For each statement, identify the associated phakomatoses:

(1) Comes in a central and peripheral variety:

•
•
•
•
•
•
•
•
•

Number of answers:

1
● For each statement, identify the associated phakomatoses

(1) • Comes in a central and peripheral variety: NF
For each statement, identify the associated phakomatoses

(1) Comes in a central and peripheral variety: NF

(2) May present with ataxia

IP: Incontinentia pigmenti  NF: Neurofibromatosis  RA: Racemose angioma
AT: Ataxia-telangiectasia  KTS: Klippel-Trénaunay syndrome
For each statement, identify the associated phakomatoses

(1) Comes in a *central* and *peripheral* variety: NF
(2) May present with ataxia AT, NF2
For each statement, identify the associated phakomatoses

1. Comes in a **central** and **peripheral** variety: NF
2. May present with ataxia AT, NF2
3. Only syndrome without skin findings:

---

**Q**

**TS:** Tuberous sclerosis  **SWS:** Sturge-Weber syndrome  **vH-L:** von Hippel-Lindau
**IP:** Incontinentia pigmenti  **NF:** Neurofibromatosis  **RA:** Racemose angioma
**AT:** Ataxia-telangiectasia  **KTS:** Klippel-Trénaunay syndrome
For each statement, identify the associated phakomatoses:

1. Comes in a **central** and **peripheral** variety: **NF**
2. May present with ataxia **AT, NF2**
3. Only syndrome without skin findings: **vH-L**
For each statement, identify the associated phakomatosis(es)

1. Comes in a *central* and *peripheral* variety: **NF**
2. May present with ataxia **AT**, **NF2**
3. Only syndrome without skin findings: **vH-L**
4. Increased risk of pheo, renal malignancies:
For each statement, identify the associated phakomatoses:

1. Comes in a **central** and **peripheral** variety: **NF**
2. May present with ataxia **AT**, **NF2**
3. Only syndrome without skin findings: **vH-L**
4. Increased risk of pheo, renal malignancies: **NF1**, **vH-L**

**TS**: Tuberous sclerosis  **SWS**: Sturge-Weber syndrome  **vH-L**: von Hippel-Lindau
**IP**: Incontinentia pigmenti  **NF**: Neurofibromatosis  **RA**: Racemose angioma
**AT**: Ataxia-telangiectasia  **KTS**: Klippel-Trénaunay syndrome
• For each statement, identify the associated phakomatoses

  (1) Comes in a central and peripheral variety: NF
  (2) May present with ataxia AT, NF2
  (1) Only syndrome without skin findings: vH-L
  (2) Increased risk of pheo, renal malignancies: NF1, vH-L
  (1) Associated with thymus hypoplasia → T-cell immune system abnormalities:

  IP: Incontinentia pigmenti  NF: Neurofibromatosis  RA: Racemose angioma
  AT: Ataxia-telangiectasia  KTS: Klippel-Trénaunay syndrome
For each statement, identify the associated phakomatosis(es)

1. Comes in a central and peripheral variety: NF
2. May present with ataxia AT, NF2
3. Only syndrome without skin findings: vH-L
4. Increased risk of pheo, renal malignancies: NF1, vH-L
5. Associated with thymus hypoplasia → T-cell immune system abnormalities: AT

IP: Incontinentia pigmenti  NF: Neurofibromatosis  RA: Racemose angioma
AT: Ataxia-telangiectasia  KTS: Klippel-Trénaunay syndrome
For each statement, identify the associated phakomatoses:

1. Comes in a central and peripheral variety: NF
2. May present with ataxia AT, NF2
3. Only syndrome without skin findings: vH-L
4. Increased risk of pheo, renal malignancies: NF1, vH-L
5. Associated with thymus hypoplasia→T-cell immune system abnormalities: AT
6. Manifests in females almost exclusively:
For each statement, identify the associated phakomatoses

1. Comes in a central and peripheral variety: NF
2. May present with ataxia AT, NF2
1. Only syndrome without skin findings: vH-L
2. Increased risk of pheo, renal malignancies: NF1, vH-L
1. Associated with thymus hypoplasia → T-cell immune system abnormalities: AT
1. Manifests in females almost exclusively: IP

A

IP: Incontinentia pigmenti  NF: Neurofibromatosis  RA: Racemose angioma
AT: Ataxia-telangiectasia  KTS: Klippel-Trénaunay syndrome
For each statement, identify the associated phakomatoses:

1. Comes in a central and peripheral variety: NF
2. May present with ataxia AT, NF2
3. Only syndrome without skin findings: vH-L
4. Increased risk of pheo, renal malignancies: NF1, vH-L
5. Associated with thymus hypoplasia → T-cell immune system abnormalities: AT
6. Manifests in females almost exclusively: IP
7. Associated with Lisch nodules:
   - NF1
   - NF2

IP: Incontinentia pigmenti  NF: Neurofibromatosis  RA: Racemose angioma  
AT: Ataxia-telangiectasia  KTS: Klippel-Trénaunay syndrome
For each statement, identify the associated phakomatoses(s)

1. Comes in a central and peripheral variety: NF
2. May present with ataxia AT, NF2
3. Only syndrome without skin findings: vH-L
4. Increased risk of pheo, renal malignancies: NF1, vH-L
5. Associated with thymus hypoplasia \( \rightarrow \) T-cell immune system abnormalities: AT
6. Manifests in females almost exclusively: IP
7. Associated with Lisch nodules: NF1, NF2 (uncommon; not expected)

IP: Incontinentia pigmenti  NF: Neurofibromatosis  RA: Racemose angioma
AT: Ataxia-telangiectasia  KTS: Klippel-Trénaunay syndrome
For each statement, identify the associated phakomatoses:

1. Comes in a central and peripheral variety: NF
2. May present with ataxia AT, NF2
1. Only syndrome without skin findings: vH-L
2. Increased risk of pheo, renal malignancies: NF1, vH-L
1. Associated with thymus hypoplasia → T-cell immune system abnormalities: AT
1. Manifests in females almost exclusively: IP
2. Associated with Lisch nodules: NF1, NF2 (uncommon; not expected)
2. Associated with port-wine stain:
   
   IP: Incontinentia pigmenti  NF: Neurofibromatosis  RA: Racemose angioma
   AT: Ataxia-telangiectasia  KTS Klippel-Trénaunay syndrome
For each statement, identify the associated phakomatosis(es)

1. Comes in a central and peripheral variety: NF
2. May present with ataxia AT, NF2
3. Only syndrome without skin findings: vH-L
4. Increased risk of pheo, renal malignancies: NF1, vH-L
5. Associated with thymus hypoplasia → T-cell immune system abnormalities: AT
6. Manifests in females almost exclusively: IP
7. Associated with Lisch nodules: NF1, NF2 (uncommon; not expected)
8. Associated with port-wine stain: SWS, KTS
For each statement, identify the associated phakomatoses:

1. Comes in a central and peripheral variety: NF
2. May present with ataxia AT, NF2
3. Only syndrome without skin findings: vH-L
4. Increased risk of pheo, renal malignancies: NF1, vH-L
5. Associated with thymus hypoplasia → T-cell immune system abnormalities: AT
6. Manifests in females almost exclusively: IP
7. Associated with Lisch nodules: NF1, NF2 (uncommon; not expected)
8. Associated with port-wine stain: SWS, KTS
9. Sporadic inheritance only:
For each statement, identify the associated phakomatoses

1. Comes in a central and peripheral variety: **NF**
2. May present with ataxia **AT**, **NF2**
3. Only syndrome without skin findings: **vH-L**
4. Increased risk of pheo, renal malignancies: **NF1**, **vH-L**
5. Associated with thymus hypoplasia → T-cell immune system abnormalities: **AT**
6. Manifests in females almost exclusively: **IP**
7. Associated with Lisch nodules: **NF1**, **NF2** (uncommon; not expected)
8. Associated with port-wine stain: **SWS**, **KTS**
9. Sporadic inheritance only: **SWS**, **RA**
For each statement, identify the associated phakomatoses

1. Comes in a central and peripheral variety: NF
2. May present with ataxia AT, NF2
3. Only syndrome without skin findings: vH-L
4. Increased risk of pheo, renal malignancies: NF1, vH-L
5. Associated with thymus hypoplasia → T-cell immune system abnormalities: AT
6. Manifests in females almost exclusively: IP
7. Associated with Lisch nodules: NF1, NF2 (uncommon; not expected)
8. Associated with port-wine stain: SWS, KTS
9. Sporadic inheritance only: SWS, RA
10. Classic skin lesion description is ‘splashed paint:’
For each statement, identify the associated phakomatosis(es)

1. Comes in a central and peripheral variety: \textit{NF}
2. May present with ataxia \textit{AT, NF2}
3. Only syndrome without skin findings: \textit{vH-L}
4. Increased risk of pheo, renal malignancies: \textit{NF1, vH-L}
5. Associated with thymus hypoplasia $\rightarrow$ T-cell immune system abnormalities: \textit{AT}
6. Manifests in females \textit{almost} exclusively: \textit{IP}
7. Associated with Lisch nodules: \textit{NF1, NF2} (uncommon; not expected)
8. Associated with port-wine stain: \textit{SWS, KTS}
9. Sporadic inheritance \textit{only}: \textit{SWS, RA}
10. Classic skin lesion description is ‘splashed paint:’ \textit{IP}
For each statement, identify the associated phakomatoses

1. Comes in a central and peripheral variety: NF
2. May present with ataxia AT, NF2
1. Only syndrome without skin findings: vH-L
2. Increased risk of pheo, renal malignancies: NF1, vH-L
1. Associated with thymus hypoplasia → T-cell immune system abnormalities: AT
1. Manifests in females almost exclusively: IP
2. Associated with Lisch nodules: NF1, NF2 (uncommon; not expected)
2. Associated with port-wine stain: SWS, KTS
2. Sporadic inheritance only: SWS, RA
1. Classic skin lesion description is ‘splashed paint:’ IP
1. Complain of decreased hearing and/or tinnitus:
For each statement, identify the associated phakomatosis(es)

1. Comes in a central and peripheral variety: NF
2. May present with ataxia AT, NF2
3. Only syndrome without skin findings: vH-L
4. Increased risk of pheo, renal malignancies: NF1, vH-L
5. Associated with thymus hypoplasia → T-cell immune system abnormalities: AT
6. Manifests in females almost exclusively: IP
7. Associated with Lisch nodules: NF1, NF2 (uncommon; not expected)
8. Associated with port-wine stain: SWS, KTS
9. Sporadic inheritance only: SWS, RA
10. Classic skin lesion description is ‘splashed paint’: IP
11. Complain of decreased hearing and/or tinnitus: NF2

Number of answers: 22

IP: Incontinentia pigmenti  NF: Neurofibromatosis  RA: Racemose angioma
AT: Ataxia-telangiectasia  KTS: Klippel-Trénaunay syndrome
For each statement, identify the associated phakomatoses

(1) Comes in a central and peripheral variety: NF
(2) May present with ataxia AT, NF2
(1) Only syndrome without skin findings: vH-L
(2) Increased risk of pheo, renal malignancies: NF1, vH-L
(1) Associated with thymus hypoplasia → T-cell immune system abnormalities: AT
(1) Manifests in females almost exclusively: IP
(2) Associated with Lisch nodules: NF1, NF2 (uncommon; not expected)
(2) Associated with port-wine stain: SWS, KTS
(2) Sporadic inheritance only: SWS, RA
(1) Classic skin lesion description is ‘splashed paint:’ IP
(1) Complain of decreased hearing and/or tinnitus: NF2
(2) Strongly associated with seizures:
For each statement, identify the associated phakomatoses

(1) Comes in a central and peripheral variety: NF
(2) May present with ataxia AT, NF2
(1) Only syndrome without skin findings: vH-L
(2) Increased risk of pheo, renal malignancies: NF1, vH-L
(1) Associated with thymus hypoplasia → T-cell immune system abnormalities: AT
(1) Manifests in females almost exclusively: IP
(2) Associated with Lisch nodules: NF1, NF2 (uncommon; not expected)
(2) Associated with port-wine stain: SWS, KTS
(2) Sporadic inheritance only: SWS, RA
(1) Classic skin lesion description is ‘splashed paint:’ IP
(1) Complain of decreased hearing and/or tinnitus: NF2
(2) Strongly associated with seizures: TS, SWS
For each statement, identify the associated phakomatosis(es)

(1) Comes in a central and peripheral variety: **NF**
(2) May present with ataxia **AT, NF2**
(1) Only syndrome without skin findings: **vH-L**
(2) Increased risk of pheo, renal malignancies: **NF1, vH-L**
(1) Associated with thymus hypoplasia→T-cell immune system abnormalities: **AT**
(1) Manifests in females almost exclusively: **IP**
(2) Associated with Lisch nodules: **NF1, NF2** (uncommon; not expected)
(2) Associated with port-wine stain: **SWS, KTS**
(2) Sporadic inheritance only: **SWS, RA**
(1) Classic skin lesion description is ‘splashed paint’: **IP**
(1) Complain of decreased hearing and/or tinnitus: **NF2**
(2) Strongly associated with seizures: **TS, SWS**
(2) Diagnostic criteria includes family history:
For each statement, identify the associated phakomatoses

1. Comes in a central and peripheral variety: NF
2. May present with ataxia AT, NF2
1. Only syndrome without skin findings: vH-L
2. Increased risk of pheo, renal malignancies: NF1, vH-L
1. Associated with thymus hypoplasia → T-cell immune system abnormalities: AT
1. Manifests in females almost exclusively: IP
2. Associated with Lisch nodules: NF1, NF2 (uncommon; not expected)
2. Associated with port-wine stain: SWS, KTS
2. Sporadic inheritance only: SWS, RA
1. Classic skin lesion description is ‘splashed paint’: IP
1. Complain of decreased hearing and/or tinnitus: NF2
2. Strongly associated with seizures: TS, SWS
2. Diagnostic criteria includes family history: NF1, NF2
Keep going…

(2) Associated with intracranial AVM:

- Retinal lesion is an astrocytic hamartoma:
- Only major syndrome without a retinal lesion:
- Associated with intracranial tumors:
- Associated with glaucoma:
- Retinal findings look like ROP:
- Eyelid findings common:
- Has a classic conjunctival finding:
- Useful mnemonic is epiloia:
- 10% of breast Ca patients are heterozygotes for this:
- Associated with cerebellar tumor:
- Retinal lesions can be ‘tapioca’ or ‘mulberry:’
- Classic eye finding is cortical cats or PSC:
- The eye findings are unilateral:

IP: Incontinentia pigmenti  NF: Neurofibromatosis  RA: Racemose angioma
AT: Ataxia-telangiectasia  KTS: Klippel-Trénaunay syndrome
Keep going…

(2) Associated with intracranial AVM: **SWS, RA**

- Associated with intracranial tumors: NF2, NF1, TS, vH-L
- Associated with glaucoma: SWS, NF1, RA
- Retinal lesions can be ‘tapioca’ or ‘mulberry:’ TS
- Classic eye finding is cortical 'cats' or PSC: NF2
- The eye findings are unilateral: RA, SWS

**TS:** Tuberous sclerosis  **SWS:** Sturge-Weber syndrome  **vH-L:** von Hippel-Lindau
**IP:** Incontinentia pigmenti  **NF:** Neurofibromatosis  **RA:** Racemose angioma
**AT:** Ataxia-telangiectasia  **KTS:** Klippel-Trénaunay syndrome
Keep going…

(2) Associated with intracranial AVM: SWS, RA

How do the intracranial AVM differ in SWS vs RA?
- **Keep going…**

(2) Associated with **intracranial AVM**: **SWS, RA**

<table>
<thead>
<tr>
<th>How do the intracranial AVM differ in SWS vs RA?</th>
</tr>
</thead>
<tbody>
<tr>
<td>--AVM are location in SWS but location in RA</td>
</tr>
</tbody>
</table>

**TS**: Tuberous sclerosis  
**SWS**: Sturge-Weber syndrome  
**vH-L**: von Hippel-Lindau  
**IP**: Incontinentia pigmenti  
**NF**: Neurofibromatosis  
**RA**: Racemose angioma  
**AT**: Ataxia-telangiectasia  
**KTS**: Klippel-Trénaunay syndrome
Keep going…

(2) Associated with **intracranial AVM**: SWS, RA

How do the intracranial AVM differ in SWS vs RA?
--AVM are **meningeal** in SWS but **parenchymal** in RA
Keep going…

(2) Associated with intracranial AVM: **SWS, RA**

*How do the intracranial AVM differ in SWS vs RA?*

--- AVM are **meningeal** in SWS but **parenchymal** in RA.

--- They are prone to bleeding in **one**, but not in **other**.
Keep going…

(2) Associated with intracranial AVM: SWS, RA

How do the intracranial AVM differ in SWS vs RA?
--AVM are meningeal in SWS but parenchymal in RA
--They are prone to bleeding in RA, but not in SWS
Keep going…

- Associated with intracranial AVM: **SWS, RA**
- Retinal lesion is an astrocytic hamartoma:
Keep going…

(2) Associated with intracranial AVM: SWS, RA

(1) Retinal lesion is an astrocytic hamartoma: TS

IP: Incontinentia pigmenti  NF: Neurofibromatosis  RA: Racemose angioma
AT: Ataxia-telangiectasia  KTS: Klippel-Trénaunay syndrome
Keep going…

(2) Associated with intracranial AVM: SWS, RA

(1) Retinal lesion is an astrocytic hamartoma: TS

(2) Only ones without a retinal lesion:
Keep going…

(2) Associated with intracranial AVM: SWS, RA
(1) Retinal lesion is an astrocytic hamartoma: TS
(2) Only ones without a retinal lesion: SWS, KTS
Keep going…

(2) Associated with intracranial AVM: SWS, RA
(1) Retinal lesion is an astrocytic hamartoma: TS
(2) Only ones without a retinal lesion: SWS, KTS

Wait a minute--what about the hemangioma in SWS?
- Keep going…

1. Associated with intracranial AVM: SWS, RA
2. Retinal lesion is an astrocytic hamartoma: TS
3. Only ones without a retinal lesion: SWS, KTS

Wait a minute--what about the hemangioma in SWS? The hemangioma is a choroidal, not retinal lesion
Keep going…

- Associated with intracranial AVM: SWS, RA
- Retinal lesion is an astrocytic hamartoma: TS
- Only ones without a retinal lesion: SWS, KTS

**Wait a minute--what about the hemangioma in SWS?**
The hemangioma is a choroidal, not retinal lesion

**Just because SWS pts don’t have a retinal lesion doesn’t mean they don’t have retinal issues.**
*What significant retinal problem can arise secondary to the diffuse choroidal hemangioma?*
Keep going…

(2) Associated with intracranial AVM: SWS, RA

(1) Retinal lesion is an astrocytic hamartoma: TS

(2) Only ones without a retinal lesion: SWS, KTS

Wait a minute--what about the hemangioma in SWS? The hemangioma is a choroidal, not retinal lesion.

Just because SWS pts don’t have a retinal lesion doesn’t mean they don’t have retinal issues. What significant retinal problem can arise secondary to the diffuse choroidal hemangioma? Exudative RD
Keep going…

(2) Associated with intracranial AVM: SWS, RA
(1) Retinal lesion is an astrocytic hamartoma: TS
(2) Only ones without a retinal lesion: SWS, KTS

Wait a minute--what about the hemangioma in SWS?
The hemangioma is a choroidal, not retinal lesion

Just because SWS pts don’t have a retinal lesion doesn’t mean they don’t have retinal issues. What significant retinal problem can arise secondary to the diffuse choroidal hemangioma?
Exudative RD

How common is exudative RD in Sturge-Weber?
Keep going…

- Associated with intracranial AVM: SWS, RA
- Retinal lesion is an astrocytic hamartoma: TS
- Only ones without a retinal lesion: SWS, KTS

Wait a minute--what about the hemangioma in SWS?
The hemangioma is a choroidal, not retinal lesion

Just because SWS pts don’t have a retinal lesion doesn’t mean they don’t have retinal issues. What significant retinal problem can arise secondary to the diffuse choroidal hemangioma?

Exudative RD

How common is exudative RD in Sturge-Weber?
About 50% of patients with a diffuse choroidal hemangioma will develop one
Keep going…

(2) Associated with intracranial AVM: SWS, RA
(1) Retinal lesion is an astrocytic hamartoma: TS
(2) Only ones without a retinal lesion: SWS, KTS
(4) Associated with intracranial tumors:

IP: Incontinentia pigmenti  NF: Neurofibromatosis  RA: Racemose angioma
AT: Ataxia-telangiectasia  KTS: Klippel-Trénaunay syndrome
Keep going…

(2) Associated with intracranial AVM: SWS, RA
(1) Retinal lesion is an astrocytic hamartoma: TS
(2) Only ones without a retinal lesion: SWS, KTS
(4) Associated with intracranial tumors: NF2, NF1, TS, vH-L
Keep going…

(2) Associated with intracranial AVM: SWS, RA
(1) Retinal lesion is an astrocytic hamartoma: TS
(2) Only ones without a retinal lesion: SWS, KTS
(4) Associated with intracranial tumors: NF2, NF1, TS, vH-L
(4) Associated with glaucoma:
Keep going…

1. Associated with intracranial AVM: SWS, RA
2. Retinal lesion is an astrocytic hamartoma: TS
3. Only ones without a retinal lesion: SWS, KTS
4. Associated with intracranial tumors: NF2, NF1, TS, vH-L
5. Associated with glaucoma: SWS, NF1, RA, KTS

Number of answers: 5
Q

- Keep going…

(2) Associated with intracranial AVM: SWS, RA

(1) Retinal lesion is an astrocytic hamartoma: TS

(2) Only ones without a retinal lesion: SWS, KTS

(4) Associated with intracranial tumors: NF2, NF1, TS, vH-L

(4) Associated with glaucoma: SWS, NF1, RA, KTS

What is the glaucoma mechanism for each of these?

--SWS:

--NF1:

--RA:

--KTS:
Keep going…

Associated with intracranial AVM: SWS, RA
Retinal lesion is an astrocytic hamartoma: TS
Only ones without a retinal lesion: SWS, KTS
Associated with intracranial tumors: NF2, NF1, TS, vH-L

Associated with glaucoma: SWS, NF1, RA, KTS

What is the glaucoma mechanism for each of these?
--SWS: ↑ EVP; ↑ ciliary-body perfusion; developmental angle anomalies
--NF1: mechanism unclear
--RA: intraretinal hemorrhage → neo → NVI → NVA → NVG
--KTS: Similar to SWS
Keep going…

(2) Associated with intracranial AVM: SWS, RA
(1) Retinal lesion is an astrocytic hamartoma: TS
(2) Only ones without a retinal lesion: SWS, KTS
(4) Associated with intracranial tumors: NF2, NF1, TS, vH-L
(4) Associated with glaucoma: SWS, NF1, RA, KTS
(1) Retinal findings look like ROP:
Keep going…

(2) Associated with intracranial AVM: SWS, RA

(1) Retinal lesion is an astrocytic hamartoma: TS

(2) Only ones without a retinal lesion: SWS, KTS

(4) Associated with intracranial tumors: NF2, NF1, TS, vH-L

(4) Associated with glaucoma: SWS, NF1, RA, KTS

(1) Retinal findings look like ROP: IP
Keep going...

(2) Associated with intracranial AVM: SWS, RA
(1) Retinal lesion is an astrocytic hamartoma: TS
(2) Only ones without a retinal lesion: SWS, KTS
(4) Associated with intracranial tumors: NF2, NF1, TS, vH-L
(4) Associated with glaucoma: SWS, NF1, RA, KTS
(1) Retinal findings look like ROP: IP
(2) Lid involvement → increased risk of glaucoma:
● Keep going…

(2) Associated with intracranial AVM: SWS, RA
(1) Retinal lesion is an astrocytic hamartoma: TS
(2) Only ones without a retinal lesion: SWS, KTS
(4) Associated with intracranial tumors: NF2, NF1, TS, vH-L
(4) Associated with glaucoma: SWS, NF1, RA, KTS
(1) Retinal findings look like ROP: IP
(2) Lid involvement → increased risk of glaucoma: NF1, SWS

IP: Incontinentia pigmenti  NF: Neurofibromatosis  RA: Racemose angioma
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Keep going…

- Associated with intracranial AVM: SWS, RA
- Retinal lesion is an astrocytic hamartoma: TS
- Only ones without a retinal lesion: SWS, KTS
- Associated with intracranial tumors: NF2, NF1, TS, vH-L
- Associated with glaucoma: SWS, NF1, RA, KTS
- Retinal findings look like ROP: IP
- Lid involvement \( \rightarrow \) increased risk of glaucoma: NF1, SWS
- Has a classic conjunctival finding:
Keep going…

(2) Associated with intracranial AVM: SWS, RA
(1) Retinal lesion is an astrocytic hamartoma: TS
(2) Only ones without a retinal lesion: SWS, KTS
(4) Associated with intracranial tumors: NF2, NF1, TS, vH-L
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(1) Retinal findings look like ROP: IP
(2) Lid involvement → increased risk of glaucoma: NF1, SWS
(2) Has a classic conjunctival finding: AT, SWS
Keep going…

(2) Associated with intracranial AVM: SWS, RA
(1) Retinal lesion is an astrocytic hamartoma: TS
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(1) Retinal findings look like ROP: IP
(2) Lid involvement → increased risk of glaucoma: NF1, SWS
(2) Has a classic conjunctival finding: AT, SWS

What is the conj finding in:
--AT?
--SWS?
Keep going…

(2) Associated with intracranial AVM: SWS, RA

(1) Retinal lesion is an astrocytic hamartoma: TS

(2) Only ones without a retinal lesion: SWS, KTS

(4) Associated with intracranial tumors: NF2, NF1, TS, vH-L

(4) Associated with glaucoma: SWS, NF1, RA, KTS

(1) Retinal findings look like ROP: IP

(2) Lid involvement → increased risk of glaucoma: NF1, SWS

(2) Has a classic conjunctival finding: AT, SWS

What is the conj finding in:
--AT? Telangiectasias
--SWS? Increased vascularity → ’pink eye’ appearance
Keep going…

1. Associated with intracranial AVM: SWS, RA
2. Retinal lesion is an astrocytic hamartoma: TS
3. Only ones without a retinal lesion: SWS, KTS
4. Associated with intracranial tumors: NF2, NF1, TS, vH-L
5. Associated with glaucoma: SWS, NF1, RA, KTS
6. Retinal findings look like ROP: IP
7. Lid involvement → increased risk of glaucoma: NF1, SWS
8. Has a classic conjunctival finding: AT, SWS
9. ‘Classic triad’ mnemonic is epiloia:
Keep going…

(2) Associated with intracranial AVM: SWS, RA
(1) Retinal lesion is an astrocytic hamartoma: TS
(2) Only ones without a retinal lesion: SWS, KTS

(4) Associated with intracranial tumors: NF2, NF1, TS, vH-L
(4) Associated with glaucoma: SWS, NF1, RA, KTS

(1) Retinal findings look like ROP: IP
(2) Lid involvement → increased risk of glaucoma: NF1, SWS
(2) Has a classic conjunctival finding: AT, SWS
(1) ‘Classic triad’ mnemonic is epiloia: TS

Number of answers

59
Keep going…

- Associated with intracranial AVM: SWS, RA
- Retinal lesion is an astrocytic hamartoma: TS
- Only ones without a retinal lesion: SWS, KTS
- Associated with intracranial tumors: NF2, NF1, TS, vH-L
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- Has a classic conjunctival finding: AT, SWS

‘Classic triad’ mnemonic is epiloia: TS

What does epiloia stand for?

- Epi
- Lo
- A

IP: Incontinentia pigmenti  NF: Neurofibromatosis  RA: Racemose angioma
AT: Ataxia-telangiectasia  KTS: Klippel-Trénaunay syndrome
Keep going…

(2) Associated with intracranial AVM: SWS, RA
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What does epiloia stand for?
-- Epilepsy
-- Low intelligence
-- Angiomas
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**Abbreviations:**

- **TS:** Tuberous sclerosis
- **SWS:** Sturge-Weber syndrome
- **vH-L:** von Hippel-Lindau
- **IP:** Incontinentia pigmenti
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