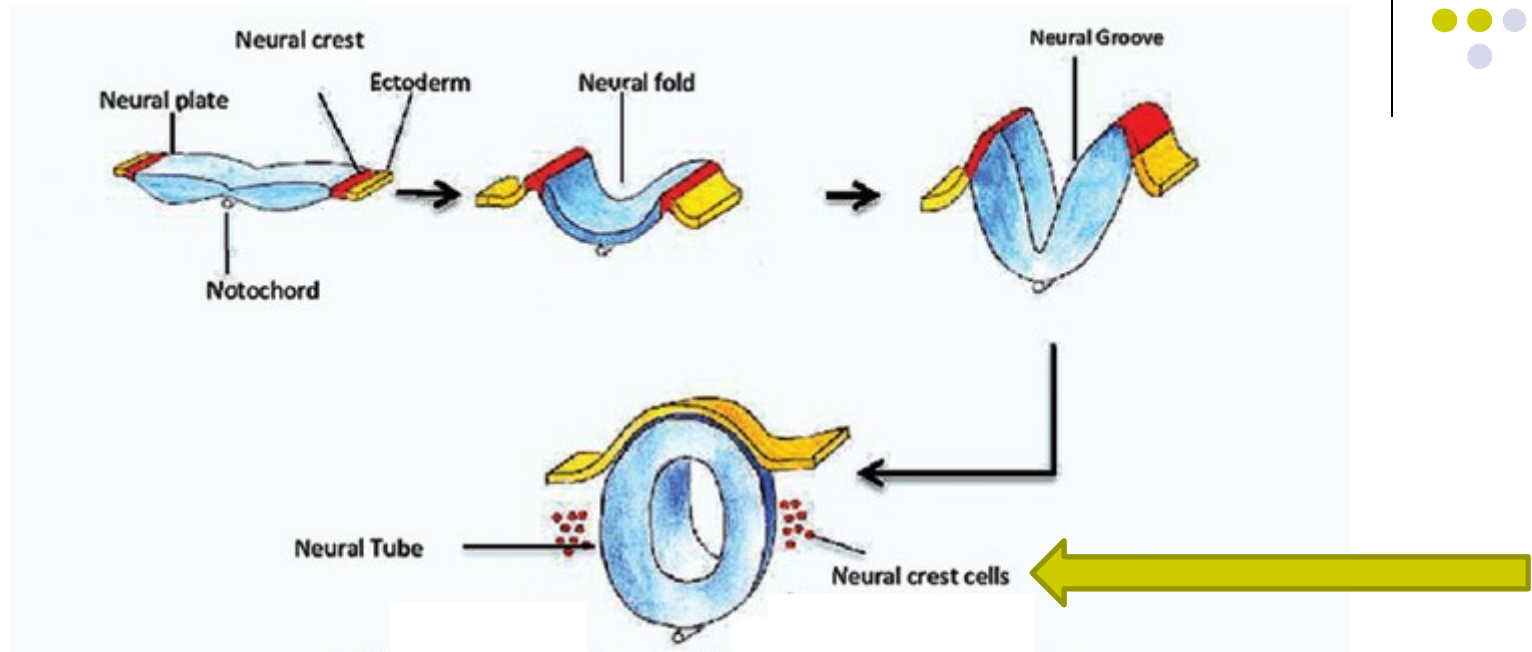
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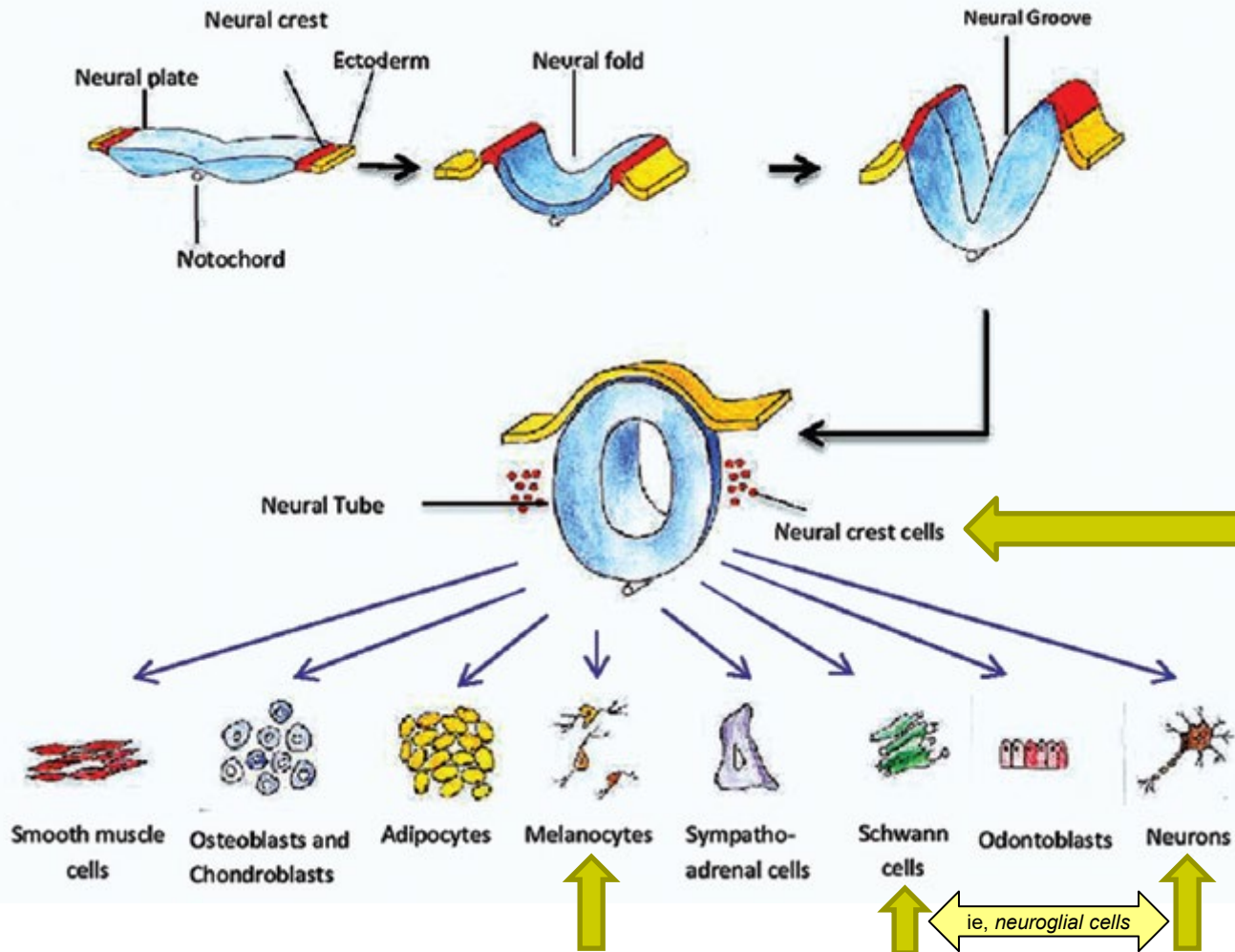
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Melanocytic Eyelid and Epibulbar Lesions



Neural crest cells...

Melanocytic Eyelid and Epibulbar Lesions



Neural crest cells...and their derivatives

Phakomatoses



NF1

--Peripheral NF

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melanocytes or **neuroglial cells**

How are these cell lines related embryologically?
Both derive from **neural-crest cells**

Given this, it should come as no surprise that NF is associated with other manifestations of disordered neural-crest embryology, including and especially:

- **two long words** (aka **something of something**)

--?

--?

Phakomatoses



NF1

--Peripheral NF

--Most lesions due to abnormal

melanocytes or **neuroglial cells**

How are these cell lines related embryologically?

Both derive from **neural-crest cells**

Given this, it should come as no surprise that NF is associated with other manifestations of disordered neural-crest embryology, including and especially:

--Oculodermal melanocytosis (aka **nevus of Ota**)

--?

--?

Phakomatoses

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--Choroidal **yikes**

--?

Phakomatoses



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--?

Phakomatoses



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--Choroidal melanoma

--Conjunctival **yikes** stemming from **abb.**

Phakomatoses



NF1

--Peripheral NF

--Most lesions due to abnormal

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- Oculodermal melanocytosis (aka **nevus of Ota**)
- Choroidal melanoma
- Conjunctival melanoma stemming from PAM

Phakomatoses



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--Peripheral NF

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How are these cell lines related embryologically?
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--Choroidal melanoma

--Conjunctival melanoma stemming from **PAM**

What does PAM stand for in this context?

Phakomatoses



NF1

--Peripheral NF

--Most lesions due to abnormal

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How are these cell lines related embryologically?
Both derive from **neural-crest cells**

Given this, it should come as no surprise that NF is associated with other manifestations of disordered neural-crest embryology, including and especially:

--Oculodermal melanocytosis (aka **nevus of Ota**)

--Choroidal melanoma

--Conjunctival melanoma stemming from **PAM**

What does PAM stand for in this context?
Primary acquired melanosis

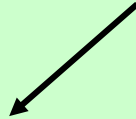
Phakomatoses



NF1

--Peripheral NF

--Most lesions due to abnormal **melanocytes** or **neuroglial** cells



Melanocytic lesions

--?
--?
--?
--?

Name four common NF1
lesions that derive from
melanocytes

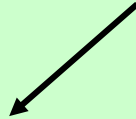
Phakomatoses



NF1

--Peripheral NF

--Most lesions due to abnormal **melanocytes** or **neuroglial** cells



Melanocytic lesions

- [] spots
- Axillary/inguinal []
- [] nodules
- Choroidal lesions

Name four common NF1 lesions that derive from melanocytes

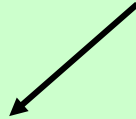
Phakomatoses



NF1

--Peripheral NF

--Most lesions due to abnormal **melanocytes** or **neuroglial** cells

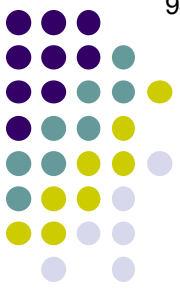


Melanocytic lesions

- Café au lait spots
- Axillary/inguinal freckles
- Lisch nodules
- Choroidal lesions

Name four common NF1 lesions that derive from melanocytes

Phakomatoses



Lisch nodules



Axillary freckling



Café au lait spots

NF1: Melanocytic lesions

Phakomatoses



NF1

--Peripheral NF

--Most lesions due to abnormal **melanocytes** or **neuroglial** cells

Melanocytic lesions

- Café au lait spots
- Axillary/inguinal freckles
- Lisch nodules
- Choroidal lesions

Neuroglial lesions

- ?
- ?
- ?
- ?

Name four
common NF1
lesions that
derive from
neuroglial cells

Phakomatoses



NF1

--Peripheral NF

--Most lesions due to abnormal **melanocytes** or **neuroglial** cells

Melanocytic lesions

- Café au lait spots
- Axillary/inguinal freckles
- Lisch nodules
- Choroidal lesions

Neuroglial lesions

- Nodular neurofibromas
- Plexiform neurofibromas
- Optic
- Prominent corneal

Name four common NF1 lesions that derive from neuroglial cells

Phakomatoses



NF1

--Peripheral NF

--Most lesions due to abnormal **melanocytes** or **neuroglial** cells

Melanocytic lesions

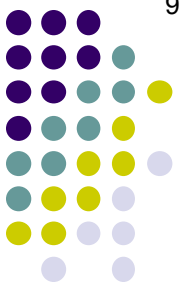
- Café au lait spots
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- Lisch nodules
- Choroidal lesions

Neuroglial lesions

- Nodular neurofibromas
- Plexiform neurofibromas
- Optic glioma
- Prominent corneal nerves

Name four
common NF1
lesions that
derive from
neuroglial cells

Phakomatoses



Optic nerve glioma



Plexiform neurofibroma



Nodular neurofibroma

NF1: Neuroglial lesions

Phakomatoses



NF1

--Peripheral NF

--Most lesions due to abnormal **melanocytes** or **neuroglial** cells

Melanocytic lesions

- Café au lait spots
- Axillary/inguinal freckles
- Lisch nodules
- Choroidal lesions

Neuroglial lesions

- Nodular neurofibromas
- Plexiform neurofibromas
- Optic glioma
- Prominent corneal nerves

In what fundamental way do these lesions differ (other than the cell type of origin, duh)?

Phakomatoses



NF1

--Peripheral NF

--Most lesions due to abnormal **melanocytes** or **neuroglial** cells

Melanocytic lesions

- Café au lait spots
- Axillary/inguinal freckles
- Lisch nodules
- Choroidal lesions

Neuroglial lesions

- Nodular neurofibromas
- Plexiform neurofibromas
- Optic glioma
- Prominent corneal nerves

In what fundamental way do these lesions differ (other than the cell type of origin, duh)?

The **m'cytic v N-G** lesions are of no clinical significance beyond establishing the diagnosis, whereas the **m'cytic v N-G** lesions are associated with significant ocular and/or systemic morbidity

Phakomatoses



NF1

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- Café au lait spots
- Axillary/inguinal freckles
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- Choroidal lesions

Neuroglial lesions

- Nodular neurofibromas
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In what fundamental way do these lesions differ (other than the cell type of origin, duh)?
The **melanocytic** lesions are of no clinical significance beyond establishing the diagnosis, whereas the **neuroglial** lesions are associated with significant ocular and/or systemic morbidity

Phakomatoses



- NF1**
--Peripheral NF
--Most lesions due to abnormal **melanocytes** or **neuroglial** cells

Melanocytic lesions

- Café au lait spots
- Axillary/inguinal freckles
- Lisch nodules
- Choroidal lesions

Neuroglial lesions

- Nodular neurofibromas
- Plexiform neurofibromas
- Optic glioma
- Prominent corneal nerves

Do the neuroglial lesions in NF1 carry a risk of malignant transformation?

Phakomatoses



- NF1**
--Peripheral NF
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Melanocytic lesions

- Café au lait spots
- Axillary/inguinal freckles
- Lisch nodules
- Choroidal lesions

Neuroglial lesions

- Nodular neurofibromas
- Plexiform neurofibromas
- Optic glioma
- Prominent corneal nerves

Do the neuroglial lesions in NF1 carry a risk of malignant transformation?

Yes, especially the [redacted], which can give rise to lesions known as

[redacted]
five words

Phakomatoses



- NF1**
--Peripheral NF
--Most lesions due to abnormal **melanocytes** or **neuroglial** cells

Melanocytic lesions

- Café au lait spots
- Axillary/inguinal freckles
- Lisch nodules
- Choroidal lesions

Neuroglial lesions

- Nodular neurofibromas
- Plexiform neurofibromas
- Optic glioma
- Prominent corneal nerves

Do the neuroglial lesions in NF1 carry a risk of malignant transformation?

Yes, especially the plexiform neurofibromas, which can give rise to lesions known as '**malignant peripheral nerve-sheath tumors**'

Phakomatoses



- NF1**
--Peripheral NF
--Most lesions due to abnormal **melanocytes** or **neuroglial** cells

Melanocytic lesions

- Café au lait spots
- Axillary/inguinal freckles
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- Choroidal lesions

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- Nodular neurofibromas
- Plexiform neurofibromas
- Optic glioma
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Yes, especially the plexiform neurofibromas, which can give rise to lesions known as '**malignant peripheral nerve-sheath tumors**'

What is the lifetime risk of such a transformation?

Phakomatoses



- NF1**
--Peripheral NF
--Most lesions due to abnormal **melanocytes** or **neuroglial** cells

Melanocytic lesions

- Café au lait spots
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- Optic glioma
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Do the neuroglial lesions in NF1 carry a risk of malignant transformation?

Yes, especially the plexiform neurofibromas, which can give rise to lesions known as '**malignant peripheral nerve-sheath tumors**'

What is the lifetime risk of such a transformation?

About 10%

Phakomatoses



NF1

~ Peripheral NF

--**Most** lesions due to abnormal **melanocytes** or **neuroglial** cells

What does 'most' mean in this context?

Phakomatoses



NF1
- Peripheral NF
--**Most** lesions due to abnormal **melanocytes** or **neuroglial** cells

What does 'most' mean in this context?

It means 'not all.' That is, there are lesions associated with NF1 that cannot be attributed to abnormalities of neural-crest derivatives.

Phakomatoses



NF1
- Peripheral NF
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Four non-neural-crest-derived malignancies are associated with NF1 (albeit uncommonly). What are they?

--?

--?

--?

--?

Phakomatoses



NF1
--Peripheral NF
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It means 'not all.' That is, there are lesions associated with NF1 that cannot be attributed to abnormalities of neural-crest derivatives.

Four non-neural-crest-derived malignancies are associated with NF1 (albeit uncommonly). What are they?

- Leukemia
- Rhabdomyosarcoma
- Pheochromocytoma
- Wilms tumor

Phakomatoses



NF1

--*Peripheral* NF

--Most lesions due to abnormal **melanocytes** or **neuroglial** cells

--Glaucoma associated with ipsilateral classic lid finding and/or less classic iris finding

Phakomatoses



NF1

--*Peripheral* NF

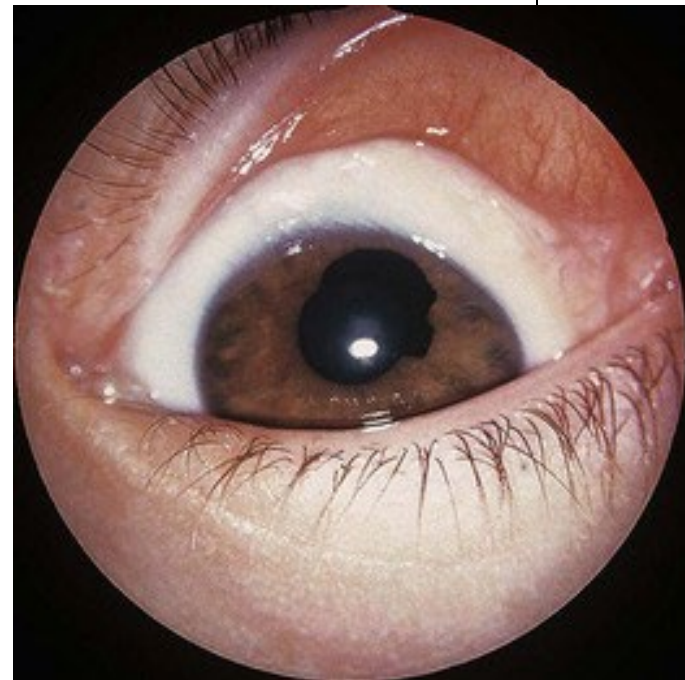
--Most lesions due to abnormal **melanocytes** or **neuroglial** cells

--Glaucoma associated with ipsilateral **upper-lid plexiform fibroma** and/or **iris ectropion**

Phakomatoses



Plexiform neurofibroma



Ectropion uveae

NF1

Phakomatoses



NF1

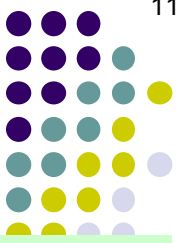
--Peripheral NF

--Most lesions due to abnormal melanocytes or neuroglial cells

--**Glaucoma associated with ipsilateral upper-lid plexiform fibroma and/or iris ectropion**

How does a plexiform fibroma and/or iris ectropion cause glaucoma?

Phakomatoses



NF1

--Peripheral NF

--Most lesions due to abnormal melanocytes or neuroglial cells

--**Glaucoma associated with ipsilateral upper-lid plexiform fibroma and/or iris ectropion**

How does a plexiform fibroma and/or iris ectropion cause glaucoma?

So far as we know, they don't. That is, while they are strongly associated with glaucoma in NF1, there is no known direct causal connection.

Phakomatoses



NF1

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How strong is the association with glaucoma; ie, what percent of NF1 cases with an upper-lid plexiform fibroma and/or ectropion will have ipsilateral glaucoma?

Phakomatoses



NF1

--Peripheral NF

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How strong is the association with glaucoma; ie, what percent of NF1 cases with an upper-lid plexiform fibroma and/or ectropion will have ipsilateral glaucoma?

About 50

Phakomatoses



NF1

--*Peripheral* NF

--Most lesions due to abnormal **melanocytes** or **neuroglial** cells

--Glaucoma associated with ipsilateral **upper-lid plexiform fibroma** and/or **iris ectropion**

--Iris lesions include classic finding, less classic and congenital less classic

Phakomatoses



NF1

--*Peripheral* NF

--Most lesions due to abnormal **melanocytes** or **neuroglial** cells

--Glaucoma associated with ipsilateral **upper-lid plexiform fibroma** and/or **iris ectropion**

--Iris lesions include **Lisch nodules**, **JXG nodules**, and congenital **ectropion**

Phakomatoses



NF1

--Peripheral NF

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--Glaucoma associated with ipsilateral upper-lid plexiform fibroma and/or iris ectropion

--Iris lesions include **Lisch nodules**, XG nodules, and congenital ectropion

What rule of thumb adheres regarding the appearance of Lisch nodules?

Phakomatoses



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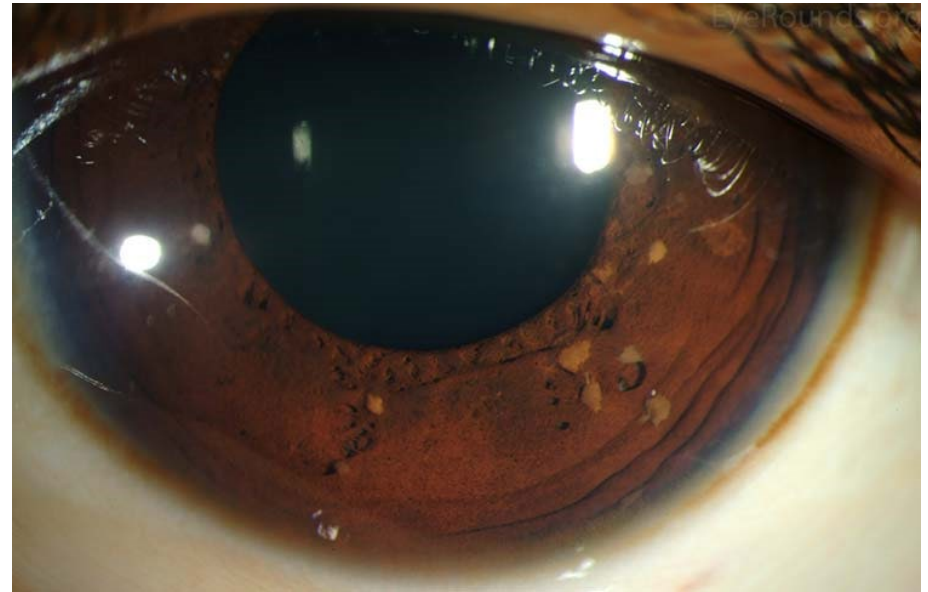
What rule of thumb adheres regarding the appearance of Lisch nodules?

Lisch nodules are **lighter** than the rest of the iris when the iris in question is dark, but **darker** than the rest when the iris is light

Phakomatoses



Darker on light iris



Lighter on dark iris

NF1: Lisch nodules

Phakomatoses



NF1

--Peripheral NF

--Most lesions due to abnormal

--Glaucoma associated with ip

--Iris lesions include **Lisch nodules**

What tops the DDx for Lisch nodules, ie, what other sort of nodule can they be confused with?

Phakomatoses



NF1

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What tops the DDx for Lisch nodules, ie, what other sort of nodule can they be confused with?

Iris mammillations

Phakomatoses



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What tops the DDx for Lisch nodules, ie, what other sort of nodule can they be confused with?

Iris **mammillations**

Mammillations? Aren't those a CNS thingamajig?

Phakomatoses



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Iris **mammillations**

Mammillations? Aren't those a CNS thingamajig?

You're thinking of the **two words**,
paired structures that are part of the limbic system

Phakomatoses



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What tops the DDX for Lisch nodules, ie, what other sort of nodule can they be confused with?

Iris **mammillations**

Mammillations? Aren't those a CNS thingamajig?

You're thinking of the **mammillary bodies**, paired structures that are part of the limbic system

Phakomatoses



NF1

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What tops the DDx for Lisch nodules, ie, what other sort of nodule can they be confused with?

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OK, then what are iris mammillations?

Phakomatoses



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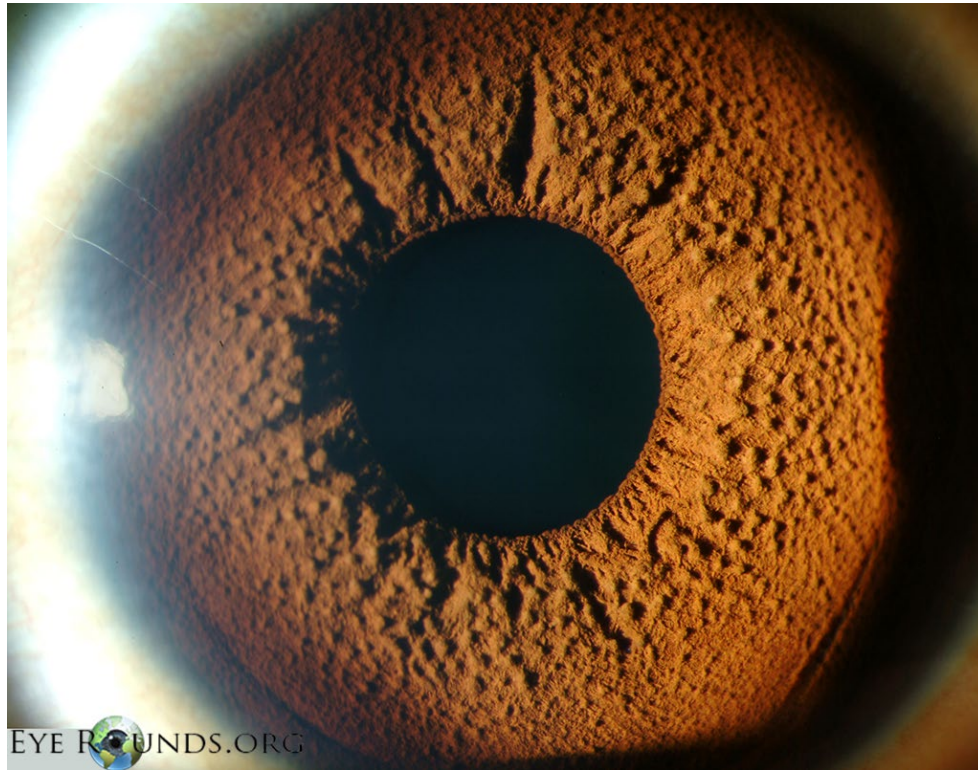
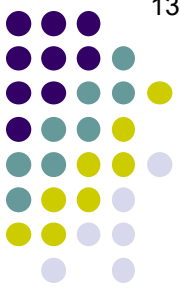
What tops the DDx for Lisch nodules, ie, what other sort of nodule can they be confused with?

Iris mammillations

OK, then what are iris mammillations?

Tiny pigmented nodules which, when present, are found in vast numbers diffusely scattered across the iris surface

Phakomatoses



Iris mammilations

Phakomatoses



NF1

--Peripheral NF

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Iris mammillations

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Tiny pigmented nodules which, when present, are found in vast numbers diffusely scattered across the iris surface

Are they unilateral, or bilateral?

Phakomatoses



NF1

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--Glaucoma associated with ip

--Iris lesions include **Lisch nodules**

What tops the DDx for Lisch nodules, ie, what other sort of nodule can they be confused with?

Iris mammillations

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Tiny pigmented nodules which, when present, are found in vast numbers diffusely scattered across the iris surface

Are they unilateral, or bilateral?

Usually unilateral, but bilaterality occurs frequently enough that it can't be used to rule them out

Phakomatoses



NF1

--Peripheral NF

--Most lesions due to abnormal

--Glaucoma associated with ip

--Iris lesions include **Lisch nodules**

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Are they associated with NF1?

Phakomatoses



NF1

--Peripheral NF

--Most lesions due to abnormal

--Glaucoma associated with ip

--Iris lesions include **Lisch nodules**

What tops the DDx for Lisch nodules, ie, what other sort of nodule can they be confused with?

Iris mammillations

OK, then what are iris mammillations?

Tiny pigmented nodules which, when present, are found in vast numbers diffusely scattered across the iris surface

Are they unilateral, or bilateral?

Usually unilateral, but bilaterality occurs frequently enough that it can't be used to rule them out

Are they associated with NF1?

Yes (albeit not nearly as strongly as Lisch nodules)

Phakomatoses



NF1

--Peripheral NF

--Most lesions due to abnormal

--Glaucoma associated with ip

--Iris lesions include **Lisch nodules**

What tops the DDx for Lisch nodules, ie, what other sort of nodule can they be confused with?

Iris mammillations

OK, then what are iris mammillations?

Tiny pigmented nodules which, when present, are found

'Tiny pigmented iris nodules associated with NF1'—given this, how on earth are you supposed to differentiate between the two?

*Are they **associated with NF1**?*

Yes (albeit not nearly as strongly as Lisch nodules)

Phakomatoses



NF1

--Peripheral NF

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Iris mammillations

OK, then what are iris mammillations?

Tiny pigmented nodules which, when present, are found

'Tiny pigmented iris nodules associated with NF1'—given this, how on earth are you supposed to differentiate between the two?

By appearance. As previously stated, Lisch nodules are lighter than the rest of the iris when the iris in question is dark, but darker than the rest when the iris is light.

*Are they **associated with NF1**?*

Yes (albeit not nearly as strongly as Lisch nodules)

Phakomatoses



NF1

--Peripheral NF

--Most lesions due to abnormal

--Glaucoma associated with ip

--Iris lesions include **Lisch nodules**

What tops the DDX for Lisch nodules, ie, what other sort of nodule can they be confused with?

Iris mammillations

OK, then what are iris mammillations?

Tiny pigmented nodules which, when present, are found

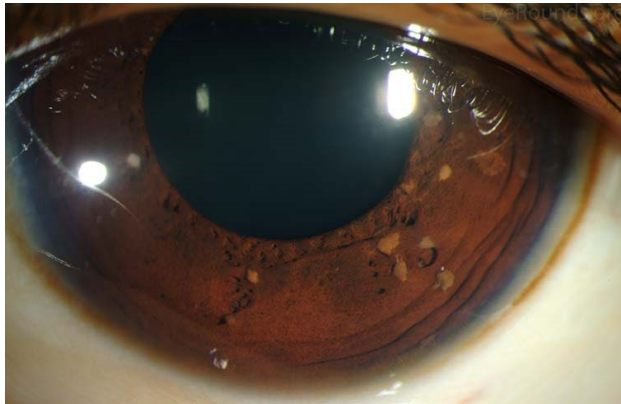
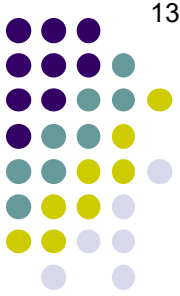
'Tiny pigmented iris nodules associated with NF1'—given this, how on earth are you supposed to differentiate between the two?

By appearance. As previously stated, Lisch nodules are lighter than the rest of the iris when the iris in question is dark, but darker than the rest when the iris is light. In contrast, iris mammillations are always the same color as the rest of the iris.

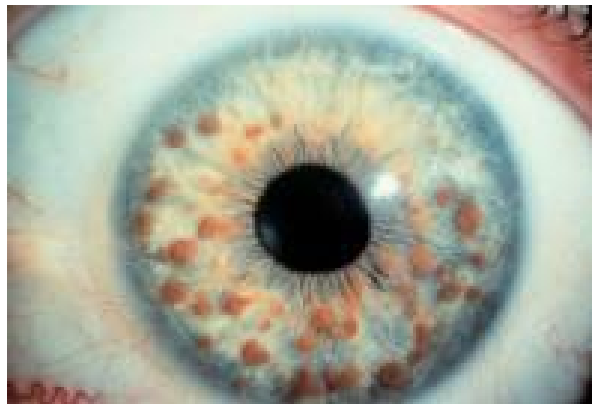
*Are they **associated with NF1**?*

Yes (albeit not nearly as strongly as Lisch nodules)

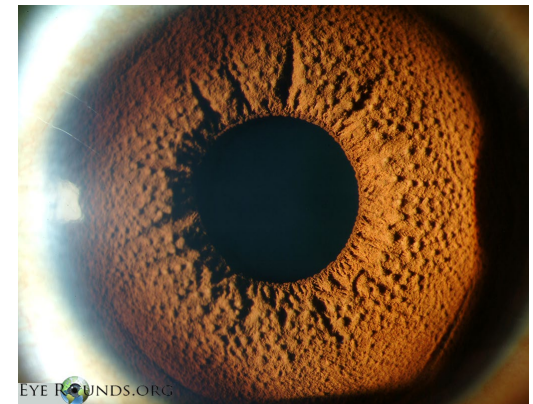
Phakomatoses



Lisch nodules lighter on dark iris



Lisch nodules darker on light iris



Mammillations same color as iris

Lisch nodules vs iris mammillations

Phakomatoses



NF1

--Peripheral NF

--Most lesions due to abnormal melanocytes or neuroglial cells

--Glaucoma associated with ipsilateral upper lid lesion (Mojavian lesion)

--Iris lesions include Lisch nodules (nodules)

JXG

What does JXG stand for in this context?

Phakomatoses



NF1

--Peripheral NF

--Most lesions due to abnormal melanocytes or neuroglial cells

--Glaucoma associated with ipsilateral upper lid lesion

--Iris lesions include Lisch nodules

JXG

What does JXG stand for in this context?

Juvenile xanthogranuloma

Phakomatoses



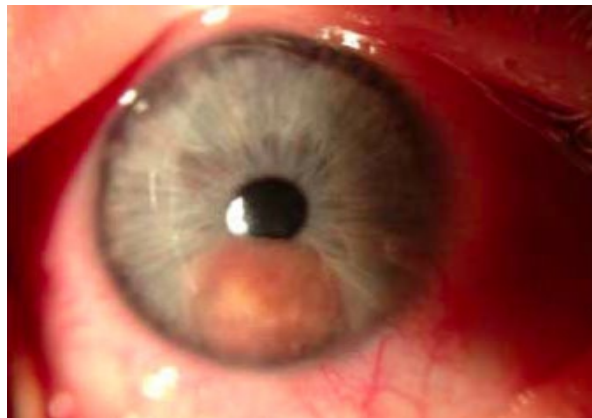
NF1

--Peripheral NF

--Most lesions due to abnormal melanocytes or neuroglial cells

--Glaucoma associated with ipsilateral upper lid plexiform fibroma and/or iris ectropion

--Iris lesions include Lisch nodules **JXG nodules** and congenital ectropion



NF1: JXG nodules

Phakomatoses



NF1

--Peripheral NF

--Most lesions due to abnormal melanocytes or neuroglial cells

--Glaucoma associated with ipsilateral upper lid plexiform fibroma and/or iris ectropion

--Iris lesions include Lisch nodules **JXG nodules** and congenital ectropion

In three words, what sort of condition is it?

It is a...

Phakomatoses



NF1

--Peripheral NF

--Most lesions due to abnormal melanocytes or neuroglial cells

--Glaucoma associated with ipsilateral upper lid plexiform fibroma and/or iris ectropion

--Iris lesions include Lisch nodules **JXG nodules** and congenital ectropion

In three words, what sort of condition is it?

It is a...nonneoplastic histiocytic proliferation

Phakomatoses



NF1

--Peripheral NF

--Most lesions due to abnormal melanocytes or neuroglial cells

--Glaucoma associated with ipsilateral upper lid plexiform fibroma and/or iris ectropion

--Iris lesions include Lisch nodules **JXG nodules** and congenital ectropion

In three words, what sort of condition is it?

It is a...nonneoplastic histiocytic proliferation

What are the two hallmarks of JXG histology?

The presence of...?

The presence of...?

Phakomatoses



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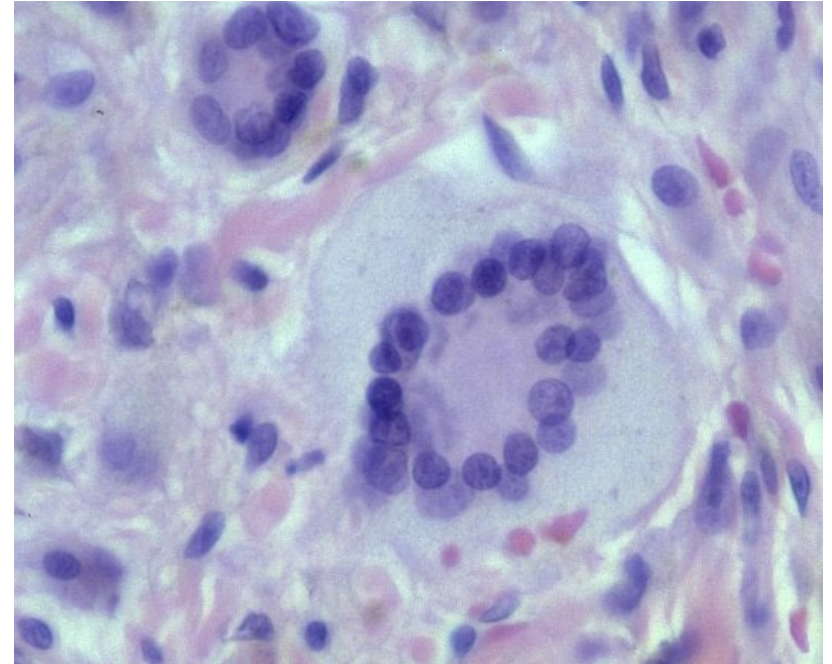
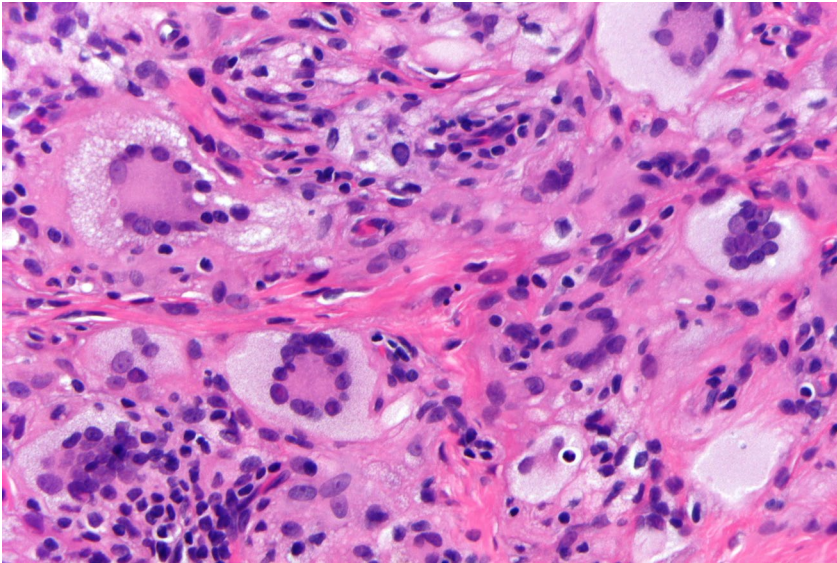
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The presence of...Touton giant cells

The presence of...?

Phakomatoses



JXG: Touton giant cells

Phakomatoses



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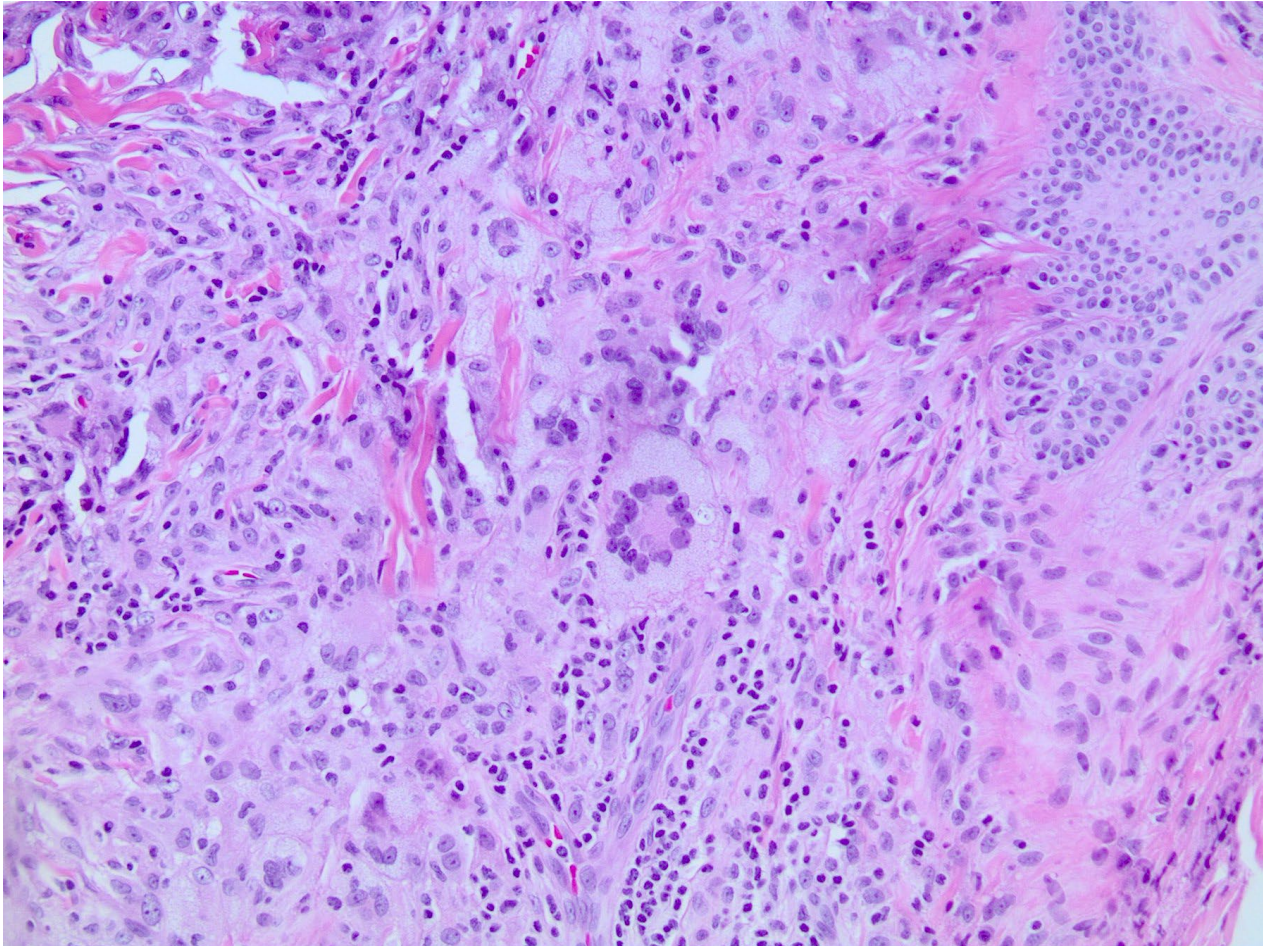
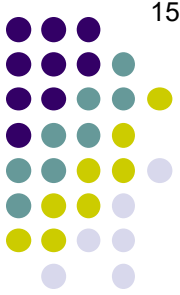
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The presence of... 'foamy macrophages'

Phakomatoses



JXG: Foamy macrophages (and a Touton giant cell)

Phakomatoses



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The majority before age 1 year, and almost all by age 2

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Phakomatoses



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Phakomatoses



JXG nodules

Phakomatoses



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1 word 1 abb.

Phakomatoses



NF1

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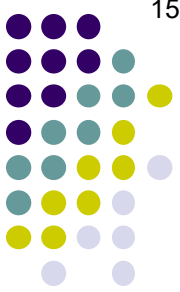
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Phakomatoses



NF1: Optic nerve gliomas bilaterally. Note the 'kinked' appearance

Phakomatoses



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Phakomatoses



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--Rule of thumb for Lisch nodule prevalence: something x something

Phakomatoses



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Phakomatoses



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--Rule of thumb for Lisch nodule prevalence: **Age in years x 10**

In other words, about 10% of 1 year olds will have Lisch nodules, 40% of 4 yo, 60% of 6 yo, etc.

By the age of 10 years, essentially 100% of NF1 pts will manifest Lisch nodules.

Phakomatoses



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Tuberous sclerosis

- Classic triad is *epiloia*

Phakomatoses



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*What does **epiloia** stand for?*

--**Epi**

--**Lo i**

--**A**

Phakomatoses



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--**Epilepsy**

--**Low intelligence**

--**Angiomas**

Phakomatoses



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What is the eponymous name of this triad?

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What % of TS pts have seizures?

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Tuberous sclerosis

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--**Low Intelligence**: **50**

--**Angiomas**

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--**Epilepsy**: 80

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--**Angiomas**: ?

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Phakomatoses



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Tuberous sclerosis

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--**Angiomas**: 75

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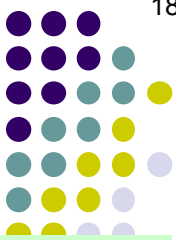
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What % of TS pts have all three?

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Vogt's triad

Phakomatoses



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Tuberous sclerosis

--Classic triad is **epiloia**

What does **epiloia** stand for?

--**Epilepsy PLUS**

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--**Angiomas**

What % of TS pts have all three? Only 30

What is the eponymous name of this triad?

Vogt's triad

Phakomatoses



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Tuberous sclerosis

- Classic triad is *epiloia*
- Skin: classic finding of face

Phakomatoses



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Tuberous sclerosis

- Classic triad is *epiloia*
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Phakomatoses



Tuberous sclerosis: Adenoma sebaceum

Phakomatoses



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- Skin: **Adenoma sebaceum** of face; and on torso

Phakomatoses



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Phakomatoses



Tuberous sclerosis: Ash leaf spots

Phakomatoses



Tuberous sclerosis: Shagreen patch

Phakomatoses



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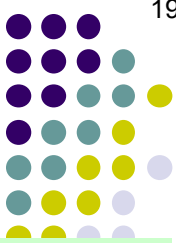
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Skin Lesions: Matching!

<i>Adenoma sebaceum</i>	?	Appear in infancy
<i>Shagreen patches</i>	?	Usually in lumbosacral region
<i>Ash-leaf spots</i>	?	Appear in childhood

Phakomatoses



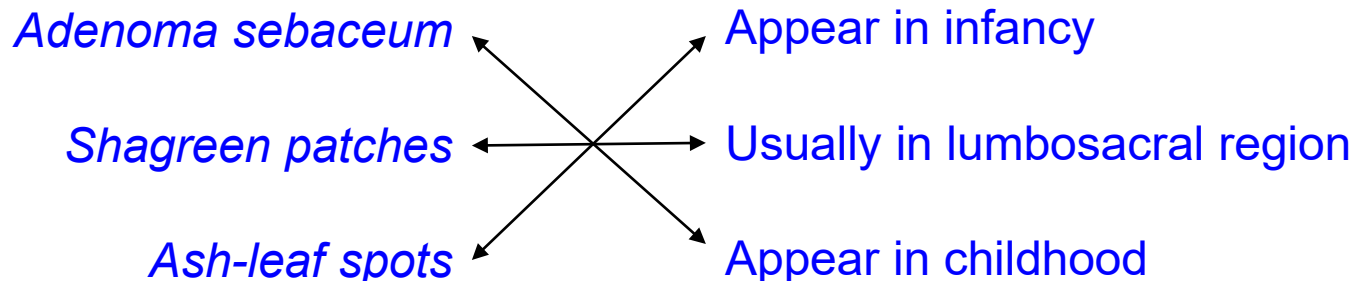
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Tuberous sclerosis

- Classic triad is *epiloia*
- Skin: **Adenoma sebaceum** of face; **ash-leaf spots** and **shagreen patches** on torso

Skin Lesions: Matching!



Phakomatoses



NF1

--*Peripheral* NF

--Most lesions due to abnormal **melanocytes** or **neuroglial** cells

--Glaucoma associated with ipsilateral **upper-lid plexiform fibroma** and/or **iris ectropion**

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Skin Lesions: Not Matching!

Adenoma sebaceum ?

Shagreen patches ?

Ash-leaf spots ?

*Which lesion(s) is/are raised,
and which is/are flat?*

Phakomatoses



NF1

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Skin Lesions: Not Matching!

Adenoma sebaceum

Shagreen patches

Ash-leaf spots

Raised

Flat

*Which lesion(s) is/are raised,
and which is/are flat?*

Phakomatoses



NF1

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Skin Lesions: Not Matching!

Adenoma sebaceum ?

Shagreen patches ?

Ash-leaf spots ?

*Which lesion(s) is/are
hyperpigmented, and which
is/are hypopigmented?*

Phakomatoses



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Adenoma sebaceum

Shagreen patches

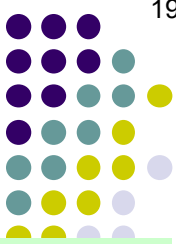
Ash-leaf spots

} Hyperpigmented

} Hypopigmented

*Which lesion(s) is/are
hyperpigmented, and which
is/are **hypopigmented**?*

Phakomatoses



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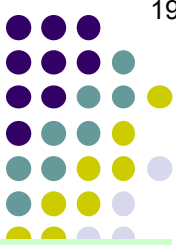
Adenoma sebaceum ?

Shagreen patches ?

Ash-leaf spots ?

*Which lesion(s) fluoresce
under a Woods lamp, and
which do/does not?*

Phakomatoses



NF1

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Skin Lesions: Not Matching!

Adenoma sebaceum

Shagreen patches

Ash-leaf spots

Don't fluoresce

Fluoresce

Which lesion(s) fluoresce under a Woods lamp, and which do/does not?

Phakomatoses



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Skin Lesions: Not Matching!

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Shagreen patches ?

Ash-leaf spots ?

*Which lesion(s) is/are
considered pathognomonic
for TS, and which is/are not?*

Phakomatoses



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Shagreen patches

Ash-leaf spots

Not

Pathognomonic

Which lesion(s) is/are considered pathognomonic for TS, and which is/are not?

Phakomatoses



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Tuberous sclerosis

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- Skin: **Adenoma sebaceum** of face; **ash-leaf spots** and **shagreen patches** on torso
- CNS:

classic finding

 other benign tumors

Phakomatoses



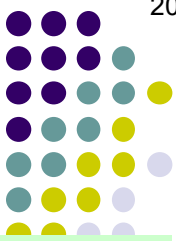
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Phakomatoses



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Tuberous sclerosis

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What is a cortical tuber?

Phakomatoses



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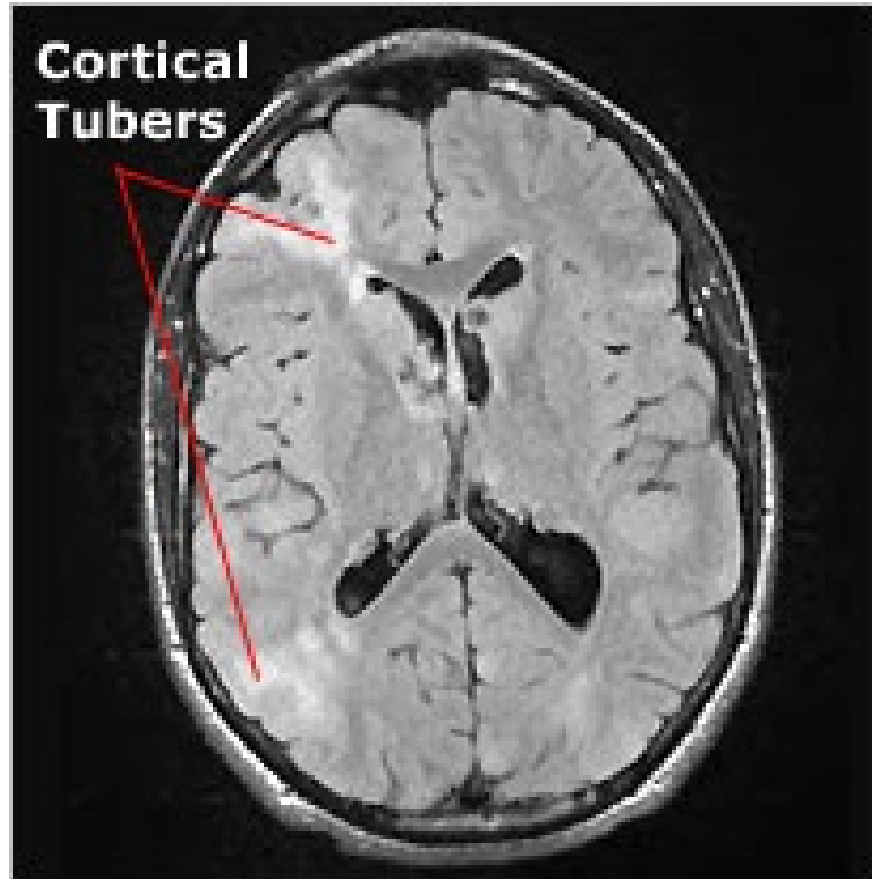
Tuberous sclerosis

- Classic triad is *epiloia*
- Skin: Adenoma sebaceum of face; ash-leaf spots and shagreen patches on torso
- CNS: **Cortical tubers**, other benign tumors

What is a cortical tuber?

A benign tumor of the brain

Phakomatoses



Tuberous sclerosis: Cortical tuber

Phakomatoses



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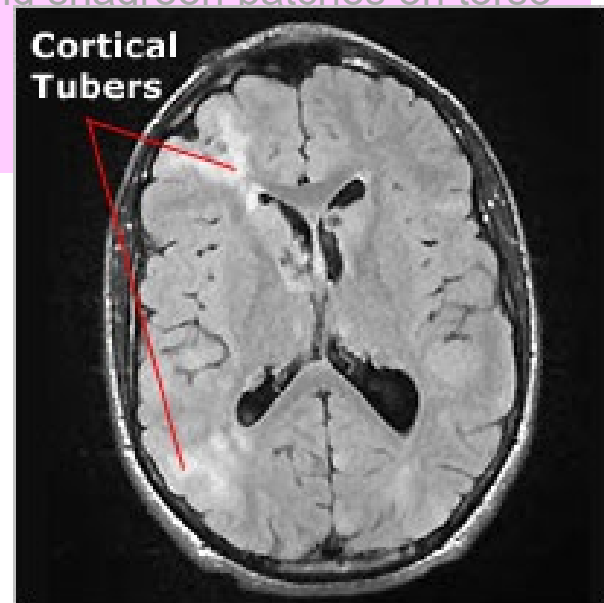
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What is a cortical tuber?

A benign tumor of the brain

Why is it called a 'tuber'?



Phakomatoses



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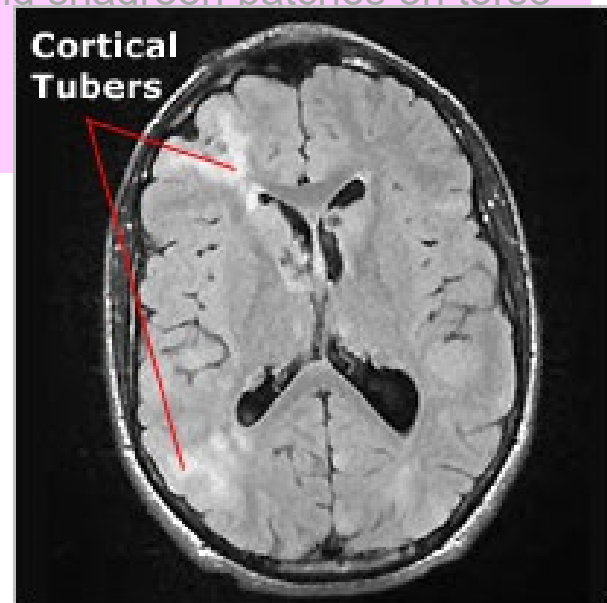
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What is a cortical tuber?

A benign tumor of the brain

Why is it called a 'tuber'?

Because it's shaped like a potato (sort of)



Phakomatoses



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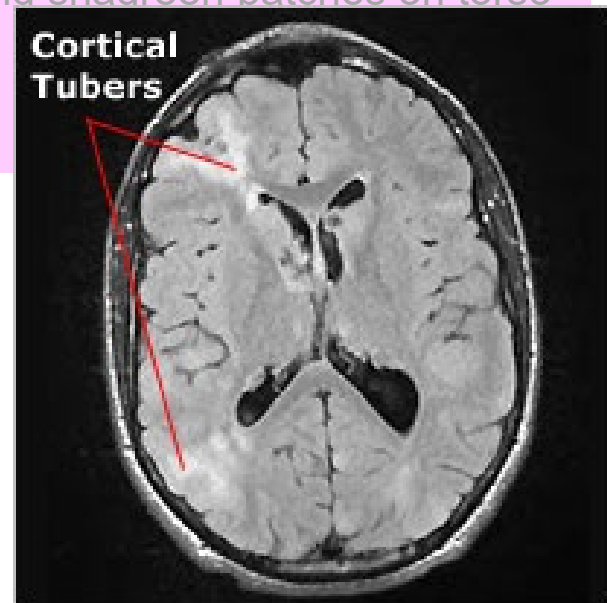
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What basic geometric shape do tubers often take?



Phakomatoses



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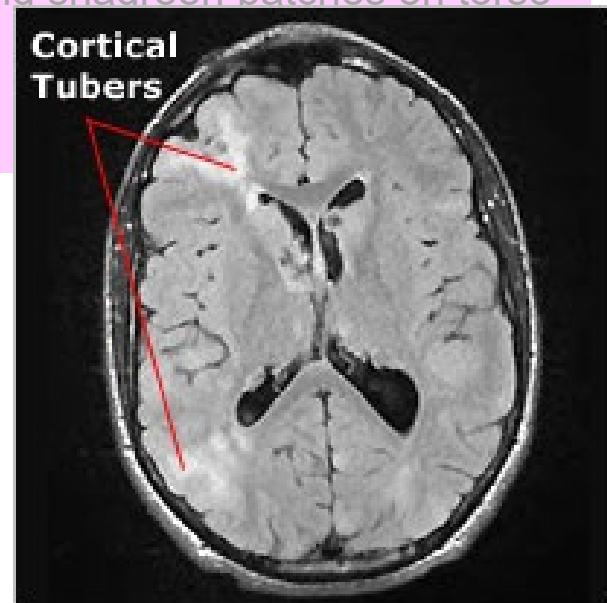
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Phakomatoses



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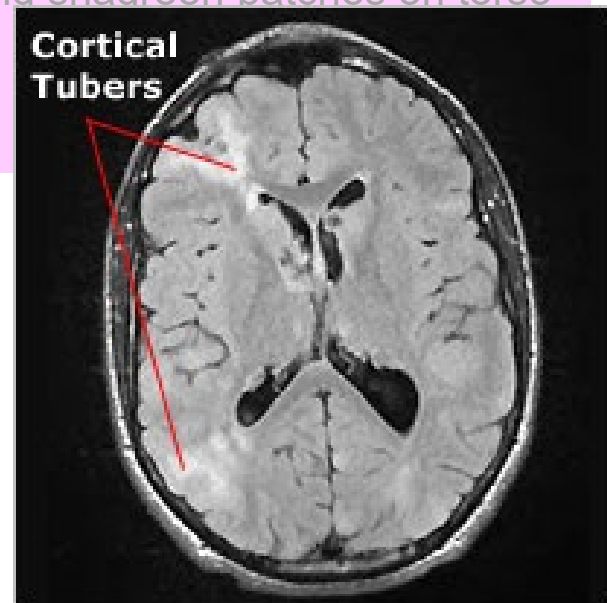
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Which way does the apex of the triangle point?



Phakomatoses



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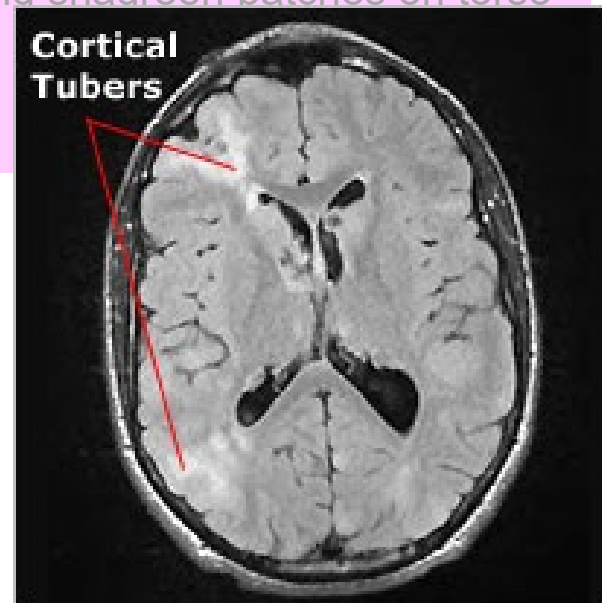
Because it's shaped like a potato (sort of)

What basic geometric shape do tubers often take?

A triangle

Which way does the apex of the triangle point?

Toward a ventricle



Phakomatoses



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Phakomatoses



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Phakomatoses



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Tuberous sclerosis

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--**Benign tumors of heart and kidney as well**

Other than their location, in what key way do the heart and kidney tumors differ?

Phakomatoses



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Other than their location, in what key way do the heart and kidney tumors differ?

The ☐/not ☐ tumors are not associated with an increased risk of morbidity/mortality, whereas the ☐/not ☐ tumors are

Phakomatoses



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Phakomatoses



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--Retinal tumor is something something

Phakomatoses



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Tuberous sclerosis

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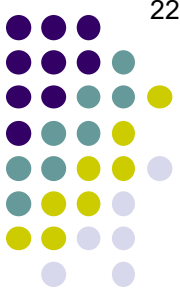
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--Retinal tumor is **astrocytic hamartoma**

Phakomatoses



Tuberous sclerosis: Astrocytic hamartoma

Phakomatoses



NF1

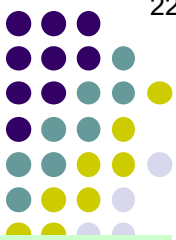
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- Retinal tumors **astrocytic hamartoma**

By what other name is the astrocytic hamartoma of the retina known?

Phakomatoses



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Retinal phakoma

Phakomatoses



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Retinal phakoma

What proportion of TS pts develop a phakoma?

Phakomatoses



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By what other name is the astrocytic hamartoma of the retina known?

Retinal phakoma

What proportion of TS pts develop a phakoma?

1/3 to 1/2

Phakomatoses



NF1

- Peripheral* NF
- Most lesions due to abnormal **melanocytes** or **neuroglial** cells
- Glaucoma associated with ipsilateral **upper-lid plexiform fibroma** and/or **iris ectropion**
- Iris lesions include **Lisch nodules**, **JXG nodules**, and congenital **ectropion**
- Optic nerve glioma: Always symptomatic by age **10 years**. Classic CT appearance: **Kinked ON**
- Rule of thumb for Lisch nodule prevalence: **Age in years x 10**

Tuberous sclerosis

- Classic triad is *epiloia*
- Skin: Adenoma sebaceum of face; ash-leaf spots
- CNS: Cortical tubers, other benign tumors
- Benign tumors of heart and kidney as well
- Retinal tumors **astrocytic hamartoma**

By what other name is the astrocytic hamartoma of the retina known?

Retinal phakoma

What proportion of TS pts develop a phakoma?
1/3 to 1/2

Can they present bilaterally?

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--Smooth, nearly flat, with poorly-defined margins

--Irregular, elevated, and sharply demarcated

Are they found in one eye?

Yes

Are they pathognomonic for TS?

No

Phakomatoses

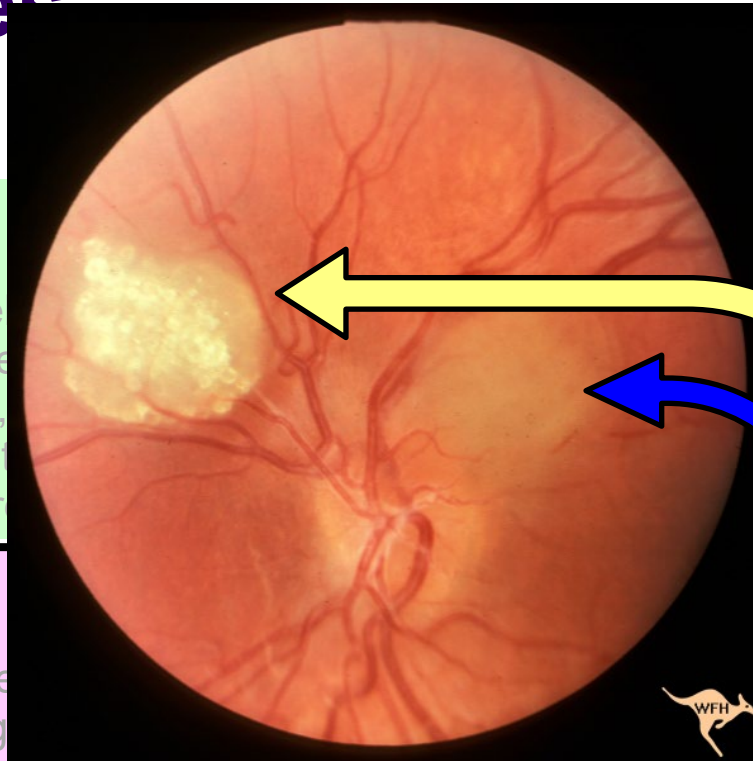


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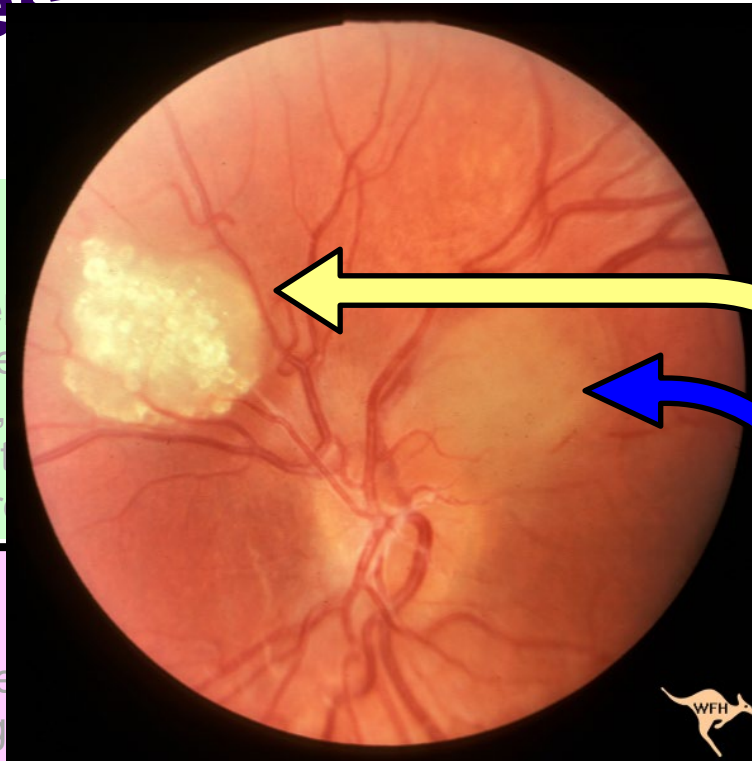
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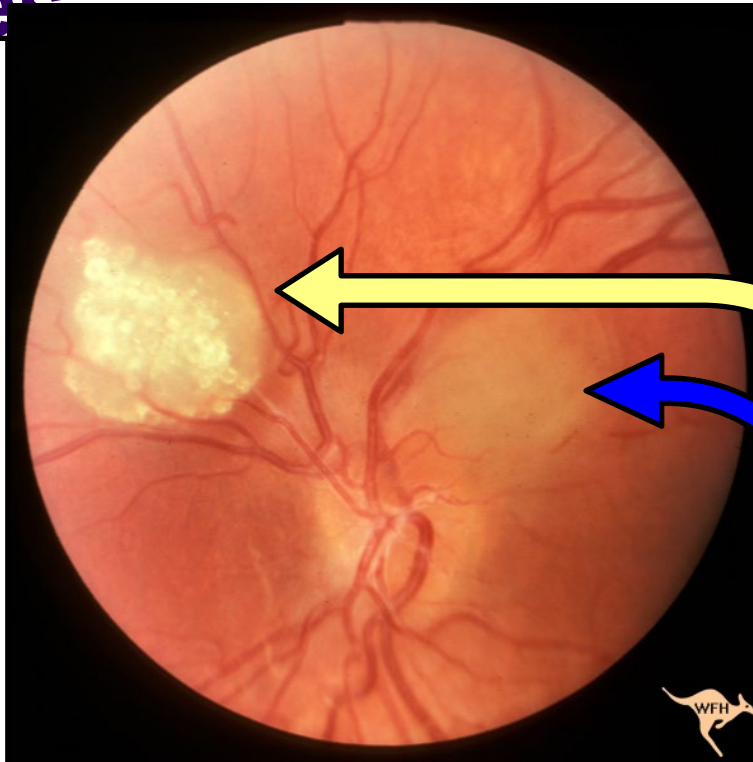
--Fruit: ?

--Foodstuff:

Phakomatoses



Mulberry



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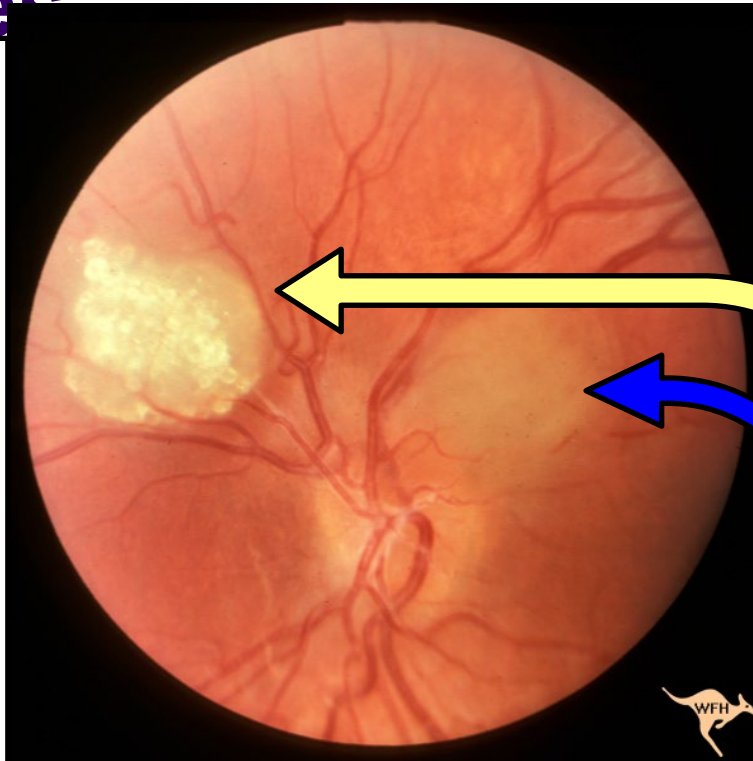
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Phakomatoses

Tapioca
(pudding)



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--Benign tumors of **heart** and **kidney** as well

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von Hippel-Lindau

--Skin:

trick question

Phakomatoses



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Phakomatoses



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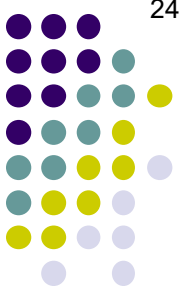
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--CNS: **Hemangioblastomas**, classically of **cerebellum** (if absent, is called **von Hippel disease**)

Phakomatoses



von Hippel-Lindau: Cerebellar hemangioblastoma

Phakomatoses



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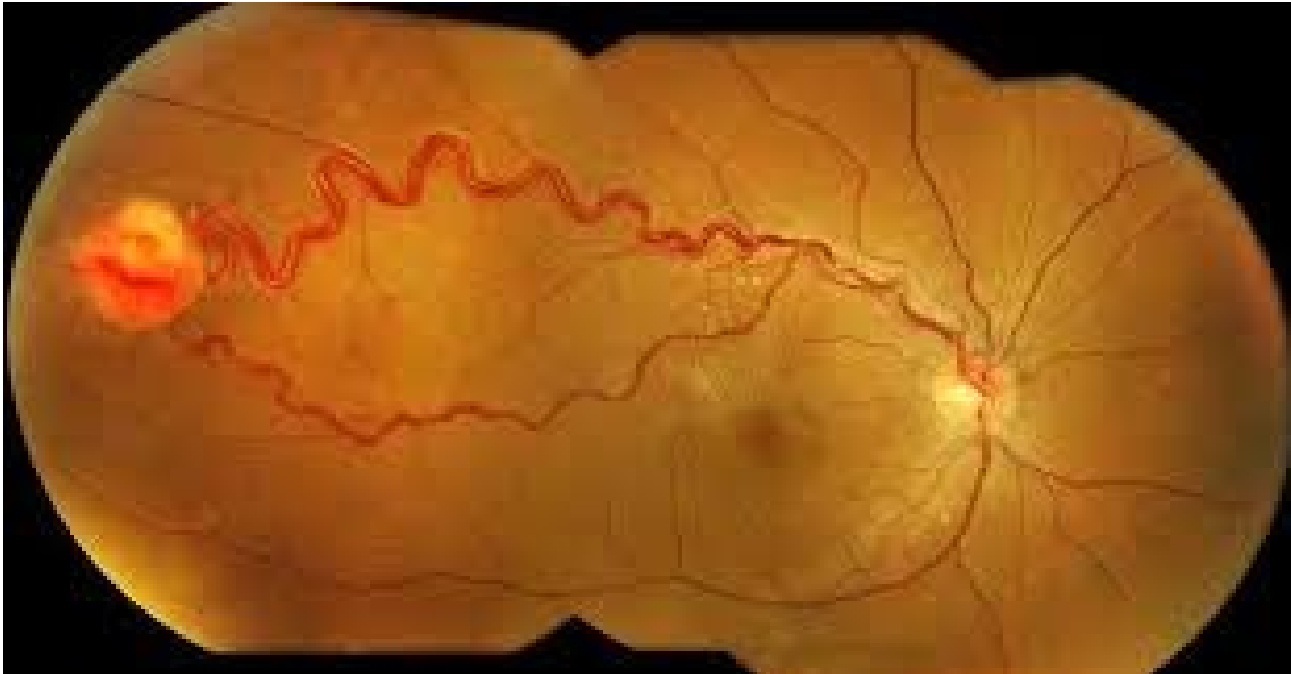
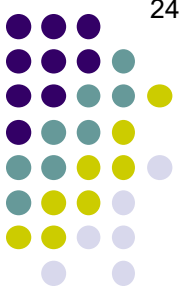
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Phakomatoses



von Hippel-Lindau: Capillary hemangioblastoma.
Note the large feeder/drainage vessels

Phakomatoses



NF1

By what other name is this lesion known (it's a subtle change)?

--F
--M
--C
--I
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--F

Tu

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Can the retinal lesions be present bilaterally?

Yes, in about ## of cases

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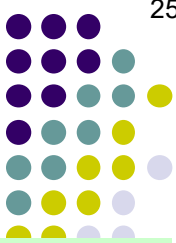
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- Cysts and tumors in multiple organs, including malignancies: Pheo, renal-cell Ca
- Retinal tumor is **capillary hemangioblastoma**; has large feeder/drainage vessels

Phakomatoses



NF1

By what other name is this lesion known (it's a subtle change)?

Capillary **hemangioma** (ie, no '-blasto-')

Can the retinal lesions be present bilaterally?

Yes, in about **1/2** of cases

Can there be multiple lesions in the same eye?

Yes, these occur in about **1/3** of cases

Are all retinal hemangio(blasto)mas associated with vHL?

--Retinal tumor is astrocytic hamartoma; can appear smooth or lumpy (*mulberry*)

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No, they can be sporadic

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Phakomatoses



NF1

--*Peripheral* NF

- Most lesions due to abnormal **melanocytes** or **neuroglial** cells
- Glaucoma associated with ipsilateral **upper-lid plexiform fibroma** and/or **iris ectropion**
- Iris lesions include **Lisch nodules**, **JXG nodules**, and congenital **ectropion**
- Optic nerve glioma: Always symptomatic by age **10 years**. Classic CT appearance: **Kinked ON**
- Rule of thumb for Lisch nodule prevalence: **Age in years x 10**

Tuberous sclerosis

- Classic triad is **epiloia**
- Skin: **Adenoma sebaceum** of face; **ash-leaf spots** and **shagreen patches** on torso
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Phakomatoses



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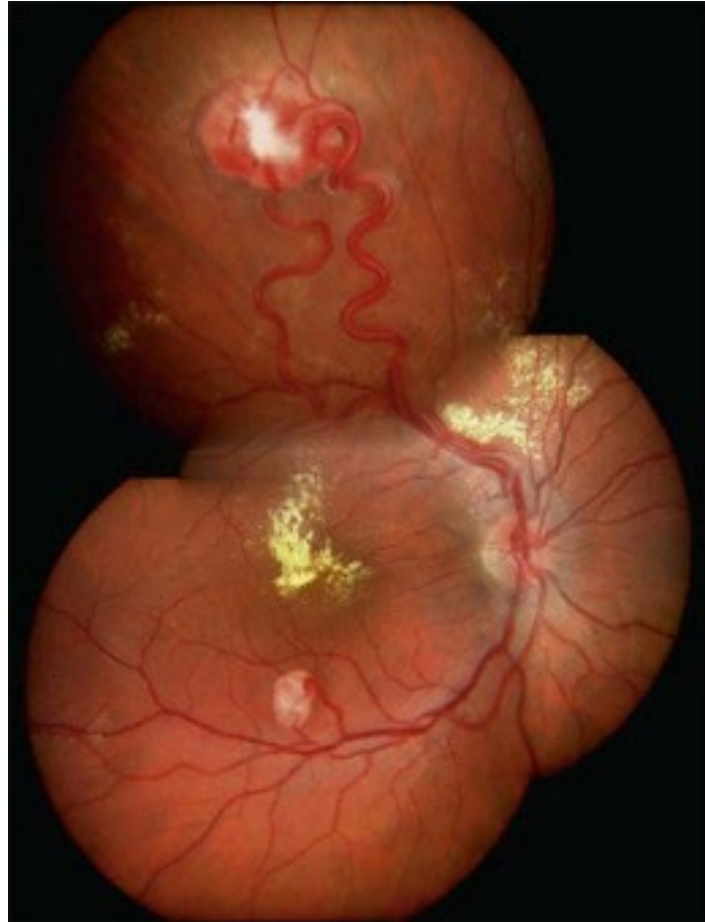
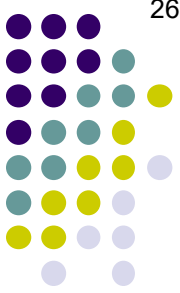
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Phakomatoses



von Hippel-Lindau: Edema

Phakomatoses



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--Management

--*Ocular*: DFE

frequency

Phakomatoses



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Phakomatoses



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--**Systemic**: Complete PE q1year with

test 1

test 2

Phakomatoses



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- Management
 - Ocular**: DFE **q1 year**
 - Systemic**: Complete PE q1year with **renal u/s**, **24° urine for VMA**

Phakomatoses



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--Tumor leaks fluid, can cause exudates, serous or cystic

--Management

--Occ

--Sys

What does VMA stand for in this context?

VMA

Phakomatoses



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--Tumor leaks fluid; can cause retinal detachment; can be treated with laser or cryo

--Management

--Ocu

--Sys

What does VMA stand for in this context?

Vanillylmandelic acid

VMA

Phakomatoses



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--Tumor leaks: can cause exudate/drainage vessels

--Management: Laser, cryo

--Occ

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VMA

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--Tumor leaks fluid; can cause retinal detachment; can be treated with laser or cryo

--Management: Genetic counseling; surveillance for tumors

--Occ

--Sys

What does VMA stand for in this context?

Vanillylmandelic acid

What is vanillylmandelic acid?

A metabolic byproduct of

VMA

Phakomatoses

270



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--Tumor leaks: can cause exudative detachment, can cause serous detachment

--Management: surgical resection, laser, cryotherapy

--Occ

--Sys

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--Tumor leaks fluid into vitreous; can cause retinal detachment or cry

--Management: Surgical resection of tumors; systemic therapy for pheo

--Occurrence: Autosomal dominant; 1:10,000

--Systemic therapy: Octreotide; somatostatin analog

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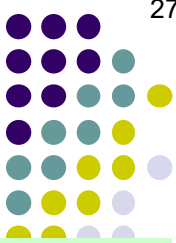
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What is the purpose of checking for VMA in the urine?

VMA

Phakomatoses



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--Management: Surgical resection of tumor; cryotherapy

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elevated vs
depressed

levels indicate the possible presence of a

VMA

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--Tumor leaks fluid into vitreous; can cause retinal detachment or vitreous hemorrhage

--Management: Surgical resection of tumors; laser therapy for retinal tumors

--Occur

--Sys

What does VMA stand for in this context?

Vanillylmandelic acid

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A metabolic byproduct of epy/norepy

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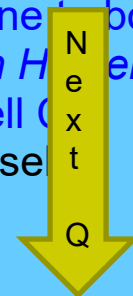
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- Management

--*Ocular*: DFE **q1 year**

--*Systemic*: Complete **PE q1 year** with renal u/s, 24^h urine for VMA; MRI brain frequency until age

yrs; after that, MRI brain frequency



Phakomatoses



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von Hippel-Lindau

--Skin: **None!** Despite this, is still considered a phakomatosis (and a classic one to boot)

--CNS: **Hemangioblastomas**, classically of **cerebellum** (if absent, is called **von Hippel disease**)

--**Cysts** and **tumors** in multiple organs, including malignancies: **Pheo**, **renal-cell Ca**

--Retinal tumor is **capillary hemangioblastoma**; has large **feeder/drainage** vessels

--Tumor leaks → **SRF** → **ERD** → decreased VA; treat with **laser** or **cryo**

--Management

--**Ocular**: DFE **q1 year**

--**Systemic**: Complete PE **q1 year** with renal u/s, 24^h urine for VMA; MRI brain **q3 years** until age 40 ; after that, MRI brain **q5 years**

Phakomatoses



NF1

--Peripheral NF

- Most lesions due to abnormal melanocytes or neuroglial cells
- Glaucoma associated with ipsilateral upper-lid plexiform fibroma and/or iris ectropion
- Iris lesions include Lisch nodules, JXG nodules, and congenital ectropion
- Optic nerve glioma: Always symptomatic by age 10 years. Classic CT appearance: Kinked ON
- Rule of thumb for Lisch nodule prevalence: Age in years x 10

Tuberous sclerosis

- Classic triad
- Skin: Adenoma
- CNS: Cortical
- Benign tumors
- Retinal tumors

Is vH-L a potentially fatal condition?

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--S It is a benign lesion. However, it is notoriously 'leaky,' and the accumulating exudate can
--C lead to compression of vital intracranial structures

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--Retinal tumor *The cerebellar hemangioma and the renal carcinoma*

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(This is a good point in the set to take a break)

Phakomatoses



NF2

Which is more common, NF1 or NF2?

Phakomatoses



NF2

Which is more common, NF1 or NF2?
NF1 is about more common

Phakomatoses



NF2

Which is more common, NF1 or NF2?
NF1 is about 10x more common

Phakomatoses



NF2

-- peripheral
vs central NF

Phakomatoses

NF2

--*Central* NF

Phakomatoses



NF2

--*Central* NF

--Classic finding: bilateral not eye

Phakomatoses

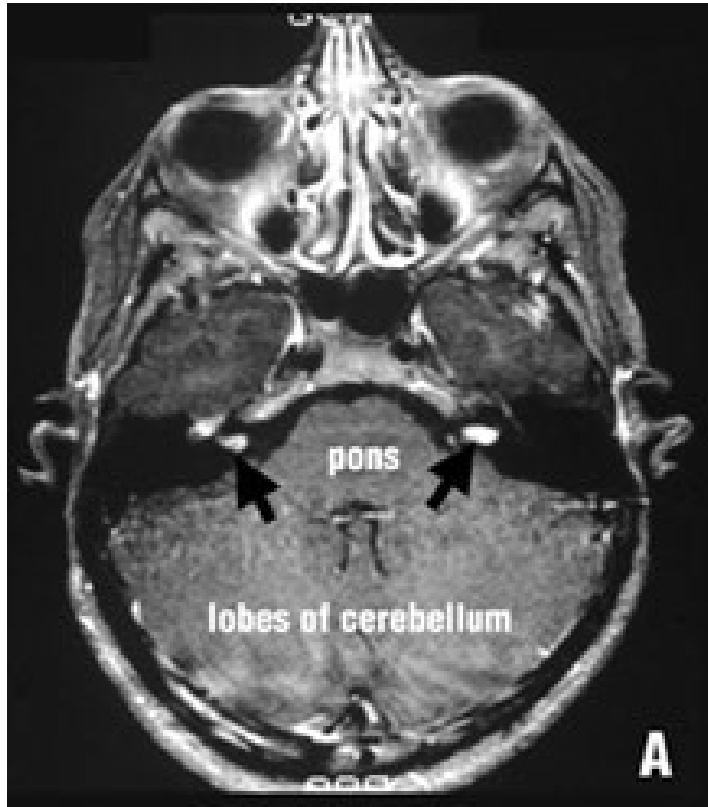
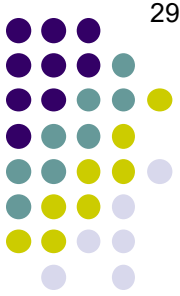


NF2

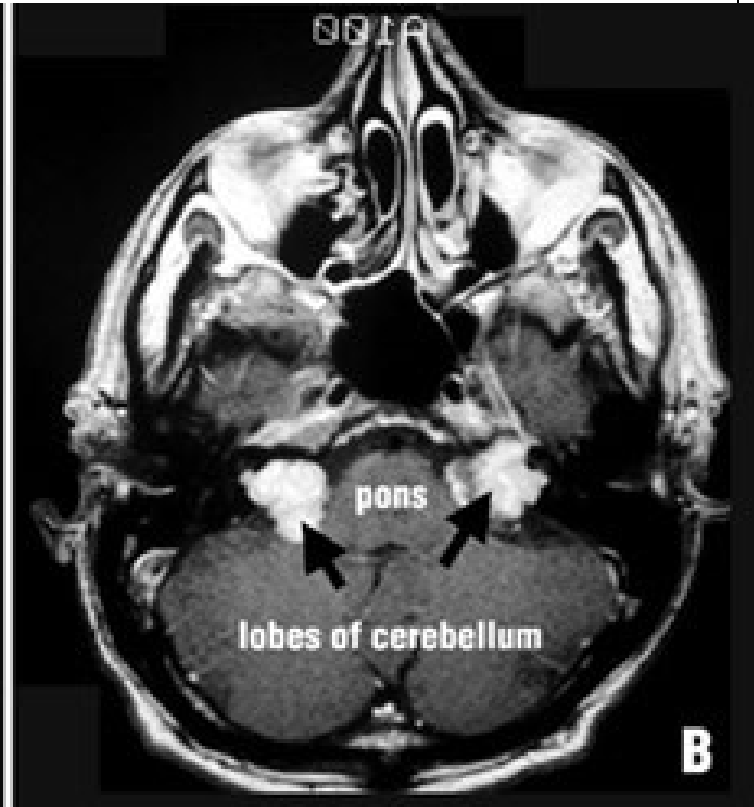
--*Central* NF

--Classic finding: bilateral *acoustic neuromas*

Phakomatoses



14 y.o. with NF2



His 50 y.o. uncle with NF2

Acoustic neuromas in NF2 (*black arrows*)

Phakomatoses



NF2

--Central NF

--Classic finding: bilateral **acoustic neuromas**

What sort of tumor is the acoustic neuroma of NF2; ie, what specific cell type is involved?

Phakomatoses



NF2

--Central NF

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What sort of tumor is the acoustic neuroma of NF2; ie, what specific cell type is involved?

A schwannoma



Phakomatoses

NF2

--Central NF

--Classic finding: bilateral **acoustic neuromas**

What are the three most common symptoms of acoustic neuroma?

#1: Reduced

#2:

#3: issues

Phakomatoses



NF2

--Central NF

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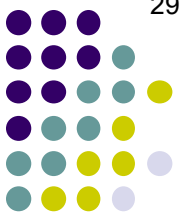
What are the three most common symptoms of acoustic neuroma?

#1: Reduced hearing

#2: Tinnitus

#3: Balance issues

Phakomatoses



NF2

--*Central* NF

--Classic finding: bilateral **acoustic neuromas**

--Eye findings: *Common:* anterior segment

Phakomatoses



NF2

--*Central* NF

--Classic finding: bilateral **acoustic neuromas**

--Eye findings: *Common:* **PSC/cortical cataracts**

Phakomatoses



NF2

- Central NF
- Classic finding: bilateral acoustic neuromas
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Are the cataracts visually significant?

Phakomatoses



NF2

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Are the cataracts visually significant?
Yes

Phakomatoses



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Are the cataracts visually significant?

Yes

Do they manifest prior to or after the acoustic neuromas?

Phakomatoses



NF2

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Are the cataracts visually significant?

Yes

Do they manifest prior to or after the acoustic neuromas?

Usually prior

Phakomatoses



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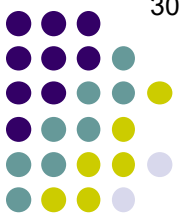
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At what age do they become clinically significant?

Phakomatoses



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Phakomatoses

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Pro tip: If you see a pt <30 years old with significant PSCs and/or cortical cataracts, consider whether s/he might have NF2!

Phakomatoses



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--Classic finding: bilateral acoustic neuromas

--Eye findings: Common: **PSC/cortical cataracts**

Are the cataracts visually significant?

Yes

Do they manifest prior to or after the acoustic neuromas?

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At what age do they become clinically significant?

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Are they unilateral, or bilateral?



Phakomatoses

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Are the cataracts visually significant?

Yes

Do they manifest prior to or after the acoustic neuromas?

Usually prior

At what age do they become clinically significant?

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Are they unilateral, or bilateral?

Both presentations are common

Phakomatoses



NF2

--*Central* NF

--Classic finding: bilateral **acoustic neuromas**

--Eye findings: *Common:* **PSC/cortical cataracts**;

Rare:

Phakomatoses



NF2

--*Central* NF

--Classic finding: bilateral **acoustic neuromas**

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Phakomatoses



NF2

--*Central* NF

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--Eye findings: *Common:* **PSC/cortical cataracts;**

Rare: **Combined hamartoma of retina and RPE;** *Rarer:*

ant seg: two words

Phakomatoses



NF2

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Melanocytic lesions

- Café au lait spots
- Axillary/inguinal freckles
- Lisch nodules
- Choroidal lesions

Neuroglial lesions

- Nodular neurofibromas
- Plexiform neurofibromas
- Optic glioma
- Prominent corneal nerves

One key difference between NF1 and NF2 is this:

In NF1, both melanocytic and neuroglial lesions are common, whereas...

Phakomatoses



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In NF2, neuroglial lesions predominate.

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Do melanocytic lesions occur in NF2 at all?



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In NF1, both melanocytic and neuroglial lesions are common, whereas...

*In NF2, neuroglial lesions **predominate**.*

Do melanocytic lesions occur in NF2 at all?

Yes. The occasional café au lait spot and/or Lisch nodule shows up now and then

Phakomatoses



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Do NF2 pts get peripheral-nerve tumors like NF1 pts?

Neuroglial lesions

--*Nodular neurofibromas?*

--*Plexiform neurofibromas?*

--*Optic glioma?*

--*Prominent corneal nerves?*

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Do NF2 pts get peripheral-nerve tumors like NF1 pts?

Yes, but at much lower rates

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--?

--?

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--**Meningiomas (intracranial)**

--**Ependymomas**



Phakomatoses

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This propensity for manifesting mainly as CNS tumors is why NF2 is referred to as 'central' NF

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NF2 is also known as MISME syndrome. MISME is an acronym. What does it stand for?

--**M**

--**I**

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--**M**

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What is an ependymoma?

is an acronym.

--**Ependymomas**

Phakomatoses



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What is an ependymoma?

A glioma consisting of ependymal cells

is an acronym.

--Ependymomas

Phakomatoses



NF2

--**Central** NF

--Classic finding: bilateral **acoustic neuromas**

--Eye findings: **Common:** PSC/cortical cataracts;

Rare: Combined hamartoma of retina and RPE; **Rarer:** Lisch nodules

Do NF2 pts get peripheral-nerve tumors like NF1 pts?

Yes, but at much lower rates

OK then, other than acoustic neuromas, what sorts of neuroglial lesions occur in NF2?

CNS neuroglial lesions; eg, spinal-cord schwannomas, intracranial meningiomas, and ependymomas

Neuroglial lesions

--Nodular neurofibromas

--Plexiform neurofibromas

--Optic glioma

--Prominent corneal nerves

--**Schwannomas** of the SC

--**Meningiomas** (intracranial)

--**Ependymomas**

What is an ependymoma?

A glioma consisting of ependymal cells

OK smart guy, what are ependymal cells?

is an acronym.

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Phakomatoses



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OK smart guy, what are ependymal cells?

The epithelial-like glial cells that form the inner lining of the cerebral ventricles and the central canal of the spinal cord

--**Ependymomas**

is an acronym.

Phakomatoses



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Do the neuroglial lesions in NF2 carry a risk of malignant transformation?

The epithelial-like glial cells that form the inner lining of the cerebral ventricles and the central canal of the spinal cord

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Phakomatoses



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Do the neuroglial lesions in NF2 carry a risk of malignant transformation?

No. Unlike in NF1, malignant transformation of benign lesions in NF2 is almost unheard of.

The epithelial-like glial cells that form the inner lining of the cerebral ventricles and the central canal of the spinal cord

--Ependymomas

Phakomatoses

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By way of a refresher: What is a hamartoma?



Phakomatoses



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By way of a refresher: What is a hamartoma?

A tumor composed of histologically clinical state cells found in their opposite clinical state location

Phakomatoses



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By way of a refresher: What is a hamartoma?

A tumor composed of histologically abnormal cells found in their normal location



Phakomatoses

NF2

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Again, a refresher: What is the name of the reverse clinical entity, ie, one with normal cells found in an abnormal location?

Lisch nodules

By way of a refresher: What is a [?]~~hamartoma~~
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Phakomatoses

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Again, a refresher: What is the name of the reverse clinical entity, ie, one with normal cells found in an abnormal location?

A **choristoma**

Lisch nodules

By way of a refresher: What is a ^{choristoma}~~hamartoma~~?
 A tumor composed of histologically ~~normal~~^{normal} cells found in ~~an abnormal~~^{an abnormal} location

Phakomatoses



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So, what combination of hamartomatous cells of the retina and RPE are involved in a combined hamartoma of the retina and RPE?



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RPE cells (duh) and retinal glial cells



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As a variably pigmented, slightly elevated retinal mass of the location retina



Phakomatoses

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Phakomatoses



Combined hamartoma of retina and RPE

Phakomatoses



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With what more sinister dz entity is it often confused?



Phakomatoses

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With what more sinister dz entity is it often confused?

Choroidal melanoma (eyes have been enucleated because of this misdiagnosis)



Phakomatoses

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With what more sinister dz can this be confused? How can one avoid making such a disastrous mistake?

Choroidal melanoma



Phakomatoses

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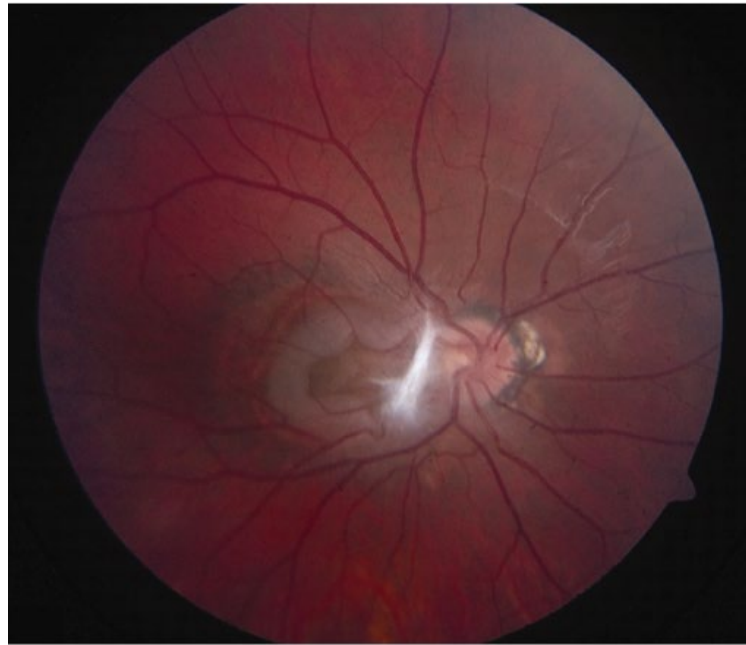
With what more sinister dz

Choroidal melanoma

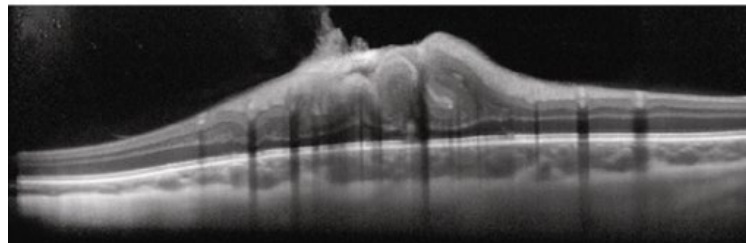
How can one avoid making such a disastrous mistake?

By taking pains to carefully determine the anatomic location of the tumor in question. Choroidal melanomas originate behind Bruch's membrane, whereas combined hamartomas of the retina and RPE are located wholly in front of it.

Phakomatoses



(a)



(b)

Combined hamartoma of retina and RPE. Note the entire lesion is above Bruchs

Phakomatoses



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Acoustic neuroma

Another eye finding associated with acoustic neuroma is corneal decompensation. By what two mechanisms might this occur?

Corneal decompensation

Phakomatoses



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Acoustic neuroma

Bag CN V_1

Another eye finding associated with acoustic neuroma is corneal decompensation. By what two mechanisms might this occur?

(No question yet—keep going)

Corneal decompensation



Phakomatoses

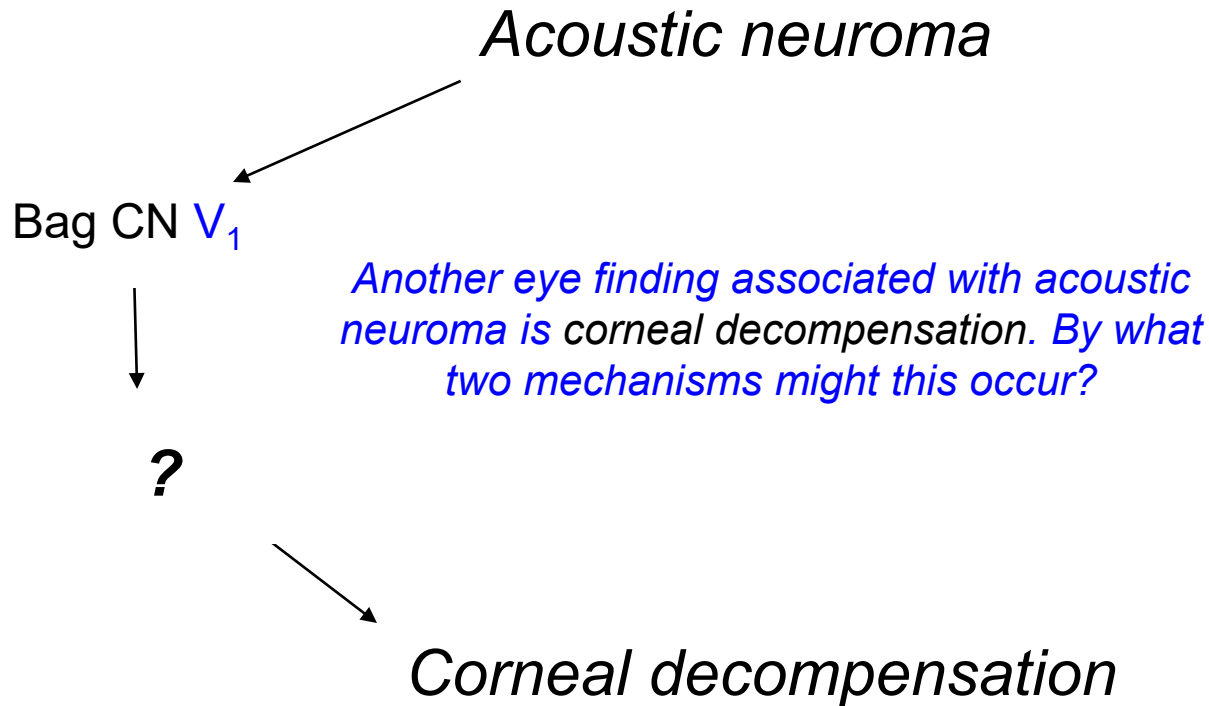
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Phakomatoses



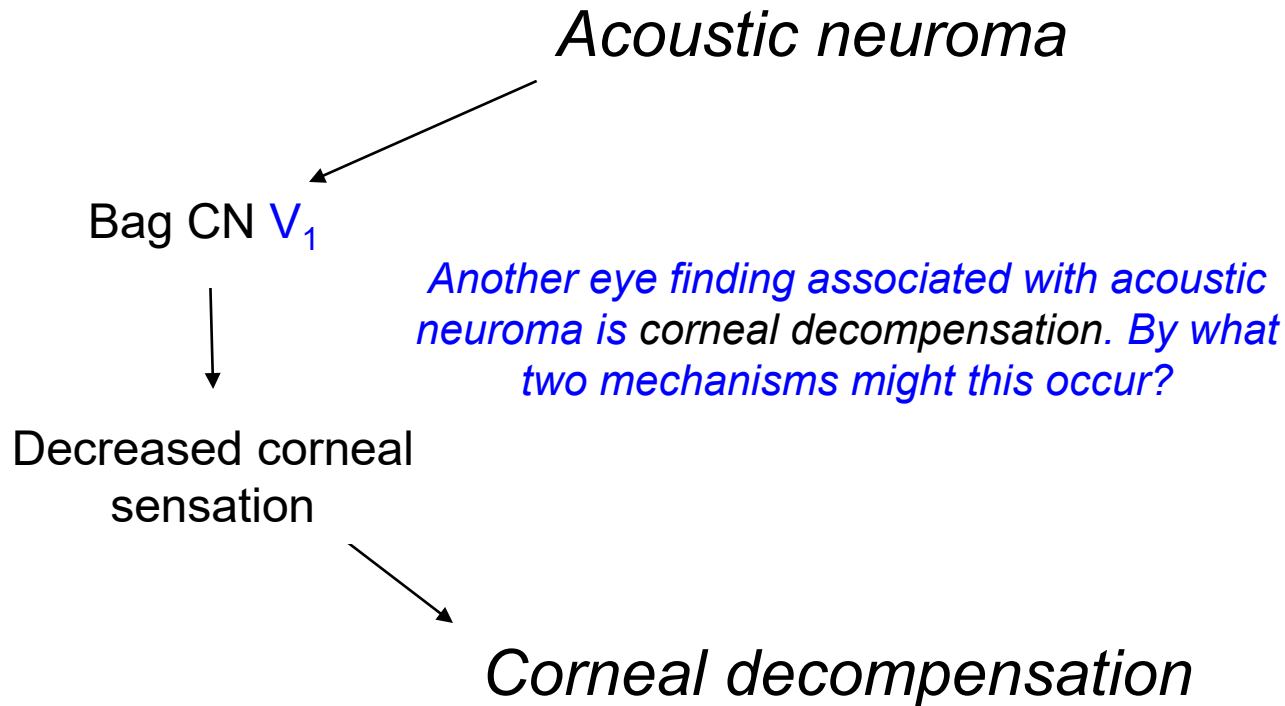
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Phakomatoses



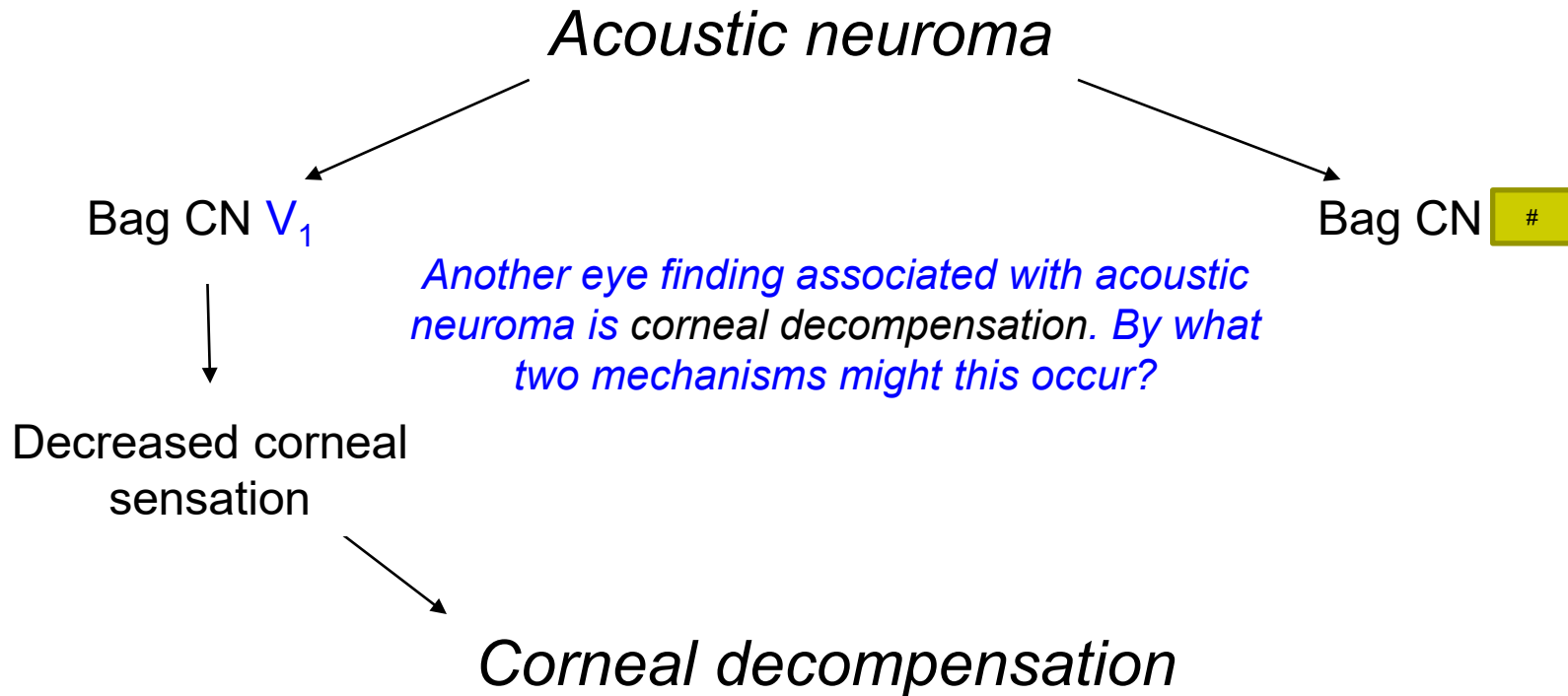
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Phakomatoses



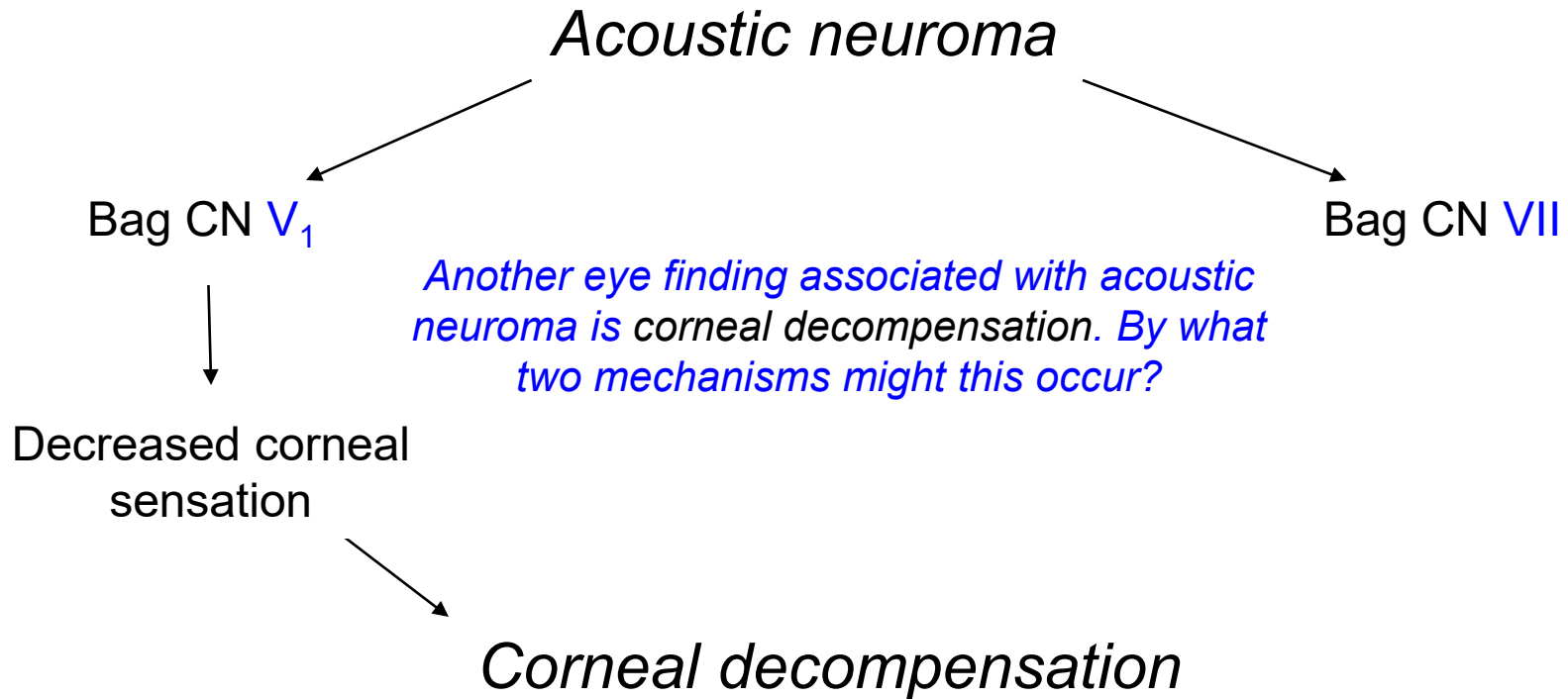
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Phakomatoses



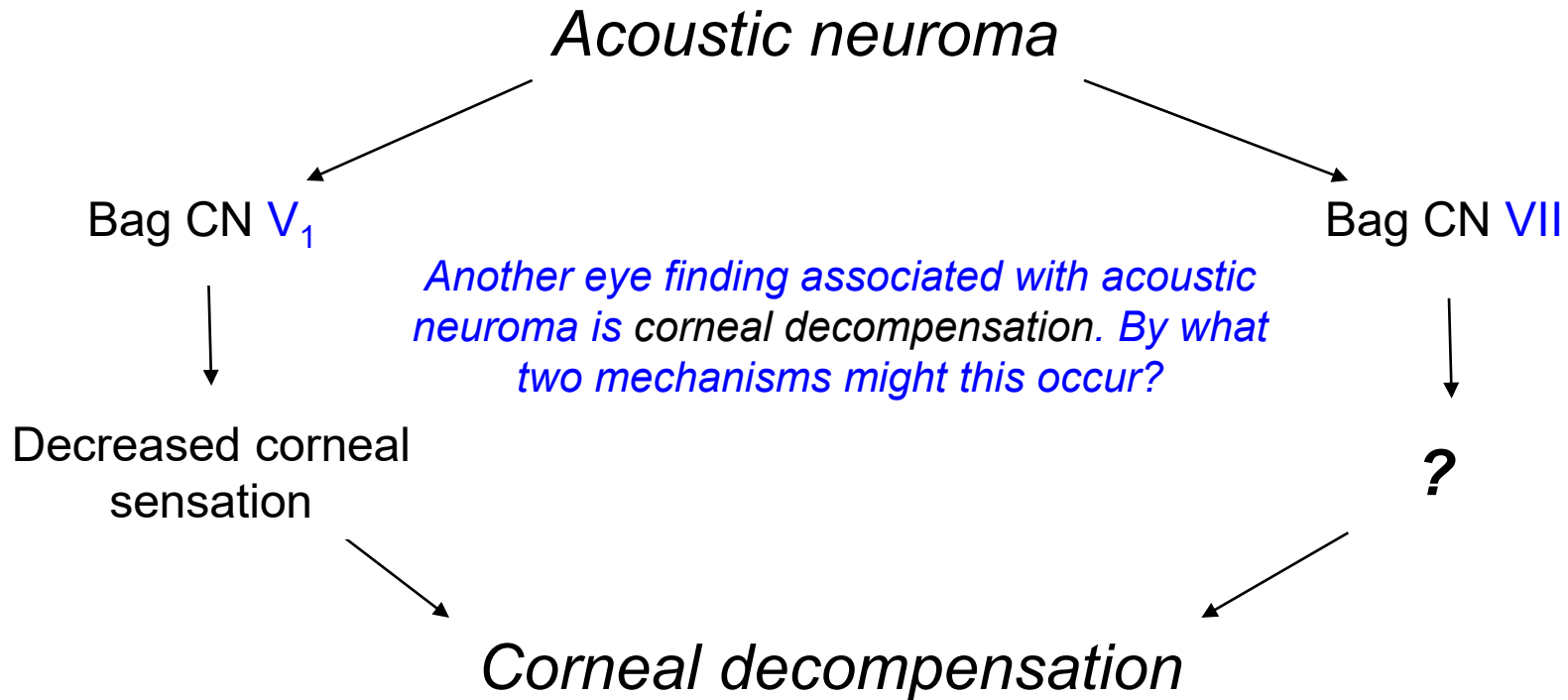
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Phakomatoses



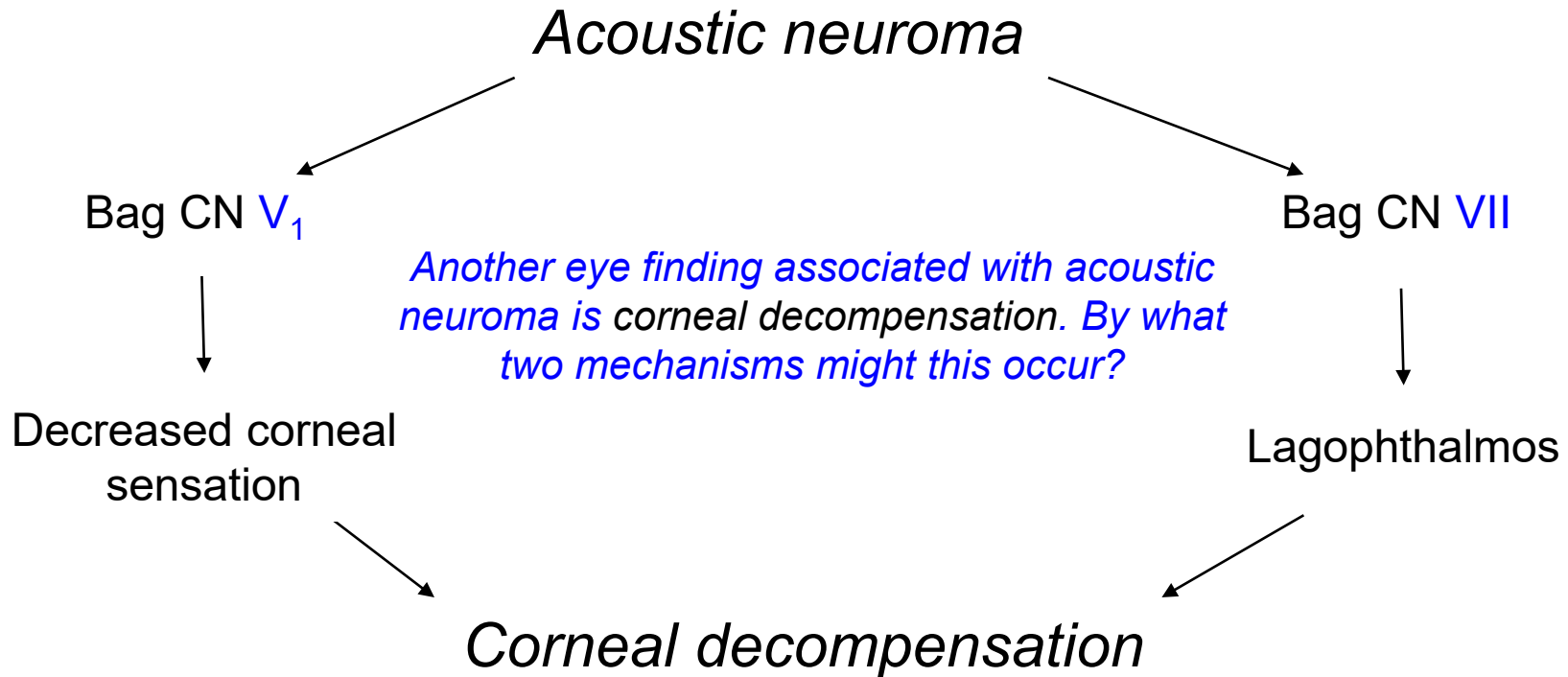
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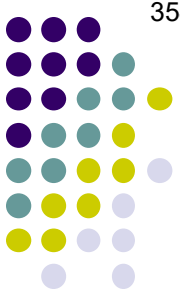
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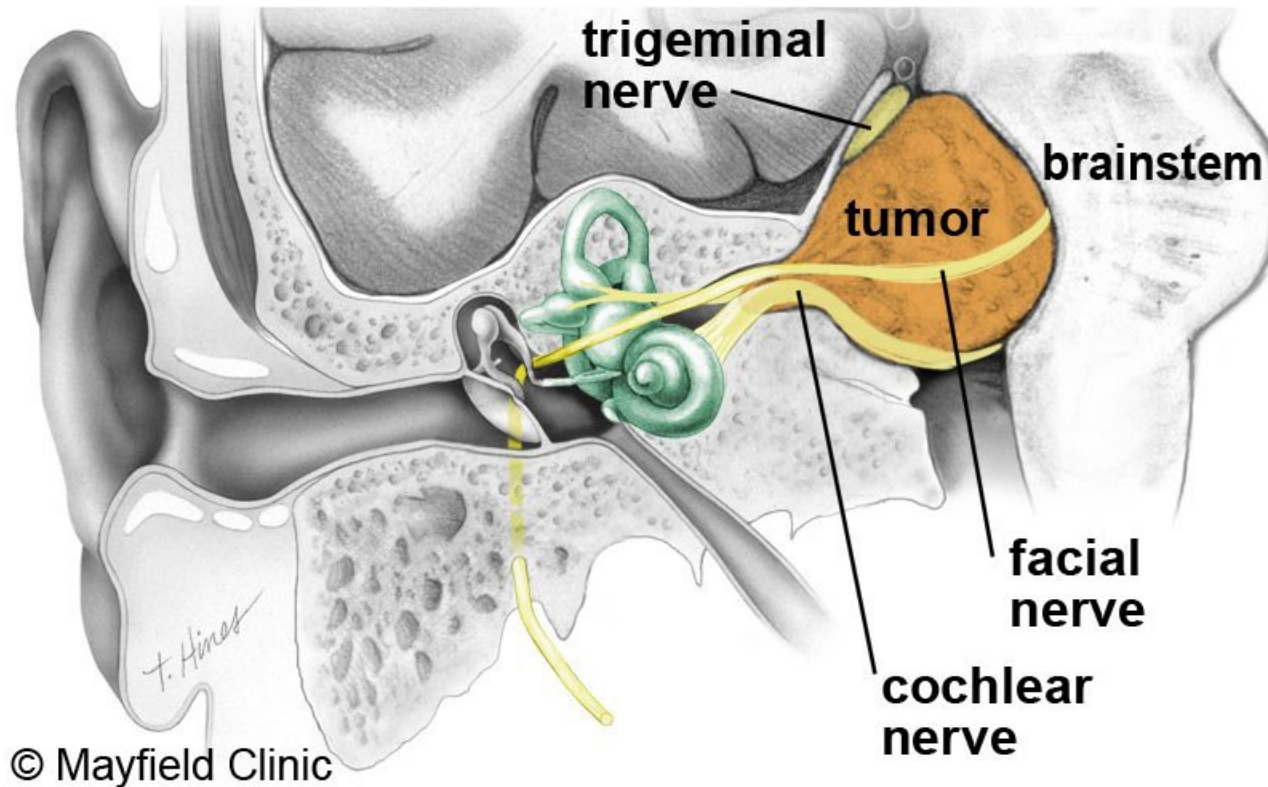
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Phakomatoses



B. acoustic neuroma



NF2: Acoustic neuroma. Note its close association with both CN5 and CN7.

Phakomatoses



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Sturge-Weber

--Classic stigmata is the

three words

Phakomatoses



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Sturge-Weber

--Classic stigmata is the **port-wine stain**

Phakomatoses



Sturge-Weber: Port-wine stain

Phakomatoses

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In one word, what sort of lesion is the port-wine stain?

Phakomatoses

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Sturge-Weber

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By what 'official' name is it known?

Phakomatoses

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Nevus flammeus

Phakomatoses

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Phakomatoses

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When does it present?

At birth

Phakomatoses



Sturge-Weber: Port-wine stain

Phakomatoses

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What is the typical pattern of distribution?

Phakomatoses

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What is the typical pattern of distribution?
It comports to the distribution of one or more divisions of

CN#?

Phakomatoses

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When does it present?
At birth

What is the typical pattern of distribution?
It comports to the distribution of one or more divisions of CN5

Phakomatoses

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Does it always present in this manner?

Phakomatoses

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No. Some cases will cross the midline of the face

Phakomatoses

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All infants with SWS have a port-wine stain. Do all infants with a port-wine stain have SWS?

Phakomatoses

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Phakomatoses

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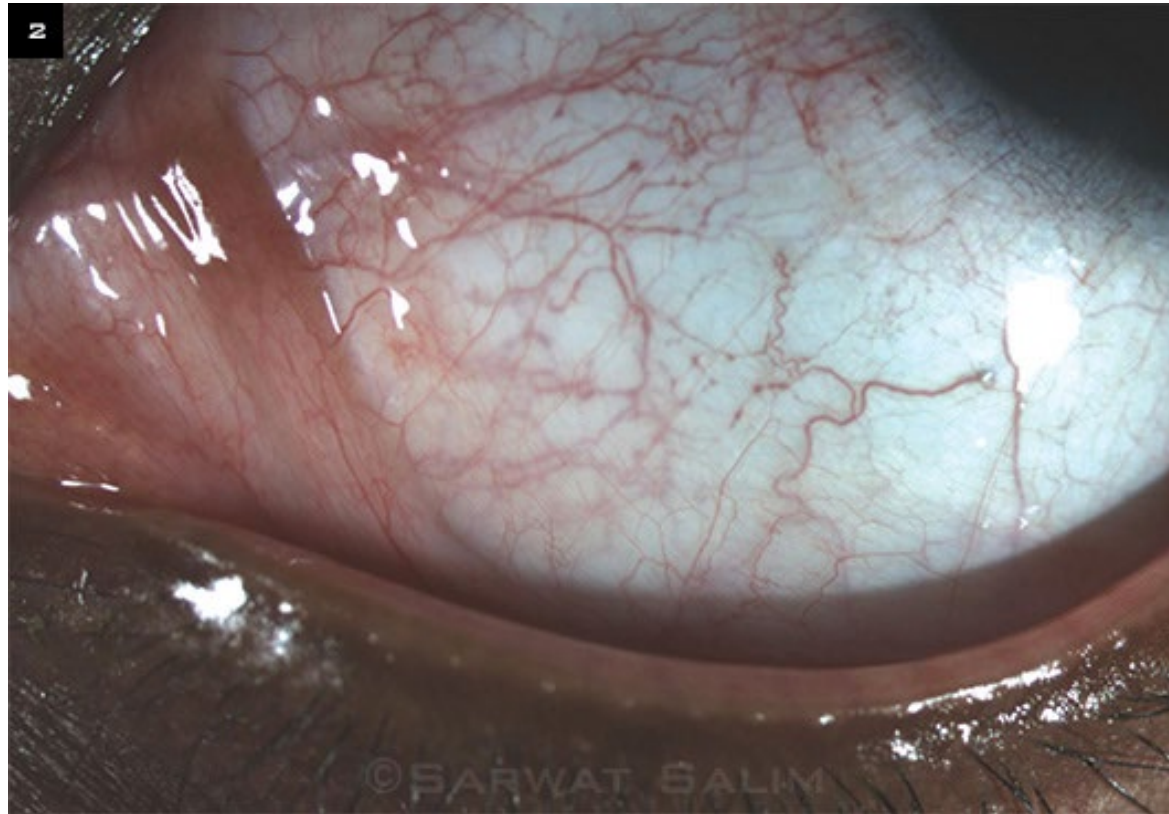
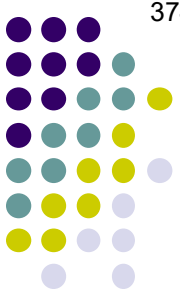
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If the port-wine stain involves the eyelid, what adjacent structure is commonly affected as well?
The conjunctiva. It will have increased vascularity and hyperemia, producing a false impression of 'pink eye.'

Phakomatoses



Sturge-Weber: Conjunctival hyperemia

Phakomatoses



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Sturge-Weber

--Classic stigmata is the **port-wine stain**

--Ipsilateral meningeal AVM → symptom/sign

Phakomatoses



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Phakomatoses



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Is the meningeal AVM prone to bleeding?



Phakomatoses

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Phakomatoses

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Sturge-Weber

--Classic stigmata is the **port-wine stain**

--Ipsilateral meningeal AVM → **seizures**

How prevalent is seizure activity in SWS?



Phakomatoses

NF2

--*Central* NF

--Classic finding: bilateral **acoustic neuromas**

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Very—estimates run as high as **90%** of cases



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lesion (something something something)



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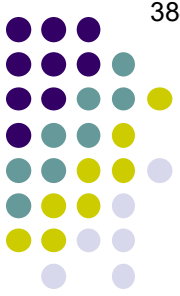
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Phakomatoses



Sturge-Weber: Tomato catsup fundus OD



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Diffuse choroidal hemangioma is present in what percent of SWS?



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Diffuse choroidal hemangioma is present in what percent of SWS?
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Yes, but it's uncommon



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--Another classic finding on DFE:

non-retinal pathology



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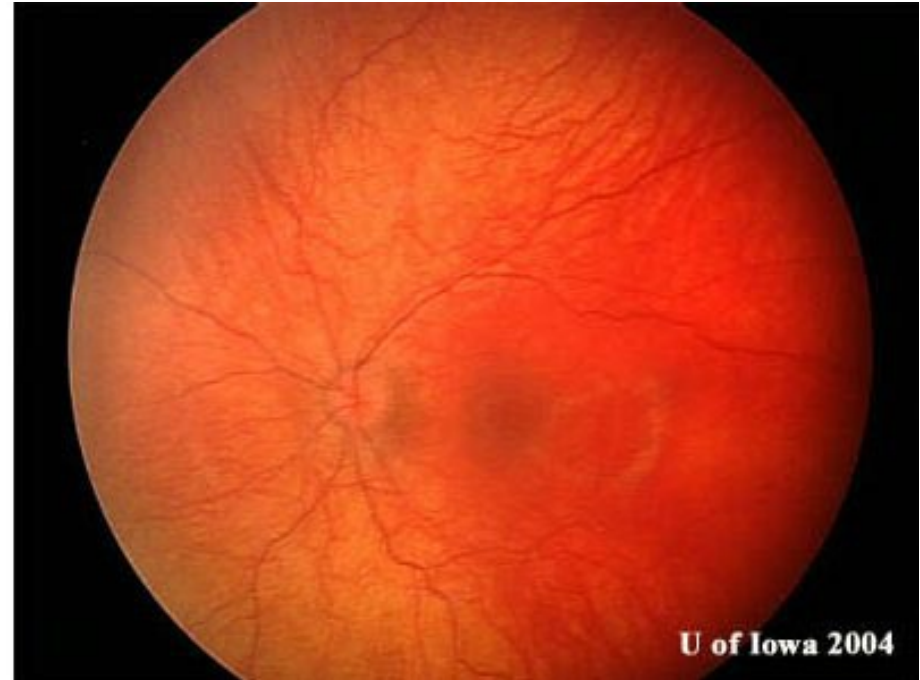
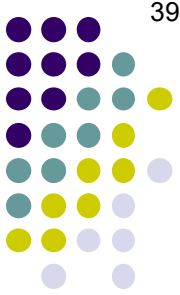
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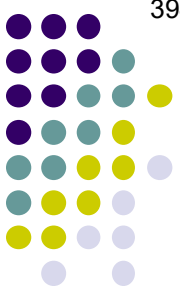
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Phakomatoses



Sturge-Weber: Note the glaucomatous cupping on the affected side

Phakomatoses



Sturge-Weber: Note the subtle PWS; also the buphthalmos and increased corneal diameter typical of congenital glaucoma



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--?

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Hints upcoming...



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← A noncirculatory anomaly



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Estimates range as high as 70%

Rule of thumb regarding the mechanism of glaucoma and SWS:

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--If glaucoma doesn't manifest until **after age 10**, increased EVP is the cause

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Phakomatoses

$$IOP = \frac{\text{Aqueous Formation Rate } (\mu\text{L/min})}{\text{Outflow Facility } (\mu\text{L/min/mmHg})} + \text{Episcleral Venous Pressure (mmHg)}$$

Recalling the Goldmann equation for IOP...

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$$\begin{array}{c} \uparrow IOP \\ \text{in SWS} \end{array} \begin{array}{c} \text{is} \\ \text{secondary} \\ \text{to} \end{array} \frac{\text{Aqueous hypersecretion}}{\text{Abnormal drainage angle}} + \uparrow \text{Episcleral Venous Pressure}$$

...we can see how all three components are involved in SWS glaucoma!

What percent of SWS pts develop glaucoma?

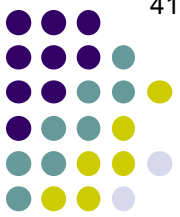
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--Glaucoma surgery: ↑ risk of massive bad surgical complication due to abnormal two words

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~~Sturge-Weber~~

syndrome

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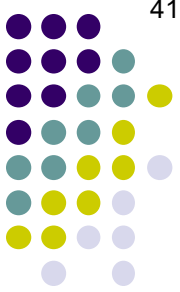
~~Sturge-Weber~~ Klippel-Trénaunay(-Weber) syndrome

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Phakomatoses



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How is Klippel-Trénaunay pronounced?



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How is Klippel-Trénaunay pronounced?
CLIP-ell try-NO-nay



Phakomatoses

NF2

--Central NF

--Classic finding: bilateral acoustic neuromas

--Eye findings: *Common*: PSC/cortical cataracts;

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In the vast majority (~90%) of cases,

arm vs
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Phakomatoses



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Which limb is involved?

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Is the limb hypertrophy present at birth?

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In the vast majority (~90%) of cases, a leg

Is the limb hypertrophy present at birth?

In most cases, yes

Are there other associations of note?

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Ataxia-telangiectasia

--Most common cause of main symptom (not ocular) in childhood



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--Classic finding of conjunctival telangiectasia typically appear between ages of # to # years



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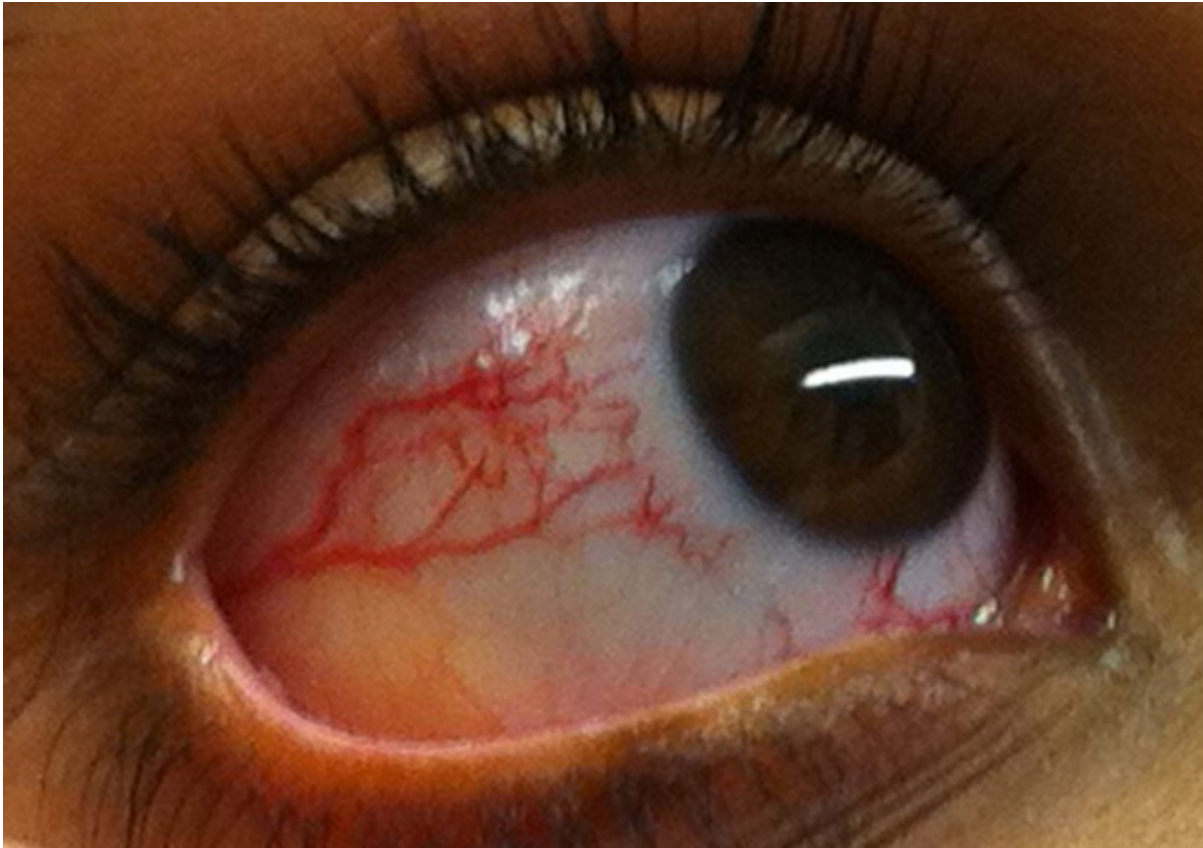
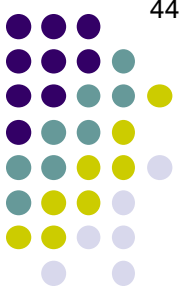
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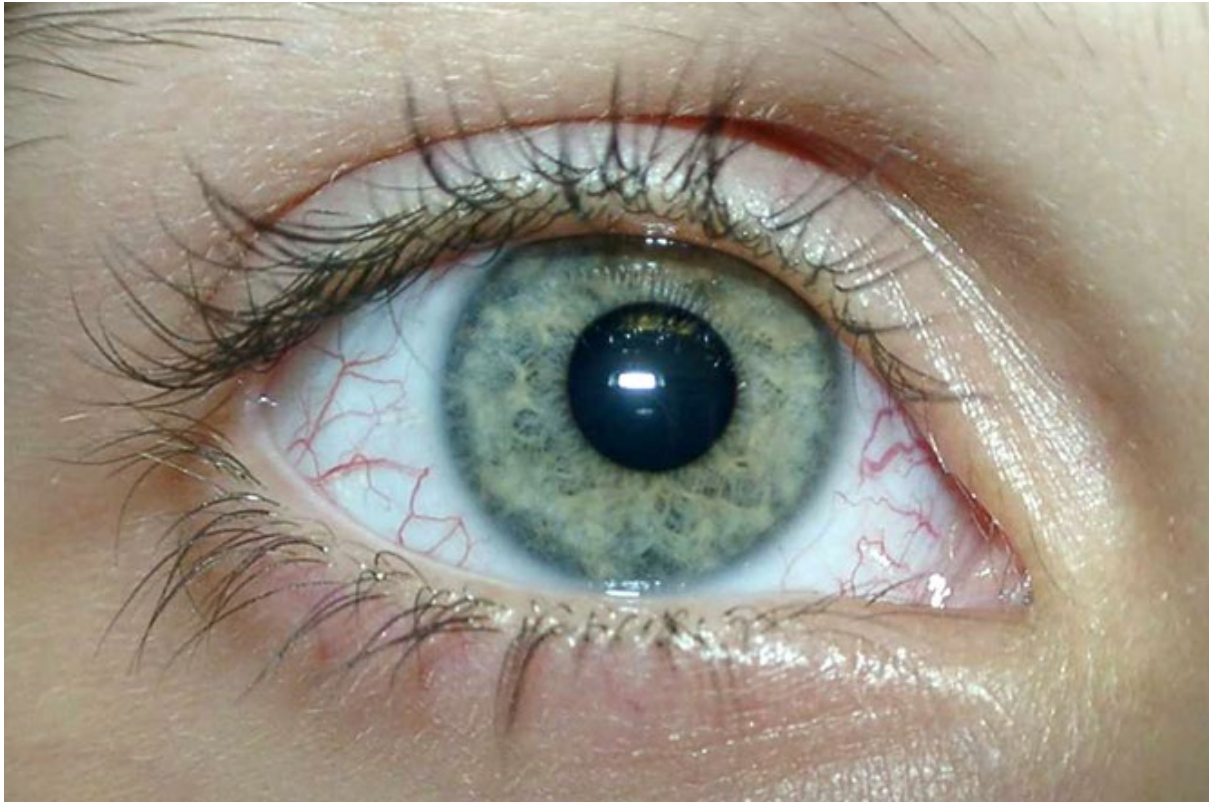
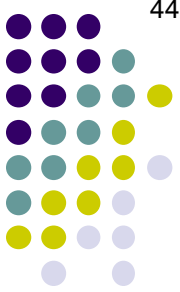
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Phakomatoses



Ataxia-telangiectasia: Conj telangiectasias

Phakomatoses



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*A-T toddlers have difficulty initiating saccades, and sometimes use a head turn/thrust to do so.
What more-common, less-devastating oculomotor disorder presents similarly?*



Phakomatoses

NF2

- Central* NF
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Rare: **Combined hamartoma of retina and RPE**; *Rarer*: **Lisch nodules**

Sturge-Weber

- Classic stigmata is the **port-wine stain**
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Congenital ocular motor apraxia (COMA). For more on COMA, see slide-set P4



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What aspects of the immune system are abnormal?

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These immunodeficiencies are due in large part to hypoplasia of what immune organ?



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What buzzword is used to define the specific sort of RT infection A-T pts are vulnerable to?

...with intact doll's eyes; strabismus; nystagmus
...to **respiratory tract** infections → risk of death in teens



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'Sinopulmonary' infections

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--Also have significantly increased risk of cancer 1 and cancer 2



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Ataxia-telangiectasia

- Most common cause of progressive ataxia in childhood
- Only phakomatoses with **immunodeficiency**
- Classic finding: **telangiectases** (dilated capillaries) → **bleeding** (esp. conjunctivae, skin, GI)
Your A-T pt may have a sinus infection. Should you get a CT to confirm?
- Other eye findings include **abnormal accommodation** with intact corneal eyes, **strabismus**, **nystagmus**
- Abnormal immune function → ↑ susceptibility to **respiratory tract** infections → **risk of death in teens**
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- Most common cause of progressive ataxia in childhood
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- Classic finding: **NO! A-T pt's DNA is extremely vulnerable to damage from ionizing radiation—X-rays should be performed only if no other imaging modality will suffice**
- Other eye findings include abnormal oculocutaneous telangiectases with intact corneal eyes, strabismus, nystagmus
- Abnormal immune function → ↑ susceptibility to respiratory tract infections → risk of death in teens
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The unfortunate truth of the matter is this:

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--In countries with less-robust healthcare systems (ie, without readily-available antibiotics), A-T pts die of sinopulmonary infections in their teens; whereas

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- in countries **with** robust healthcare systems, sinopulmonary infections can be kept at bay long enough for A-T pts to die of cancer (usually leukemia or lymphoma).
- Abnormal immune function → ↑ susceptibility to respiratory tract infections → risk of death in teens
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--Heterozygotes (% of population) have increased risk of as well

non-ocular prob



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For what cancer are A-T heterozygotes at particular risk?

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- Skin manifestation: **Telangiectasias**



Phakomatoses

NF2

- Central NF
- Classic finding: bilateral acoustic neuromas
- Eye findings: *Common*: PSC/cortical cataracts;
Rare: Combined hamartoma of retina and RPE; *Rarer*: Lisch nodules

Sturge-Weber

- Classic stigmata is the **port-wine stain**
- Ipsilateral meningeal A **At what age do cutaneous telangiectasias begin to appear?**
- Classic *tomato catsup* 3-5 years (ie, at about the same time the conj ones do)
- Another classic finding
- Glaucoma surgery: ↑ risk of retinal vasculature

Ataxia-telangiectasia

- Most common cause of ataxia
- Only phakomatosis with ataxia
- Classic finding of conjunctival telangiectasia typically **appear between ages of 3-5 years**
- Other eye findings include abnormal saccades with intact doll's eyes; strabismus; nystagmus
- Abnormal immune function → ↑ susceptibility to respiratory tract infections → risk of death in teens
- Also have significantly increased risk of leukemia and lymphoma
- Heterozygotes (~2 % of population) have increased risk of malignancy as well
- Skin manifestation: **Telangiectasias**

Phakomatoses



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3-5 years (ie, at about the same time the conj ones do)

At what location do they typically appear first?

ma

y

l vasculature

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Phakomatoses



Ataxia-telangiectasia: Facial telangiectasias



Phakomatoses

NF2

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The malar region of the face

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y

l vasculature

Ataxia-telangiectasia

Do they remain localized to the malar region throughout life?

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Phakomatoses

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At what location do they typically appear first?

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ma

y

l vasculature

Ataxia-telangiectasia

Do they remain localized to the malar region throughout life?

No, they typically spread across the face and neck, and new

'crops' will appear on the limbs

--Most common cause c

--Only phakomatosis wi

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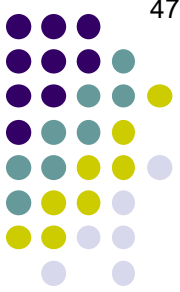
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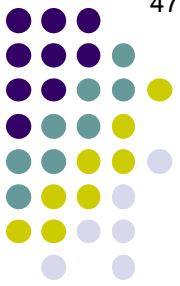
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Phakomatoses



Ataxia-telangiectasia: Telangiectasias

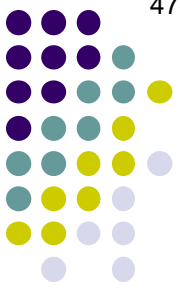
Phakomatoses



Incontinentia pigmenti

--Skin normal at birth, but abnormality 1 and abn 2 develop by age ; only later develops the classic description appearance

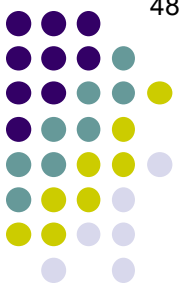
Phakomatoses



Incontinentia pigmenti

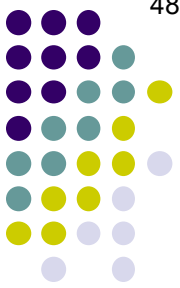
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Phakomatoses



Incontinentia pigmenti: Splashed-paint appearance

Phakomatoses

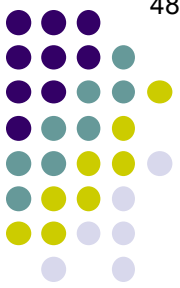


Incontinentia pigmenti

--Skin normal at birth, but **erythema** and **bullae** develop by **1 week** ; only later develops the classic 'splashed paint' appearance

--Eye finding: x/x will have peripheral retina problem that looks just like a more common dz

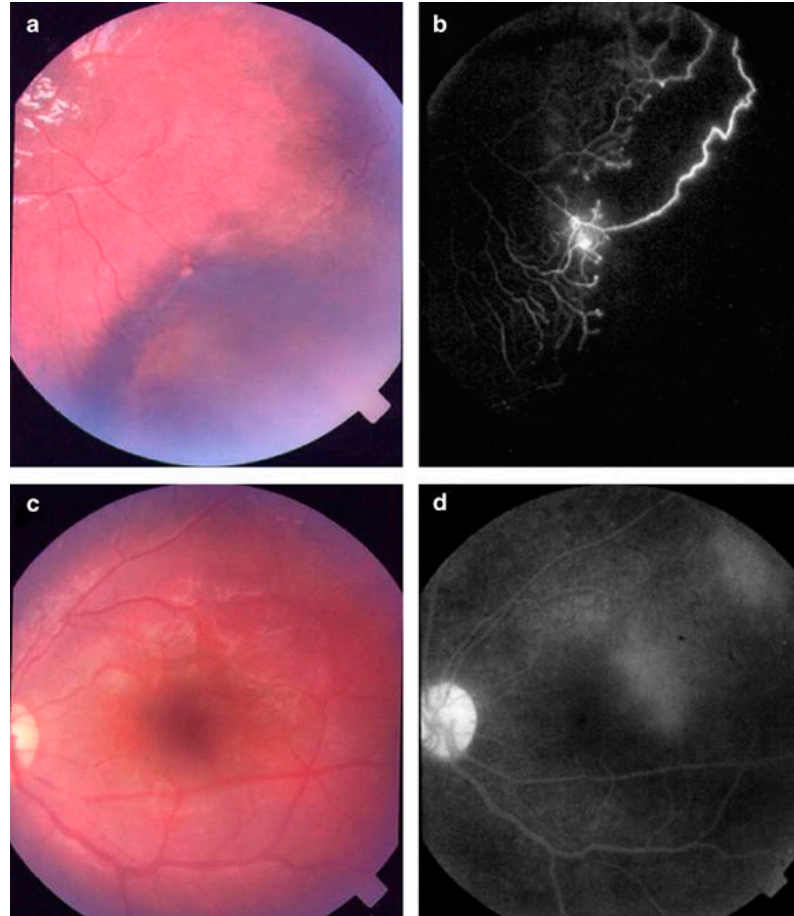
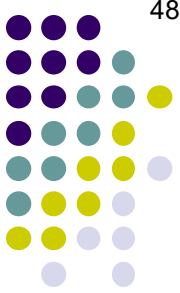
Phakomatoses



Incontinentia pigmenti

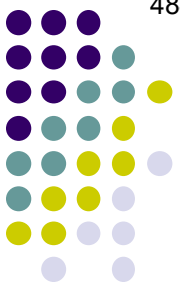
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-

Phakomatoses



Incontinentia pigmenti: ROP-like retinal appearance

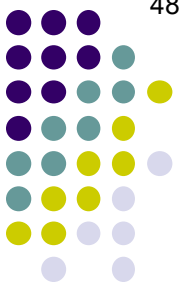
Phakomatoses



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-

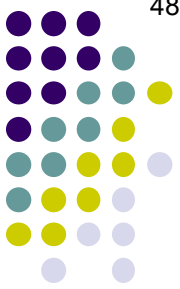
Phakomatoses



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Phakomatoses

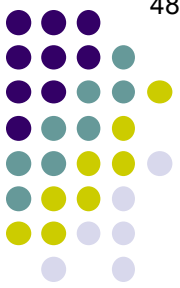


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- Skin normal at birth, but erythema and bullae develop by 1 week ; only later develops the classic 'splashed paint' appearance
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How is the peripheral proliferative retinopathy managed?

Phakomatoses



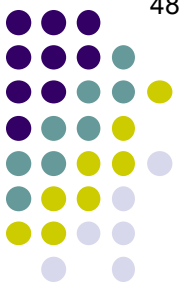
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How is the peripheral proliferative retinopathy managed?

Basically, in the same manner as ROP

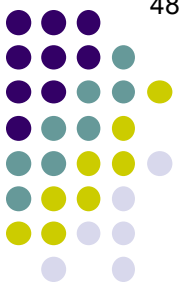
Phakomatoses



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 - Eye findings are usually **unilateral**
 - 2/3 will also have abnormal mouth issue
-

Phakomatoses



Incontinentia pigmenti

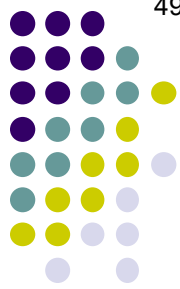
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-

Phakomatoses



Incontinentia pigmenti: Abnormal dentition

Phakomatoses



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Speaking of eye dentistry: When you learn that a pt has teephus issues, four conditions should spring immediately to mind. One is incontinentia pigmenti; what are the other three?

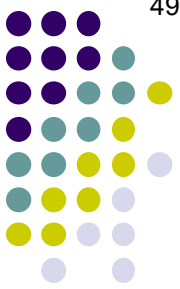
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Phakomatoses



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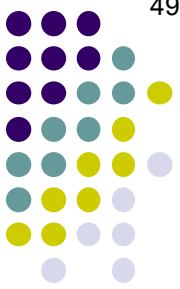
--Incontinentia pigmenti

-- eponym-eponym
(syndrome)

-- eponym syndrome

-- syphilis

Phakomatoses



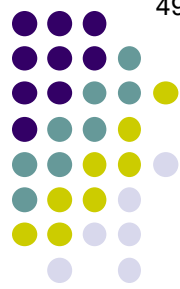
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Phakomatoses



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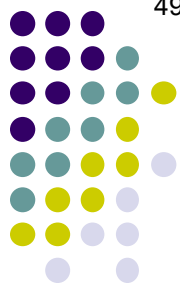
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An...

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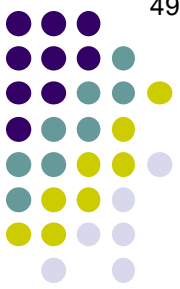
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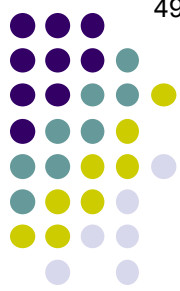
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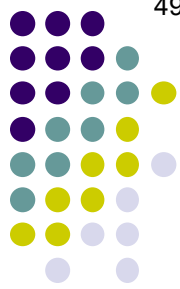
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Is Axenfeld-Reiger a peripheral, or central dysgenesis?

Phakomatoses



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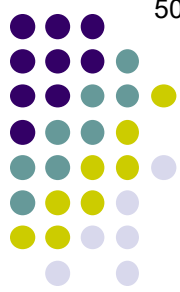
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Phakomatoses



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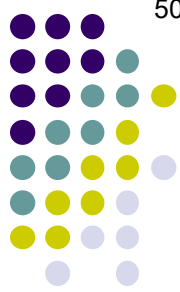
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Peripheral

There is one other major peripheral dysgenesis covered in the BCSC—what is it?

Phakomatoses

501



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Aniridia

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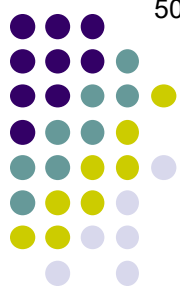
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Likewise, there are two major central dysgeneses—what are they?

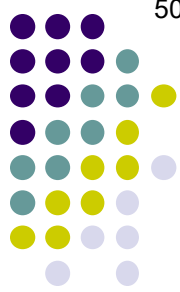
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Peters anomaly

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Peripheral dysgeneses and **central dysgeneses**

--Incontinentia pigmenti

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What features define Axenfeld-Rieger syndrome?

--?

--?

--?

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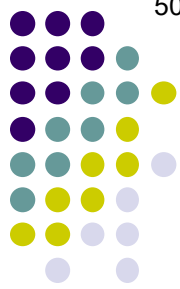
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Phakomatoses



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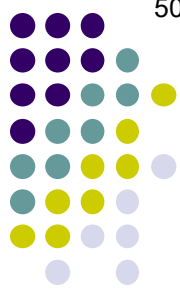
Is Axenfeld-Reiger a peripheral, or central dysgenesis?

Peripheral

There is one other major peripheral dysgenesis covered in the BCSC—what is it?

Aniridia

Phakomatoses



Incontinentia pigmenti

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In three words, what sort of condition is Axenfeld-Reiger?

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Peripheral dysgeneses and **central dysgeneses**

--Incontinentia pigmenti

--**Axenfeld-Reiger**

--Gardner syndrome

--Congenital

What features define Axenfeld-Rieger syndrome?

--Posterior embryotoxon

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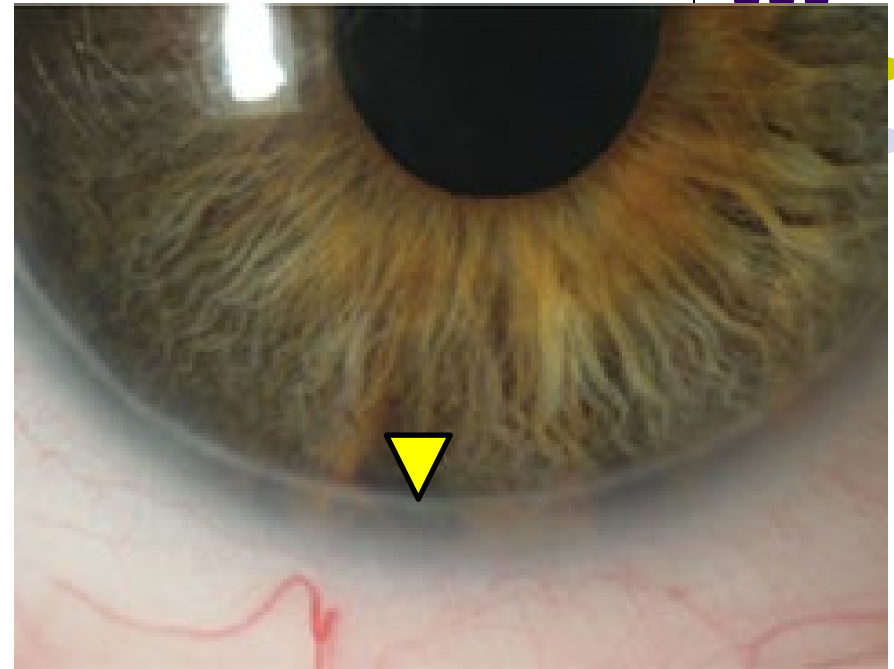
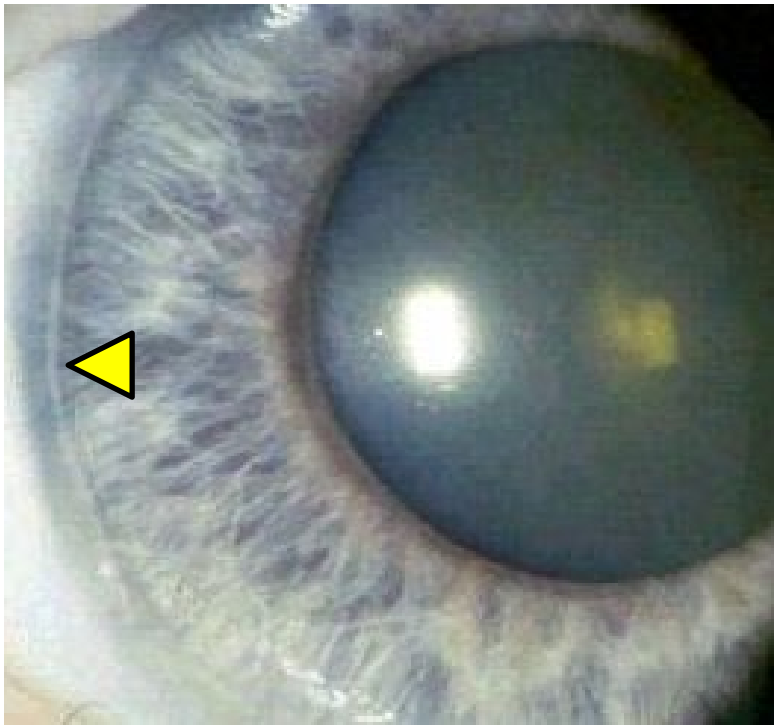
Posterior embryotoxon

What is a posterior embryotoxon?

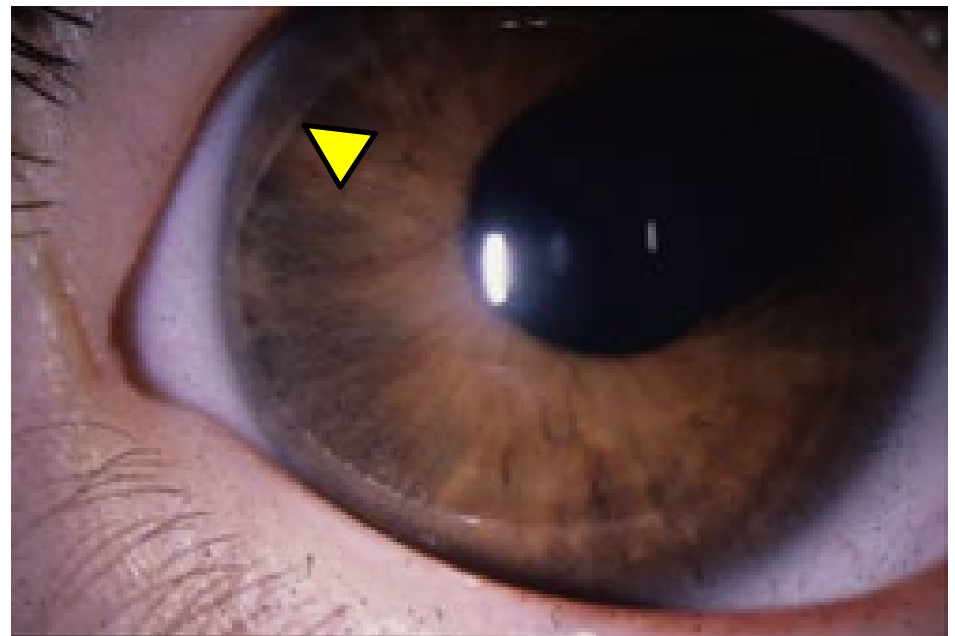
An anteriorly displaced and thickened Schwalbe's line

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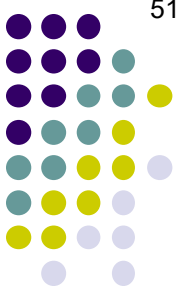
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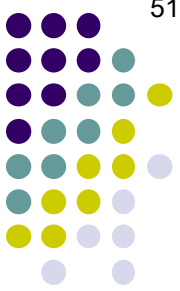
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Is it always a harbinger of significant pathology?

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two words

Next Q

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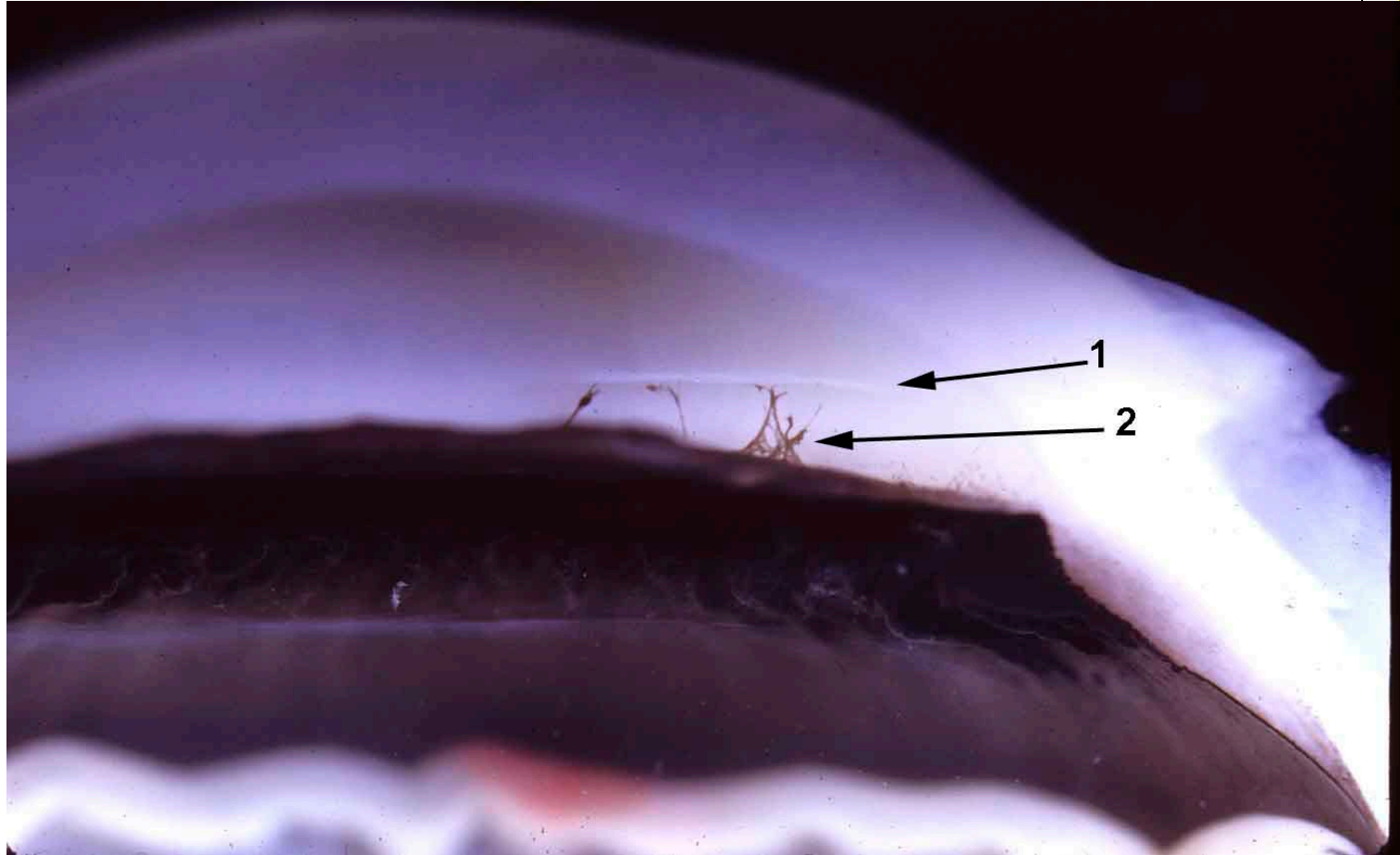
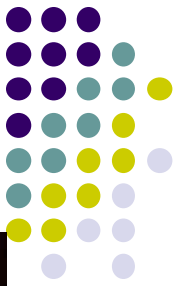
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Phakomatoses



Abnormal iris strands (2) attached to posterior embryotoxon (1) in A-R

Phakomatoses



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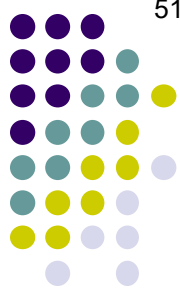
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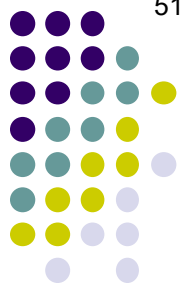
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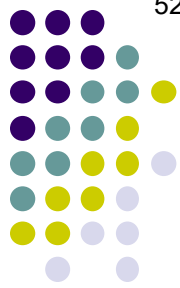
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Rare

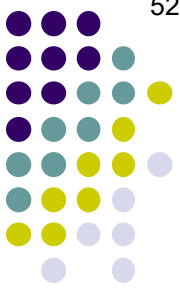
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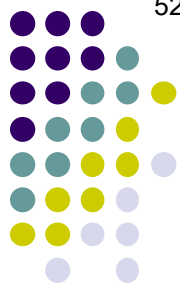
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They develop innumerable colonic polyps at a young age

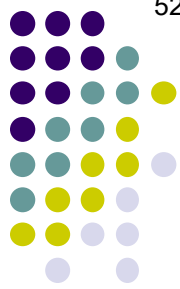
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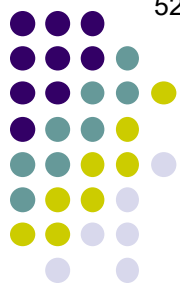
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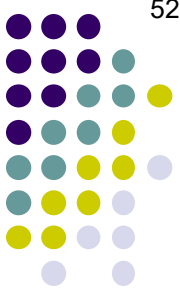
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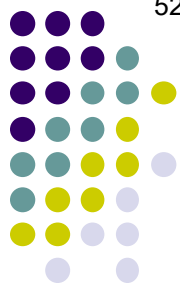
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Phakomatoses



Gardner syndrome: Colonic polyps

Phakomatoses



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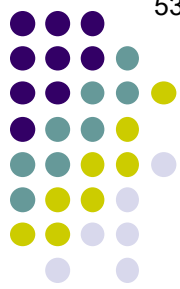
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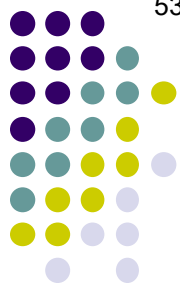
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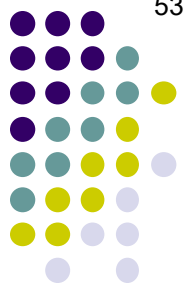
CHRPE



CHRPE-like lesions of Gardner syndrome

For more on Gardner syndrome, see slide-set P3

Phakomatoses



Incontinentia pigmenti

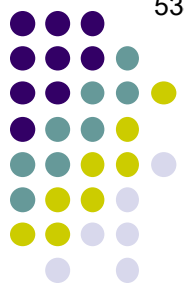
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What is the eponymous name for the abnormal dentition of congenital syphilis?

Phakomatoses



Incontinentia pigmenti

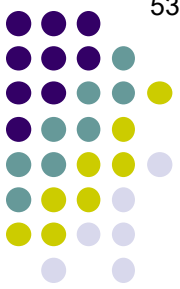
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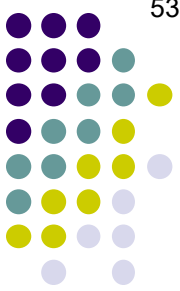
Speaking of eye dentistry: When you learn that a pt has teephus issues, four conditions should spring immediately to mind. One is incontinentia pigmenti; what are the other three?

- Incontinentia pigmenti
- Axenfled-Reiger**
- Gardner syndrome**
- Congenital syphilis**

What is the eponymous name for the abnormal dentition of congenital syphilis?
Hutchinson teeth

What description is commonly applied to the appearance of Hutchinson teeth?

Phakomatoses



Incontinentia pigmenti

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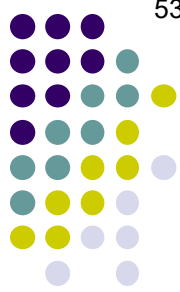
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'Peg shaped'

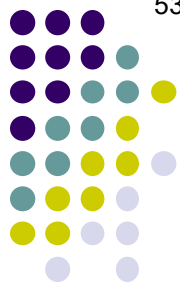
Phakomatoses



Congenital syphilis: Hutchinson teeth

For more on congenital syphilis, see slide-set U16

Phakomatoses



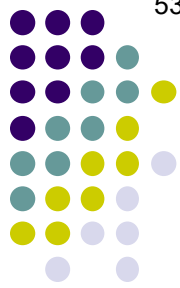
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Racemose angioma

- Characterized by AVM of **organ 1** and **organ 2**

Phakomatoses



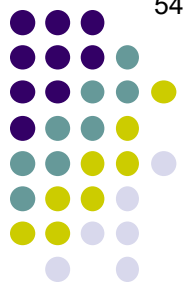
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Racemose angioma

- Characterized by AVM of **eye** and **brain**

Phakomatoses



Incontinentia pigmenti

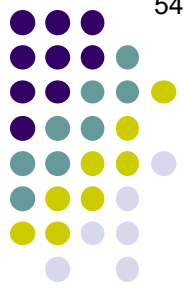
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Racemose angioma

- Characterized by **AVM** of eye and brain

In basic terms, what is an AVM?

Phakomatoses



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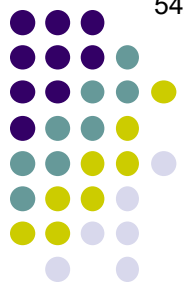
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Phakomatoses



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Racemose angioma

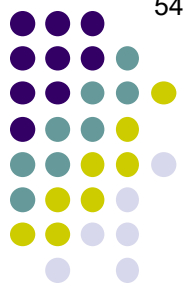
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Phakomatoses



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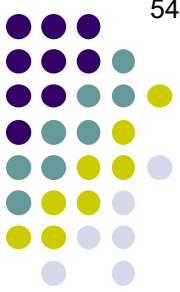
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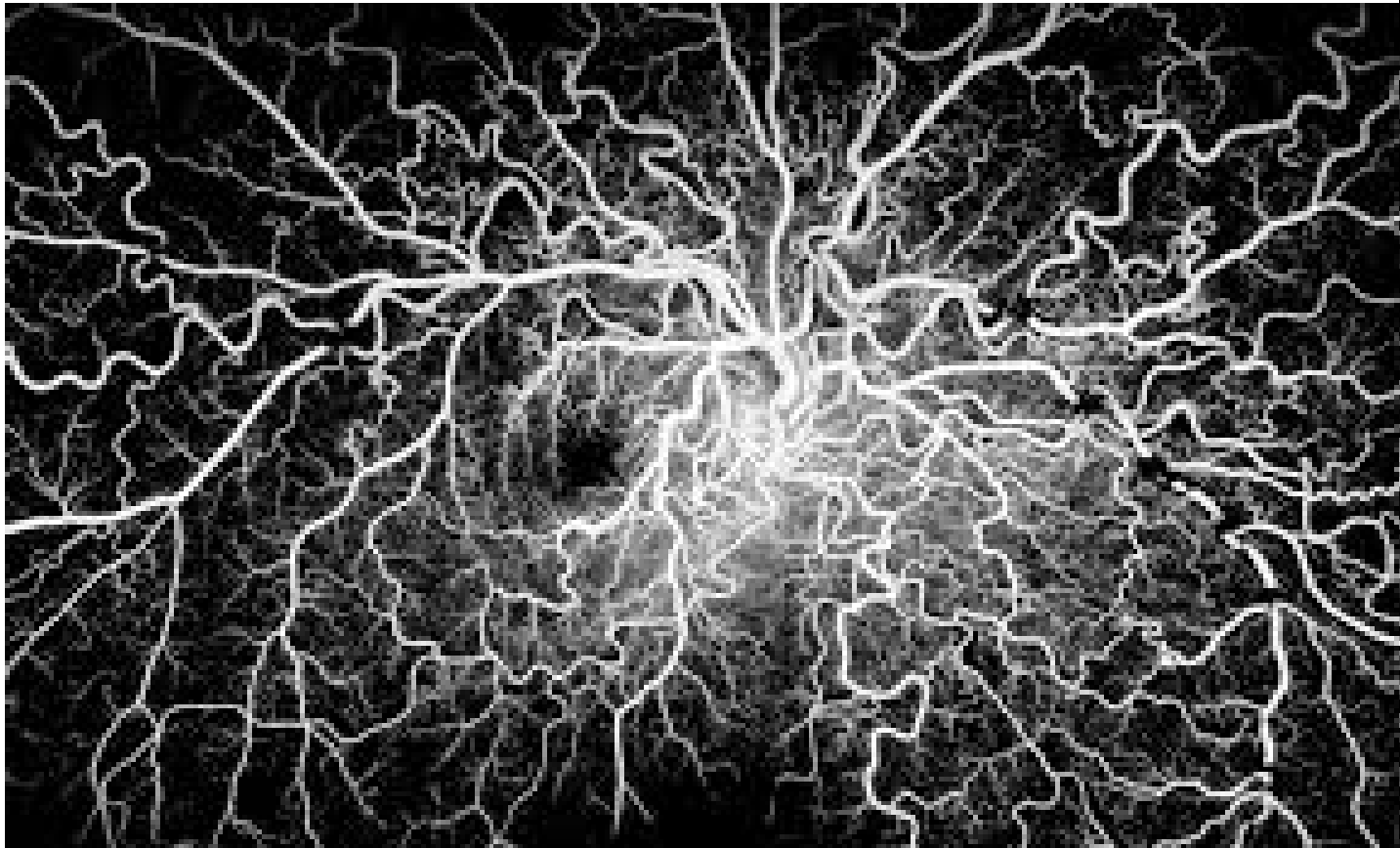
Unilateral

Phakomatoses



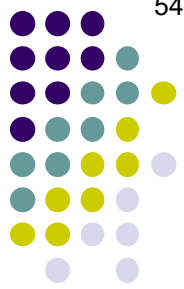
Racemose angioma

Phakomatoses



Racemose angioma

Phakomatoses



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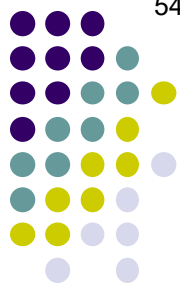
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Unilateral

How about the AVM of the brain?

Phakomatoses



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How about the AVM of the brain?

Also unilateral

Phakomatoses



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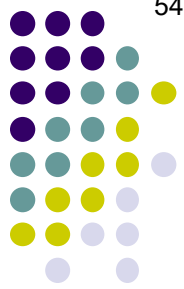
Unilateral

How about the AVM of the brain?

Also unilateral

Are the eye and brain AVM ipsilateral or contralateral with respect to one another?

Phakomatoses



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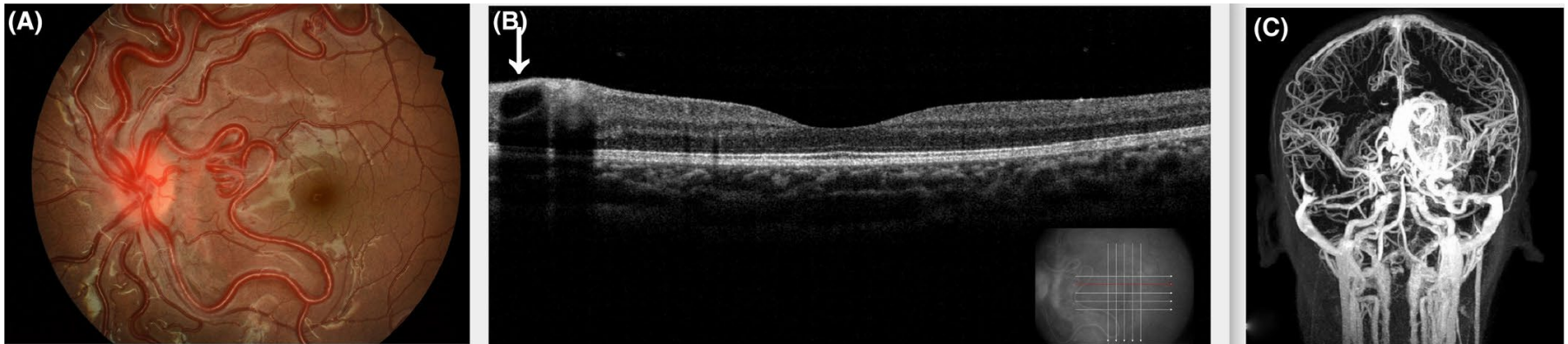
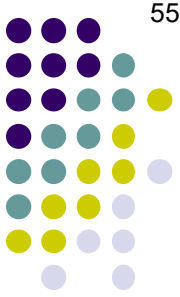
How about the AVM of the brain?

Also unilateral

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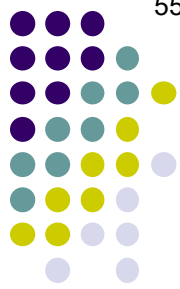
Ipsilateral

Phakomatoses



- A, FP demonstrates racemose angioma OS
- B, The vascular lumen (arrow) is visible on OCT
- C, The MRA shows the associated AVM on the left side

Phakomatoses



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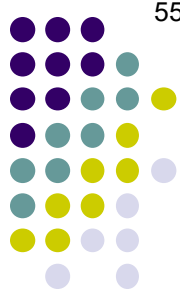
Ipsilateral

Where specifically are the AVM located in RA?

--The eye AVM are usually in the...?

--The brain AVM are usually in the...

Phakomatoses



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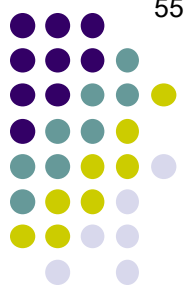
--The eye AVM are usually in the..

temporal?
nasal?
superior?
inferior?

retina

--The brain AVM are usually...

Phakomatoses



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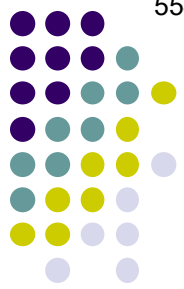
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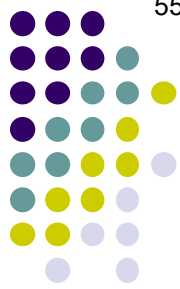
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- The brain AVM are usually...in the

cortex?
brainstem?
forebrain?
midbrain?

Phakomatoses



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Also unilateral

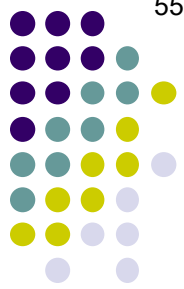
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Phakomatoses



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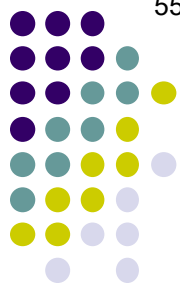
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Racemose angioma

- Characterized by **AVM** of eye and brain

Is there some sort of fundamental relationship between the AVM of the eye and brain in RA, or is their co-existence simply a matter of happenstance?

Phakomatoses



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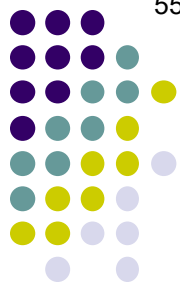
Racemose angioma

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Is there some sort of fundamental relationship between the AVM of the eye and brain in RA, or is their co-existence simply a matter of happenstance?

There is definitely a fundamental relationship between the two. This relationship stems from an abnormality of the cerebral vascular plexus of the embryo. We know this because pathologic exam has in some cases revealed the presence of a direct connection between the AVM in the eye and the AVM in the brain!

Phakomatoses



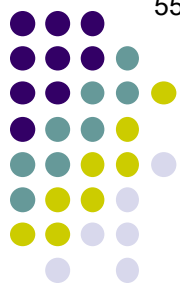
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Racemose angioma

- Characterized by AVM of **eye** and **brain**
- Brain AVM frequently bleed, leading to **bad** and **worse**

Phakomatoses



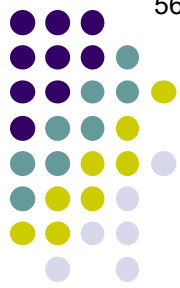
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Phakomatoses



Incontinentia pigmenti

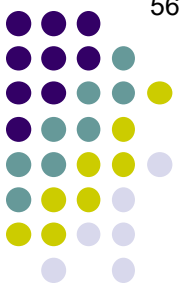
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At what age do RA pts begin to suffer these brain bleeds?

Phakomatoses



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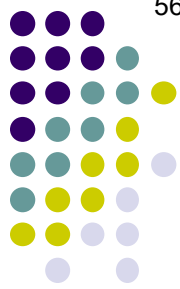
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At what age do RA pts begin to suffer these brain bleeds?

Usually at some point from the teen years into their 20s

Phakomatoses



Incontinentia pigmenti

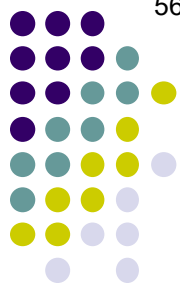
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What about seizures? How prevalent is seizure activity in RA?

Phakomatoses



Incontinentia pigmenti

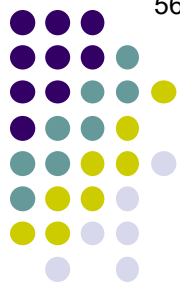
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What about seizures? How prevalent is seizure activity in RA?
Not very—estimates run as low as 5% of cases

Phakomatoses



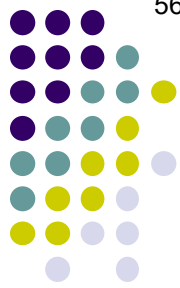
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Racemose angioma

- Characterized by AVM of **eye** and **brain**
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- Retinal AVM **do/don't** leak on FA

Phakomatoses



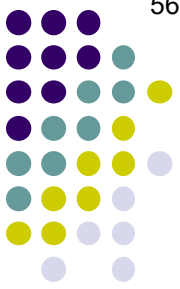
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Phakomatoses



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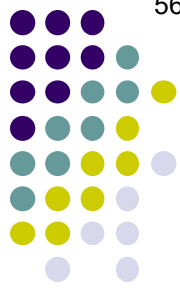
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Does this mean RA pts don't have eye/vision trouble related to their condition?

Phakomatoses



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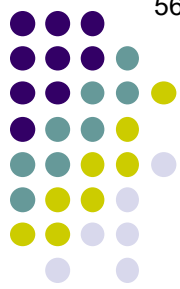
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Far from it. Like the AVM found in the brain, the AVM in the eye tend to bleed, thus predisposing these pts to retinal and/or vitreous hemorrhages. Some pts develop retinal ischemia, resulting in neovascularization and ultimately NVG.

Phakomatoses



Incontinentia pigmenti

- Skin normal at birth, but **erythema** and **bullae** develop by **1 week** ; only later develops the classic 'splashed paint' appearance
- Eye finding: **1/3** will have peripheral **proliferative retinopathy** that looks just like **ROP**
- Eye findings are usually **unilateral**
- 2/3 will also have abnormal **dentition**

Racemose angioma

- Characterized by AVM of eye and brain
- Brain AVM frequently bleed, lead to hydrocephalus
- Retinal AVM don't leak on FA

--*Skin finding = ?*

What about skin findings? If this condition is a phakomatosis (aka a neurocutaneous syndrome), shouldn't the skin be affected as well?

Phakomatoses



Incontinentia pigmenti

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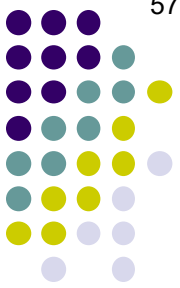
Racemose angioma

- Characterized by AVM of eye and brain
- Brain AVM frequently bleed, lead to seizures
- Retinal AVM don't leak on FA

--Skin finding = ?

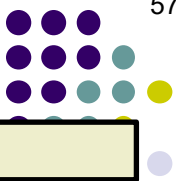
What about skin findings? If this condition is a phakomatosis (aka a neurocutaneous syndrome), shouldn't the skin be affected as well?

It should be, and in fact it is—at least 50% of RA pts manifest angiomas, vascular nevi, etc (usually on the face). However, the skin findings are not a prominent feature of the condition.



As promised, next is a TLDR. There are two versions. The first lists the characteristics of the phakomatoses and asks you to provide their names; the second does the opposite. For each version, toggle back and forth between the Q&A slides until you've got them all.

Phakomatoses aka neuro-oculocutaneous syndromes: TLDR



?	
Neuro	Optic nerve glioma
Oculo	Lisch nodules; upper-lid plexiform neurofibroma
Cutaneous	Café-au-lait spots

?	
Neuro	Bilateral acoustic neuromas
Oculo	Early PSCs
Cutaneous	Occasional café-au-lait spots

?	
Neuro	Seizures
Oculo	Diffuse choroidal hemangioma
Cutaneous	Port-wine stain

?	
Neuro	Cortical tubers
Oculo	Astrocytic hamartoma
Cutaneous	Adenoma sebaceum; ash-leaf spots; shagreen patches

?	
Neuro	Cerebellar hemangioblastoma
Oculo	Capillary hemangioblastoma
Cutaneous	None

?	
Neuro	Seizures
Oculo	Unilateral ROP-like appearance
Cutaneous	Erythema/bullae: 'Splashed paint'

?	
Neuro	Ataxia
Oculo	Conj telangiectasias
Cutaneous	Telangiectasias

?	
Neuro	A-V malformation
Oculo	A-V malformation
Cutaneous	Not much

Phakomatoses aka neuro-oculocutaneous syndromes: TLDR



NF1: 'Peripheral' NF

Neuro	Optic nerve glioma
Oculo	Lisch nodules; upper-lid plexiform neurofibroma
Cutaneous	Café-au-lait spots

NF2: 'Central' NF

Neuro	Bilateral acoustic neuromas
Oculo	Early PSCs
Cutaneous	Occasional café-au-lait spots

Sturge-Weber

Neuro	Seizures
Oculo	Diffuse choroidal hemangioma
Cutaneous	Port-wine stain

Tuberous sclerosis: 'EPILOA'

Neuro	Cortical tubers
Oculo	Astrocytic hamartoma
Cutaneous	Adenoma sebaceum; ash-leaf spots; shagreen patches

von Hippel-Lindau

Neuro	Cerebellar hemangioblastoma
Oculo	Capillary hemangioblastoma
Cutaneous	None

Incontinentia pigmenti

Neuro	Seizures
Oculo	Unilateral ROP-like appearance
Cutaneous	Erythema/bullae: 'Splashed paint'

Ataxia-telangiectasia (Louis-Bar)

Neuro	Ataxia
Oculo	Conj telangiectasias
Cutaneous	Telangiectasias

Racemose angioma (Wyburn-Mason)

Neuro	A-V malformation
Oculo	A-V malformation
Cutaneous	Not much



(Next, Version 2)

Phakomatoses aka neuro-oculocutaneous syndromes: TLDR



Incontinentia pigmenti

Neuro	?
Oculo	?
Cutaneous	?

Sturge-Weber

Neuro	?
Oculo	?
Cutaneous	?

Racemose angioma (Wyburn-Mason)

Neuro	?
Oculo	?
Cutaneous	?

von Hippel-Lindau

Neuro	?
Oculo	?
Cutaneous	?

Tuberous sclerosis: 'EPILOA'

Neuro	?
Oculo	?
Cutaneous	?

Ataxia-telangiectasia (Louis-Bar)

Neuro	?
Oculo	?
Cutaneous	?

NF2: 'Central' NF

Neuro	?
Oculo	?
Cutaneous	?

NF1: 'Peripheral' NF

Neuro	?
Oculo	?
Cutaneous	?

Phakomatoses aka neuro-oculocutaneous syndromes: TLDR



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