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

Before you begin: There’s a natural break around slide 282; I placed a *break time!* slide at that location.
Phakomatoses are known also as what sort of syndrome?
Phakomatoses are known also as what sort of syndrome?
Neuro-oculocutaneous syndromes
Phakomatoses are known also as what sort of syndrome?
Neuro-oculocutaneous syndromes

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

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

Neuro-oculocutaneous syndromes

In general terms, how do phakomatoses present?

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

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

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

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

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

Are the lesions in phakomatoses predominantly choristomas or hamartomas?

**Most (but not all) are hamartomas (some are choristomas)**

What's the difference between a hamartoma and a choristoma?

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

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A hamartoma is a nest of abnormal cells in a normal location, whereas a choristoma is a nest of relatively-normal cells in an abnormal location.

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

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*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?
Neuro-oculocutaneous syndromes

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

Are the lesions in phakomatoses predominantly choristomas or hamartomas?
Hamartomas

Is there a single, universally accepted definition of the term phakomatosis?
Unfortunately not, and for this reason, the conditions so labelled will vary from source to source
Neurofibromatosis type 1: von Rechlinghausen syndrome

Tuberous sclerosis: Bournville disease

Sturge-Weber syndrome: Encephalotrigeminal angiomatosis

von Hippel-Lindau: Retinal angiomatosis

Incontinentia pigmenti: Bloch-Sulzberger syndrome

Neurofibromatosis type 2: None in common use of which I am aware

Racemose angioma: Wyburn-Mason syndrome

Ataxia-telangiectasia: Louis-Bar syndrome

Abbreviations used henceforth

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

NF1: von Rechlinghausen syndrome

Abbreviations used henceforth

Start here with the other name for NF1
A phakomatosis by any other name...by what other name is each syndrome known?

Abbreviations used henceforth

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

Next for TS
A phakomatosis by any other name…by what other name is each syndrome known?

Abbreviations used henceforth

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

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

1. Neurofibromatosis type 1: von Rechlinghausen syndrome
2. Tuberous sclerosis: Bournville disease
3. Sturge-Weber syndrome:
   - Etc.
A phakomatosis by any other name...by what other name is each syndrome known?

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

**TS**  
- Tuberous sclerosis: Bournville disease

**SWS**  
- Sturge-Weber syndrome: Encephalotrigeminal angiomatosis
  
  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: von Rechlinghausen syndrome
- **TS**: Tuberous sclerosis: Bournville disease
- **SWS**: Sturge-Weber syndrome: Encephalotrigeminal angiomatosis
- **vH-L**: von Hippel-Lindau:
  - 
  - 
  - 

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

Abbreviations used henceforth

- **NF1**: Neurofibromatosis type 1: von Rechlinghausen syndrome
- **TS**: Tuberous sclerosis: Bournville disease
- **SWS**: Sturge-Weber syndrome: Encephalotrigeminal angiomatosis
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Abbreviations used henceforth
A phakomatosis by any other name...by what other name is each syndrome known?

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

Abbreviations used henceforth

- Neurofibromatosis type 1: **von Rechlinghausen syndrome**
- Tuberous sclerosis: **Bournville disease**
- Sturge-Weber syndrome: **Encephalotrigeminal angiomatosis**
- von Hippel-Lindau: **Retinal angiomatosis**
- Incontinentia pigmenti: **Bloch-Sulzberger syndrome**
- Neurofibromatosis type 2:
  - **NF2**
  - **RA**

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

NF1: Neurofibromatosis type 1: von Rechlinghausen syndrome
TS: Tuberous sclerosis: Bournville disease
SWS: Sturge-Weber syndrome: Encephalotrigeminal angiomatosis
vH-L: von Hippel-Lindau: Retinal angiomatosis
IP: Incontinentia pigmenti: Bloch-Sulzberger syndrome
NF2: Neurofibromatosis type 2: MISME syndrome
A phakomatosis by any other name...by what other name is each syndrome known?

- Neurofibromatosis type 1: von Rechlinghausen syndrome
- Tuberous sclerosis: Bournville disease
- Sturge-Weber syndrome: Encephalotrigeminal angiomatosis
- von Hippel-Lindau: Retinal angiomatosis
- Incontinentia pigmenti: Bloch-Sulzberger syndrome
- Neurofibromatosis type 2: MISME syndrome
- Racemose angioma: Wyburn-Mason syndrome
- Ataxia-telangiectasia: Louis-Bar 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?

- Neurofibromatosis type 1: von Rechlinghausen syndrome
- Tuberous sclerosis: Bournville disease
- Sturge-Weber syndrome: Encephalotrigeminal angiomatosis
- von Hippel-Lindau: Retinal angiomatosis
- Incontinentia pigmenti: Bloch-Sulzberger syndrome
- Neurofibromatosis type 2: **MISME syndrome**

**Abbreviations used henceforth**

**Abbreviations used henceforth**

MISME is an acronym. What does it stand for?

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

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

- **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)
- **AT**: Ataxia-telangiectasia (Louis-Bar syndrome)

A phakomatosis by any other name...by what other name is each syndrome known?
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- Neurofibromatosis type 1: von Rechlinghausen syndrome
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- von Hippel-Lindau: Retinal angiomatosis
- Neurofibromatosis type 2: MISME syndrome
- Racemose angioma: Wyburn-Mason syndrome
- Ataxia-telangiectasia: Louis-Bar syndrome
Phakomatoses: Inheritance patterns

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

- Ataxia-telangiectasia: \textit{X-linked dominant}
- Incontinentia pigmenti: sporadic/nonhereditary
- Sturge-Weber: Racemose angioma
Phakomatoses: Inheritance patterns

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

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

- This one is **AR**...
  - ?

- Incontinentia pigmenti

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

- These four are **AD**...
  - NF2
  - NF1
  - von Hippel-Lindau
  - Tuberous sclerosis
- This one is **AR**...
  - Ataxia-telangiectasia

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

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

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

- This one is **X-linked dominant**...
  - ?

- and these two are sporadic/nonhereditary
  - Sturge-Weber
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Phakomatoses: Inheritance patterns

- These four are *AD*...
  - NF2
  - NF1
  - von Hippel-Lindau
  - Tuberous sclerosis
- This one is *AR*...
  - Ataxia-telangiectasia
- This one is *X-linked dominant*...
  - Incontinentia pigmenti
Phakomatoses: Inheritance patterns

- These four are AD…
  - NF2
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- This one is AR…
  - Ataxia-telangiectasia
- This one is X-linked dominant…
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And these two are sporadic/nonhereditary
- ?
- ?
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- These four are **AD**
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  - Sturge-Weber
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Phakomatoses: Inheritance patterns

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

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

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

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

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

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

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

What does *X-linked dominant* transmission mean? It means the condition manifests in every conception possessing at least one X chromosome (ie, everyone). But almost all IP pts are female. If IP is X-linked dominant, why don't male infants present with it? The mutation causing IP is lethal to males in utero. That's about as 'manifest' as it gets.
Phakomatoses: Inheritance patterns

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

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

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

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

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

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

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

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

These four are AD...

- NF2
- NF1
- von Hippel-Lindau
- Tuberous sclerosis

This one is AR...

- Ataxia-telangiectasia

This one is X-linked dominant...

- Incontinentia pigmenti

And these two are sporadic/nonhereditary

- Sturge-Weber
- Racemose angioma

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**...

- Incontinentia pigmenti

And these two are **sporadic/nonhereditary**

- Sturge-Weber
- Racemose angioma
Phakomatoses: Inheritance patterns

These four are AD...

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- 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

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:

--?

--?

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.

There are two ways by which a male child could be liveborn with IP:

- the (phenotypically) male child possesses two X chromosomes (eg, Klinefelter syndrome, XXY) and is therefore heterozygous for IP; or
Phakomatoses: Inheritance patterns

- These four are AD...

- NF2
- NF1
- von Hippel-Lindau
- Tuberous sclerosis

- This one is X-linked dominant...

- Incontinentia pigmenti

- And these two are sporadic/nonhereditary
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  - Racemose angioma

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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.

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--?
Phakomatoses: Inheritance patterns

These four are AD...

- NF2
- NF1
- von Hippel-Lindau
- Tuberous sclerosis

This one is AR...

- Ataxia-telangiectasia

This one is X-linked dominant...

- Incontinentia pigmenti

And these two are sporadic/nonhereditary

- Sturge-Weber
- Racemose angioma

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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’

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.

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--If the (phenotypically) male child possesses two X chromosomes (eg, Klinefelter syndrome, XXY) and is therefore heterozygous for IP; or
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The mutation causing IP is lethal to males in utero. That’s about as ‘manifest’ as it gets.
Phakomatoses: Inheritance patterns

- NF1
- NF2
- Tuberous sclerosis

- NF1
- Ataxia-telangiectasia

- von Hippel-Lindau
- Incontinentia pigmenti

- Sturge-Weber
- Racemose angioma

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

- Incontinentia pigmenti

And these two are **sporadic/nonhereditary**

- Sturge-Weber
- Racemose angioma

---

‘An x-linked recessive condition, lethal in genetically typical males such that all pts are female except for Klinefelter males.’ There is only **one other condition** in the BCSC that fits this description—what is it?

**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

- NF2
- NF1
- von Hippel-Lindau
- Tuberous sclerosis

This one is AD...

- Ataxia-telangiectasia
- Incontinentia pigmenti
- These two are sporadic/nonhereditary
- Sturge-Weber
- Racemose angioma

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.

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 (e.g., 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.

An X-linked recessive condition, lethal in genetically typical males such that all pts are female except for Klinefelter males. There is only one other condition in the BCSC that fits this description—what is it?

Aicardi syndrome

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

- These four are AD...
  - NF2
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  - Tuberous sclerosis

- This one is AR...
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- CNS issue #1
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CNS issue #2 (two words)
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Aicardi syndrome has its own slide-set (FELT20)—see it for more on this condition

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Why is the sporadic-occurrence rate of A-T essentially zero? Because it is AR, it can occur sporadically only if someone heterozygous for it happens to suffer a mutation of the other copy of the responsible gene—an unlikely event.
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Why is the sporadic-occurrence rate of A-T essentially zero? Because it is AR, it can occur sporadically only if someone heterozygous for it happens to suffer a mutation of the other copy of the responsible gene—an unlikely event.
Phakomatoses

**NF1**

---

**NF**

中央 vs. 外周
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
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How are these cell lines related embryologically? Both derive from neural-crest cells
Phakomatoses

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

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

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NCCs are a subtype of embryo cell type
Phakomatoses

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Briefly, what’s the backstory on neural crest cells—what are they, how do they develop? NCCs are a subtype of neuroectodermal cells. Early in embryogenesis, some of the neuroectodermal cells located along the dorsal aspect of the ventral structure (two words) are induced to transition into NCCs.
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Melanocytic Eyelid and Epibulbar Lesions

Neural crest cells…
Melanocytic Eyelid and Epibulbar Lesions

Neural crest cells… and their derivatives
Phakomatoses

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

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

something of something
Phakomatoses

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

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Name four common NF1 lesions that derive from melanocytes

Melanocytic lesions
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Phakomatoses

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- Axillary/inguinal freckles
- Lisch nodules
- Choroidal lesions
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Phakomatosis

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Phakomatoses

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Phakomatoses

NF1: Neuroglial lesions

Plexiform neurofibroma

Optic nerve glioma

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

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

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**What is the lifetime risk of such a transformation?**
About 10%
Phakomatoses

NF1 - Peripheral NF

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What does ‘most’ mean in this context?

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

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

- Leukemia
- Rhabdomyosarcoma
- Pheochromocytoma
- Wilms tumor
Phakomatoses

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--- classic lid finding
--- less classic iris finding
**Phakomatoses**

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Phakomatoses

Plexiform neurofibroma

Ectropion uveae

NF1
Phakomatoses

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*How does a plexiform fibroma and/or iris ectropion cause 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.
Phakomatoses

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

About 50
Phakomatoses

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

What rule of thumb adheres regarding the appearance of Lisch nodules? Lisch nodules are lighter than the rest of the iris when the iris in question is dark, but darker than the rest when the iris is light.
Phakomatoses

Darker on light iris

Lighter on dark iris

NF1: Lisch nodules
Phakomatoses

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

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Iris mammillations

OK, then what are iris mammillations?
Tiny pigmented nodules which, when present, are found in vast numbers diffusely scattered across the iris surface

Are they unilateral, or bilateral?
Usually unilateral, but bilaterality occurs frequently enough that it can’t be used to rule them out

Are they associated with NF1?
Yes (albeit not nearly as strongly as Lisch nodules)
Phakomatoses

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Mammillations? Aren’t those a CNS thingamajig?

You’re thinking of the mammillary bodies, paired structures that are part of the limbic system.
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OK, then what are iris mammillations?

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

Are they unilateral, or bilateral?

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

Are they associated with NF1?

Yes (albeit not nearly as strongly as Lisch nodules)
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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**Rule of thumb for Lisch nodule prevalence:** Age in years x 10

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

*Iris mammillations* which, when present, are found in vast numbers diffusely scattered across the iris surface

**OK, then what are iris mammillations?**

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Are they associated with NF1?

Yes (albeit not nearly as strongly as Lisch nodules)

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

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

In contrast, iris mammillations are always the same color as the rest of the iris.
Phakomatoses

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OK, then what are iris mammillations?

Tiny pigmented nodules which, when present, are found in large numbers diffusely scattered across the iris surface. Are they unilateral, or bilateral? Usually unilateral, but bilaterality occurs frequently enough that it can’t be used to rule them out.

Are they **associated with NF1**? Yes (albeit not nearly as strongly as Lisch nodules)

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Are they associated with NF1?

Yes (albeit not nearly as strongly as Lisch nodules)
Phakomatoses

Lisch nodules darker on light iris

Mammillations same color as iris

Lisch nodules vs iris mammillations
Phakomatoses

NF1
-- Peripheral NF
-- Most lesions due to abnormal melanocytes or neuroglial cells
-- Glaucoma associated with ipsilateral upper lid 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

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

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

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

How does JXG usually present? (Hint: It's not ophthalmic)
As orange skin papules
Phakomatoses

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In three words, what sort of condition is it?
It is a…nonneoplastic histiocytic proliferation
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The presence of…?
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Phakomatoses

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JXG: Foamy macrophages (and a Touton giant cell)
Phakomatoses

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**JXG nodules**

*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…Touton giant cells
The presence of…'foamy macrophages’

*At what age does JXG present?*

---

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

As orange skin papules
NF1
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Phakomatoses

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Phakomatoses

NF1: Optic nerve gliomas bilaterally. Note the ‘kinked’ appearance
Phakomatoses

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What percent of NF1 pts develop a glioma of the optic pathway (ie, nerve or chiasm)?
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Of those, about how many will be symptomatic?
Phakomatoses

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*What about NF1 pts with chiasmal gliomas—do they fare better than their non-NF1 counterparts?*
Phakomatoses

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-- Rule of thumb for Lisch nodule prevalence: \( \text{Age in years} \times 10 \text{ years} \)
Phakomatoses

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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
--A
Phakomatoses

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

What does **epiloia** stand for?
--**Epi**lepsy
--**Low** intelligence
--**Angiomas**
Phakomatoses

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

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

*What is the eponymous name of this triad?*
Phakomatoses

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

*What does epiloia stand for?*
--*Epi*lepsy
--*Lo*w *intelligence*
--*Angiomas*

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

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

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

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

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

What % of TS pts have seizures?
Phakomatoses

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

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

*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**: 80%
  --- **LOW INTELLIGENCE**: ?
  --- **ANGIOMAS**

--- *What % of TS pts have cognitive impairment?*

--- *Vogt’s triad*

--- What is *the eponymous name of this triad?*
Phakomatoses

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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

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

What % of TS pts have cognitive impairment?
Phakomatoses

**NF1**
-- *Peripheral NF*
-- Most lesions due to abnormal melanocytes or neuroglial cells
-- Glaucoma associated with ipsilateral upper-lid plexiform fibroma and/or iris ectropion
-- Iris lesions include Lisch nodules, JXG nodules, and congenital ectropion
-- Optic nerve glioma: Always symptomatic by age 10 years. Classic CT appearance: Kinked ON
-- Rule of thumb for Lisch nodule prevalence: Age in years x 10

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

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

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

**Vogt’s triad**
NF1
--Peripheral NF
--Most lesions due to abnormal melanocytes or neuroglial cells
--Glaucoma associated with ipsilateral upper-lid plexiform fibroma and/or iris ectropion
--Iris lesions include Lisch nodules, JXG nodules, and congenital ectropion
--Optic nerve glioma: Always symptomatic by age 10 years. Classic CT appearance: Kinked ON
--Rule of thumb for Lisch nodule prevalence: Age in years x 10

Tuberous sclerosis
--Classic triad is *epiloia*

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

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

**NF1**
--Peripheral NF
--Most lesions due to abnormal melanocytes or neuroglial cells
--Glaucoma associated with ipsilateral upper-lid plexiform fibroma and/or iris ectropion
--Iris lesions include Lisch nodules, JXG nodules, and congenital ectropion
--Optic nerve glioma: Always symptomatic by age 10 years. Classic CT appearance: Kinked ON
--Rule of thumb for Lisch nodule prevalence: Age in years x 10

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

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

*What % of TS pts have all three? Vogt’s triad*
Phakomatoses

**NF1**
--Peripheral NF
--Most lesions due to abnormal melanocytes or neuroglial cells
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--Rule of thumb for Lisch nodule prevalence: Age in years x 10

**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**

--- *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

--- 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*) classic finding
Phakomatoses

**NF1**

-- *Peripheral NF*
-- Most lesions due to abnormal melanocytes or neuroglial cells
-- Glaucoma associated with ipsilateral upper-lid plexiform fibroma and/or iris ectropion
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**Tuberous sclerosis**

-- Classic triad is *epiloia*
-- Skin: Adenoma sebaceum of face
Phakomatoses

Tuberous sclerosis: Adenoma sebaceum
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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-- Rule of thumb for Lisch nodule prevalence: Age in years x 10

**Tuberous sclerosis**

-- Classic triad is *epiloia*
-- Skin: Adenoma sebaceum of face; ditto and ditto 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)
Phakomatoses

**NF1**
--*Peripheral* NF
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--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
Tuberous sclerosis: Ash leaf spots
Phakomatoses

Tuberous sclerosis: Shagreen patch
Phakomatoses

**NF1**
- *Peripheral NF*
- Most lesions due to abnormal melanocytes or neuroglial cells
- Glaucoma associated with ipsilateral upper-lid plexiform fibroma and/or iris ectropion
- Iris lesions include Lisch nodules, JXG nodules, and congenital ectropion
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- Rule of thumb for Lisch nodule prevalence: Age in years x 10

**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
- **Ash-leaf spots**
  - Appear in childhood
- **Shagreen patches**
  - Usually in lumbosacral region
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: Matching!**

- Adenoma sebaceum
- Shagreen patches
- Ash-leaf spots

- Appear in infancy
- Usually in lumbosacral region
- 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: Not Matching!**

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

*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

--- 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*)

--- **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
--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

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
--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

---

**Skin Lesions: Not Matching!**

- **Adenoma sebaceum**
  - Hyperpigmented

- **Shagreen patches**
  - Hypopigmented

- **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
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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
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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>
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<td>?</td>
</tr>
<tr>
<td>Ash-leaf spots</td>
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Which lesion(s) fluoresce under a Woods lamp, and which do/does not?
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

**Skin Lesions: Not Matching!**

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

*Which lesion(s) fluoresce under a Woods lamp, and which do/does not?*
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**
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**Skin Lesions: Not Matching!**

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*Which lesion(s) is/are considered pathognomonic for TS, and which is/are not?*
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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-- Rule of thumb for Lisch nodule prevalence: Age in years x 10

**Tuberous sclerosis**

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

---

**Skin Lesions: Not Matching!**

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

---

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

**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*

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--CNS: Cortical tubers, other benign tumors
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
--- 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
Phakomatoses

**NF1**
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**Tuberous sclerosis**
-- Classic triad is *epiloia*
-- Skin: Adenoma sebaceum of face; ash-leaf spots and shagreen patches on torso
-- CNS: **Cortical tubers**, other benign tumors

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

Tuberous sclerosis: Cortical tuber
Phakomatoses

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

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

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Toward a ventricle
Phakomatoses

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**Tuberous sclerosis**
--Classic triad is *epioloia*
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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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--*Peripheral* NF
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--- *Tuberous sclerosis*

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--CNS: Cortical tubers, other benign tumors

--Benign tumors of heart and kidney as well

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

**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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--CNS: Cortical tubers, other benign tumors

---Benign tumors of heart and kidney as well

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

The **kidney** tumors are not associated with an increased risk of morbidity/mortality, whereas the **heart** tumors are
Phakomatoses

**NF1**
---Peripheral NF
---Most lesions due to abnormal melanocytes or neuroglial cells
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**Tuberous sclerosis**
---Classic triad is epiloia
---Skin: Adenoma sebaceum of face; ash-leaf spots and shagreen patches on torso
---CNS: Cortical tubers, other benign tumors
---Benign tumors of heart and kidney as well
---Retinal tumor is

*something 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
--Iris lesions include Lisch nodules, JXG nodules, and congenital ectropion
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--Rule of thumb for Lisch nodule prevalence: Age in years x 10

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Phakomatoses

Tuberous sclerosis: Astrocytic hamartoma
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?  
Yes

Are the pathognomonic for TS?  
No
**Phakomatoses**

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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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Phakomatoses

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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: ?

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

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

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--Smooth, nearly flat, with poorly-defined margins
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--- The appearance of this lesion-type has been likened to that of a fruit, and a foodstuff.
--- Fruit: ‘Mulberry’
--- Foodstuff: ‘Tapioca’
**Phakomatoses**

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**Tuberous sclerosis**

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

--CNS: Cortical tubers, other benign tumors

--Benign tumors of heart and kidney as well

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

**von Hippel-Lindau**

--Skin: *trick question*

--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, 24h urine for VMA; MRI brain q3 years until age 40; after that, MRI brain q5 years
Phakomatoses

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**Tuberous sclerosis**
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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: Cerebellar hemangioblastoma
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--Finding in multiple organs, including malignancies: 2 different malignancies
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von Hippel-Lindau: Capillary hemangioblastoma.
Note the large feeder/drainage vessels
Phakomatoses

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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

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

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

Capillary hemangioblastoma
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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**By what other name is this lesion known (it’s a subtle change)?**
*Capillary hemangioma* (ie, no ‘-blasto-’)

**Can the retinal lesions be present bilaterally?**
Yes, in about **1/2** of cases

**Can there be multiple lesions in the same eye?**
Yes, these occur in about **1/3** of cases

**Are all retinal hemangio(blasto)mas associated with vHL?**
No, they can be sporadic
Phakomatoses

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-- Tumor leaks $\rightarrow$ SRF $\rightarrow$ ERD $\rightarrow$ decreased VA

**Management**

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

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Phakomatoses

von Hippel-Lindau: Edema
Phakomatoses

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![test 1](#) ![test 2](#)
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What does VMA stand for in this context?

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A metabolic byproduct of eppy/noreppy
What is the purpose of checking for VMA in the urine?
Elevated levels indicate the possible presence of a pheo
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What does VMA stand for in this context? Vanillylmandelic acid
Phakomatoses

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What is vanillylmandelic acid?
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Vanillylmandelic acid (VMA) is a metabolic byproduct of eppy (epinephrine) and noreppy (norepinephrine). It is elevated in conditions such as pheochromocytoma (pheo) and renal-cell carcinoma (renal-cell Ca).
Phakomatoses

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Is vH-L a potentially fatal condition?
Yes
Two components are most likely to result in death. What are they?
The cerebellar hemangioma and the renal carcinoma
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The cerebellar hemangioma??!! I thought that was a benign lesion. How could it be fatal?

The **cerebellar hemangioma** and the renal carcinoma

The cerebellar hemangioma?

The cerebellum is a critical part of the brain responsible for control of movement and balance. A cerebellar hemangioma is a benign tumor, but if it is large or causes significant compression, it can lead to serious complications. The leaky nature of the tumor can lead to subarachnoid hemorrhage, obstructive hydrocephalus, or other complications that can be life-threatening.
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The cerebellar hemangioma??!! I thought that was a benign lesion. How could it be fatal?
It is a benign lesion. However, it is notoriously ‘leaky,’ and the accumulating exudate can lead to compression of vital intracranial structures

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

NF2

Which is more common, NF1 or NF2?

NF1 is about 10x more common
Phakomatoses

NF2

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

- 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**

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

NF2
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--Central NF

Common: PSC/cortical cataracts; Rare: combined hamartoma of retina and RPE; Rarer: Lisch nodules
Phakomatoses

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Phakomatoses

**NF2**

--- *Central* NF

--Classic finding: bilateral *acoustic neuromas*

**Eye findings:**

Common: PSC/cortical cataracts;

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Rarer: Lisch nodules
Phakomatoses

14 y.o. with NF2

His 50 y.o. uncle with NF2

Acoustic neuromas in NF2 (*black arrows*)
Phakomatoses

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

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

NF2
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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**
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What are the three most common symptoms of acoustic neuroma?

1. Reduced hearing
2. Tinnitus
3. Balance issues
Phakomatoses

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What are the three most common symptoms of acoustic neuroma?
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**Are the cataracts visually significant?**

Yes

**Do they manifest prior to or after the acoustic neuromas?**
Usually prior

**At what age do they become clinically significant?**
Usually in the 30s

**Are they unilateral, or bilateral?**
Both presentations are common
Phakomatoses

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

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Yes

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

At what age do they become clinically significant?
Usually in the 30s

Are they unilateral, or bilateral?
Phakomatoses

**NF2**
--Central NF
--Classic finding: bilateral acoustic neuromas
--Eye findings: Common: **PSC/cortical cataracts**

**Are the cataracts visually significant?**
Yes

**Do they manifest prior to or after the acoustic neuromas?**
Usually prior

**At what age do they become clinically significant?**
Usually in the 30s

**Are they unilateral, or bilateral?**
Both presentations are common
Phakomatoses

**NF2**

--**Central** NF

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

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

*Rare*: posterior segment

*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
Phakomatoses

**NF2**

---**Central NF**

--Classic finding: bilateral acoustic neuromas

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

---ant seg: two words
Phakomatoses

NF2
--Central NF
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Phakomatoses

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--**Central** NF
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---

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

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

---

*One key difference between NF1 and NF2 is this:*
In NF1, both melanocytic and neuroglial lesions are common, whereas…
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.
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?
Phakomatoses

**NF2**

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**Melanocytic lesions**

--Café au lait spots

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---

**Neuroglial lesions**

--Nodular neurofibromas

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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

**NF2**

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

--- Neuroglial lesions
--- Nodular neurofibromas
--- Plexiform neurofibromas
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Neuroglial lesions

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Do NF2 pts get peripheral-nerve tumors like NF1 pts?
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OK then, other than acoustic neuromas, what sorts of neuroglial lesions occur in NF2?
Phakomatoses

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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

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

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

**Schwannomas of the SC**

**Meningiomas (intracranial)**

**Ependymomas**

---

**Phakomatoses**

**Central NF**

- Classic finding: bilateral acoustic neuromas
- Eye findings: Common: PSC/cortical cataracts;
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This propensity for manifesting mainly as CNS tumors is why NF2 is referred to as ‘central’ NF

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

Yes, but at much lower rates.
**Phakomatoses**

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

--M
--I
--S
--M
--E
Phakomatoses

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What is an ependymoma?

A glioma consisting of ependymal cells

OK smart guy, what are ependymal cells?
The epithelial-like glial cells that form the inner lining of the cerebral ventricles and the central canal of the spinal cord.
**Phakomatoses**

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

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Do the neuroglial lesions in NF2 carry a risk of malignant transformation? No. Unlike in NF1, malignant transformation of benign lesions in NF2 is almost unheard of.

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

--Ependymomas

Phakomatoses

Neuroglial lesions
--Schwannomas of the SC
--Meningiomas (intracranial)
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Melanocytic lesions
--Café au lait spots
--Axillary/inguinal freckles
--Lisch nodules
--Choroidal lesions

Neuroglial lesions
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By way of a refresher: What is a hamartoma?
Phakomatoses

**NF2**
--Central NF
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--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 opposite 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
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Eye findings:
Common: PSC/cortical cataracts;
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By way of a refresher: What is a hamartoma?
A tumor composed of histologically normal cells found in their normal location.

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 choristoma?
A tumor composed of histologically abnormal cells found in an abnormal 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?
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
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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 location
Phakomatoses

NF2
--Central NF
--Classic finding: bilateral acoustic neuromas
--Eye findings: Common: PSC/cortical cataracts;
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By way of a refresher: What is a hamartoma?
A tumor composed of histologically abnormal cells found in their normal location

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

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

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

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

With what more sinister dz entity is it often confused?
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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.
Combined hamartoma of retina and RPE. Note the entire lesion is above Bruchs
NF2
--Central NF
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\textbf{Acoustic neuroma}

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

\textbf{Corneal decompensation}
Phakomatoses

NF2
--Central NF
--Classic finding: bilateral **acoustic neuromas**
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**Acoustic neuroma**

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

(No question yet—keep going)

**Corneal decompensation**
Phakomatoses

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

--- Acoustic neuroma

--- Bag CN V1

--- ?

--- Corneal decompensation

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

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---

**Acoustic neuroma**

Bag CN V₁

Decreased corneal sensation

Corneal decompensation

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

NF2
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**Acoustic neuroma**

---

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

---

**Decreased corneal sensation**

---

**Corneal decompensation**
Phakomatoses

**NF2**

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**Acoustic neuroma**

- Bag CN V<sub>1</sub>
  - Decreased corneal sensation

**Corneal decompensation**

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

- Bag CN VII
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

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

- Decreased corneal sensation
- Lagophthalmos
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 $V_1$
- Decreased corneal sensation

Bag CN VII
- Lagophthalmos

**Corneal decompensation**

Another eye finding associated with acoustic neuroma is corneal decompensation. By what two mechanisms might this occur?
NF2: Acoustic neuroma. Note its close association with both CN5 and CN7.
Phakomatoses

**NF2**
--Central NF
--Classic finding: bilateral acoustic neuromas
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**Sturge-Weber**
--Classic stigmata is the...
Phakomatoses

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

Sturge-Weber: Port-wine stain
Phakomatoses

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

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

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

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Phakomatoses

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

**By what ‘official’ name is it known?**
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**When does it present?**
At birth

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Sturge-Weber
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Sturge-Weber: Port-wine stain
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Phakomatoses

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

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The conjunctiva. It will have increased vascularity and hyperemia, producing a false impression of ‘pink eye.’
Phakomatoses

Sturge-Weber: Conjunctival hyperemia
Phakomatoses

**Phakomatoses**

**Sturge-Weber**
--Classic stigmata is the *port-wine stain*
--Ipsilateral meningeal AVM \(\rightarrow\) seizures

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

Glaucoma surgery: ↑ risk of massive choroidal effusion due to abnormal choroidal vasculature

**Symptom/Sign**

375
Phakomatoses

**NF2**
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---

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

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**Sturge-Weber**
--Classic stigmata is the **port-wine stain**
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*Is the meningeal AVM prone to bleeding?*
Phakomatoses

NF2
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Sturge-Weber
--Classic stigmata is the port-wine stain
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Is the meningeal AVM prone to bleeding? No
Sturge-Weber
--Classic stigmata is the port-wine stain
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How prevalent is seizure activity in SWS?

Phakomatoses

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How prevalent is seizure activity in SWS?
Very—estimates run as high as \( % \) of cases
Phakomatoses

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**Sturge-Weber**
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Very—estimates run as high as 90% of cases
Phakomatoses

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**Sturge-Weber**
--Classic stigmata is the port-wine stain
--Ipsilateral meningeal AVM $\rightarrow$ seizures
--Classic fundus appearance is due to a lesion (something something something)
Phakomatoses

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

Sturge-Weber: Tomato catsup fundus OD
Phakomatosis

Sturge-Weber
--Classic stigmata is the port-wine stain
--Ipsilateral meningeal AVM $\rightarrow$ seizures
--Classic tomato catsup fundus appearance is due to a diffuse choroidal hemangioma

Diffuse choroidal hemangioma is present in what percent of SWS?

- About 50%
- Yes, but it's uncommon
- No

NF2
--Central NF
--Classic finding: bilateral acoustic neuromas
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**NF2**
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**Diffuse choroidal hemangioma is present in what percent of SWS?**
About 50%

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

**Does the choroidal hemangioma have malignant potential?**
No
Phakomatoses

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

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Phakomatoses

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

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

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*What percent of SWS pts develop glaucoma?*
Phakomatoses

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

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

*Is there a relationship between the port-wine stain and risk of glaucoma?*

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

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Yes. If the port-wine stain involves the **structure**, the risk is **increased** vs. **decreased**
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--?
--?  
*Hints upcoming…*
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Rule of thumb regarding the mechanism of glaucoma and SWS:

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**Rule of thumb regarding the mechanism of glaucoma and SWS:**
--If glaucoma is **evident at birth**, the angle is to blame; but
--If glaucoma doesn’t manifest until **after age 10**, increased EVP is the cause

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Recalling the Goldmann equation for IOP...

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

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Recalling the Goldmann equation for IOP...

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

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

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

**Klippel-Trénaunay syndrome**

--Associated with glaucoma?
  - Yes
--Nonhereditary?
  - Yes
--Associated with meningeal AVMs/seizures?
  - No
--Associated with diffuse choroidal hemangioma?
  - No

OK then, with what is it associated?

Vascular lesions (including PWS) of the trunk and an extremity, along with hypertrophy of the limb
Phakomatoses

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How is Klippel-Trénaunay pronounced?
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Are there other associations of note?
Phakomatoses

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--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 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? **No**
--associated with diffuse choroidal hemangioma? **No**

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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Phakomatoses

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Yes—vascular lesions of the trunk and a single limb, along with marked hypertrophy of that limb

Which limb is involved?
In the vast majority (~90%) of cases, arm vs leg
Phakomatoses

**Sturge-Weber**

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

--Ipsilateral meningeal AVM $\rightarrow$ *seizures*

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--- associated with meningeal AVMs/seizures? No

--- associated with diffuse choroidal hemangioma? No

--- vascular lesions of the trunk and a single limb, along with marked hypertrophy of that limb?

--Which limb is involved?

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

---

**NF2**

--Central NF

--Classic finding: bilateral acoustic neuromas

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Klippel-Trénaunay syndrome
Phakomatoses

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Which limb is involved?
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Is the limb hypertrophy present at birth?
Usually
Phakomatoses

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

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

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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 eye part
Phakomatoses

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

**Phakomatoses**

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

Ataxia-telangiectasia: Conj telangiectasias
Phakomatoses

**Phakomatoses**

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**Sturge-Weber**

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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 telangiectasias typically appear between ages of **3-5 years**
--Other eye findings include **atrophy** with intact ~

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**NF2**

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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?

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- Most common cause of progressive ataxia in childhood
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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). For more on COMA, see slide-set P4
Phakomatoses

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--Classic finding of conjunctival telangiectasia typically appear between ages of 3-5 years
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--Abnormal immune function $\rightarrow$ ↑ susceptibility to nonocular system infections $\rightarrow$ risk of death in teens
Phakomatoses

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--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

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**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?**
--?  
--?

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

**NF2**
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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
Phakomatoses

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**What aspects of the immune system are abnormal?**
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Abnormally high, or low?
Phakomatoses

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**What aspects of the immune system are abnormal?**
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**Abnormally high, or low?** Low
Phakomatoses

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Phakomatoses

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

**Respiratory tract infections**
Phakomatoses

**Sturge-Weber**
--Classic stigmata is the **port-wine stain**
--Ipsilateral meningeal AVM $\rightarrow$ **seizures**
--Classic *tomato catsup* fundus appearance is due to a diffuse choroidal hemangioma
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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

**NF2**
--**Central** NF
--Classic finding: bilateral **acoustic neuromas**
--Eye findings: **Common:** PSC/cortical cataracts;
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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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**The unfortunate truth of the matter is this:**
--Abnormal immune function → ↑ susceptibility to respiratory tract infections → risk of death in teens
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**The unfortunate truth of the matter is this:**
--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
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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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-- Heterozygotes (2% of population) have increased risk of non-ocular prob as well
**Phakomatoses**

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

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| not surprisingly… |
Phakomatoses

**Phakomatoses**

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**Skin manifestation**: Telangiectasias

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---At what age do cutaneous telangiectasias begin to appear?
---3-5 years (ie, at about the same time the conj ones do)
---At what location do they typically appear first?
The malar region of the face
---Do they remain localized to the malar region throughout life?
No, they typically spread across the face and neck, and new ‘crops’ will appear on the limbs
Phakomatoses

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

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**Skin manifestation:** Telangiectasias
Phakomatoses

Ataxia-telangiectasia: Facial telangiectasias
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Ataxia-telangiectasia: Telangiectasias
**Incontinentia pigmenti**

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

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Phakomatoses

*Incontinentia pigmenti*

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

--Eye finding: 1/3 will have peripheral proliferative retinopathy that looks just like ROP

----Eye findings are usually unilateral

--2/3 will also have abnormal dentition
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: \( \frac{1}{3} \) will have peripheral retina problem that looks just like a more common dz
**Phakomatoses**

*Incontinentia pigmenti*

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

-- Eye finding: 1/3 will have peripheral **proliferative retinopathy** that looks just like ROP

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Phakomatoses

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

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

How is the peripheral proliferative retinopathy managed?
**Incontinentia pigmenti**
--Skin normal at birth, but erythema and bullae develop by 1 week; only later develops the classic ‘splashed paint’ appearance
--Eye finding: 1/3 will have **peripheral proliferative retinopathy** that looks **just like ROP**
----Eye findings are usually unilateral

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 ‘splashed paint’ appearance
--Eye finding: 1/3 will have peripheral proliferative retinopathy that looks just like ROP
----Eye findings are usually unilateral
--2/3 will also have abnormal mouth issue
Phakomatoses

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

Incontinentia pigmenti: Abnormal dentition
Phakomatoses

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--Skin normal at birth, but erythema and bullae develop by 1 week; only later develops the classic 'splashed paint' appearance
--Eye finding: 1/3 will have peripheral proliferative retinopathy that looks just like ROP
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*Speaking of eye dentistry: When you learn that a pt has teephus issues, four conditions should spring immediately to mind. One is incontinentia pigmenti; what are the other three?*
--Incontinentia pigmenti
--?
--?
--?
Phakomatoses

**Incontinentia pigmenti**

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

-- Eye finding: 1/3 will have peripheral proliferative retinopathy that looks just like ROP

--- Eye findings are usually unilateral

-- 2/3 will also have abnormal **dentition**

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

-- Incontinentia pigmenti

-- Axenfeld-Reiger

-- Gardner syndrome

-- Congenital syphilis
Phakomatoses

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--Skin normal at birth, but erythema and bullae develop by 1 week; only later develops the classic ‘splashed paint’ appearance
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In three words, what sort of condition is Axenfeld-Reiger?
An…
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--Skin normal at birth, but erythema and bullae develop by 1 week; only later develops the classic 'splashed paint' appearance
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An...anterior-segment dysgenesis

The anterior-segment dysgeneses are divvied into two groups—what are they?
Peripheral dysgeneses and central dysgeneses
Phakomatoses

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

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

--Incontinentia pigmenti
--Axenfeld-Reiger
--Gardner syndrome
--Congenital syphilis

Is Axenfeld-Reiger a peripheral, or central dysgenesis?

In three words, what sort of condition is Axenfeld-Reiger?
An...anterior-segment dysgenesis

The anterior-segment dysgeneses are divvied into two groups—what are they?

Peripheral dysgeneses and central dysgeneses
Incontinentia pigmenti

--Skin normal at birth, but erythema and bullae develop by 1 week; only later develops the classic 'splashed paint' appearance
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--Gardner syndrome
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In three words, what sort of condition is Axenfeld-Reiger?
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The anterior-segment dysgeneses are divvied into two groups—what are they?

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Is Axenfeld-Reiger a peripheral, or central dysgenesis?
Peripheral
**Phakomatoses**

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--Skin normal at birth, but erythema and bullae develop by 1 week; only later develops the classic ‘splashed paint’ appearance
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----Eye findings are usually unilateral
--2/3 will also have abnormal dentition

---Eye findings should spring immediately to mind when you learn a patient has epiphora. Name three conditions that are important to consider.

---Incontinentia pigmenti
---Axenfeld-Reiger
---Gardner syndrome
---Congenital syphilis

**Is Axenfeld-Reiger a peripheral, or central dysgenesis?**
Peripheral

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

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Aniridia
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Peripheral dysgeneses and central dysgeneses

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

Is Axenfeld-Reiger a peripheral, or central dysgenesis?
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Aniridia
Phakomatoses

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Peripheral

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

Anirida, Peters anomaly, Posterior keratoconus

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

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Is Axenfeld-Reiger a peripheral, or central dysgenesis?
Peripheral

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

What features define Axenfeld-Rieger syndrome?
--Posterior embryotoxon with attached iris strands
--?
**Incontinentia pigmenti**  
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--Gardner syndrome  
--Congenital syphilis  

In three words, what sort of condition is Axenfeld-Reiger?  
An…anterior-segment dysgenesis  

The anterior-segment dysgeneses are divvied into two groups—what are they?

**Peripheral dysgeneses** and central dysgeneses

Is Axenfeld-Reiger a peripheral, or central dysgenesis?  
Peripheral

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**What features define Axenfeld-Rieger syndrome?**  
--Posterior embryotoxon  
-- THEME
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Speaking of eye dentistry: When you learn that a pt has teephus issues, four conditions should spring immediately to mind. One is incontinentia pigmenti; what are the other three?

--Incontinentia pigmenti
--Axenfeld-Reiger
--Gardner syndrome
--Congenital syphilis

In three words, what sort of condition is Axenfeld-Reiger?
An…anterior-segment dysgenesis

The anterior-segment dysgeneses are divvied into two groups—what are they?

Peripheral dysgeneses
Central dysgeneses

Is Axenfeld-Reiger a peripheral, or central dysgenesis?
Peripheral

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

Is a posterior embryotoxon always a harbinger of significant pathology?
No; it is found in about 15% of otherwise normal eyes

What is a posterior embryotoxon?
An anteriorly displaced and thickened Schwalbe’s line
Incontinentia pigmenti
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--Gardner syndrome
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In three words, what sort of condition is Axenfeld-Reiger? An…anterior-segment dysgenesis

The anterior-segment dysgeneses are divvied into two groups—what are they?
--Peripheral dysgeneses and central dysgeneses

--Incontinentia pigmenti
--Axenfeld-Reiger
--Gardner syndrome
--Congenital syphilis

Peripheral dysgeneses

Is Axenfeld-Reiger a peripheral, or central dysgenesis? Peripheral

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

Peripheral dysgeneses

Posterior embryotoxon

What is a posterior embryotoxon?
An anteriorly displaced and thickened Schwalbe’s line

Axenfeld-Reiger

Peripheral dysgeneses

Aniridia

Peripheral dysgeneses

Axenfeld-Reiger

Aniridia

Peripheral dysgeneses

Axenfeld-Reiger

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Posterior embryotoxon
Phakomatoses

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--Axenfeld-Reiger
--Gardner syndrome
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What sort of condition is Axenfeld-Reiger?
An anterior-segment dysgenesis

The anterior-segment dysgeneses are divvied into two groups—what are they?
--Peripheral dysgeneses
--Central dysgeneses

Is Axenfeld-Reiger a peripheral, or central dysgenesis?
Peripheral

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

What features define Axenfeld-Rieger syndrome?
--Posterior embryotoxon
--?
--?

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**Phakomatoses**

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An…anterior-segment dysgenesis

The anterior-segment dysgeneses are divvied into two groups—what are they?
--Peripheral dysgeneses
--Central dysgeneses

Axenfeld-Reiger

Is Axenfeld-Reiger a peripheral, or central dysgenesis?
Peripheral

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

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In three words, what sort of condition is Axenfeld-Reiger?
An…anterior-segment dysgenesis

The anterior-segment dysgeneses are divvied into two groups—what are they?
Peripheral and central dysgeneses

--Incontinentia pigmenti
--Axenfeld-Reiger
--Anirida
--Gardner syndrome
--Congenital syphilis

Are Axenfeld-Reiger a peripheral, or central dysgenesis?
Peripheral

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

Is Axenfeld-Reiger an anterior-segment dysgenesis?
Yes

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--Gardner syndrome
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An…anterior-segment dysgenesis

The anterior-segment dysgeneses are divvied into two groups—what are they?
Peripheral dysgeneses and central dysgeneses

--Axenfeld-Reiger
--Aniridia Peters
--Posterior keratoconus

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

Is Axenfeld-Reiger a peripheral, or central dysgenesis?
Peripheral

What features define Axenfeld-Rieger syndrome?
--Posterior embryotoxon with attached

There is one other major peripheral dysgenesis covered in the BCSC—what is it?
Aniridia
Incontinentia pigmenti
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Anirida Peters anomaly
Posterior keratoconus

Is Axenfeld-Reiger a peripheral, or central dysgenesis?
Peripheral

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Aniridia
Abnormal iris strands (2) attached to posterior embryotoxon (1) in A-R Phakomatoses
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**Peripheral dysgeneses** and **central dysgeneses**

Is Axenfeld-Reiger a peripheral, or central dysgenesis?
Peripheral

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

Anirida Peters

What features define Axenfeld-Rieger syndrome?
--Posterior embryotoxon with attached iris strands
--Iris
--?
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--Central dysgeneses

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

Is Axenfeld-Reiger a peripheral, or central dysgenesis?
Peripheral

What features define Axenfeld-Reiger syndrome?
--Posterior embryotoxon with attached iris strands
--Iris hypoplasia
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---Peripheral dysgeneses
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Is Axenfeld-Reiger a peripheral, or central dysgenesis?
Peripheral

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

What features define Axenfeld-Rieger syndrome?
---Posterior embryotoxon with attached iris strands
---Iris hypoplasia
---Angle abnormalities

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

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What is the noneponymous name of this syndrome?

Familial adenomatous polyposis

Is it common, or rare?

Rare

What is the main issue facing these pts? (It's not ophthalmic.)

They develop innumerable colonic polyps at a young age, and are at extremely high risk of developing colon cancer by age 40 or so

Why are we talking about it, ie, what is its ocular involvement?

Pts have CHRPE-like lesions in their retina
Phakomatoses

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

What is the noneponymous name of this syndrome?
Familial adenomatous polyposis

Is it common, or rare?
Rare

What is the main issue facing these pts? (It’s not ophthalmic.)
They develop innumerable colonic polyps at a young age, and are at extremely high risk of developing colon cancer by age 40 or so

Why are we talking about it, ie, what is its ocular involvement?
Pts have CHRPE-like lesions in their retina

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Gardner syndrome
Phakomatoses

Gardner syndrome: Colonic polyps
Phakomatoses

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CHRPE-like lesions of Gardner syndrome

For more on Gardner syndrome, see slide-set P3
Phakomatoses

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

'Hutchinson teeth'

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

'Peg shaped'
Phakomatoses

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Congenital syphilis: Hutchinson teeth

For more on congenital syphilis, see slide-set U16
**Phakomatoses**

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**Racemose angioma**
--Characterized by AVM of **organ 1** and **organ 2**
Racemose angioma
--Characterized by AVM of eye and brain

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In basic terms, what is an AVM?

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

How about the AVM of the brain?

Also unilateral

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

Ipsilateral

Where specifically are the AVM located in RA?

--The eye AVM are usually found in the temporal retina
--The brain AVM are usually in the midbrain
**Phakomatoses**

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-- Brain AVM frequently bleed, leading to hemiparesis and death
-- Retinal AVM don’t leak on FA

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Phakomatoses
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A, FP demonstrates racemose angioma OS
B, The vascular lumen (arrow) is visible on OCT
C, The MRA shows the associated AVM on the left side
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superior?
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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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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!
Racemose angioma
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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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--Characterized by AVM of *eye* and *brain*
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**Racemose angioma**

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

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Racemose angioma
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At what age do RA pts begin to suffer these brain bleeds? Usually at some point from the teen years into their 20s
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What about seizures? How prevalent is seizure activity in RA?

Not very—estimates run as low as 5% of cases
**Phakomatoses**

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--Brain AVM frequently bleed, leading to **hemiparesis** and **death**

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

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

**Incontinentia pigmenti**
- Skin normal at birth, but erythema and bullae develop by 1 week; only later develops the classic ‘splashed paint’ appearance
- Eye finding: 1/3 will have peripheral proliferative retinopathy that looks just like ROP
- Eye findings are usually unilateral
- 2/3 will also have abnormal dentition
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_Does this mean RA pts don't have eye/vision trouble related to their condition?_
Phakomatoses

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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.
**Phakomatoses**

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**Skin finding = ?**

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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?**
**Phakomatoses**

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

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Phakomatoses *aka* neuro-oculocutaneous syndromes: TLDR

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(Next, Version 2)
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