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

In general terms, how do phakomatoses present?
Phakomatoses are known also as what sort of syndrome?
Neuro-oculocutaneous syndromes

In general terms, how do phakomatoses present?
With multiple lesions in two or more organ systems, usually including the CNS, eyes, and skin.
Phakomatoses are known also as what sort of syndrome?

**Neuro**-oculocutaneous syndromes

In general terms, how do phakomatoses present?
With multiple lesions in two or more organ systems, usually including the CNS, and...
Phakomatoses are known also as what sort of syndrome?
**Neuro-oculocutaneous syndromes**

*In general terms, how do phakomatoses present?*
With multiple lesions in two or more organ systems, usually including the CNS, eyes and
Phakomatoses are known also as what sort of syndrome?

**Neuro-oculocutaneous** syndromes

In general terms, how do phakomatoses present?
With multiple lesions in two or more organ systems, usually including the CNS, eyes and skin
**Phakomatoses are known also as what sort of syndrome?**
Neuro-oculocutaneous syndromes

**In general terms, how do phakomatoses present?**
With multiple lesions in two or more organ systems, usually including the CNS, eyes, and skin

**Are the lesions in phakomatoses predominantly choristomas or hamartomas?**
Phakomatoses are known also as what sort of syndrome?
Neuro-oculocutaneous syndromes

In general terms, how do phakomatoses present?
With multiple lesions in two or more organ systems, usually including the CNS, eyes and skin

Are the lesions in phakomatoses predominantly choristomas or hamartomas?
Most (but not all) are hamartomas (some are choristomas)
Phakomatoses are known also as what sort of syndrome? Neuro-oculocutaneous syndromes

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Are the lesions in phakomatoses predominantly choristomas or hamartomas? Most (but not all) are hamartomas (some are choristomas)

What’s the difference between a hamartoma and a choristoma?
Phakomatoses are known also as what sort of syndrome? Neuro-oculocutaneous syndromes

In general terms, how do phakomatoses present? With multiple lesions in two or more organ systems, usually including the CNS, eyes, and skin.

Are the lesions in phakomatoses predominantly choristomas or hamartomas? Most (but not all) are hamartomas (some are choristomas)

What’s the difference between a hamartoma and a choristoma?
- A hamartoma is a nest of abnormal cells in a normal location, whereas a choristoma is a nest of relatively-normal cells in an abnormal location.
Phakomatoses are known also as what sort of syndrome?
Neuro-oculocutaneous syndromes

In general terms, how do phakomatoses present?
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Are the lesions in phakomatoses predominantly choristomas or hamartomas?
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In general terms, how do phakomatoses present?
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Are the lesions in phakomatoses predominantly choristomas or hamartomas?
Hamartomas

Is there a single, universally accepted definition of the term phakomatosis?
Phakomatoses are known also as what sort of syndrome?
Neuro-oculocutaneous syndromes

In general terms, how do phakomatoses present?
With multiple lesions in two or more organ systems, usually including the CNS, eyes, and skin

Are the lesions in phakomatoses predominantly choristomas or hamartomas?
Hamartomas

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

Abbreviations used henceforth

NF1 • Neurofibromatosis type 1:

•

•

•

•

•

•

•

•

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

1. Neurofibromatosis type 1: von Rechlinghausen syndrome

Abbreviations used henceforth

NF1
A phakomatosis by any other name... by what other name is each syndrome known?

- **NF1**
  - Neurofibromatosis type 1: von Rechlinghausen syndrome

- **TS**
  - Tuberous sclerosis:
    - 
    - 
    - 
    - 
    - 
    - 
    - 
    - 
    - 

**Abbreviations used henceforth**

- NF1
- TS
A phakomatosis by any other name... by what other name is each syndrome known?

Abbreviations used henceforth

NF1 • Neurofibromatosis type 1: von Rechlinghausen syndrome

TS • Tuberous sclerosis: Bournville disease

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

Abbreviations used henceforth

**NF1**
- Neurofibromatosis type 1: von Rechlinghausen syndrome

**TS**
- Tuberous sclerosis: Bournville disease

**SWS**
- Sturge-Weber syndrome:
Neurofibromatosis type 1: von Rechlinghausen syndrome
Tuberous sclerosis: Bournville disease
Sturge-Weber syndrome: Encephalotrigeminal angiomatosis
von Hippel-Lindau: Retinal angiomatosis
Incontinentia pigmenti: Bloch-Sulzberger syndrome
Neurofibromatosis type 2: None in common use of which I am aware
Racemose angioma: Wyburn-Mason syndrome
Ataxia-telangiectasia: Louis-Bar syndrome

Abbreviations used henceforth:
NF1, Neurofibromatosis type 1
TS, Tuberous sclerosis
SWS, Sturge-Weber syndrome
vH-L, von Hippel-Lindau
IP, Incontinentia pigmenti
NF2, Neurofibromatosis type 2
RAAT, Racemose angioma
AT, Ataxia-telangiectasia

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

Other names you might encounter for SWS:
Encephalofacial angiomatosis
Cerebrofacial angiomatosis
A phakomatosis by any other name... by what other name is each syndrome known?

NF1 • Neurofibromatosis type 1: von Rechlinghausen syndrome

TS • Tuberous sclerosis: Bournville disease

SWS • Sturge-Weber syndrome: Encephalotrigeminal angiomatosis

vH-L • von Hippel-Lindau:
  •
  •
  •
  •
  •

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

Abbreviations used henceforth

NF1  •  Neurofibromatosis type 1: von Rechlinghausen syndrome
TS   •  Tuberous sclerosis: Bournville disease
SWS  •  Sturge-Weber syndrome: Encephalotrigeminal angiomatosis
vH-L •  von Hippel-Lindau: Retinal angiomatosis
Neurofibromatosis type 1: von Rechlinghausen syndrome

Tuberous sclerosis: Bournville disease

Sturge-Weber syndrome: Encephalotrigeminal angiomatosis

von Hippel-Lindau: Retinal angiomatosis

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Abbreviations used henceforth

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SWS  • Sturge-Weber syndrome: Encephalotrigeminal angiomatosis

vH-L • von Hippel-Lindau: Retinal angiomatosis

IP   • Incontinentia pigmenti:
A phakomatosis by any other name...by what other name is each syndrome known?

Abbreviations used henceforth:

- NF1: Neurofibromatosis type 1: von Rechlinghausen syndrome
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A phakomatosis by any other name...by what other name is each syndrome known?

Abbreviations used henceforth

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TS  ● Tuberous sclerosis: Bournville disease

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vH-L  ● von Hippel-Lindau: Retinal angiomatosis

IP  ● Incontinentia pigmenti: Bloch-Sulzberger syndrome

NF2  ● Neurofibromatosis type 2:
  ●
  ●

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

Abbreviations used henceforth

- **NF1**  Neurofibromatosis type 1: von Rechlinghausen syndrome
- **TS**  Tuberous sclerosis: Bournville disease
- **SWS**  Sturge-Weber syndrome: Encephalotrigeminal angiomatosis
- **vH-L**  von Hippel-Lindau: Retinal angiomatosis
- **IP**  Incontinentia pigmenti: Bloch-Sulzberger syndrome
- **NF2**  Neurofibromatosis type 2: MISME syndrome

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

Abbreviations used henceforth

NF1: Neurofibromatosis type 1: von Rechlinghausen syndrome
TS: Tuberous sclerosis: Bournville disease
SWS: Sturge-Weber syndrome: Encephalotrigeminal angiomatosis
vH-L: von Hippel-Lindau: Retinal angiomatosis
IP: Incontinentia pigmenti: Bloch-Sulzberger syndrome

NF2: Neurofibromatosis type 2: **MISME syndrome**

*MISME is an acronym. What does it stand for?*

---M
---I
---S
---M
---E
Abbreviations used henceforth

NF1  ● Neurofibromatosis type 1: von Rechlinghausen syndrome

TS  ● Tuberous sclerosis: Bournville disease

SWS  ● Sturge-Weber syndrome: Encephalotrigeminal angiomatosis

vH-L  ● von Hippel-Lindau: Retinal angiomatosis

IP  ● Incontinentia pigmenti: Bloch-Sulzberger syndrome

NF2  ● Neurofibromatosis type 2: **MISME syndrome**

*MISME is an acronym. What does it stand for?*

-- Multiple
-- Inherited
-- Schwannomas,
-- Meningiomas (and)
-- Ependymomas
A phakomatosis by any other name…by what other name is each syndrome known?

Abbreviations used henceforth

NF1: Neurofibromatosis type 1: von Rechlinghausen syndrome
TS: Tuberous sclerosis: Bournville disease
SWS: Sturge-Weber syndrome: Encephalotrigeminal angiomatosis
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Abbreviations used henceforth

NF1  ● Neurofibromatosis type 1: von Rechlinghausen syndrome
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Abbreviations used henceforth
A phakomatosis by any other name... by what other name is each syndrome known?

- **NF1**: Neurofibromatosis type 1: von Rechlinghausen syndrome
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- **vH-L**: von Hippel-Lindau: Retinal angiomatosis
- **IP**: Incontinentia pigmenti: Bloch-Sulzberger syndrome
- **NF2**: Neurofibromatosis type 2: MISME syndrome
- **RA**: Racemose angioma: Wyburn-Mason syndrome
- **AT**: Ataxia-telangiectasia:
A phakomatosis by any other name... by what other name is each syndrome known?

NF1 ● Neurofibromatosis type 1: von Rechlinghausen syndrome
TS ● Tuberous sclerosis: Bournville disease
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NF2 ● Neurofibromatosis type 2: MISME syndrome
RA ● Racemose angioma: Wyburn-Mason syndrome
AT ● Ataxia-telangiectasia: Louis-Bar syndrome
Phakomatoses: Inheritance patterns

- These four are AD...
  
  - NF1
  - NF2
  - von Hippel-Lindau
  - Tuberous sclerosis

- This one is AR...
  
  - Ataxia-telangiectasia

- These two are sporadic/nonhereditary
  
  - Sturge-Weber
  - Racemose angioma
Phakomatoses: Inheritance patterns

- These four are AD...
  - NF2
  - NF1
  - von Hippel-Lindau
  - Tuberous sclerosis
**Phakomatoses: Inheritance patterns**

- These four are *AD*…
  - NF2
  - NF1
  - von Hippel-Lindau
  - Tuberous sclerosis
- This one is *AR*…
● **Phakomatoses: Inheritance patterns**

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- This one is *AR*...
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- This one is *X-linked dominant*...
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- This one is \textit{X-linked dominant}...
  - Incontinentia pigmenti
- And these two are \textit{sporadic/nonhereditary}
Phakomatoses: Inheritance patterns

- These four are **AD**...
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  - NF1
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  - Tuberous sclerosis
- This one is **AR**...
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- And these two are **sporadic/nonhereditary**
  - Sturge-Weber
  - Racemose angioma
**Phakomatoses: Inheritance patterns**

- These four are AD...
  - NF2
  - NF1
  - von Hippel-Lindau

What does X-linked dominant transmission mean?

- This one is **X-linked dominant**...
  - Incontinentia pigmenti

- And these two are **sporadic/nonhereditary**
  - Sturge-Weber
  - Racemose angioma
Phakomatoses: Inheritance patterns

- These four are AD...
  - NF2
  - NF1
  - von Hippel-Lindau

*What does X-linked dominant transmission mean?*
It means the condition manifests in every conception possessing at least one X chromosome (ie, everyone)

- This one is **X-linked dominant**...
  - Incontinentia pigmenti

- And these two are **sporadic/nonhereditary**
  - Sturge-Weber
  - Racemose angioma
Phakomatoses: Inheritance patterns

These four are AD...
- NF2
- NF1
- von Hippel-Lindau

**What does X-linked dominant transmission mean?**
It means the condition manifests in every conception possessing at least one X chromosome (ie, everyone)

*But almost all IP pts are female. If IP is X-linked dominant, why don’t male infants present with it?*

This one is **X-linked dominant**...
- Incontinentia pigmenti

And these two are **sporadic/nonhereditary**
- Sturge-Weber
- Racemose angioma
Phakomatoses: Inheritance patterns

These four are **AD**...
- NF2
- NF1
- von Hippel-Lindau

**What does X-linked dominant transmission mean?**
It means the condition manifests in every conception possessing at least one X chromosome (i.e., everyone)

But almost all IP pts are female. If IP is X-linked dominant, why don't male infants present with it? The mutation causing IP is lethal to males in utero. That's about as 'manifest' as it gets.

**This one is X-linked dominant...**
- Incontinentia pigmenti

And these two are **sporadic/nonhereditary**
- Sturge-Weber
- Racemose angioma
Phakomatoses: Inheritance patterns

- These four are AD...

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?

But almost all IP pts are female. If IP is X-linked dominant, why don’t male infants present with it? The mutation causing IP is lethal to males in utero. That’s about as ‘manifest’ as it gets.

- This one is X-linked dominant...
  - Incontinentia pigmenti

- And these two are sporadic/nonhereditary
  - Sturge-Weber
  - Racemose angioma
Phakomatoses: Inheritance patterns

These four are AD...

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? There are two ways by which a male child could be liveborn with IP:
--If the (phenotypically) male child possesses two X chromosomes (eg, Klinefelter syndrome, XXY) and is therefore heterozygous for IP; or
--it can occur in males via a sporadic post-zygotic mutation that renders the male child an IP ‘mosaic’

It means the condition manifests in every conception possessing at least one X chromosome (ie, everyone)

But almost all IP pts are female. If IP is X-linked dominant, why don’t male infants present with it? The mutation causing IP is lethal to males in utero. That’s about as ‘manifest’ as it gets.

This one is X-linked dominant...

Incontinentia pigmenti

And these two are sporadic/nonhereditary

Sturge-Weber

Racemose angioma
### Phakomatoses: Inheritance patterns

- These four are **AD**...
  - **NF2**
  - **NF1**
  - **von Hippel-Lindau**
  - **Tuberous sclerosis**

- This one is **AR**...
  - **Ataxia-telangiectasia**

- This one is **X-linked dominant**...
  - **Incontinentia pigmenti**

- And these two are **sporadic/nonhereditary**
  - **Sturge-Weber**
  - **Racemose angioma**

---

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

<table>
<thead>
<tr>
<th>Condition</th>
<th>% Sporadic</th>
</tr>
</thead>
<tbody>
<tr>
<td>NF2</td>
<td>?</td>
</tr>
<tr>
<td>NF1</td>
<td>?</td>
</tr>
<tr>
<td>von Hippel-Lindau</td>
<td>?</td>
</tr>
<tr>
<td>Tuberous sclerosis</td>
<td>?</td>
</tr>
<tr>
<td>Ataxia-telangiectasia</td>
<td>?</td>
</tr>
<tr>
<td>Incontinentia pigmenti</td>
<td>?</td>
</tr>
</tbody>
</table>
Phakomatoses:

- These four are **AD**:
  - NF2: 50%
  - NF1: 50%
  - von Hippel-Lindau: 20%
  - Tuberous sclerosis: 80%

- This one is **AR**:
  - Ataxia-telangiectasia: ~0%

- This one is **X-linked dominant**:
  - Incontinentia pigmenti: 60%

- And these two are sporadic/nonhereditary:
  - Sturge-Weber
  - Racemose angioma

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

These four are AD...

<table>
<thead>
<tr>
<th>Condition</th>
<th>% Sporadic</th>
</tr>
</thead>
<tbody>
<tr>
<td>NF2</td>
<td>50</td>
</tr>
<tr>
<td>NF1</td>
<td>50</td>
</tr>
<tr>
<td>von Hippel-Lindau</td>
<td>20</td>
</tr>
<tr>
<td>Tuberous sclerosis</td>
<td>80</td>
</tr>
</tbody>
</table>

This one is AR...

<table>
<thead>
<tr>
<th>Condition</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ataxia-telangiectasia</td>
</tr>
</tbody>
</table>

This one is X-linked dominant...

<table>
<thead>
<tr>
<th>Condition</th>
</tr>
</thead>
<tbody>
<tr>
<td>Incontinentia pigmenti</td>
</tr>
</tbody>
</table>

And these two are sporadic/nonhereditary

<table>
<thead>
<tr>
<th>Condition</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sturge-Weber</td>
</tr>
<tr>
<td>Racemose angioma</td>
</tr>
</tbody>
</table>

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

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

These four are AD...

- NF2 50%
- NF1 50%
- von Hippel-Lindau 20%
- Tuberous sclerosis 80%

This one is AR...

- Ataxia-telangiectasia ~0%

This one is X-linked dominant...

- Incontinentia pigmenti 60%

And these two are sporadic/nonhereditary

- Sturge-Weber
- Racemose angioma

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

Why is the sporadic-occurrence rate of A-T essentially zero? Because it is an autosomal-recessive condition, and thus can occur sporadically only if someone heterozygous for it happens to suffer a mutation of the other copy of the responsible gene--a very unlikely event.
### Study Guide: Phakomatoses

<table>
<thead>
<tr>
<th>NF1</th>
<th>Central vs Peripheral</th>
</tr>
</thead>
<tbody>
<tr>
<td>NF</td>
<td></td>
</tr>
</tbody>
</table>

**NF1**

-- Peripheral

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
NF1
--Peripheral NF
**Study Guide: Phakomatoses**

**NF1**
-- *Peripheral* NF
-- Most lesions due to abnormal [one cell type] or [different cell type] cells

---

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

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

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**NF1**

--*Peripheral* NF

--Most lesions due to abnormal *melanocytes* or *neuroglial* cells
**Study Guide: Phakomatoses**

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

**melanocytes**  **neuroglial cells**

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

**NF1**
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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
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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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 yikes
Study Guide: 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
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- Rule of thumb for Lisch nodule prevalence: Age in years x 10

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How are these cell lines related embryologically?
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Given this, it should come as no surprise that NF is associated with other manifestations of disordered neural-crest embryology, including and especially:

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Study Guide: Phakomatoses

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--Iris lesions include Lisch nodules, JXG nodules, and congenital ectropion
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How are these cell lines related embryologically?
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--Conjunctival yikes stemming from abb.
Study Guide: 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, JXG nodules, and congenital ectropion
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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
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What does PAM stand for in this context?
**Study Guide: Phakomatoses**

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

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**What does PAM stand for in this context?**
Primary acquired melanosis
**Study Guide: Phakomatoses**

**NF1**
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**Melanocytic lesions**
- 
- 
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- 

Name four common NF1 lesions that derive from melanocytes
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Study Guide: Phakomatoses

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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%
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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.

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

--Leukemia--Rhabdomyosarcoma--Pheochromocytoma--Wilms tumor
NF1

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*How does a plexiform fibroma and/or iris ectropion cause glaucoma?*
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About 50
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What percent of NF1 pts develop a glioma of the optic pathway (ie, nerve or chiasm)?

About 15
Of those, about how many will be symptomatic?
About 1/3
With what symptoms will they present?
Vision loss and/or proptosis
Are optic-nerve gliomas typically life-threatening?
No
What about NF1 pts with chiasmal gliomas—do they fare better than their non-NF1 counterparts?
Much better
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In other words, about 10% of 1 year olds will have Lisch nodules, 40% of 4 y.o.s, 60% of 6 y.o., etc. By the age of 10 years, essentially 100% of NF1 pts will manifest Lisch nodules.
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**Tuberous sclerosis**
-- Classic triad is *epiloia*
**Study Guide: Phakomatoses**

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--Lo
--A

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--Epilepsy
--Low intelligence
--Angiomas
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### Tuberous sclerosis

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**Vogt’s triad**
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**Tuberous sclerosis**
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  - Epilepsy?: What % of TS pts have seizures?
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*aWhat does epiloia stand for?*
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--Low intelligence: 50
--Angiomas: ?

What % of TS pts have facial angifibromas; ie, adenoma sebaceum?

What is the eponymous name of this triad?
Vogt’s triad
Study Guide: 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*

**What does epiloia stand for?**
--*Epi*lepsy: 80
--*Lo*w *intelligence*: 50
--*An*giomas: 75

**What % of TS pts have facial angifibromas; ie, adenoma sebaceum?**

**What is the eponymous name of this triad? Vogt’s triad**
Study Guide: Phakomatoses

NF1
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Tuberous sclerosis
--Classic triad is epiloia

What does epiloia stand for?
--Epilepsy PLUS
--Low intelligence PLUS
--Angiomas

What % of TS pts have all three?

What is the eponymous name of this triad?
Vogt’s triad
**Study Guide: Phakomatoses**

**NF1**
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**Tuberous sclerosis**
-- Classic triad is *epiloia*

--- What does *epiloia* stand for?
- **Epilepsy PLUS**
- **Low intelligence PLUS**
- **Angiomas**

--- What % of TS pts have all three? Only 30

--- What is the eponymous name of this triad? Vogt’s triad
Study Guide: Phakomatoses

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**Tuberous sclerosis**
--Classic triad is *epiloia*
--Skin: classic finding of face; ditto and ditto on torso
Study Guide: Phakomatoses

**NF1**
-- *Peripheral* NF
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**Tuberous sclerosis**
-- Classic triad is *epiloia*
-- Skin: Adenoma sebaceum of face; ash-leaf spots and shagreen patches on torso
Study Guide: Phakomatoses

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

- **Adenoma sebaceum**  
  ?  
  Appear in infancy

- **Shagreen patches**  
  ?  
  Usually in lumbosacral region

- **Ash-leaf spots**  
  ?  
  Appear in childhood
**Study Guide: Phakomatoses**

**NF1**
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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!**

- **Adenoma sebaceum**: Appear in infancy
- **Shagreen patches**: Usually in lumbosacral region
- **Ash-leaf spots**: Appear in childhood
NF1
--Peripheral NF
--Most lesions due to abnormal melanocytes or neuroglial cells
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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: Not Matching!

Adenoma sebaceum  ?

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

Ash-leaf spots  ?
**Study Guide: 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: Not Matching!**

- **Adenoma sebaceum**
  - Raised

- **Shagreen patches**

- **Ash-leaf spots**
  - Flat

Which lesion(s) is/are raised, and which is/are flat?
Study Guide: 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: Not Matching!**

- Adenoma sebaceum
- Shagreen patches
- Ash-leaf spots

*Which lesion(s) is/are hyperpigmented, and which is/are hypopigmented?
**NF1**
--*Peripheral* NF
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--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
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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: Not Matching!**

- *Adenoma sebaceum*
  - Hyperpigmented
- *Shagreen patches*
- *Ash-leaf spots*
  - Hypopigmented
Study Guide: Phakomatoses

**NF1**
-- *Peripheral* NF
-- Most lesions due to abnormal melanocytes or neuroglial cells
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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: Not Matching!**

- Adenoma sebaceum ?
- Shagreen patches ?
- Ash-leaf spots ?

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

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

---

**Skin Lesions: Not Matching!**

- *Adenoma sebaceum*: Don’t fluoresce
- *Shagreen patches*: Don’t fluoresce
- *Ash-leaf spots*: Fluoresce

*Which lesion(s) fluoresce under a Woods lamp, and which do/does not?*
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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Tuberous sclerosis
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--Skin: Adenoma sebaceum of face; ash-leaf spots and shagreen patches on torso

Skin Lesions: Not Matching!

Adenoma sebaceum
Shagreen patches
Ash-leaf spots

Which lesion(s) is/are considered pathognomonic for TS, and which is/are not?
**NF1**
-- *Peripheral NF*
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**Tuberous sclerosis**
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-- Skin: *Adenoma sebaceum* of face; *ash-leaf spots* and *shagreen patches* on torso

**Skin Lesions: Not Matching!**

- *Adenoma sebaceum*: Not
- *Shagreen patches*: Not
- *Ash-leaf spots*: Pathognomonic

*Which lesion(s) is/are considered pathognomonic for TS, and which is/are not?*
**NF1**
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**Tuberous sclerosis**
--Classic triad is *epiloia*
--Skin: Adenoma sebaceum of face; ash-leaf spots and shagreen patches on torso
--CNS: [classic finding] other benign tumors
**NF1**
--- *Peripheral* NF
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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
**Study Guide: 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?
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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
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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

Why is it called a ‘tuber’?
**Study Guide: Phakomatoses**

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**Tuberous sclerosis**
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- Skin: Adenoma sebaceum of face; ash-leaf spots and shagreen patches on torso
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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)
Study Guide: Phakomatoses

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*What is a cortical tuber?*
A benign tumor of the brain

*Why is it called a ‘tuber’?*
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*What basic geometric shape do tubers often take?*
NF1
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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

Why is it called a ‘tuber’?
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What basic geometric shape do tubers often take?
A triangle
**NF1**
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**Tuberous sclerosis**
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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)

**What basic geometric shape do tubers often take?**
A triangle

**Which way does the apex of the triangle point?**
Study Guide: Phakomatoses

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A benign tumor of the brain

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What basic geometric shape do tubers often take?
A triangle

Which way does the apex of the triangle point?
Toward a ventricle
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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
--Benign tumors of not eye and not eye as well
# Study Guide: Phakomatoses

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Study Guide: Phakomatoses

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*Other than their location, in what key way do the heart and kidney tumors differ?*
**Study Guide: 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
NF1
--Peripheral NF
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Tuberous sclerosis
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--CNS: Cortical tubers, other benign tumors
--Benign tumors of heart and kidney as well

Other than their location, in what key way do the heart and kidney tumors differ? The kidney tumors are not associated with an increased risk of morbidity/mortality, whereas the heart tumors are
**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

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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

--Benign tumors of heart and kidney as well

--Retinal tumor is something something
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**Tuberous sclerosis**
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--Skin: Adenoma sebaceum of face; ash-leaf spots and shagreen patches on torso
--CNS: Cortical tubers, other benign tumors
--Benign tumors of heart and kidney as well
--Retinal tumor is astrocytic hamartoma
NF1
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--CNS: Cortical tubers, other benign tumors
--Benign tumors of heart and kidney as well
--Retinal tumor is *astrocytic hamartoma*

By what other name is the astrocytic hamartoma of the retina known?
**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
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--Benign tumors of heart and kidney as well
--Retinal tumor is *astrocytic hamartoma*

*By what other name is the astrocytic hamartoma of the retina known?*
Retinal phakoma
Study Guide: 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
-- Benign tumors of heart and kidney as well
-- Retinal tumor is **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?
Yes

Can multiple phakomas be found in one eye?
Yes

Are the pathognomonic for TS?
No
**Study Guide: Phakomatoses**

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

---CNS: Cortical tubers, other benign tumors

---Benign tumors of heart and kidney as well

---Retinal tumor is astrocytic hamartoma

---Retinal phakoma

*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?*
**NF1**
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Can multiple phakomas be found in one eye?
**Study Guide: Phakomatoses**

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-- Irregular, elevated, and sharply demarcated
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--Fruit:
--Foodstuff:
Study Guide: Phakomatoses

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--Fruit: *‘Mulberry’*
--Foodstuff: *‘Tapioca’*

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**Study Guide: Phakomatoses**

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### von Hippel-Lindau
--- Skin: trick question
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**von Hippel-Lindau**
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--CNS: (tumor type) (tumor location) (if absent, is called not von Hippel-Lindau syndrome)
**Study Guide: Phakomatoses**

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

  *(short for Pheochromocytoma)*
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--Retinal tumor is **something something**; has large **vessels**
Study Guide: Phakomatoses

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**Tuberous sclerosis**
- Classic triad is epiloia (but all 3 present in only ~30%)
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- Retinal tumor is *capillary hemangioblastoma*; has large feeder/drainage vessels

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 hemangioblastomas associated with vHL?
- No, they can be sporadic
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Study Guide: Phakomatoses

NF1
---NF1, also known as neurofibromatosis type 1, is a genetic disorder that affects multiple systems in the body. The hallmark of NF1 is the presence of café-au-lait spots, neurofibromas, and Lisch nodules.
---NF1 is inherited in an autosomal dominant pattern, meaning that if a parent has NF1, there is a 50% chance that each child will inherit the condition.
---Iris lesions include Lisch nodules, JXG nodules, and congenital ectropion.
---Optic nerve glioma: Always symptomatic by age 10 years. Classic CT appearance: Kinked ON.
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**Can there be multiple lesions in the same eye?**
- Yes, these occur in about # of cases

**Tuberous Sclerosis**
- Classic triad is epiloia (but all 3 present in only ~30%)
  - Skin: Adenoma sebaceum of face; ash-leaf spots and shagreen patches on torso
  - CNS: Cortical tubers, other benign tumors
    - Benign tumors of heart and kidney as well
  - Retinal tumor is astrocytic hamartoma; can appear smooth or lumpy (mulberry)

**von Hippel-Lindau**
- Skin: None! Despite this, is still considered a phakomatosis (and a classic one to boot)
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By what other name is this lesion known (it's a subtle change)?
- Capillary hemangioma (ie, no ‘-blasto-’)

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Study Guide: Phakomatoses

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

No, they can be sporadic

Tuberous sclerosis

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Study Guide: Phakomatoses

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**Study Guide: Phakomatoses**

**NF1**
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--Most lesions due to abnormal melanocytes or neuroglial cells
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--Iris lesions include Lisch nodules, JXG nodules, and congenital ectropion
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--Tumor leaks $\rightarrow$ [abb.] $\rightarrow$ [abb.] $\rightarrow$ decreased VA; treat with [______] or [______]
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(*) subretinal fluid *) exudative retinal detachment*)
## Study Guide: Phakomatoses

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--- Management
- *Ocular*: DFE

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**Study Guide: Phakomatoses**

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--Management
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--- Management  
  --- *Ocular*: DFE q1 year  
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**Study Guide: Phakomatoses**

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(Vanillylmandelic acid)
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Is vH-L a potentially fatal condition?
Yes
Two components are most likely to result in death. What are they?
The cerebellar hemangioma and the renal carcinoma
# Study Guide: Phakomatoses

## NF1

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Study Guide: Phakomatoses

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**Is vH-L a potentially fatal condition?**

*Yes*

Two components are most likely to result in death. What are they?

The *cerebellar hemangioma* and the renal carcinoma.

The *cerebellar hemangioma*??!! I thought that was a benign lesion. How could it be fatal?

It is a benign lesion. However, it is notoriously 'leaky,' and the accumulating exudate can lead to compression of vital intracranial structures.
**NF1**

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**Is vH-L a potentially fatal condition?**

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**The cerebellar hemangioma** and the renal carcinoma

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**The cerebellar hemangioma??!! I thought that was a benign lesion. How could it be fatal?**

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

---

**How could the renal carcinoma be fatal?**

-- The tumor leaks → SRF → ERD → decreased VA; treat with laser or cryo.
Study Guide: Phakomatoses

**NF2**

*Which is more common, NF1 or NF2?*


**Study Guide: Phakomatoses**

**NF2**

*Which is more common, NF1 or NF2?*

NF1 is about 10x more common
**Study Guide: Phakomatoses**

**NF2**

---

NF

- **Peripheral vs Central**

---

**Central NF**

- Classic finding: bilateral acoustic neuromas

- **Eye findings:**
  - **Common:** PSC/cortical cataracts
  - **Rare:** combined hamartoma of retina and RPE
  - **Rarer:** Lisch nodules

---

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NF2
--Central NF

---Central NF

Common:
PSC/cortical cataracts;

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Rarer:
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**NF2**

--*Central* NF

--Classic finding: bilateral not eye
**Study Guide: Phakomatoses**

**NF2**

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Study Guide: 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?*
**NF2**
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What sort of tumor is the acoustic neuroma of NF2; ie, what specific cell type is involved?
A schwannoma
NF2
--Central NF
--Classic finding: bilateral acoustic neuromas

What are the three most common symptoms of acoustic neuroma?

#1: Reduced hearing
#2: Tinnitus
#3: Balance issues
Study Guide: 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
Study Guide: Phakomatoses

**NF2**

--- Central NF

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

-- Eye findings: *Common*: anterior segment
Study Guide: Phakomatoses

**NF2**

-- *Central* NF  
-- Classic finding: bilateral *acoustic neuromas*  
-- Eye findings: *Common:* PSC/cortical cataracts
**NF2**
--Central NF
--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?
Usually prior
At what age do they become clinically significant?
Usually in the 30s
Are they unilateral, or bilateral?
Both presentations are common
**Study Guide: Phakomatoses**

**NF2**
--Central NF
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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!
**Study Guide: Phakomatoses**

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-- Classic finding: bilateral *acoustic neuromas*

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  - *Common*: PSC/cortical cataracts;
  - *Rare*: posterior segment
**Study Guide: Phakomatoses**

**NF2**
-- *Central* NF
-- Classic finding: bilateral acoustic neuromas
-- Eye findings: *Common*: PSC/cortical cataracts;
  *Rare*: Combined hamartoma of retina and RPE
NF2

--Central NF

--Classic finding: bilateral acoustic neuromas

--Eye findings: Common: PSC/cortical cataracts;
Rare: Combined hamartoma of retina and RPE; Rarer:
**NF2**

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**One key difference between NF1 and NF2 is this:**

In NF1, both melanocytic and neuroglial lesions are common, whereas…
**Study Guide: Phakomatoses**

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**Neuroglial lesions**
--Nodular neurofibromas
--Plexiform neurofibromas
--Optic glioma
--Prominent corneal nerves

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

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In NF1, both melanocytic and neuroglial lesions are common, whereas…
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Do melanocytic lesions occur in NF2 at all?
Study Guide: Phakomatoses

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One key difference between NF1 and NF2 is this:

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
Study Guide: 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**

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**Neuroglial lesions**

-- Nodular neurofibromas?
-- Plexiform neurofibromas?
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---

Do NF2 pts get peripheral-nerve tumors like NF1 pts?
Study Guide: Phakomatoses

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Do NF2 pts get peripheral-nerve tumors like NF1 pts? Yes, but at much lower rates

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--- 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?

--- Neuroglial lesions
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Study Guide: Phakomatoses

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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

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**Schwannomas of the SC**
--Meningiomas (intracranial)
--Ependymomas
Neuroglial lesions
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This propensity for manifesting mainly as CNS tumors is why NF2 is referred to as ‘central’ NF like NF1 pts?
Neuroglial lesions
--Nodular neurofibromas
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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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--E
Study Guide: Phakomatoses

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Study Guide: 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

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

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

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*PSC* = posterior subcapsular cataract
*RPE* = retinal pigmented epithelium
**Study Guide: Phakomatoses**

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A tumor composed of histologically normal cells found in their clinical state
**Study Guide: Phakomatoses**

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What is the name of the reverse clinical entity, i.e., one with normal cells found in an abnormal location?

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Rarer:
Combined hamartoma of retina and RPE
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That a lesion is a hamartoma (or choristoma) indicates what about its onset?

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

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

So, what combination of hamartomatous cells of the retina and RPE are involved in a combined hamartoma of the retina and RPE?
What is a hamartoma?
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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?
RPE cells (duh) and retinal glial cells
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How does it present clinically?
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RPE cells (duh) and retinal glial cells

How does it present clinically?
As a variably pigmented, slightly elevated retinal mass of the retina...
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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?
RPE cells (duh) and retinal glial cells

How does it present clinically?
As a variably pigmented, slightly elevated retinal mass of the peripapillary retina
What is a hamartoma?
A tumor composed of histologically abnormal cells found in their normal location

So, what combination of hamartomatous cells of the retina and RPE are involved in a combined hamartoma of the retina and RPE?
RPE cells (duh) and retinal glial cells

How does it present clinically?
As a variably pigmented, slightly elevated retinal mass of the peripapillary retina

With what more sinister dz entity is it often confused?
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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?*
RPE cells (duh) and retinal glial cells

*How does it present clinically?*
As a variably pigmented, slightly elevated retinal mass of the peripapillary retina

*With what more sinister dz entity is it often confused?*
Choroidal melanoma--eyes have been enucleated because of this misdiagnosis
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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?
RPE cells (duh) and retinal glial cells

How does it present clinically?
As a variably pigmented, slightly elevated retinal mass of the peripapillary retina

With what more sinister dz entity is it often confused?
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.
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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**
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**Acoustic neuroma**

Bag CN V1

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

Bag CN VII

Corneal decompensation
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**Acoustic neuroma**

- Bag CN V1
  - Decreased corneal sensation
- Bag CN VII
  - Lagophthalmos

*Another eye finding associated with acoustic neuroma is corneal decompensation. By what two mechanisms might this occur?*
**Study Guide: Phakomatoses**

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**Sturge-Weber**
-- Classic stigmata is the *port-wine stain*
-- Ipsilateral meningeal AVM $\rightarrow$ seizures
-- Classic *tomato catsup fundus appearance* is due to a diffuse choroidal hemangioma
-- Another classic finding on DFE: Glaucomatous cupping in the ipsilateral ONH only
-- Glaucoma surgery: ↑ risk of massive choroidal effusion due to abnormal choroidal vasculature
**NF2**

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

An angioma

By what ‘official’ name is it known?

Nevus flammeus

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

Does it always present in this manner?

No. Some cases will cross the midline of the face

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

No

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

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**Study Guide: Phakomatoses**

**NF2**
-- *Central* NF
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**Sturge-Weber**
-- Classic stigmata is the *port-wine stain*

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

*By what ‘official’ name is it known?*
Nevus flammeus
**NF2**

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**Sturge-Weber**
-- Classic stigmata is the *port-wine stain*
-- Ipsilateral meningeal AVM → symptom/sign
### NF2

- **Central NF**
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### Sturge-Weber

- Classic stigmata is the *port-wine stain*
- Ipsilateral meningeal AVM → **seizures**
**Study Guide: Phakomatoses**

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**Sturge-Weber**
-- Classic stigmata is the *port-wine stain*
-- *Ipsilateral meningeal AVM → seizures*

*Is the meningeal AVM prone to bleeding?*
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*Is the meningeal AVM prone to bleeding?*
*No*
**Study Guide: Phakomatoses**

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How prevalent is seizure activity in SWS?
Study Guide: 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?*
Very--estimates run as high as 90% of cases
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**Sturge-Weber**
--Classic stigmata is the **port-wine stain**
--Ipsilateral meningeal AVM $\rightarrow$ **seizures**
--Classic **mmmm...** fundus appearance is due to a

lesion (something something something)
**Study Guide: Phakomatoses**

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**Sturge-Weber**
--Classic stigmata is the **port-wine stain**
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--Classic **tomato catsup** fundus appearance is due to a **diffuse choroidal hemangioma**
Phakomatoses

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Diffuse choroidal hemangioma is present in what percent of SWS?

About 50%

Can the choroidal hemangioma be present bilaterally?
Yes, but it's uncommon

Does the choroidal hemangioma have malignant potential?
No
Sturge-Weber
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**Study Guide: Phakomatoses**

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*What percent of SWS pts develop glaucoma?*
**Study Guide: Phakomatoses**

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Estimates run as high as 70
Study Guide: Phakomatoses

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---

*What percent of SWS pts develop glaucoma?*
Estimates run as high as 70

*Is there a relationship between the port-wine stain and risk of glaucoma?*
Yes. If the port-wine stain involves the **eyelid** the risk is **increased**; if it involves any other structure the risk is **decreased**
**Study Guide: Phakomatoses**

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Elevated IOP in SWS stems from three different mechanisms. What are they?
--
--
--

*Hint forthcoming…*
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A noncirculatory anomaly
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Elevated IOP in SWS stems from three different mechanisms. What are they?
--Increased episcleral venous pressure (EVP)
--Increased ciliary-body perfusion → aqueous hypersecretion
--Developmental abnormality of the drainage angle

2° to ocular circulatory anomalies
A noncirculatory anomaly
**Study Guide: Phakomatoses**

\[
IOP = \frac{\text{Aqueous Formation Rate (µL/min)}}{\text{Outflow Facility (µL/min/mmHg)}} + \text{Episceral Venous Pressure (mmHg)}
\]

*Recalling the Goldmann equation for IOP…*

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**What percent of SWS pts develop glaucoma?**
Estimates run as high as 70%

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\[ IOP = \frac{\text{Aqueous Formation Rate (µL/min)}}{\text{Outflow Facility (µL/min/mmHg)}} + \text{Episcleral Venous Pressure (mmHg)} \]

*Recalling the Goldmann equation for IOP…*

- \[ \uparrow IOP \text{ in SWS} \] is secondary to 
  - **Aqueous hypersecretion**
  - **Abnormal drainage angle** + \[ \uparrow \text{Episcleral Venous Pressure} \]

---

**What percent of SWS pts develop glaucoma?**
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**Sturge-Weber**
-- Classic stigmata is the *port-wine stain*
-- Ipsilateral meningeal AVM → *seizures*
-- Classic *tomato catsup* fundus appearance is due to a *diffuse choroidal hemangioma*
-- Another classic finding on DFE: *Glaucotamous cupping in the ipsilateral ONH only*
-- Glaucoma surgery: ↑ risk of massive *bad surgical complication due to abnormal* two words
**NF2**

-- *Central* NF

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**Sturge-Weber**

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

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-- Classic *tomato catsup* fundus appearance is due to a *diffuse choroidal hemangioma*

-- Another classic finding on DFE: Glaucomatous cupping in the ipsilateral ONH only

-- Glaucoma surgery: ↑ risk of massive *choroidal effusion* due to abnormal *choroidal vasculature*
**Sturge-Weber syndrome**
--Classic stigmata is the *port-wine stain*
--Ipsilateral meningeal AVM → *seizures*
--Classic *tomato catsup* fundus appearance is due to a diffuse choroidal hemangioma
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--Glaucoma surgery: ↑ risk of massive choroidal effusion due to abnormal choroidal vasculature

There is another phakomatosis--less well-known than SWS--that also presents with a *port-wine stain*. What is it?

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**Klippel-Trénaunay syndrome**
There is another phakomatosis--less well-known than SWS--that also presents with a *port-wine stain*. What is it?
**Study Guide: Phakomatoses**

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**Sturge-Weber**
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**Study Guide: Phakomatoses**

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How is Klippel-Trénaunay pronounced?
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Phakomatoses

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Yes--vascular lesions of the trunk and a single limb, along with marked hypertrophy of that limb
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In the vast majority (~90%) of cases, a leg

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Is the limb hypertrophy present at birth? Usually
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-- Most common cause of **main symptom** in childhood
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# Study Guide: Phakomatoses

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## Ataxia-telangiectasia
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-- Other eye findings include:

  - EOM problem 1
  - EOM test
  - EOM prob 2
  - EOM prob 3

  with intact
### NF2

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*Ataxia-telangiectasia* toddlers have difficulty initiating saccades, and sometimes use a head turn/thrust to do so.

**What more-common, less-devastating oculomotor disorder presents similarly?**
### Study Guide: Phakomatoses

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**What aspects of the immune system are abnormal?**

-- T-cells are abnormal in both function and number
-- Immunoglobulin levels are abnormal

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

**The thymus**
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-- T-cells are abnormal in both function and *number*
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-- Abnormal immune function $\rightarrow$
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---Abnormally high, or low? **Low**
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*Your A-T pt may have a sinus infection. Should you get a CT to confirm? 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
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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 less-robust healthcare systems (ie, without readily-available antibiotics), A-T pts die of sinopulmonary infections in their teens; whereas
-- 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).

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For what cancer are A-T heterozygotes at particular risk?
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*For what cancer are A-T heterozygotes at particular risk? Breast*
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-- Skin manifestation: not surprisingly...
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--Classic stigmata is the **port-wine stain**
--Ipsilateral meningeal AVM → seizures
--Classic **tomato catsup** fundus appearance is due to a diffuse choroidal hemangioma
--Another classic finding on DFE: Glaucomatous cupping in the ipsilateral ONH only
--Glaucoma surgery: ↑ risk of massive choroidal effusion due to abnormal choroidal vasculature

**Ataxia-telangiectasia**
--Most common cause of progressive ataxia in childhood
--Only phakomatosis with no abnormalities of the fundus
--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**
**Study Guide: 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

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At what age do cutaneous telangiectasias begin to appear?
3-5 years (ie, at about the same time the conj ones do)

At what location do they typically appear first?
The malar region of the face

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

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--Classic finding: bilateral acoustic neuromas
--Eye findings: **Common:** PSC/cortical cataracts;
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Incontinentia pigmenti

--Skin normal at birth, but abnormality 1 and abnormality 2 develop by age X; only later develops the classic appearance
**Incontinentia pigmenti**

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

--Skin normal at birth, but *erythema* and *bullae* develop by 1 week; only later develops the classic ‘splashed paint’ appearance
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--- Eye findings are usually unilateral

*How is the peripheral proliferative retinopathy managed?*
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\textbf{How is the peripheral proliferative retinopathy managed?}
\textbf{basically, in the same manner as ROP}
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--2/3 will also have abnormal *mouth issue*
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Name two other congenital eye syndromes associated with abnormal dentition:

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In three words, what sort of condition is Axenfeld-Reiger?
It is an...
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Hutchinson’s teeth


**Study Guide: Phakomatoses**

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**Racemose angioma**
-- Characterized by AVM of **eye** and **brain**
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**In basic terms, what is an AVM?**

- A direct communication between the arterial and venous sides of the circulation; ie, without benefit of an intervening capillary bed

---

In RA, are the AVM of the eye unilateral or bilateral?
- Unilateral

How about the AVM of the brain?
- Also unilateral

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

Where specifically are the AVM located in RA?
- The eye AVM are usually found in the temporal retina
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Study Guide: Phakomatoses

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**Study Guide: Phakomatoses**

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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?
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?
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!

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

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**Study Guide: Phakomatoses**

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*At what age do RA pts begin to suffer these brain bleeds?*
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**Racemose angioma**
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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
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What about seizures? How prevalent is seizure activity in RA?
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What about seizures? How prevalent is seizure activity in RA?
Not very--estimates run as low as 5% of cases
Study Guide: Phakomatoses

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**Study Guide: Phakomatoses**

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Does this mean RA pts don’t have eye/vision trouble related to their condition?
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--Retinal AVM **don’t** leak on FA

*Does this mean RA pts don’t have eye/vision trouble related to their condition?*
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.*
**Study Guide: 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, leading to hemiparesis and death
--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?**


**Study Guide: Phakomatoses**

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**Racemose angioma**
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-- 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.