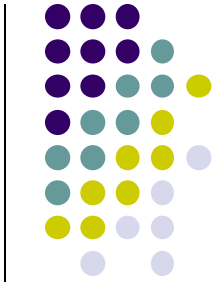
Q

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 phakomatosis(es)*

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

Number of answers

A

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

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

Number of answers

Q

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

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

(2) May present with ataxia

Number of answers

A

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

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

(2) May present with ataxia AT, NF2

Number of answers

Q

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- *For each statement, identify the associated phakomatosis(es)*
  - (1) Comes in a *central* and *peripheral* variety: NF
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  - (1) Only syndrome without skin findings:

Number of answers

A

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Number of answers

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Number of answers

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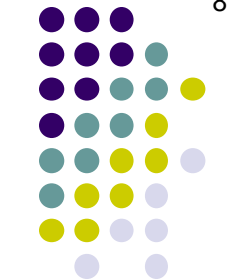

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Number of answers

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Number of answers

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(1) Associated with thymus hypoplasia→T-cell immune system abnormalities:

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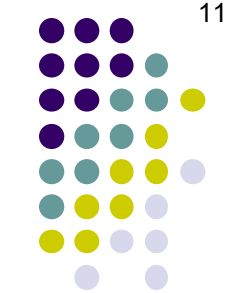

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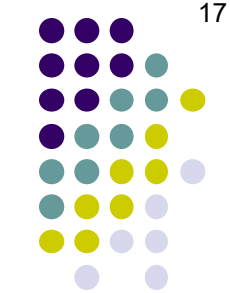

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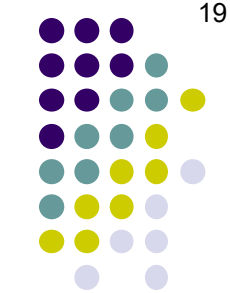

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(2) Sporadic inheritance *only*: SWS, RA
(1) Classic skin lesion description is 'splashed paint:' IP
(1) Complain of decreased hearing and/or tinnitus:

A

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Q

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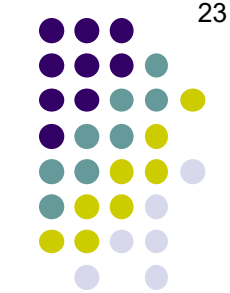

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Number of answers

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(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:

A

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(2) Strongly associated with seizures: TS, SWS

Q

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(2) Strongly associated with seizures: TS, SWS
(2) Diagnostic criteria includes family history:

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(1) Presents with hypertrophy of a single limb:

Number of answers

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

Is the involved limb usually an arm, or a leg?

Number of answers

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(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

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How do the intracranial AVM differ in SWS vs RA?

--

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

Number of answers

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TS: Tuberous sclerosis **SWS**: Sturge-Weber syndrome **vH-L**: von Hippel-Lindau
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Wait a minute--what about the hemangioma in SWS?
The hemangioma is a *choroidal*, not retinal lesion

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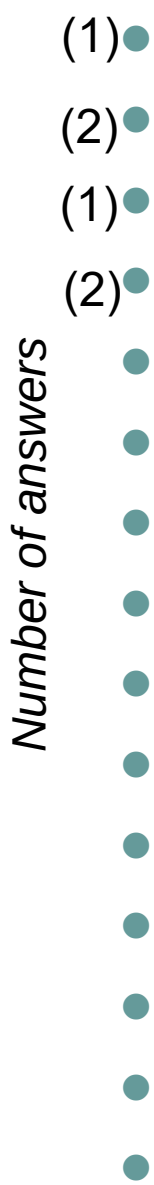

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Is choroidal hemangioma a feature of KTS?

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

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

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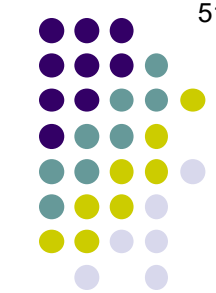

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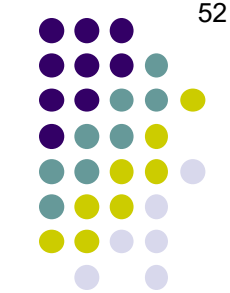

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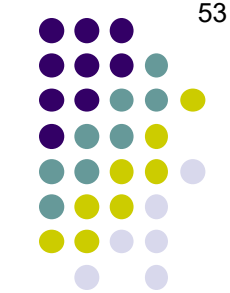

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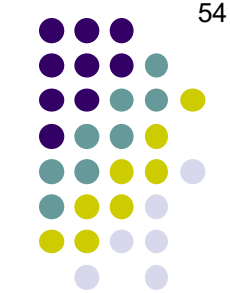

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What is the glaucoma mechanism for each of these?
--**SWS**:
--**NF1**:
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What is the glaucoma mechanism for each of these?
--**SWS**: ↑ EVP; ↑ ciliary-body perfusion; developmental angle anomalies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

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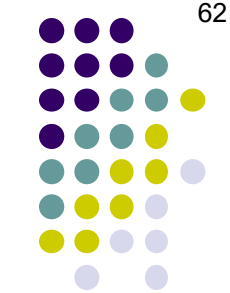

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--SWS? Increased vascularity→**'pink eye' appearance**

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What does **epiloia** *stand for?*

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Q/A

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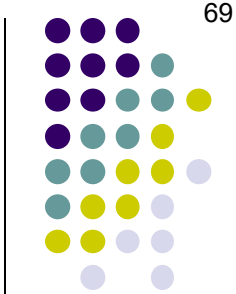

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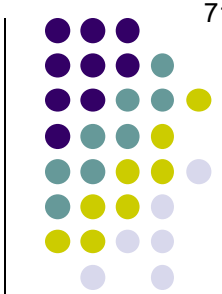

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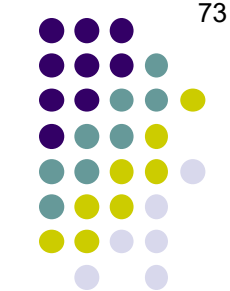

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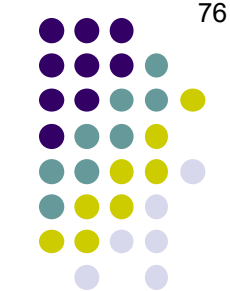

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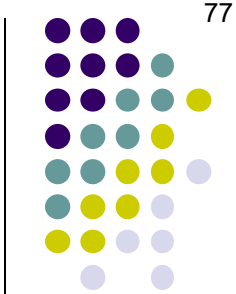

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(1) Classic eye finding is cortical cats or PSC: NF2
(2) The eye findings are unilateral:

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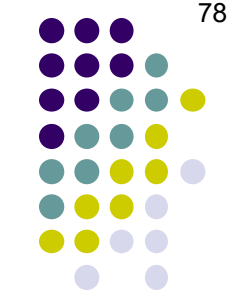

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