For each statement, identify the associated phakomatosis(es)

(1) Comes in a central and peripheral variety:

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
For each statement, identify the associated phakomatoses

(1) Comes in a central and peripheral variety: NF
(2) May present with ataxia
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:

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
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:

---

**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

**Abbreviations:**
- 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
4. Increased risk of pheo, renal malignancies: NF1, vH-L
5. Associated with thymus hypoplasia → T-cell immune system abnormalities:
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
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:
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

Number of answers: 12

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
   - RA
8. Associated with port-wine stain: SWS
9. Sporadic inheritance: SWS, RA
10. Classic skin lesion description is 'splashed paint:' IP
11. Complain of decreased hearing and/or tinnitus: NF2
12. Associated with seizures: TS, SWS, RA
13. Diagnostic criteria includes family history: NF1, NF2

Number of answers: 13
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)
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:
   - 
   - 
   - 
   - 
   - 

NUM: Number of answers
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
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 pheochromocytoma, 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*:
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
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:’
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:’ IP

Number of answers: 10

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’: IP
11. 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
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
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
12. Strongly associated with seizures:

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
12. Strongly associated with seizures: TS, SWS

- For each statement, identify the associated phakomatosis(es)

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

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 $\rightarrow$ 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**
Presents with hypertrophy of a single limb:

- 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
- Usefulness 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

Number of answers:

1. TS: Tuberous sclerosis  
2. SWS: Sturge-Weber syndrome  
3. vH-L: von Hippel-Lindau  
4. IP: Incontinentia pigmenti  
5. NF: Neurofibromatosis  
6. RA: Racemose angioma  
7. AT: Ataxia-telangiectasia  
8. KTS: Klippel-Trénaunay syndrome  

A

(1) Presents with hypertrophy of a single limb: **KTS**

**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
(1) Presents with hypertrophy of a single limb: KTS

Is the involved limb usually an arm, or a leg?
(1) Presents with hypertrophy of a single limb: KTS

Is the involved limb usually an arm, or a leg? It is almost always (>90%) a leg
(1) Presents with hypertrophy of a single limb: **KTS**

(2) Associated with intracranial AVM:
(1) Presents with hypertrophy of a single limb: KTS
(2) Associated with intracranial AVM: SWS, RA
(1) Presents with hypertrophy of a single limb: KTS
(2) Associated with intracranial AVM: SWS, RA

How do the intracranial AVM differ in SWS vs RA?
--
--
(1) Presents with hypertrophy of a single limb: **KTS**

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

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

--- AVM are **location** in SWS but **location** in RA ---
(1) Presents with hypertrophy of a single limb: KTS
(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
(1) Presents with hypertrophy of a single limb: KTS
(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**
(1) Presents with hypertrophy of a single limb: **KTS**

(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**
(1) Presents with hypertrophy of a single limb: KTS
(2) Associated with intracranial AVM: SWS, RA
(1) Retinal lesion is an astrocytic hamartoma:
(1) Presents with hypertrophy of a single limb: **KTS**
(2) Associated with intracranial AVM: **SWS, RA**
(1) Retinal lesion is an astrocytic hamartoma: **TS**
(1) Presents with hypertrophy of a single limb: **KTS**
(2) Associated with intracranial AVM: **SWS, RA**

(1) Retinal lesion is an astrocytic hamartoma: **TS**
(2) Only ones without a retinal lesion:

**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
(1) Presents with hypertrophy of a single limb: KTS
(2) Associated with intracranial AVM: SWS, RA
(1) Retinal lesion is an astrocytic hamartoma: TS
(2) Only ones without a retinal lesion: SWS, KTS
Q

(1) Presents with hypertrophy of a single limb: KTS
(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
(1) Presents with hypertrophy of a single limb: KTS
(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
(1) Presents with hypertrophy of a single limb: KTS
(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

Is choroidal hemangioma a feature of KTS?
(1) Presents with hypertrophy of a single limb: KTS

(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

Is choroidal hemangioma a feature of KTS? No, and this (along with limb hypertrophy) is one of the key points of differentiation between them
Q

(1) Presents with hypertrophy of a single limb: KTS
(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?
Present with hypertrophy of a single limb: KTS

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
**Q**

(1) Present with hypertrophy of a single limb: KTS

(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?
(1) Presents with hypertrophy of a single limb: KTS
(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?
About 50% of patients with a diffuse choroidal hemangioma will develop one
Q
(1) Presents with hypertrophy of a single limb: KTS
(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:
(1) Presents with hypertrophy of a single limb: **KTS**
(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**
Q

(1) Presents with hypertrophy of a single limb: KTS
(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:

Number of answers
(1) Presents with hypertrophy of a single limb: KTS
(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) Presents with hypertrophy of a single limb: KTS
(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:
IP: Incontinentia pigmenti  NF: Neurofibromatosis  RA: Racemose angioma  
AT: Ataxia-telangiectasia  KTS: Klippel-Trénaunay syndrome

(1) Presents with hypertrophy of a single limb: KTS
(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: ↑ EVP; ↑ ciliary-body perfusion; developmental angle anomalies
--NF1: mechanism unclear
--RA: intraretinal hemorrhage → neo → NVI → NVA → NVG
--KTS: Similar to SWS
(1) Presents with hypertrophy of a single limb: KTS
(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:
<table>
<thead>
<tr>
<th>Number of answers</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Presents with hypertrophy of a single limb: KTS</td>
</tr>
<tr>
<td>2</td>
<td>Associated with intracranial AVM: SWS, RA</td>
</tr>
<tr>
<td>1</td>
<td>Retinal lesion is an astrocytic hamartoma: TS</td>
</tr>
<tr>
<td>2</td>
<td>Only ones without a retinal lesion: SWS, KTS</td>
</tr>
<tr>
<td>4</td>
<td>Associated with intracranial tumors: NF2, NF1, TS, vH-L</td>
</tr>
<tr>
<td>4</td>
<td>Associated with glaucoma: SWS, NF1, RA, KTS</td>
</tr>
<tr>
<td>1</td>
<td>Retinal findings look like ROP: IP</td>
</tr>
</tbody>
</table>

IP: Incontinentia pigmenti  NF: Neurofibromatosis  RA: Racemose angioma
AT: Ataxia-telangiectasia  KTS: Klippel-Trénaunay syndrome
(1) Presents with hypertrophy of a single limb: **KTS**
(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:
**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

(1) Presents with hypertrophy of a single limb: **KTS**
(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**
(1) Presents with hypertrophy of a single limb: KTS
(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:

\[\text{Number of answers} = 60\]

**Abbreviations:**
- **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
(1) Presents with hypertrophy of a single limb: KTS
(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
Q

(1) Presents with hypertrophy of a single limb: KTS
(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

Number of answers: 62

What is the conj finding in:
--AT?
--SWS?

IP: Incontinentia pigmenti  NF: Neurofibromatosis  RA: Racemose angioma
AT: Ataxia-telangiectasia  KTS: Klippel-Trénaunay syndrome
(1) Presents with hypertrophy of a single limb: KTS
(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

Number of answers
(2)

Has a classic conjunctival finding: AT, SWS

What is the conj finding in:
--AT? Telangiectasias
--SWS? Increased vascularity → ‘pink eye’ appearance
(1) Presents with hypertrophy of a single limb: KTS
(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:
(1) Presents with hypertrophy of a single limb: **KTS**

(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**
Presents with hypertrophy of a single limb: KTS
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 → increased risk of glaucoma: NF1, SWS
Has a classic conjunctivval finding: AT, SWS
‘Classic triad’ mnemonic is epiloia: TS

What does epiloia stand for?

--

--

--

---

---
Q/A

(1) Presents with hypertrophy of a single limb: KTS
(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

‘Classic triad’ mnemonic is epiloia: TS

What does epiloia stand for?
--Epi
--Lo i
--A
(1) Presents with hypertrophy of a single limb: KTS
(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

What does epiloia stand for?
--Epilepsy
--Low intelligence
--Angiomas
Presents with hypertrophy of a single limb: KTS
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 → increased risk of glaucoma: NF1, SWS
Has a classic conjunctival finding: AT, SWS
‘Classic triad’ mnemonic is epiloia: TS
Many breast Ca patients are heterozygotes for this:

IP: Incontinentia pigmenti  NF: Neurofibromatosis  RA: Racemose angioma  
AT: Ataxia-telangiectasia  KTS: Klippel-Trénaunay syndrome
(1) Presents with hypertrophy of a single limb: KTS
(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
(1) Many breast Ca patients are heterozygotes for this: AT
Q

1. Presents with hypertrophy of a single limb: KTS
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
1. Many breast Ca patients are heterozygotes for this: AT
1. Associated with cerebellar tumor:
   •
   •
   •
A

(1) Presents with hypertrophy of a single limb: KTS
(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
(1) Many breast Ca patients are heterozygotes for this: AT
(1) Associated with cerebellar tumor: vH-L

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

(1) Presents with hypertrophy of a single limb: KTS
(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
(1) Many breast Ca patients are heterozygotes for this: AT
(1) Associated with cerebellar tumor: vH-L
(1) Retinal lesions can be ‘smooth’ or ‘tapioca/mulberry’:

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

1. Presents with hypertrophy of a single limb: **KTS**
2. Associated with intracranial AVM: **SWS, RA**
3. Retinal lesion is an astrocytic hamartoma: **TS**
4. 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**
1. Many breast Ca patients are heterozygotes for this: **AT**
1. Associated with cerebellar tumor: **vH-L**
1. Retinal lesions can be ‘smooth’ or ‘tapioca/mulberry’: **TS**
(1) Presents with hypertrophy of a single limb: KTS

(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

(1) Many breast Ca patients are heterozygotes for this: AT

(1) Associated with cerebellar tumor: vH-L

(1) Retinal lesions can be ‘smooth’ or ‘tapioca/mulberry’: TS

(1) Classic eye finding is cortical cats or PSC:
(1) Presents with hypertrophy of a single limb: KTS
(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
(1) Many breast Ca patients are heterozygotes for this: AT
(1) Associated with cerebellar tumor: vH-L
(1) Retinal lesions can be ‘smooth’ or ‘tapioca/mulberry’: TS
(1) Classic eye finding is cortical cats or PSC: NF2
(1) Presents with hypertrophy of a single limb: **KTS**
(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**
(1) Many breast Ca patients are heterozygotes for this: **AT**
(1) Associated with cerebellar tumor: **vH-L**
(1) Retinal lesions can be ‘smooth’ or ‘tapioca/mulberry’: **TS**
(1) Classic eye finding is cortical cats or PSC: **NF2**
(2) The eye findings are unilateral:
- Presents with hypertrophy of a single limb: KTS
- 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 → increased risk of glaucoma: NF1, SWS
- Has a classic conjunctival finding: AT, SWS
- ‘Classic triad’ mnemonic is epiloia: TS
- Many breast Ca patients are heterozygotes for this: AT
- Associated with cerebellar tumor: vH-L
- Retinal lesions can be ‘smooth’ or ‘tapioca/mulberry’: TS
- Classic eye finding is cortical cats or PSC: NF2
- The eye findings are unilateral: RA, SWS