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

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With multiple lesions in two or more organ systems, usually including the , 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, and...
Phakomatoses are known also as what sort of syndrome?
Neuro-oculo-cutaneous syndromes

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

Neuro-oculocutaneous syndromes

In general terms, how do phakomatoses present?
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Phakomatoses are known also as what sort of syndrome?
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In general terms, how do phakomatoses present?
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A hamartoma is a nest of abnormal cells in a normal location, whereas
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That a lesion is a hamartoma or choristoma indicates what about its onset?
That it is congenital
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

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That a lesion is a hamartoma or choristoma indicates what about its status vis a vis malignancy?
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That a lesion is a hamartoma or choristoma indicates what about its onset? That it is congenital.

That a lesion is a hamartoma or choristoma indicates what about its status vis a vis malignancy? That it is benign.
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?
Phakomatoses are known also as what sort of syndrome?
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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?

Abbreviations used henceforth

NF1
- Neurofibromatosis type 1: von Rechlinghausen syndrome
A phakomatoses 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:
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
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:
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

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?

Abbreviations used henceforth

NF1 • Neurofibromatosis type 1: \textit{von Rechlinghausen syndrome}

TS • Tuberous sclerosis: \textit{Bournville disease}

SWS • Sturge-Weber syndrome: \textit{Encephalotrigeminal angiomatosis}

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

vH-L - von Hippel-Lindau: Retinal angiomatosis
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:

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

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 angiomaosis

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
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

MISME is an acronym. What does it stand for?

---M
---I
---S
---M
---E
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?*

---

**Multiple**

---

**Inherited**

---

**Schwannomas**,

---

**Meningiomas (and)**

---

**Ependymomas**
Q

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

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vH-L ● von Hippel-Lindau: Retinal angioma­matosis

IP ● Incontinentia pigmenti: Bloch-Sulzberger syndrome

NF2 ● Neurofibromatosis type 2: MISME syndrome

RA ● Racemose angioma:
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
- **RA** • Racemose angioma: Wyburn-Mason 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
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- **IP**: Incontinentia pigamenti: 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?

**Abbreviations used henceforth**

- **NF1**: Neurofibromatosis type 1: von Rechlinghausen syndrome
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- **IP**: Incontinentia pigmenti: Bloch-Sulzberger syndrome
- **NF2**: Neurofibromatosis type 2: MISME syndrome
- **RA**: Racemose angioma: Wyburn-Mason syndrome
- **AT**: Ataxia-telangiectasia: Louis-Bar syndrome
Phakomatoses: Inheritance patterns

- These four are \textit{AD}...

- \textbf{NF2}

- \textbf{NF1}

- \textbf{von Hippel-Lindau}

- \textbf{Tuberous sclerosis}

- This one is \textit{AR}...

- \textbf{Ataxia-telangiectasia}

- This one is \textit{X-linked dominant}

- \textbf{Incontinentia pigmenti}

- And these two are \textit{sporadic/nonhereditary}

- \textbf{Sturge-Weber}

- \textbf{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

- These four are **AD**...
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  - Tuberous sclerosis

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Phakomatoses: Inheritance patterns

- These four are \textit{AD}...
  - NF2
  - NF1
  - von Hippel-Lindau

What does \textit{X-linked dominant} transmission mean?

- This one is \textit{X-linked dominant}...
  - Incontinentia pigmenti

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

- These four are AD...
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  - 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
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  - Racemose angioma
Phakomatoses: Inheritance patterns

- These four are \textit{AD}...
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  - NF1
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\textit{What does X-linked dominant transmission mean?}

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

\textit{But almost all IP pts are female. If IP is X-linked dominant, why don't male infants present with it?}

- This one is \textbf{X-linked dominant}...
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- And these two are \textit{sporadic/nonhereditary}
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  - NF1
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**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?

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

These four are **AD**...

- NF1
- NF2
- 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

**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.
Phakomatoses:

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>

51% Sporadic
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>
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<tr>
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<th>% Sporadic</th>
</tr>
</thead>
<tbody>
<tr>
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<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**...

- **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?
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?

% Sporadic

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

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.
Phakomatoses

NF1

- Central vs Peripheral NF

NF1

-- 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
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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- 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
Phakomatoses

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

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

How are these cell lines related embryologically?
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
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**

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
--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**

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**)
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

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 yikes
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

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--Conjunctival yikes stemming from abb.

*How are these cell lines related embryologically?*
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[Diagram showing melanocytes and neuroglial cells]
Phakomatoses

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--Conjunctival melanoma stemming from PAM
Phakomatoses

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What does PAM stand for in this context?
Phakomatoses

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Both derive from **neural-crest cells**

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--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
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

Café au lait spots

NF1: Melanocytic lesions
**Phakomatoses**

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**Neuroglial lesions**
--
--
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Name four common NF1 lesions that derive from neuroglial cells
Phakomatoses

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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

Name four common NF1 lesions that derive from neuroglial cells.
Phakomatoses

NF1: Neuroglial lesions

- Plexiform neurofibroma
- Nodular neurofibroma
- Optic nerve glioma
- NF1: Neuroglial lesions
**Phakomatoses**

**NF1**

*Peripheral NF*

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

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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**

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

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

Melanocytic lesions

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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'.
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What is the lifetime risk of such a transformation?
**Phakomatoses**

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- Peripheral NF
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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%
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

-- 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.

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

*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

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

NF1
--Peripheral NF
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--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

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*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.

*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

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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.

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
**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

---

**Rule of thumb for Lisch nodule prevalence:** Age in years $\times 10$
Phakomatoses

**NF1**

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What rule of thumb adheres regarding eh 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

NF1: Lisch nodules

Darker on light iris

Lighter on dark iris
Phakomatoses

NF1
--Peripheral NF
--Most lesions due to abnormal melanocytes or neuroglial cells
--Glaucoma associated with ipsilateral upper lid dermatochalasis, fibrous tissue and 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

What does JXG stand for in this context?

JXG: Juvenile xanthogranuloma
Phakomatoses

**NF1**
--*Peripheral NF*
--Most lesions due to abnormal melanocytes or neuroglial cells
--Glaucoma associated with ipsilateral upper lid subcutaneous fibromas and 'prism' ectropion
--Iris lesions include Lisch nodules, JXG nodules, and congenital ectropion

**Optic nerve glioma:** Always symptomatic by age 10 years. Classic CT appearance: Kinked Optic Nerve.

---

What does JXG stand for in this context? *Juvenile xanthogranuloma*
Phakomatoses

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NF1: JXG nodules
Phakomatoses

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In three words, what sort of condition is it?
It is a…

JXG nodules
NF1
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In three words, what sort of condition is it?
It is a…nonneoplastic histiocytic proliferation
**Phakomatoses**

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*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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What are the two hallmarks of JXG histology?
The presence of…Touton giant cells
The presence of…’foamy macrophages’
Phakomatoses

NF1: Touton giant cells
Phakomatoses

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At what age does JXG present?
Phakomatoses

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At what age does JXG present?
The majority before age 1 year, and almost all by age 2
Phakomatoses

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*How does JXG usually present? (Hint: It’s not ophthalmic)*
Phakomatoses

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**How does JXG usually present?** *(Hint: It’s not ophthalmic)*
As orange skin papules
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*
--Optic nerve glioma: Always symptomatic by age [ ] (years) Classic CT appearance:
Phakomatoses

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(ON = optic nerve)
NF1: Optic nerve gliomas bilaterally. Note the ‘kinked’ appearance
What percent of NF1 pts develop a glioma of the optic pathway (ie, nerve or chiasm)?

---

**Phakomatoses**

**NF1**

---

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What percent of NF1 pts develop a glioma of the optic pathway (ie, nerve or chiasm)?
About 15

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
Phakomatoses

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Phakomatoses

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About 15

**Of those, about how many will be symptomatic?**
About 1/3

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Vision loss and/or proptosis

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**Much** better
# Phakomatoses

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- 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: $\text{Age in years} \times \text{something} \times \text{something}$
**Phakomatosis**

**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*
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**

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

What does *epiloia* stand for?
--Epi
--Lo   i
--A
Phakomatoses

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

*What does epiloia stand for?*
--Epi*lepsy*
--Lo*w* i*ntelligence*
--An*giomas*
Phakomatoses

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

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

*What is the eponymous name of this triad?*

Vogt's triad
Phakomatoses

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**Tuberous sclerosis**
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*What does epiloia stand for?*
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--- Low intelligence
--- Angiomas

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

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

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

What % of TS pts have seizures?

What is the eponymous name of this triad?
Vogt's triad
Phakomatoses

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

*What does epiloia stand for?*
--Epilepsy: 80
--Low intelligence
--Angiomas

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*What is the eponymous name of this triad? Vogt's triad*
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; can appear smooth or lumpy (*mulberry*)

**What does epiloia stand for?**
--Epilepsy: 80
--Low intelligence: ?
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**What is the eponymous name of this triad?**
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Phakomatoses

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Tuberous sclerosis
--Classic triad is *epiloia*

What does *epiloia* stand for?
--Epilepsy: 80
--Low intelligence: 50
--Angiomas

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

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

*What does epiloia stand for?*
-- **Epi**lepsy: 80
-- **Lo**w **i**ntelligence: 50
-- **A**ngiomas: ?

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

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

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

*What does epiloia stand for?*
--Epilepsy: 80
--Low intelligence: 50
--Angiomas: 75

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

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**Phakomatoses**

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

*What does epiloia stand for?*
-- *Epi*lepsy PLUS
-- *Lo*w *i ntelligence* PLUS
-- *A*ngiomas

*What % of TS pts have all three?*

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

**NF1**
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**Tuberous sclerosis**
--Classic triad is epiloia
--Skin: classic finding of face; ditto and ditto on torso
Phakomatoses

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--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
Phakomatoses

Tuberous sclerosis: Adenoma sebaceum
Phakomatoses

Tuberous sclerosis: Ash leaf spots
Phakomatoses

Tuberous sclerosis: Shagreen patch
Phakomatoses

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-- *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 |
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

**Skin Lesions: Matching!**

- *Adenoma sebaceum* → Appear in infancy
- *Shagreen patches* → Usually in lumbosacral region
- *Ash-leaf spots* → Appear in childhood
Phakomatoses

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**Tuberous sclerosis**
-- Classic triad is *epiloia*
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**Skin Lesions: Not Matching!**

*Adenoma sebaceum*  ?

*Shagreen patches*  ?

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

- **Adenoma sebaceum**: Raised
- **Shagreen patches**: Flat
- **Ash-leaf spots**: Flat

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

Which lesion(s) is/are hyperpigmented, and which is/are hypopigmented?
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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**Tuberous sclerosis**
--Classic triad is *epiloia*
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**Skin Lesions: Not Matching!**

<table>
<thead>
<tr>
<th>Lesion</th>
<th>Fluorescence</th>
</tr>
</thead>
<tbody>
<tr>
<td>Adenoma sebaceum</td>
<td>?</td>
</tr>
<tr>
<td>Shagreen patches</td>
<td>?</td>
</tr>
<tr>
<td>Ash-leaf spots</td>
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</tbody>
</table>

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

Skin Lesions: Not Matching!

\[
\text{Adenoma sebaceum} \quad \text{Don’t fluoresce} \\
\text{Shagreen patches} \\
\text{Ash-leaf spots} \quad \text{Fluoresce}
\]

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

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

**Skin Lesions: Not Matching!**

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

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--*Peripheral* NF
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- **Shagreen patches**
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**Tuberous sclerosis**
-- Classic triad is *epiloia*
-- Skin: Adenoma sebaceum of face; ash-leaf spots and shagreen patches on torso
-- CNS: Other benign tumors
**Phakomatoses**

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

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

Tuberous sclerosis: Cortical tuber
Phakomatoses

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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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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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*What basic geometric shape do tubers often take?*
Phakomatoses

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A triangle
Phakomatoses

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A benign tumor of the brain

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A triangle

**Which way does the apex of the triangle point?**
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

*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?*
Toward a ventricle
**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

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--Classic triad is *epiloia*
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--CNS: Cortical tubers, other benign tumors
--Benign tumors of *not eye* and *not eye* as well
Phakomatoses

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*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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---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*
Phakomatoses

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--Retinal tumor is something something
Phakomatoses

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--Skin: Adenoma sebaceum of face; ash-leaf spots and shagreen patches on torso
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--Benign tumors of heart and kidney as well
--Retinal tumor is astrocytic hamartoma
Phakomatoses

Tuberous sclerosis: Astrocytic hamartoma
Phakomatoses

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--- CNS: Cortical tubers, other benign tumors
--- Benign tumors of heart and kidney as well
--- Retinal tumors are **astrocytic hamartoma**

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

**NF1**
- *Peripheral* NF
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*By what other name is the astrocytic hamartoma of the retina known?*
Retinal phakoma
Phakomatoses

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--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

---

Yes
---

Yes

---

No
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--- By what other name is the astrocytic hamartoma of the retina known?

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--- What proportion of TS pts develop a phakoma?

   1/3 to 1/2
Phakomatoses

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Can they present bilaterally?
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

**Can they present bilaterally?**
Yes

**Can multiple phakomas be found in one eye?**
Phakomatoses

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**Are they pathognomonic for TS?**
Phakomatoses

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1/3 to 1/2

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Phakomas typically present with one of two appearances—what are they?
-- Smooth, nearly flat, with poorly-defined margins
-- Irregular, elevated, and sharply demarcated
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**By what other name is the astrocytic hamartoma of the retina known?**
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**What proportion of TS pts develop a phakoma?**
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**Can multiple phakomas be found in one eye?**
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The appearance of this lesion-type has been likened to that of a fruit, and a foodstuff. What are they?
--Fruit:
--Foodstuff:
Phakomatoses

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1/3 to 1/2
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The appearance of this lesion-type has been likened to that of a fruit, and a foodstuff. What are they?
--Fruit: ‘Mulberry’
--Foodstuff: Mulberry
**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?
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Can they present bilaterally?
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Can multiple phakomas be found in one eye?
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Are the pathognomonic for TS?
- No

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- Smooth, nearly flat, with poorly-defined margins
- Irregular, elevated, and sharply demarcated

The appearance of this lesion-type has been likened to that of a fruit, and a foodstuff. What are they?
- Fruit: ‘Mulberry’
- Foodstuff: ‘Tapioca’
Phakomatoses

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-- Benign tumors of heart and kidney as well
-- Retinal tumor is astrocytic hamartoma; can appear smooth or lumpy (*mulberry*)

**von Hippel-Lindau**
-- Skin: trick question

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**Phakomatoses**

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

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**von Hippel-Lindau**
- Skin: None! Despite this, is still considered a phakomatosis (and a classic one to boot)
- CNS: Tumor type, classically of tumor location (if absent, is called not von Hippel-Lindau syndrome)
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*)
von Hippel-Lindau: Cerebellar hemangioblastoma
**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*)
-- In multiple organs, including malignancies: 2 different malignancies
Phakomatoses

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

( short for *Pheochromocytoma* )
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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**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; can appear smooth or lumpy (*mulberry*)

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  - Ocular: DFE q1 year
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von Hippel-Lindau: Capillary hemangioblastoma
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von Hippel-Lindau
--Capillary hemangioblastoma
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Can the retinal lesions be present bilaterally?
Yes, in about # of cases
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**von Hippel-Lindau**

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**Capillary hemangioblastoma**; has large feeder/drainage vessels
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**Are all retinal hemangio(blasto)mas associated with vHL?**
Phakomatoses

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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
Phakomatoses

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-- Tumor leaks → abb. → abb. → decreased VA; treat with or

### Abbreviations
- SRF
- ERD
**Phakomatoses**

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- Tumor leaks → SRF → ERD → decreased VA; treat with laser or cryo
  
  *(subretinal fluid)  (exudative retinal detachment)*
Phakomatoses

von Hippel-Lindau: Edema
Phakomatoses

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--Tumor leaks $\rightarrow$ *SRF* $\rightarrow$ *ERD* $\rightarrow$ decreased VA; treat with *laser* or *cryo*
--Management
  --*Ocular*: DFE [frequency]
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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
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° urine for VMA; MRI brain q3 years until age 40; after that, MRI brain q5 years

*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
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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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.
Phakomatoses

NF2

Which is more common, NF1 or NF2?

Central NF

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

**NF2**

*Which is more common, NF1 or NF2?*

NF1 is about 10x more common
Phakomatoses

**NF2**

-- Central NF

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

**Rare:**
- combined hamartoma of retina and RPE;

**Rarer:**
- Lisch nodules
Phakomatoses

**NF2**
--Central NF
Phakomatoses

**NF2**

--- *Central* NF

--- Classic finding: bilateral not eye

--- Eye findings:

- **Common:** PSC/cortical cataracts
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Phakomatoses

NF2
--Central NF
--Classic finding: bilateral acoustic neuromas
Phakomatoses

Tuberous sclerosis: Astrocytic hamartoma. Note large feeder/drainage vessels

14 y.o. with NF2

His 50 y.o. uncle with NF2
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
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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**

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**
--Central NF
--Classic finding: bilateral acoustic neuromas
--Eye findings: Common: **PSC/cortical cataracts**

Are the cataracts visually significant?
Yes
Phakomatoses

NF2
--Central NF
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Are the cataracts visually significant?
Yes

Do they manifest prior to or after the acoustic neuromas?
Phakomatoses

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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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At what age do they become clinically significant?
Phakomatoses

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Usually in the 30s
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Yes

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At what age do they become clinically significant?
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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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--Eye findings: Common: **PSC/cortical cataracts**

**Are the cataracts visually significant?**
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**Do they manifest prior to or after the acoustic neuromas?**
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**Are they unilateral, or bilateral?**
Phakomatoses

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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:* Lisch nodules

*posterior segment*
Phakomatoses

**NF2**

-- *Central* NF

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Phakomatoses

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Phakomatoses

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Phakomatoses

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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.
Phakomatoses

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Do melanocytic lesions occur in NF2 at all?
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
Phakomatoses

**NF2**
--*Central* NF
--Classic finding: bilateral acoustic neuromas
--Eye findings: Common: PSC/cortical cataracts;
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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?
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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OK then, other than acoustic neuromas, what sorts of neuroglial lesions occur in NF2?

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**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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**Neuroglial lesions**
-- Nodular neurofibromas
-- Plexiform neurofibromas
-- Optic glioma
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-- Schwannomas of the SC
-- Meningiomas (intracranial)
-- Ependymomas
Phakomatoses

NF2

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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
--S
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A glioma consisting of ependymal cells

What is an ependymoma?
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OK smart guy, *what are* ependymal cells?

--- **Ependymomas**

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

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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

--- **Neuroglial lesions**
--- Nodular neurofibromas
--- Plexiform neurofibromas
--- Optic glioma
--- Prominent corneal nerves
--- **Schwannomas of the SC**
--- Meningiomas (intracranial)
--- Ependymomas

--- **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

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

By way of a refresher: What is a hamartoma?
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

*By way of a refresher: What is a hamartoma?*
A tumor composed of histologically abnormal cells found in their clinical state.
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

By way of a refresher: What is a hamartoma?
A tumor composed of histologically abnormal cells found in their normal location
Phakomatoses

NF2
--Central NF
--Classic finding: bilateral acoustic neuromas

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

By way of a refresher: What is a hamartoma?
A tumor composed of histologically abnormal cells found in their normal location

Lisch nodules
Phakomatoses

NF2
--Central NF
--Classic finding: bilateral acoustic neuromas

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

By way of a refresher: What is a hamartoma?
A tumor composed of histologically abnormal cells found in their normal location
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

*By way of a refresher: 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?*
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

*By way of a refresher: 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
By way of a refresher: 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?
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

By way of a refresher: 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 retina
By way of a refresher: 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
Phakomatoses

Combined hamartoma of retina and RPE
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

By way of a refresher: 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?
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

By way of a refresher: 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?
Choroidal melanoma--eyes have been enucleated because of this misdiagnosis
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

By way of a refresher: 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?
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

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

*By way of a refresher: 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?*
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

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

---

**Acoustic neuroma**

?  

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

?  

?  

**Corneal decompensation**  

?
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

---

**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
Philomatoses

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

---

**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?
Phakomatoses

B. acoustic neuroma

NF2: Acoustic neuroma
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
Phakomatoses

**NF2**
-- *Central* NF
-- Classic finding: bilateral **acoustic neuromas**
-- Eye findings: *Common*: PSC/cortical cataracts;
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**Sturge-Weber**
-- Classic stigmata is the **port-wine stain**
Phakomatoses

Sturge-Weber: Port-wine stain
Phakomatoses

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

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

---

**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'
Phakomatoses

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

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

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Sturge-Weber
--Classic stigmata is the **port-wine stain**
Phakomatoses

NF2
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Sturge-Weber
--Classic stigmata is the port-wine stain

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

| NF2 |
|-----|---|
| **Central NF** |  |
| --Classic finding: bilateral **acoustic neuromas** |  |
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| Sturge-Weber |
|--------------|---|
| --Classic stigmata is the **port-wine stain** |  |

**In one word, what sort of lesion is the port-wine stain?**
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**All infants with SWS have a port-wine stain. Do all infants with a port-wine stain have SWS?**
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Sturge-Weber
--Classic stigmata is the port-wine stain

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

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’
Phakomatoses

Sturge-Weber: Port-wine stain
Phakomatoses

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

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

---

**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

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It comports to the distribution of one or more divisions of CN5.

---

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

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In one word, what sort of lesion is the port-wine stain?
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CN#?
**Phakomatoses**

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

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

---

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**When does it present?**
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It comports to the distribution of one or more divisions of CN5

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Phakomatoses

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

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**Sturge-Weber**
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Phakomatoses

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

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

By what ‘official’ name is it known?
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Phakomatoses

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--Eye findings: Common: PSC/cortical cataracts; Occasional: Lisch nodules; Rare: Combined hamartoma of retina and RPE

Sturge-Weber
--Classic stigmata is the port-wine stain
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--Classic stigmata is the port-wine stain

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’
Sturge-Weber: Conjunctival hyperemia
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
--Iipsilateral meningeal AVM → symptom/sign
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 AVM → *seizures*
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 AVM → seizures

*Is the meningeal AVM prone to bleeding?*
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 AVM $\rightarrow$ seizures

*Is the meningeal AVM prone to bleeding?*

No
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 AVM → seizures

*How prevalent is seizure activity in SWS?*
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 AVM \(\rightarrow\) seizures

*How prevalent is seizure activity in SWS?*
Very--estimates run as high as 90% of cases
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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**Sturge-Weber**
- Classic stigmata is the **port-wine stain**
- Ipsilateral meningeal AVM → **seizures**
- Classic fundus appearance is due to a lesion (something something something)
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 AVM → *seizures*
-- Classic *tomato catsup* fundus appearance is due to a *diffuse choroidal hemangioma*
Phakomatoses

Sturge-Weber: Tomato catsup fundus OD
**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 AVM → seizures
--Classic **tomato catsup** fundus appearance is due to a **diffuse choroidal hemangioma**

*Diffuse choroidal hemangioma is present in what percent of SWS?*
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 AVM → seizures
--Classic tomato catsup fundus appearance is due to a diffuse choroidal hemangioma

**Diffuse choroidal hemangioma is present in what percent of SWS?**
**About 50%**
**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**

---

**Diffuse choroidal hemangioma is present in what percent of SWS?**

**About 50%**

**Can the choroidal hemangioma be present bilaterally?**

---

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

**Diffuse choroidal hemangioma is present in what percent of SWS?**
About 50%

**Can the choroidal hemangioma be present bilaterally?**
Yes, but it’s uncommon
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 AVM → seizures
--Classic tomato catsup fundus appearance is due to a diffuse choroidal hemangioma

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?
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 AVM → seizures
--Classic *tomato catsup* fundus appearance is due to a *diffuse choroidal hemangioma*

---

**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*
Phakomatoses

**NF2**
--Central NF
--Classic finding: bilateral **acoustic neuromas**
--Eye findings: *Common:* PSC/cortical cataracts;
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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: non-retinal pathology
Phakomatoses

**NF2**
-- *Central* NF
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Sturge-Weber: Note the glaucomatous cupping on the affected side
Phakomatoses

Sturge-Weber: Note the subtle PWS; also the buphthalmos, and enlarged cornea typical of congenital glaucoma
**Phakomatoses**

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*What percent of SWS pts develop glaucoma?*
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*What percent of SWS pts develop glaucoma?*
Estimates run as high as 70
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Is there a relationship between the port-wine stain and risk of glaucoma?
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*What percent of SWS pts develop glaucoma?*
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*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.
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Elevated IOP in SWS stems from three different mechanisms. What are they?
--
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Hint forthcoming…
Phakomatoses

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2° to ocular circulatory anomalies

A noncirculatory anomaly
Phakomatoses

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Yes. If the port-wine stain involves the **eyelid**, the risk is **increased**

**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
Phakomatoses

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**Central NF**

-- Classic finding: bilateral acoustic neuromas

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

**Sturge-Weber**

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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: $\uparrow$ risk of massive choroidal effusion due to abnormal choroidal vasculature

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

Recalling the Goldmann equation for IOP...
Phakomatoses

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

Recalling the Goldmann equation for IOP...

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

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

What percent of SWS pts develop glaucoma? 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

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**Phakomatoses**

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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?
Phakomatoses

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Phakomatoses

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How is Klippel-Trénaunay pronounced?
Phakomatoses

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Are there other associations of note?
Phakomatoses

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Are there other associations of note?
Yes--vascular lesions of the trunk and a single limb, along with marked hypertrophy of that limb
Phakomatoses

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**Which limb is involved?**

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

Is the limb hypertrophy present at birth?
Usually

Are there other associations of note?
Yes—vascular lesions of the trunk and **a single limb**, along with marked hypertrophy of that limb.
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Is the limb hypertrophy present at birth?

Are there other associations of note?
Yes--vascular lesions of the trunk and a single limb, along with marked hypertrophy of that limb
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 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

There is another phakomatosis--less well-known than SWS--that also presents with a port-wine stain. What is it?
Klippel-Trénaunay syndrome (sometimes you'll see Klippel-Trénaunay-Weber syndrome). KTS is the essential rule-out on the DDx for SWS.

Like SWS, is KTS...
--associated with glaucoma? Yes
--nonhereditary? Yes
--associated with meningeal AVMs/seizures? Yes
--associated with diffuse choroidal hemangiomas? No

Which limb is involved?
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?
Yes--vascular lesions of the trunk and a single limb, along with marked hypertrophy of that limb
Phakomatoses

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**Ataxia-telangiectasia**
--Most common cause of **main symptom** in childhood
**Phakomatoses**

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**Ataxia-telangiectasia**
--Most common cause of progressive ataxia in childhood
Phakomatoses

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**Ataxia-telangiectasia**
-- Most common cause of **progressive ataxia** in childhood
-- Only phakomatosis with no abnormalities of the eye part
Phakomatoses

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--*Central* NF
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--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*
Phakomatoses

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**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 \# to \# years
Phakomatoses

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**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**
Phakomatoses

Ataxia-telangiectasia: Conj telangiectasias
Phakomatoses

Ataxia-telangiectasia: Conj telangiectasias
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**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:  
  - EOM problem 1  
  - EOM test  
  - EOM prob 2  
  - EOM prob 3  
  - EOM prob 4
**Phakomatoses**

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-- *Central* NF
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-- Only phakomatosis with no abnormalities of the *fundus*
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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

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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?*  
**Congenital ocular motor apraxia (COMA)**
Phakomatoses

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--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 nonocular system infections → risk of death in teens
Phakomatoses

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-- Abnormal immune function → ↑ susceptibility to respiratory tract infections → risk of death in teens
-- Also have significantly increased risk of leukemia and lymphoma (cause of death in up to ½)
-- Heterozygotes (~2% of population) have increased risk of malignancy as well

*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?

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

*Abnormally high, or low?*
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**

---

**Abnormally high, or low? Low**
**Phakomatoses**

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- Classic finding of conjunctival telangiectasia
- Other eye findings include abnormal saccades with intact doll’s eyes; strabismus; nystagmus
- Abnormal immune function
  - T-cells are abnormal in both function and number
  - Immunoglobulin levels are abnormal
- Abnormal immune function → What aspects of the immune system are abnormal?
  - Immunoglobulin levels are abnormal

**What aspects of the immune system are abnormal?**
- T-cells are abnormal in both function and number
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**Ataxia-telangiectasia**
- What aspects of the immune system are abnormal?
- Immunoglobulin levels are abnormal

**Ataxia-telangiectasia**
- These immunodeficiencies are due in large part to hypoplasia of what immune organ?
Phakomatoses

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The thymus
Phakomatoses

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**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
--Some have intact doll's eyes; strabismus; nystagmus
--Abnormal immune function → ↑ susceptibility to respiratory tract infections → risk of death in teens

What buzzword is used to define the specific sort of RT infection A-T pts are vulnerable to? **respiratory tract infections**
Phakomatoses

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

‘Sinopulmonary’ infections
Phakomatoses

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**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 cancer 1 and cancer 2
Phakomatoses

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*Your A-T pt may have a sinus infection. Should you get a CT to confirm?*
Phakomatoses

**NF2**
--*Central* NF
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**Ataxia-telangiectasia**
--Most common cause of progressive ataxia in childhood
--Only phakomatosis without abnormalities of the fundus
--Classic finding: 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*

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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
Phakomatoses

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--**Central** NF
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--Another classic finding on DFE: Glaucomatous cupping in the ipsilateral ONH only
--Glaucoma surgery: ↑ risk of massive **choroidal effusion** due to abnormal **choroidal vasculature**

*The unfortunate truth of the matter is this:*

--Abnormal immune function → ↑ susceptibility to respiratory tract infections → risk of death in teens
--Also have significantly increased risk of leukemia and lymphoma
**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**
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**The unfortunate truth of the matter is this:**
-- In countries with less-robust healthcare systems (ie, without readily-available antibiotics), A-T pts die of sinopulmonary infections in their teens; whereas

-- Abnormal immune function → ↑ susceptibility to **respiratory tract** infections → risk of death in teens
-- Also have significantly increased risk of leukemia and lymphoma
Phakomatoses

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*The unfortunate truth of the matter is this:*
--- 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).
--- Abnormal immune function → ↑ susceptibility to respiratory tract infections → risk of death in teens
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Phakomatoses

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**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 non-ocular prob as well
Phakomatoses

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Phakomatoses

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

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

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-- Also have significantly increased risk of leukemia and lymphoma
-- Heterozygotes ( ~2 % of population) have increased risk of malignancy as well
-- Skin manifestation: not surprisingly...
**Phakomatoses**

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-- 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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--Skin manifestation: Telangiectasias

**At what age do cutaneous telangiectasias begin to appear?**
3-5 years (ie, at about the same time the conjunctival 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
Phakomatoses

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Ataxia-telangiectasia: Facial telangiectasias
Phakomatoses

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Ataxia-telangiectasia: Telangiectasias
Phakomatoses

**Incontinentia pigmenti**

--Skin normal at birth, but abnormality 1 and abnormality 2 develop by age 1; 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
Phakomatoses

Incontinentia pigmenti: 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

--Eye finding: 1/3 will have peripheral **proliferative retinopathy** that looks just like ROP
Incontinentia pigmenti: ROP-like retinal appearance
Phakomatoses

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How is the peripheral proliferative retinopathy managed?
Incontinentia pigmenti
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---- Eye findings are usually unilateral

**How is the peripheral proliferative retinopathy managed?**
Basically, in the same manner as ROP
**Incontinentia pigmenti**
--Skin normal at birth, but **erythema** and **bullae** develop by **1 week**; only later develops the classic ‘spashed paint’ appearance
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----Eye findings are usually **unilateral**
--2/3 will also have abnormal **mouth issue**
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Phakomatoses

Incontinentia pigmeni: Abnormal dentition
Phakomatoses

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Name two other congenital eye syndromes associated with abnormal dentition:
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--Congenital syphilis
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In three words, what sort of condition is Axenfeld-Reiger?
It is an...
Phakomatoses

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

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Phakomatoses

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What is the eponymous name for abnormal dentition in congenital syphilis?
**Phakomatoses**

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**Hutchinson’s teeth**
Phakomatoses

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_Racemose angioma_
--Characterized by AVM of _eye_ and _brain_
Phakomatoses

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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

Racemose angioma
--Characterized by AVM of eye and brain
**Phakomatoses**

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---In **RA**, are the **AVM** of the eye unilateral or bilateral?
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Racemose angioma
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In RA, are the AVM of the eye unilateral or bilateral?
Unilateral
Phakomatoses

Racemose angioma
Phakomatoses

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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

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Unilateral

**How about the AVM of the brain?**
**Phakomatoses**

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

---

**In basic terms, what is an AVM?**
A direct communication between the arterial and venous sides of the circulation; i.e., without benefit of an intervening capillary bed

**In RA, are the AVM of the eye unilateral or bilateral?**
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**How about the AVM of the brain?**
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Phakomatoses

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Phakomatoses

A, The color fundus photo of the left eye shows the racemose angioma of the retina.
B, The vascular lumen (arrow) was documented with the optical coherence tomography scan.
C, The MRI angiogram of the brain shows the arteriovenous malformation on the left side.

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

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

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Not very--estimates run as low as 5% of cases

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**Phakomatoses**

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