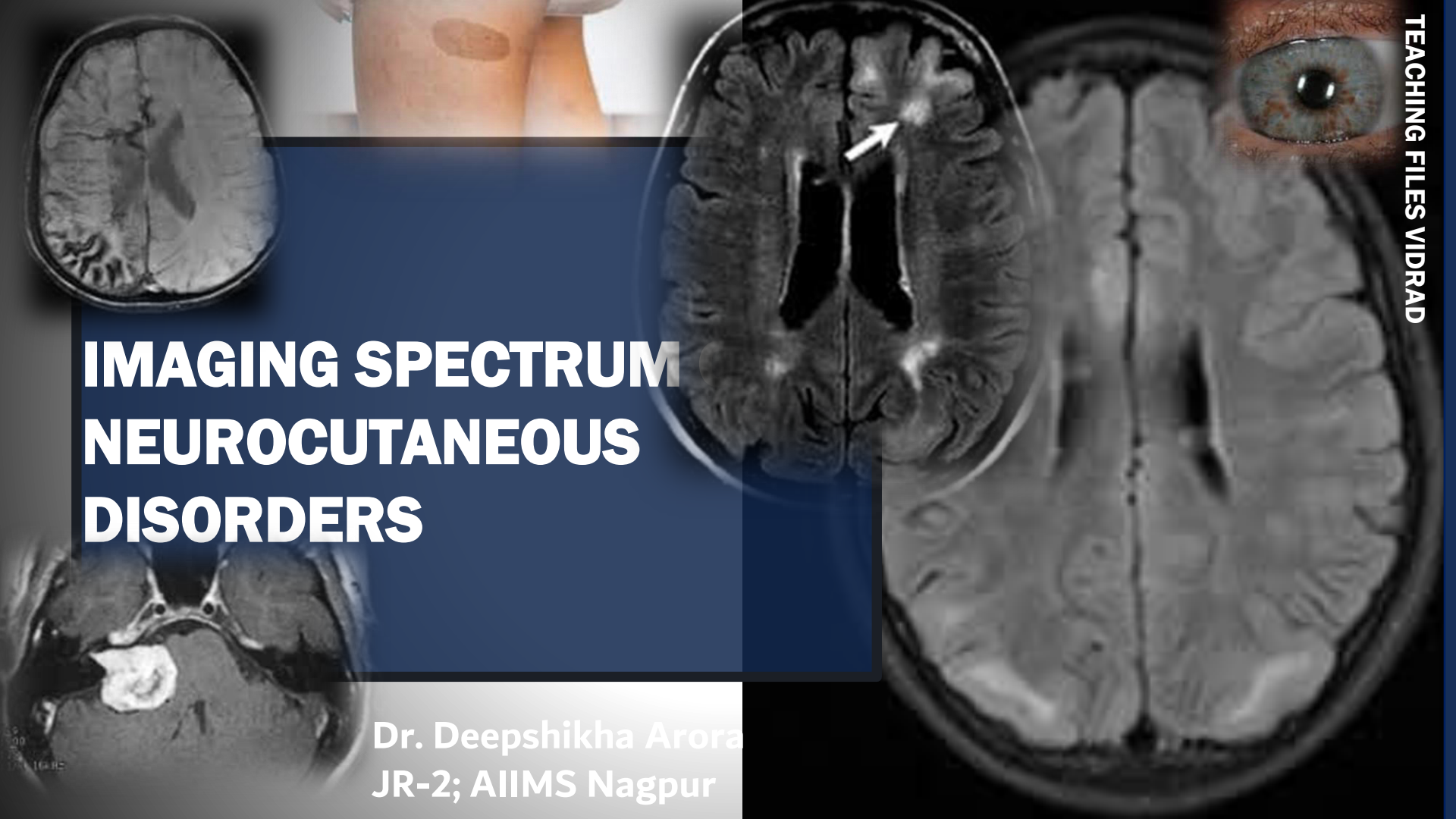


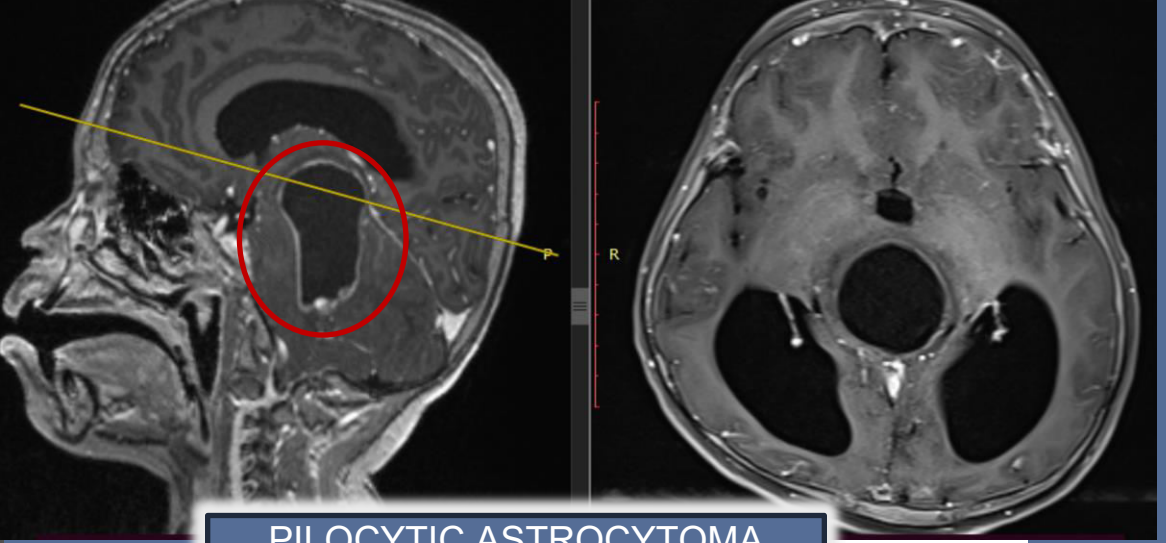
IMAGING SPECTRUM NEUROCUTANEOUS DISORDERS

Dr. Deepshikha Arora
JR-2; AIIMS Nagpur

TEACHING FILES VIDRAD

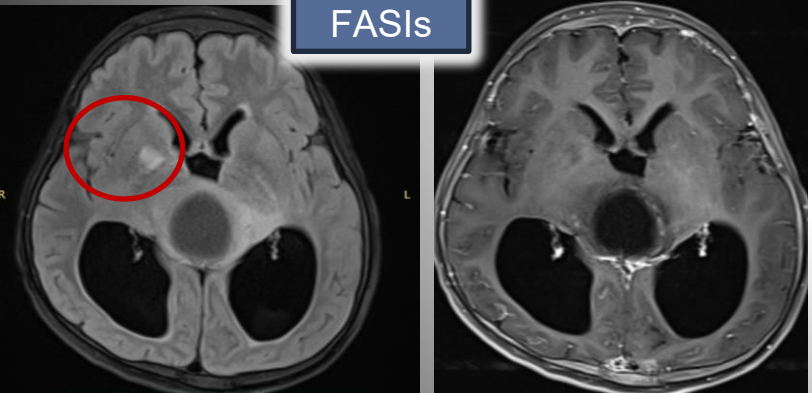
CASE 1

12 YEAR OLD MALE ; PRESENTS WITH MULTIPLE CUTANEOUS NEUROFIBROMAS, IMBALANCE AND DIMINUTION OF VISION IN BOTH EYES

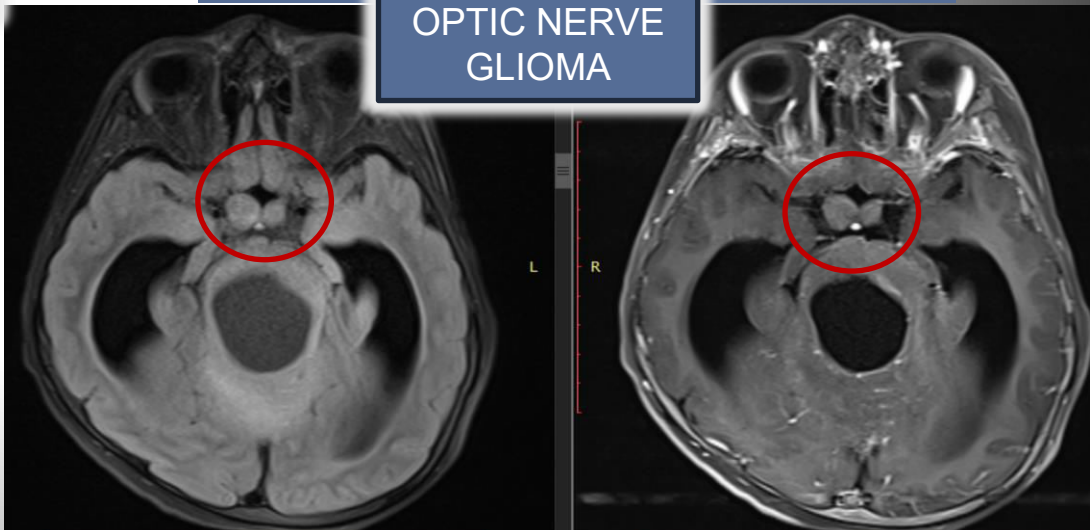


PILOCYTIC ASTROCYTOMA

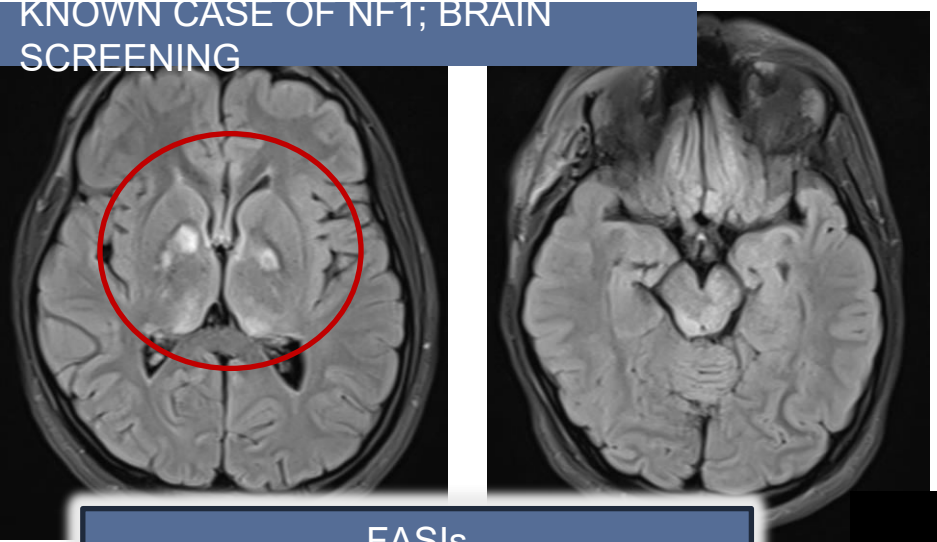
FASIs



OPTIC NERVE GLIOMA



KNOWN CASE OF NF1; BRAIN SCREENING



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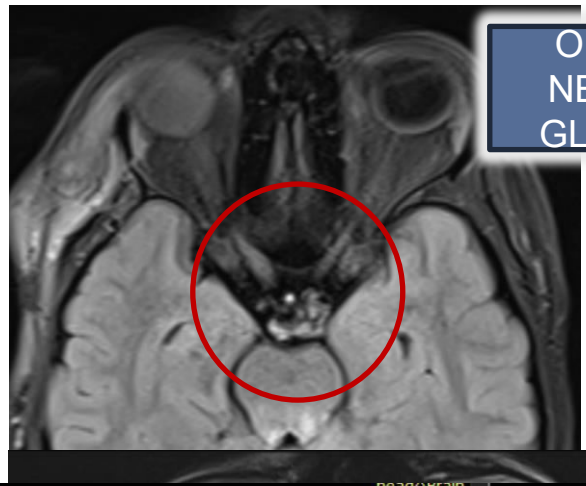

CASE 2

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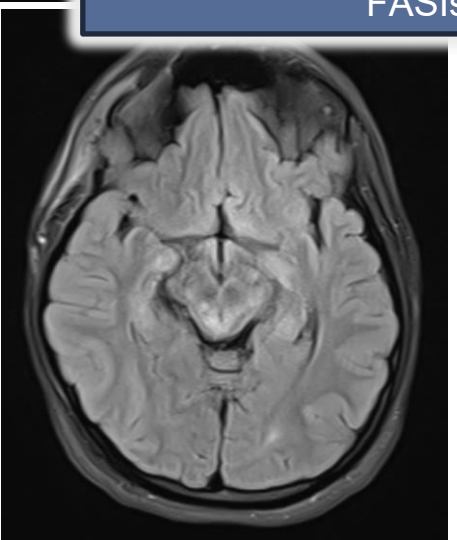
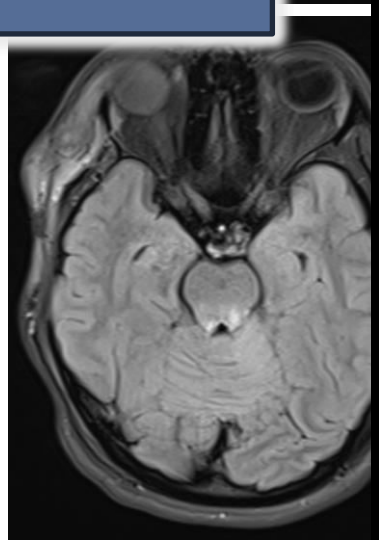
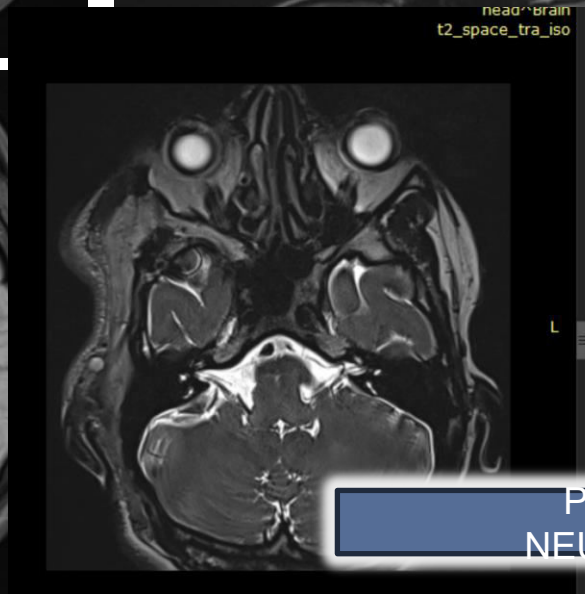
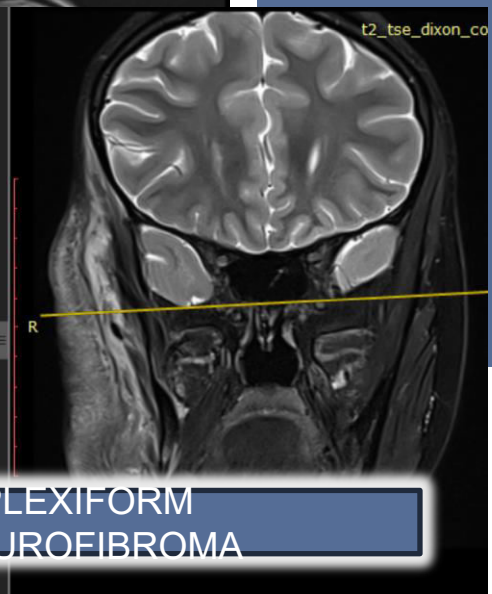


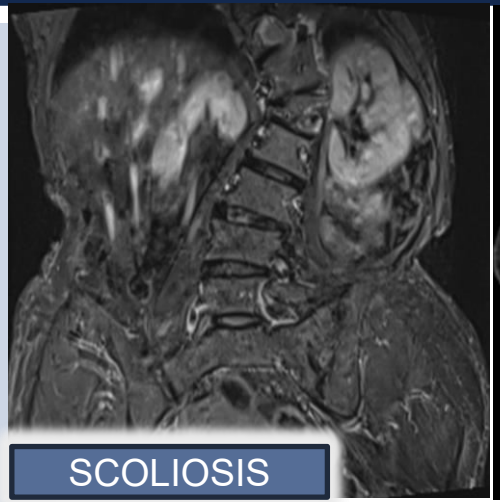




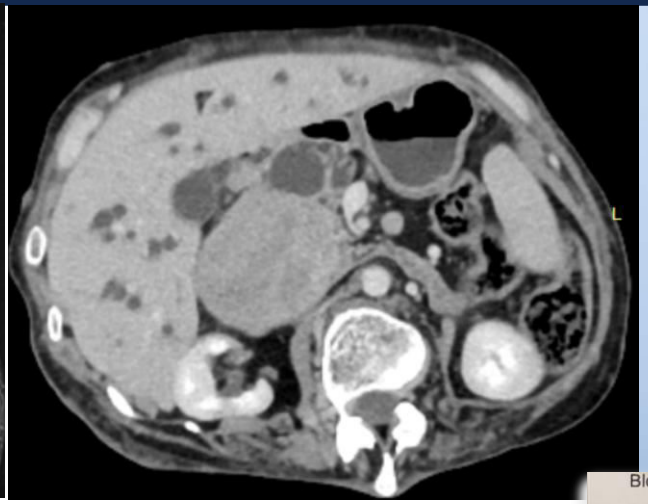


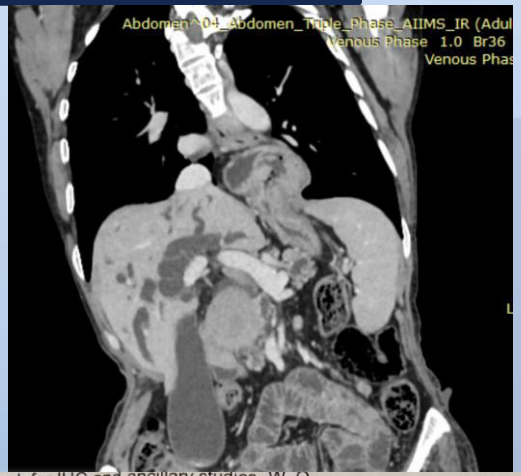
PLEXIFORM NEUROFIBROMA

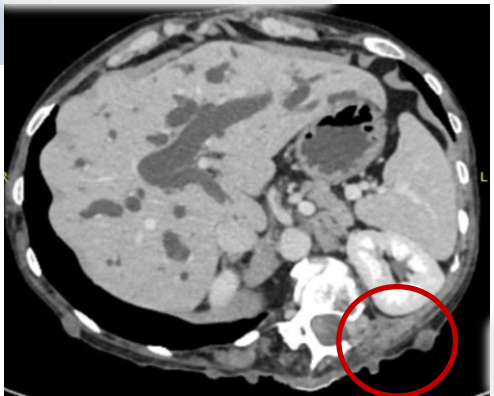
CASE 3- Known case of NF1

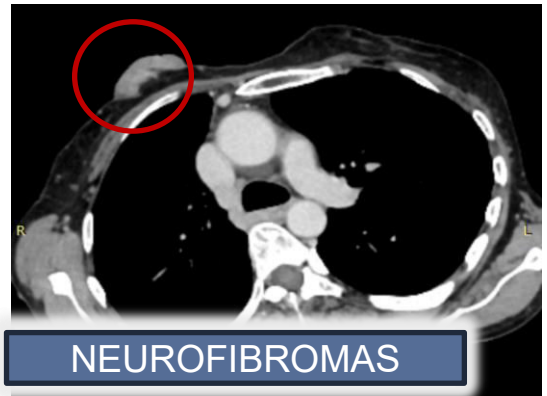


SCOLIOSIS









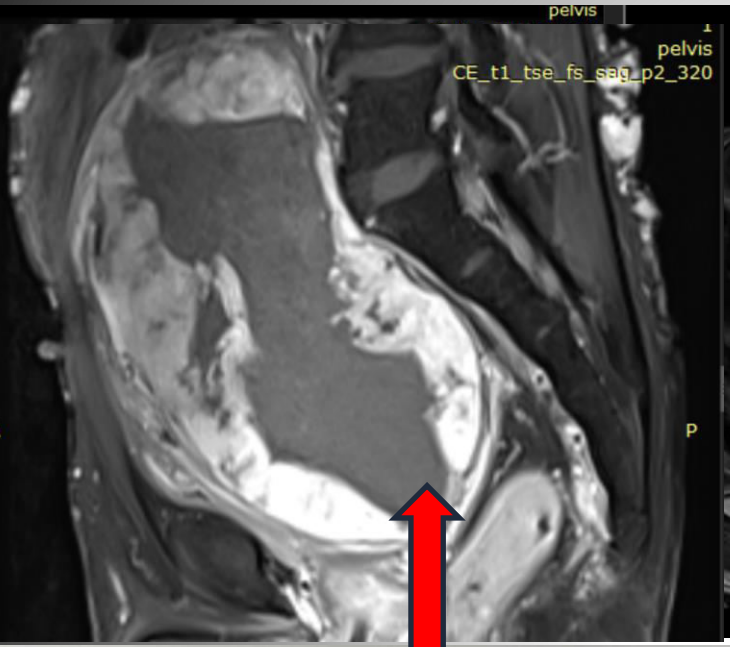
NEUROFIBROMAS

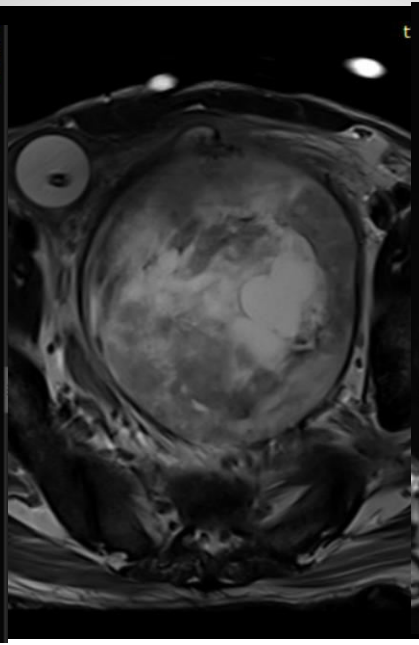
Block for IHC and ancillary studies- W, Q.

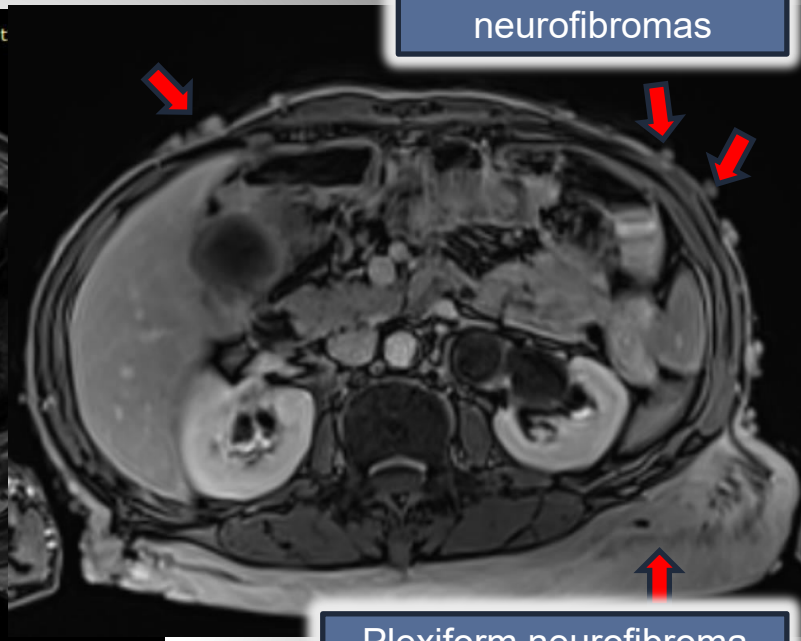
Datasheet of Gastrointestinal stromal Tumor (GIST) :

Procedure : Whipple's Procedure
Tumor Site : Periampullary subserosal nodule (1.5 cm.), 2 subserosal nodule in jejunum/D3 near distal resection (1.5 and 1 cm) margins
Tumor focality:- Multifocal
Histologic Type : *Gastrointestinal stromal tumor (GIST), Spindle type*
Mitotic rate: < 1/5 mm2
Necrosis: Not identified
Histologic grade: G1, low grade
Risk assessment: None.
Margins : proximal and distal resection margins are free of tumor.
Regional lymph nodes: 0/6
Pathologic Stage Classification (pTNM, AJCC 8TH Edition) : pT1 N0
Treatment effect:- No known presurgical treatment.
Block for IHC and ancillary studies- W, Q

CASE 4- Known case of NF1







neurofibromas

Plexiform neurofibroma

Specimen :-	Pelvic mass.
Gross :-	Received multiple linear whitish tassel cores larger measuring 1 cm in length, smaller 0.2 cm. Submitted entirely in 1 block. (Specimen Grossed by:- Dr. Ashwin)
Microscopy :-	Section studied comprises of three linear core showing bundle and fascicles of spindle cells. Spindle cells are elongated, slender, weavy with scant to moderate eosinophilic cytoplasm. Also seen scattered inflammatory infiltrate comprising of lymphocyte throughout the tumor area. Background fibrocollagenous with myxoid change. No increased mitotic activity or area of necrosis noted.
Diagnosis :-	Benign spindle tumor favouring neurofibroma.
Remarks :-	Immunohistochemistry for confirmation.

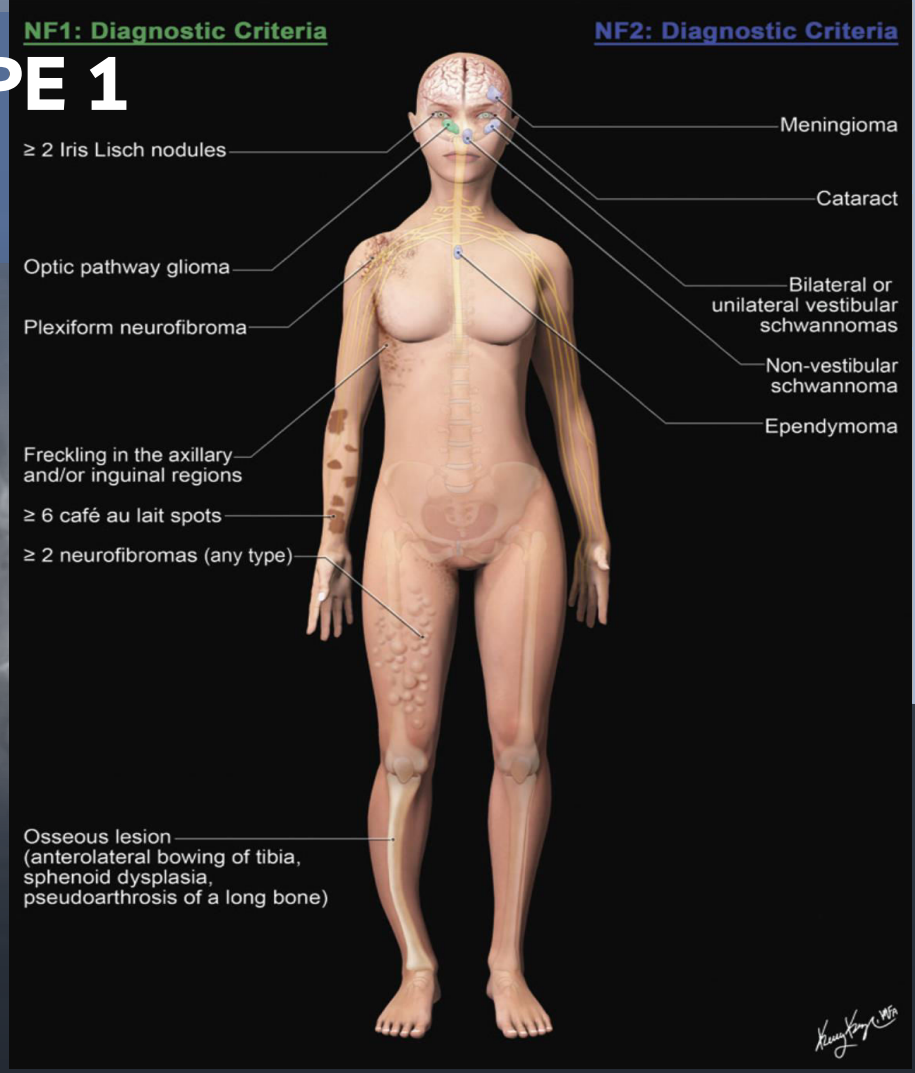
NEUROFIBROMATOSIS TYPE 1

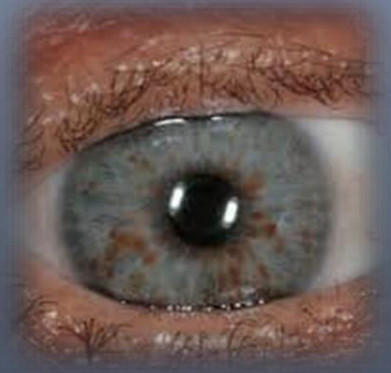
Table 1: Revised Diagnostic Criteria for NF1

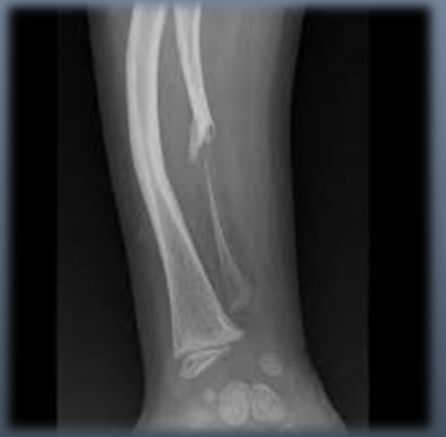
A: The diagnostic criteria for NF1 are met in an individual who does not have a parent diagnosed with NF1 if two or more of the following are present:

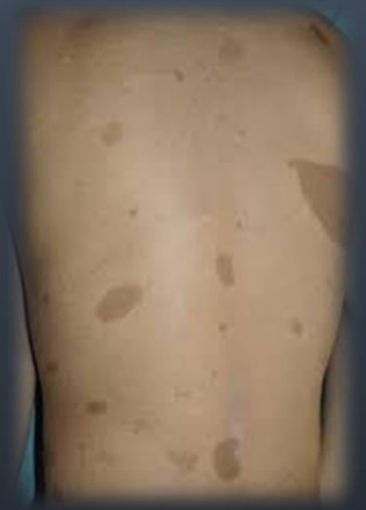
- Six or more café-au-lait macules over 5 mm in greatest diameter in prepubertal individuals and over 15 mm in greatest diameter in postpubertal individuals
- Freckling in the axillary or inguinal region
- Two or more neurofibromas of any type or one plexiform neurofibroma
- Optic pathway glioma
- Two or more iris Lisch nodules identified by slit lamp examination or two or more choroidal abnormalities
- A distinctive osseous lesion such as sphenoid dysplasia, anterolateral bowing of the tibia, or pseudarthrosis of a long bone
- A heterozygous pathogenic *NF1* variant with a variant allele fraction of 50% in apparently normal tissue such as white blood cells

B: A child of a parent who meets the diagnostic criteria specified in A merits a diagnosis of NF1 if one or more of the criteria in A are present









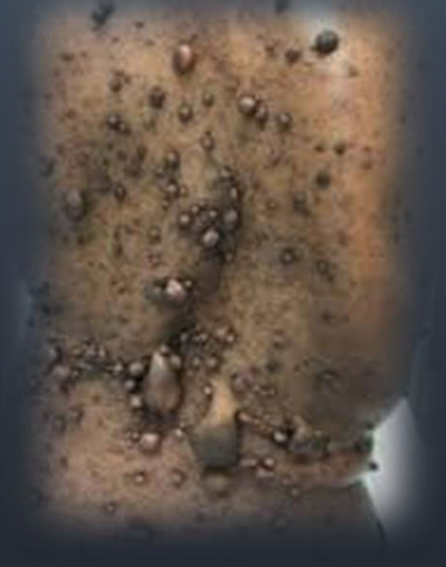
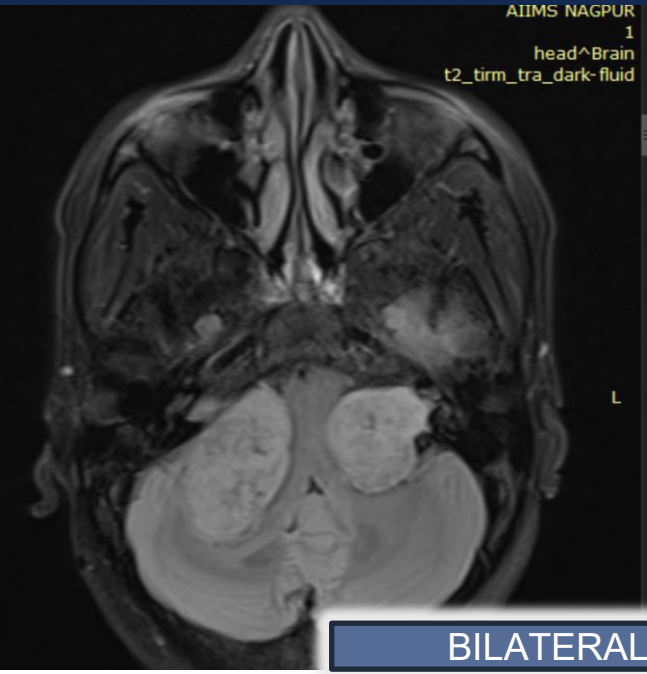


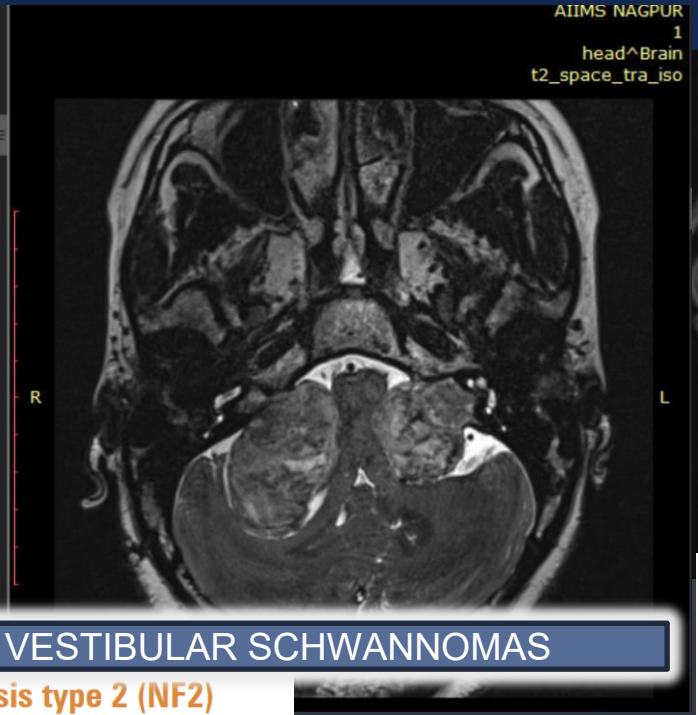
Table 3: Imaging Features of Common NF1-related Manifestations

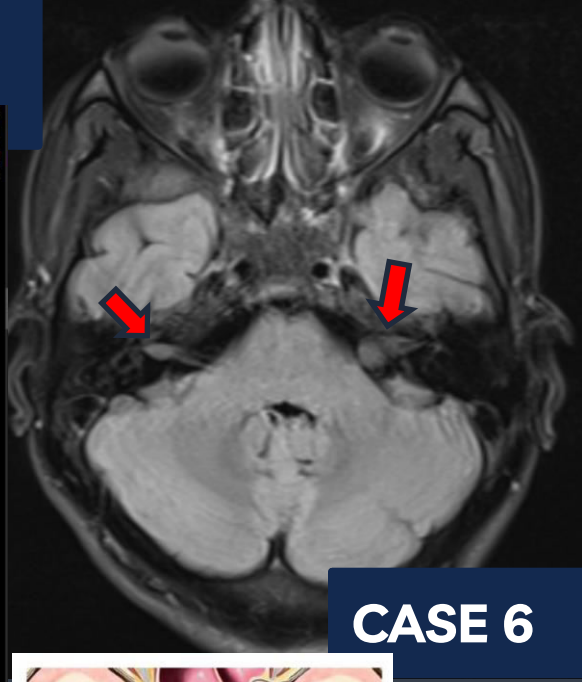
Common Manifestations	Commonly Used Imaging Modality or Modalities	Imaging Features
Neurofibromas	US, CT, MRI	US: well-defined oval hypoechoic masses contiguous with peripheral nerves CT: soft-tissue masses with low attenuation MRI: target sign (T2 hyperintense peripheral rim and hypointense central component), enhancement of central component
Plexiform neurofibromas	MRI	Multinodular confluent masses with multiple target signs and mass effect on surrounding structures
Malignant PNSTs	MRI	Similar to plexiform neurofibromas with suggestive features: large size, peripheral enhancement pattern, perilesion edemalike zone, intratumor cystic changes
Sphenoid wing dysplasia	CT, MRI	Hypoplastic sphenoid wing, widened middle cranial fossa, flattened posterior orbit, possible meningocele and meningoencephalocele
Focal areas of signal intensity (FASIs)	MRI	Focal or diffuse T2 hyperintensity in the basal ganglia, cerebellum, or brainstem without enhancement or mass effect
Optic pathway glioma	MRI	T1-hypointense, T2-hyperintense, and homogeneously enhancing enlargement of the optic nerve sheath complex
Non–optic pathway glioma	MRI	T1-hypointense or -isointense, T2-hyperintense, and variably enhancing intraparenchymal lesion, usually in the brainstem or cerebellum; may cause obstructive hydrocephalus
Dural ectasia, meningocele	CT, MRI	Well-circumscribed paravertebral mass following CSF signal intensity, often in the anterior or anterolateral aspect of the vertebral column
Interstitial lung disease	CT	Bilateral, symmetric, basal-predominant linear and ground-glass opacities; apical-predominant cysts and centrilobular nodules
GIST	CT, MRI	Submucosal bowel wall tumor with an endophytic, exophytic, or combined growth pattern
Pheochromocytoma or paraganglioma	CT, MRI	CT: well-circumscribed homogeneously enhancing adrenal gland mass (pheochromocytoma) or extra-adrenal mass (paraganglioma) MRI: often T2 hyperintense; possible T2 intermediate signal intensity due to hemorrhage, cystic changes, or myxoid degeneration
Rhabdomyosarcoma	CT, MRI	Heterogeneous enhancement with local invasion of organs and destruction of bone
Scoliosis	XR, CT	Lateral spinal curvature with sharply angulated segments of four to six vertebrae
Bone dysplasia	XR, CT	Posterior vertebral body scalloping; thinning of the pedicles, transverse processes, laminae; neural foramen enlargement; rib deformities; anterolateral tibial bowing, fracture, and pseudoarthrosis
Nonossifying fibromas	XR, CT, MRI	Slightly expansile cortically based lesions in the metaphysis of long bones with thin sclerotic borders and narrow zone of transition
Vascular dysplasia	US, CT, MRI	Narrowing and aneurysmal dilatation of various arteries (eg, renal artery, abdominal aorta, and terminal internal carotid artery)

Note.—CSF = cerebrospinal fluid, GIST = gastrointestinal stromal tumor, XR = radiography.

CASE 5: Gradually progressive hearing loss since 3 years; imbalance; Romberg+







CASE 6

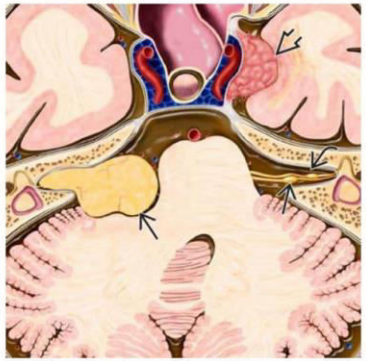
BILATERAL VESTIBULAR SCHWANNOMAS

Table 2: Diagnostic criteria of Neurofibromatosis type 2 (NF2)

A diagnosis of NF2 requires any one of the following two conditions:

i. Bilateral Vestibular schwannomas (VS)

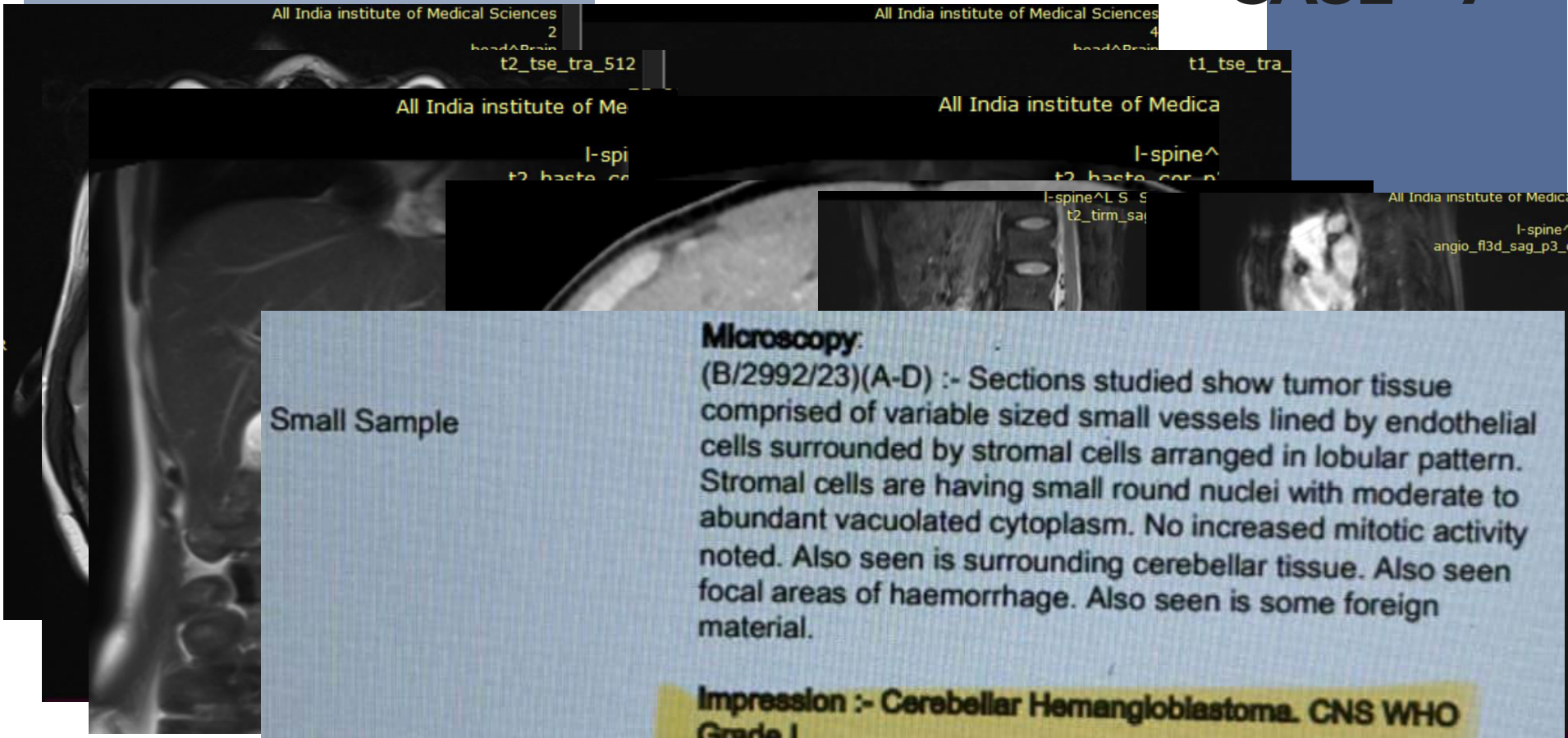
ii. Family history of NF2 plus 1) Unilateral VS or 2) Any two of: meningioma, glioma, neurofibroma, schwannoma, or juvenile posterior subcapsular lenticular opacities



Graphic depicts classic NF2 with bilateral vestibular schwannomas (VSs)→, facial schwannoma ↗, and meningioma ↷.

A 38 year old female; presenting with altered sensorium,headache, vomiting and diminution of vision in right eye

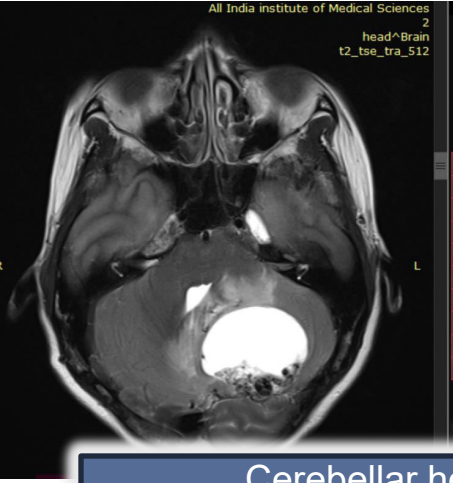
CASE - 7

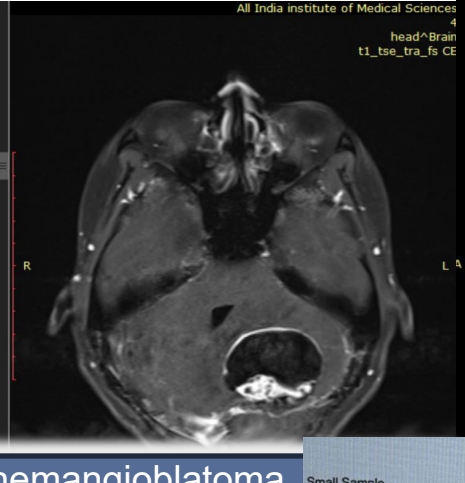


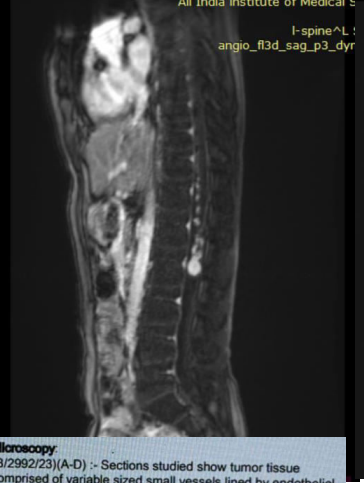
Small Sample

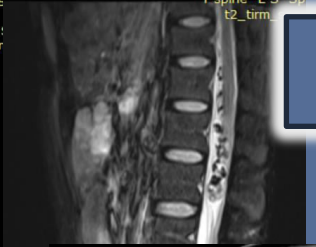
Microscopy:
(B/2992/23)(A-D) :- Sections studied show tumor tissue comprised of variable sized small vessels lined by endothelial cells surrounded by stromal cells arranged in lobular pattern. Stromal cells are having small round nuclei with moderate to abundant vacuolated cytoplasm. No increased mitotic activity noted. Also seen is surrounding cerebellar tissue. Also seen focal areas of haemorrhage. Also seen is some foreign material.

Impression :- Cerebellar Hemangioblastoma. CNS WHO Grade I.



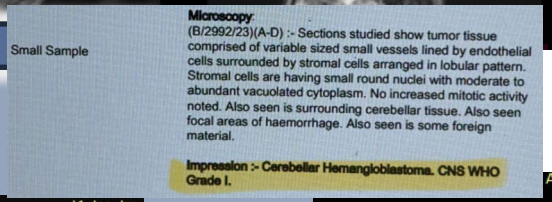






spinal hemangioblatomas

Cerebellar hemangioblatoma


Small Sample

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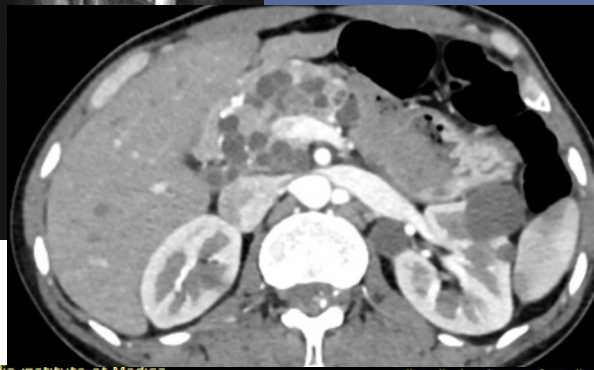
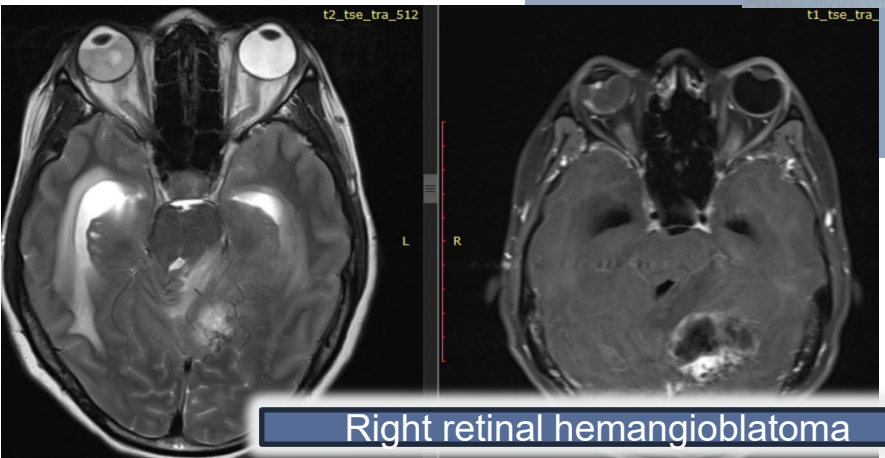
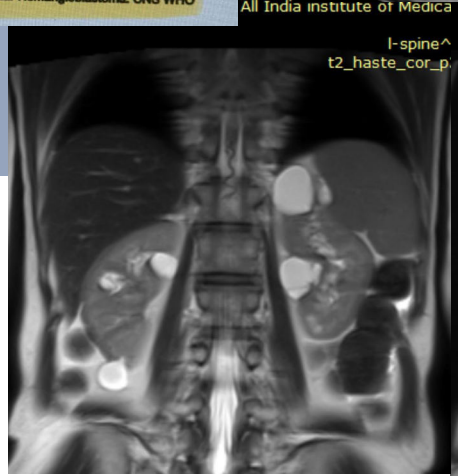
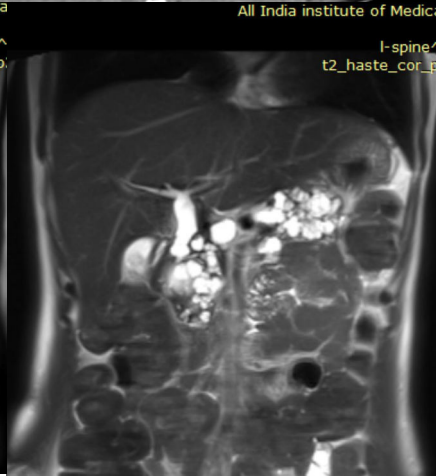


VON HIPPEL LINDAU



Right retinal hemangioblatoma





VON HIPPEL LINDAU DISEASE

VHL: PATHOLOGY

CNS Neoplasms

- HBs (60-80%)
  - Retinal HBs ("angiomas") (50%)
- ELSTs (10-15%)

Visceral Lesions

- Renal lesions (2/3 of all VHL patients)
  - Cysts (50-75%)
  - Clear cell renal carcinomas (25-45%)
- Adrenal PCC (10-20%)
  - Hallmark of type 2 VHL
- Pancreatic cysts (35-70%), nonsecretory islet cell tumors (5-10%)
- Epididymal cysts, cystadenomas (60% of male patients; often bilateral)
- Broad ligament cystadenomas (female patients; rare)

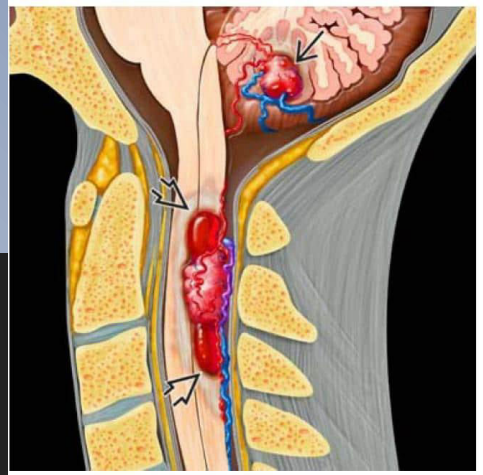
VHL: DIAGNOSTIC CLINICAL FEATURES

No Family History of VHL

- ≥ 2 CNS HBs **or**
- 1 CNS HB + visceral tumor

Positive Family History of VHL

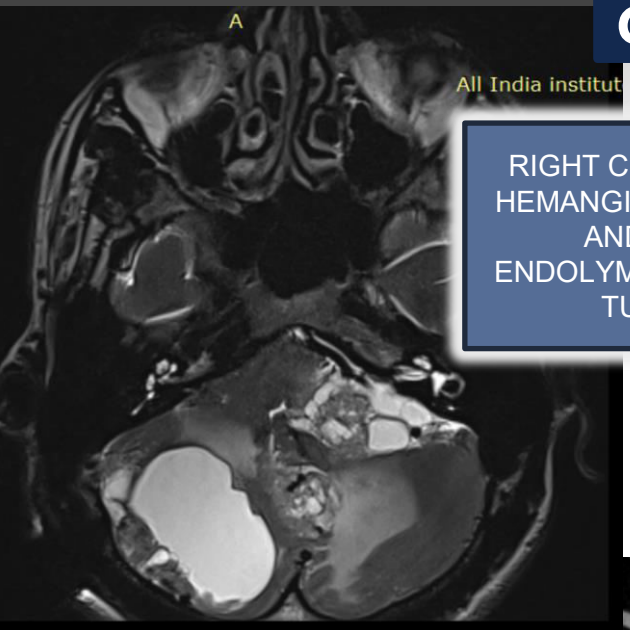
- 1 CNS HB **or**
- PCC **or**
- Clear cell renal carcinoma



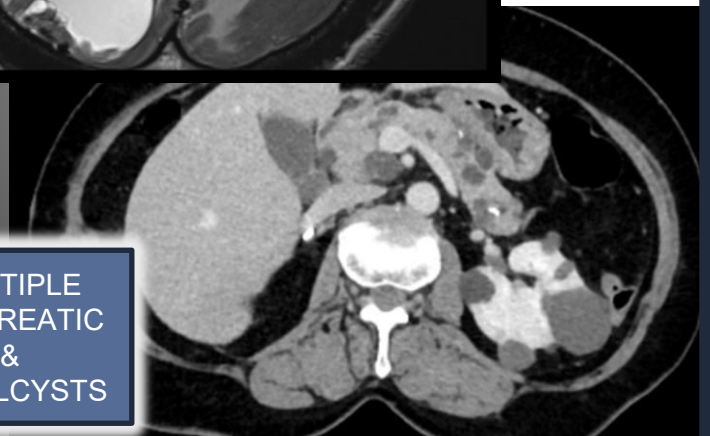
Two HBs in VHL show spinal cord tumor has an associated cyst ⇨, causing myelopathy. Small cerebellar HB → would be asymptomatic.

VON HIPPEL LINDAU DISEASE

CASE 8



RIGHT CEREBELLAR HEMANGIOBLASTOMA AND LEFT ENDOLYMPHATIC SAC TUMOR



MULTIPLE PANCREATIC & RENALCYSTS

VHL: IMAGING

Multiple Hemangioblastomas (Diagnostic of VHL)

- 2/3 cystic, 1/3 solid
- Nodule abuts pia
- 50% in cord (dorsal > ventral surface)
  - Multiple tiny "tumorlets" along cord common
  - Disseminated leptomeningeal hemangioblastomatosis

Retinal "Angiomas"

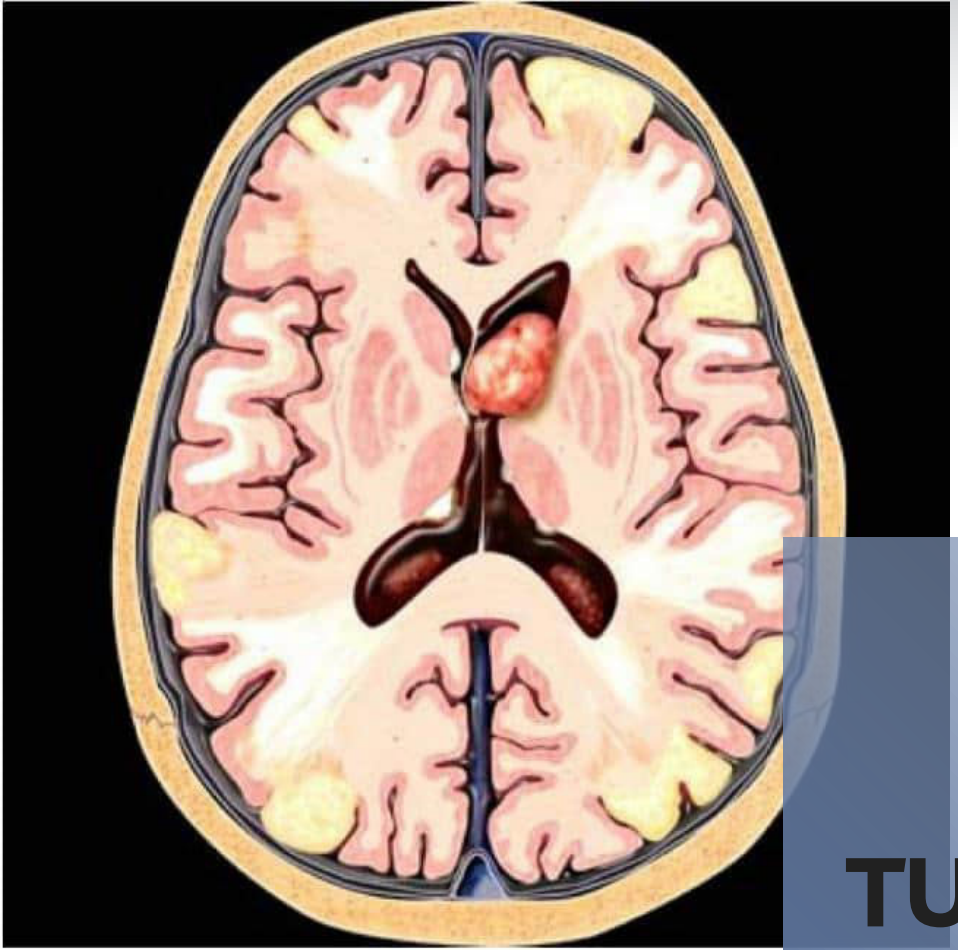
- Hemorrhagic retinal detachment
  - V-shaped hyperdense posterior globe
- ± enhancing "dots" (tiny HBs)

Uni- or Bilateral Endolymphatic Sac Tumors

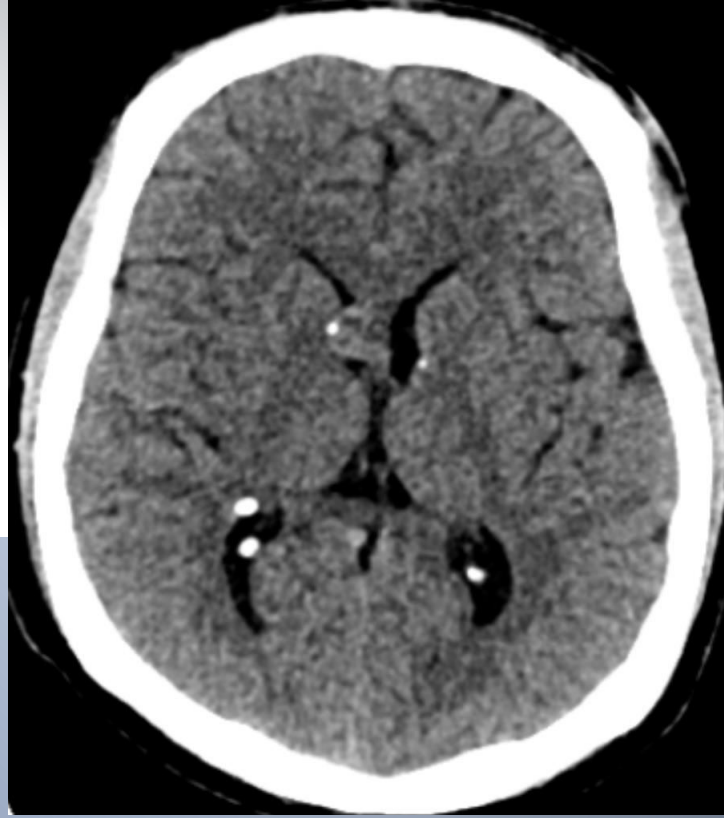
- Dorsal temporal bone
  - Between IAC, sigmoid sinus
- Infiltrative, lytic, intratumoral bone spicules
- T1 iso-/hyperintense; T2 hyperintense
- Strong enhancement

Differential Diagnosis

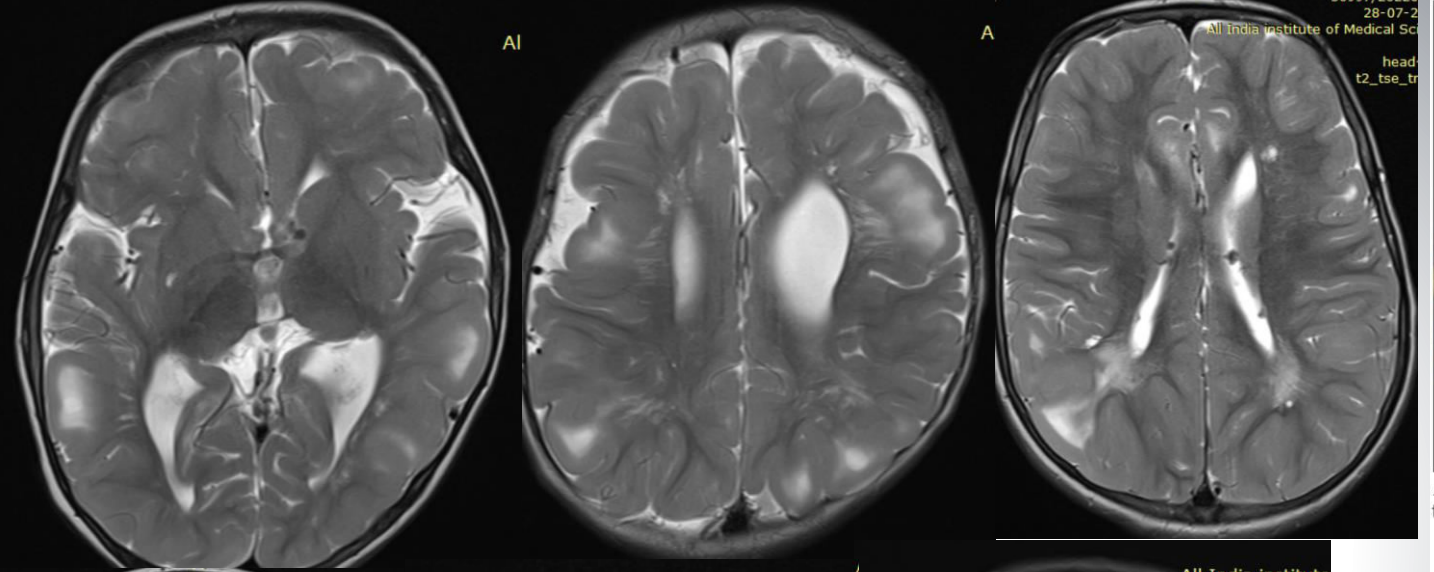
- Solitary HB

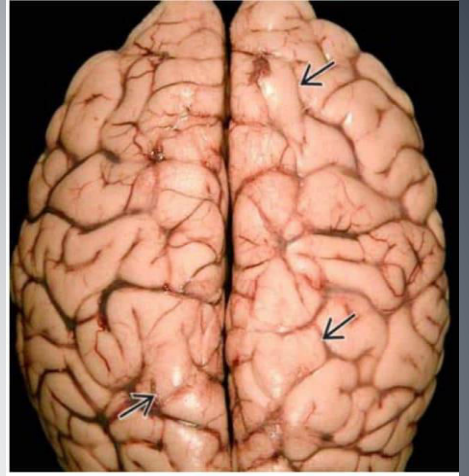


Graphic shows pathologic findings of tu
sclerosis complex (TSC).

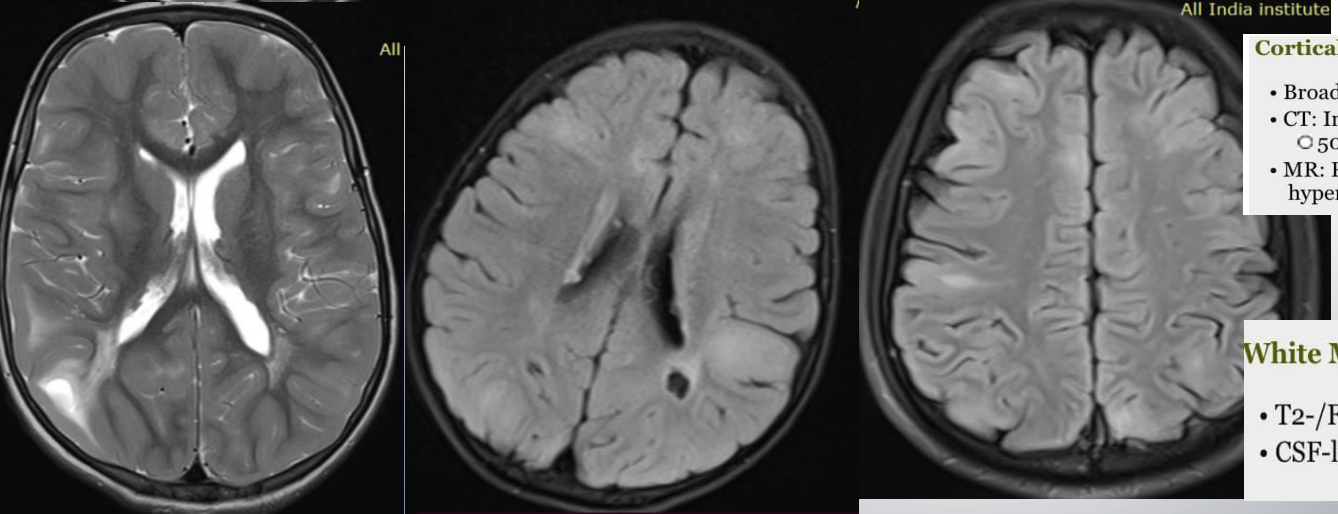


TUBEROUS SCLEROSIS





Autopsy of TSC shows multiple expanded gyri with the potato-like appearance characteristic of cortical tubers →. (Courtesy R. Hewlett, MD.)

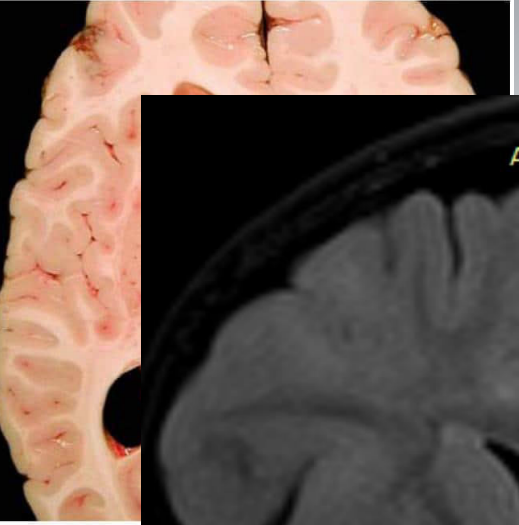


Cortical Tubers

- Broad, expanded gyrus
- CT: Initially hypodense; calcification increases with age
  - 50% of patients eventually develop ≥ 1 calcified tuber(s)
- MR: Periphery isointense, subcortical portion T2/FLAIR hyperintense

White Matter Lesions

- T2-/FLAIR-hyperintense radial lines/wedges
- CSF-like cysts in deep periventricular WM



xial cut sectio
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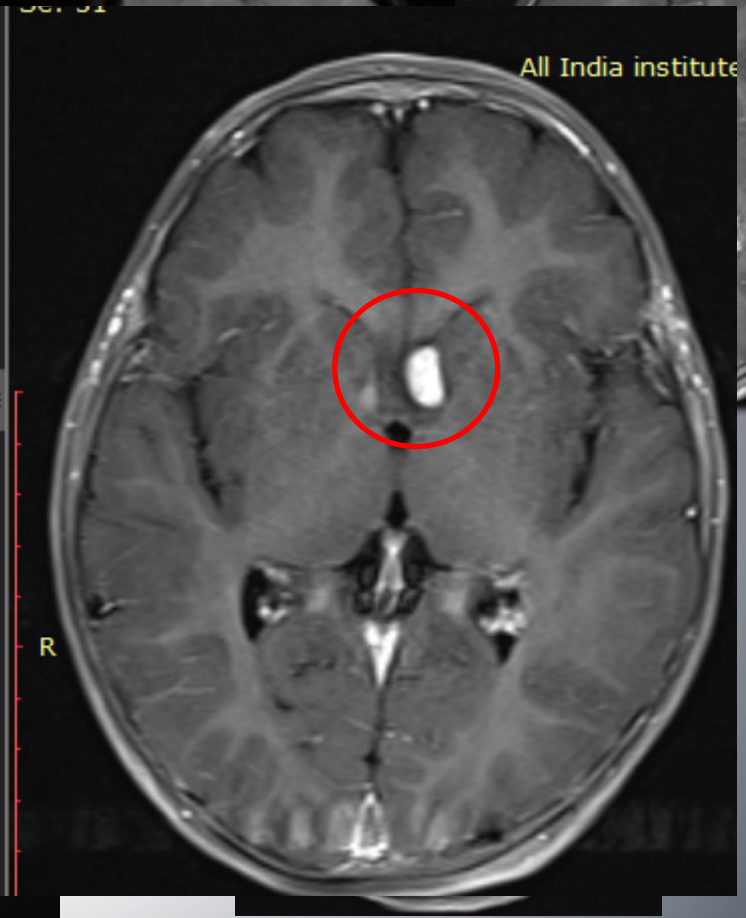
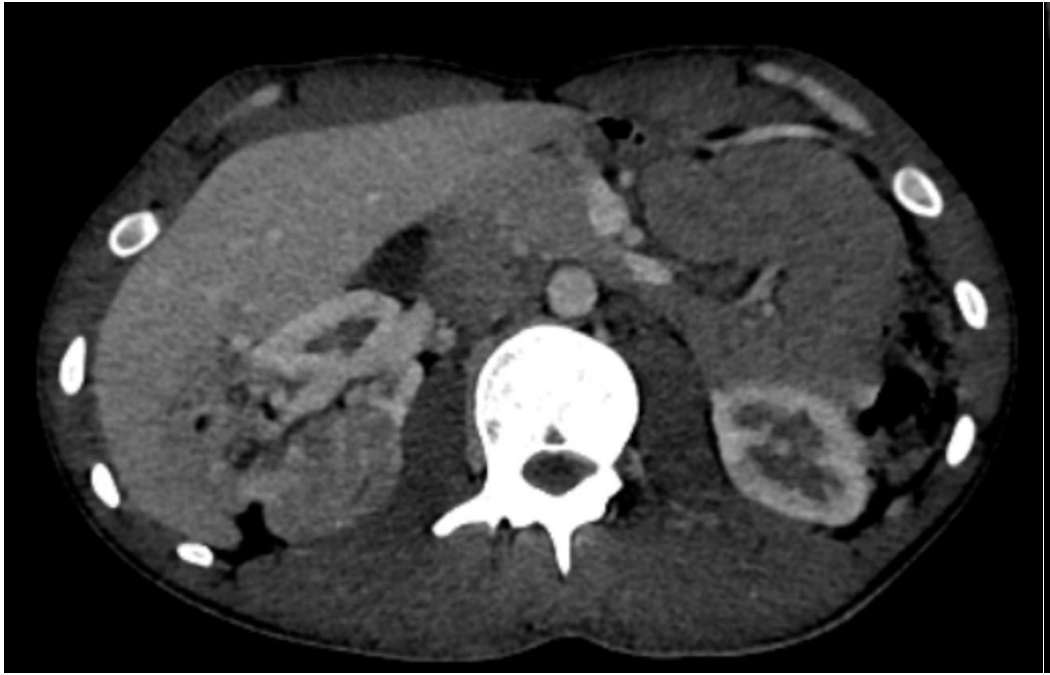
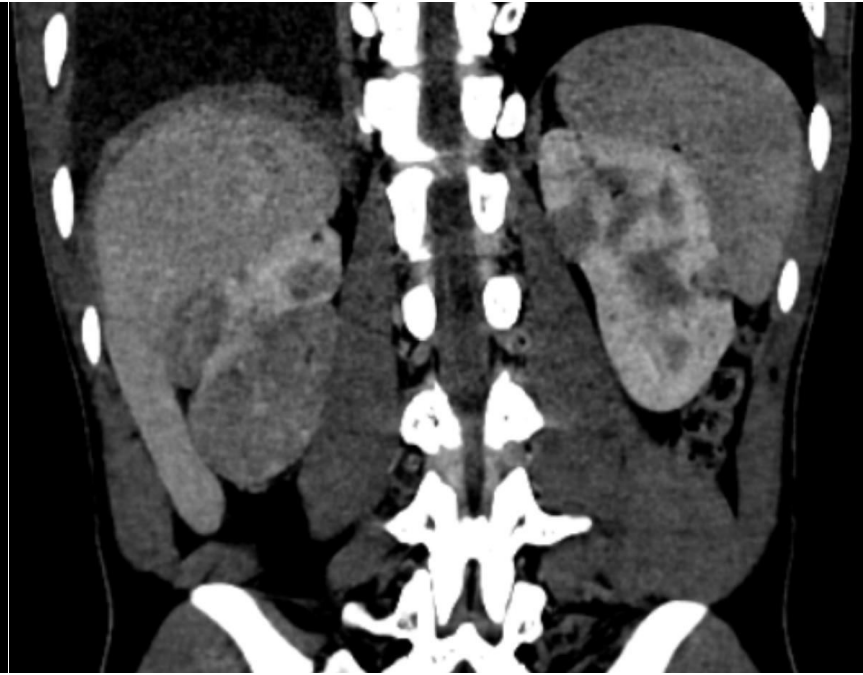


Subependyn

- CT: Calcification rare in 1st year; increases with age
  - 50% eventually calcify; do not enhance
- MR: T1 hyper-, T2 hypointense; 50% enhance

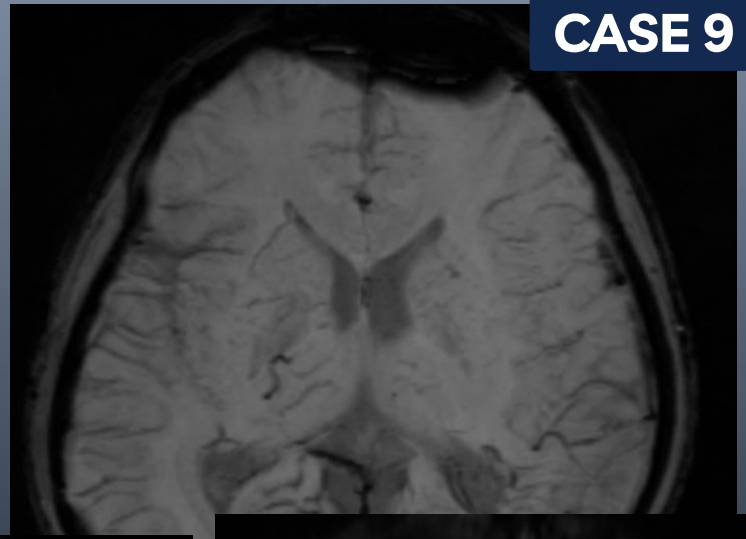


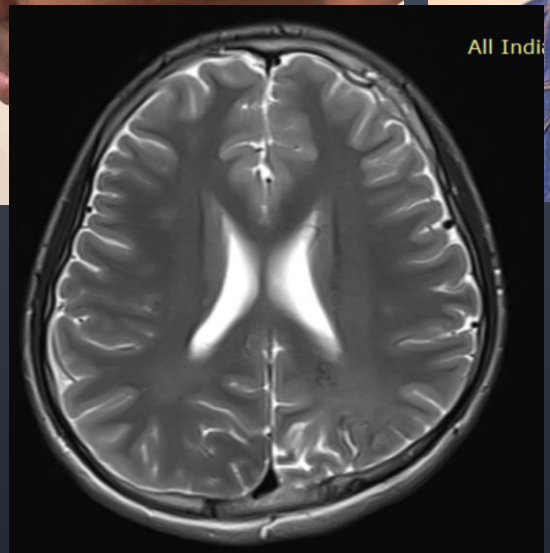


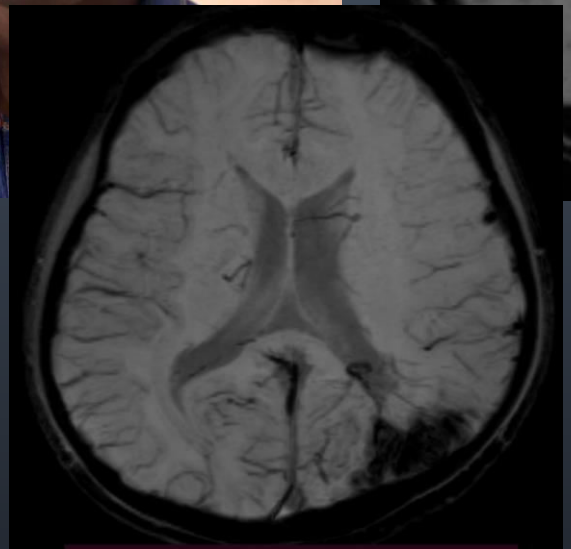
ANGIOMYOLIPOMAS

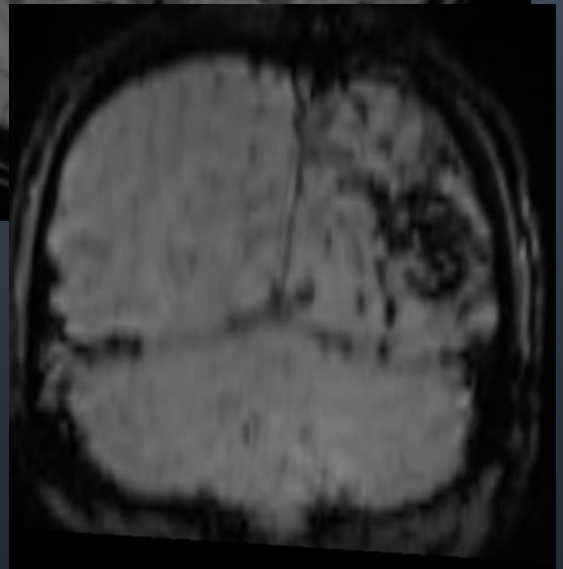
Major Criteria	Minor Criteria
1. Facial angiofibromas or forehead plaques 2. Nontraumatic ungual or periungual fibroma 3.Hypomelanotic macules (3 sau >) 4. Shagreen patch 5. Multiple nodular retinal hamartomas 6. Cortical tubers 7. Subependymal nodules 8. Subependymal giant cell astrocytoma 9. Unique/multiple cardiac rhabdomyoma 10. Unique/multiple renal angiomyolipoma	1. Alterations of dental enamel 2. Hamartomatous rectal polyps 3. Bone cysts 4. Cerebral white matter migration tracts 5. Gingival fibromas 6. Non renal hamartoamas 7. Retinal achromic patch 8. "Confetti" skin lesions 9. Multiple renal cysts
Positive TS diagnosis ***- 2 major criteriaoru*** ***- 1 major criterion + 2 minor criteria***	

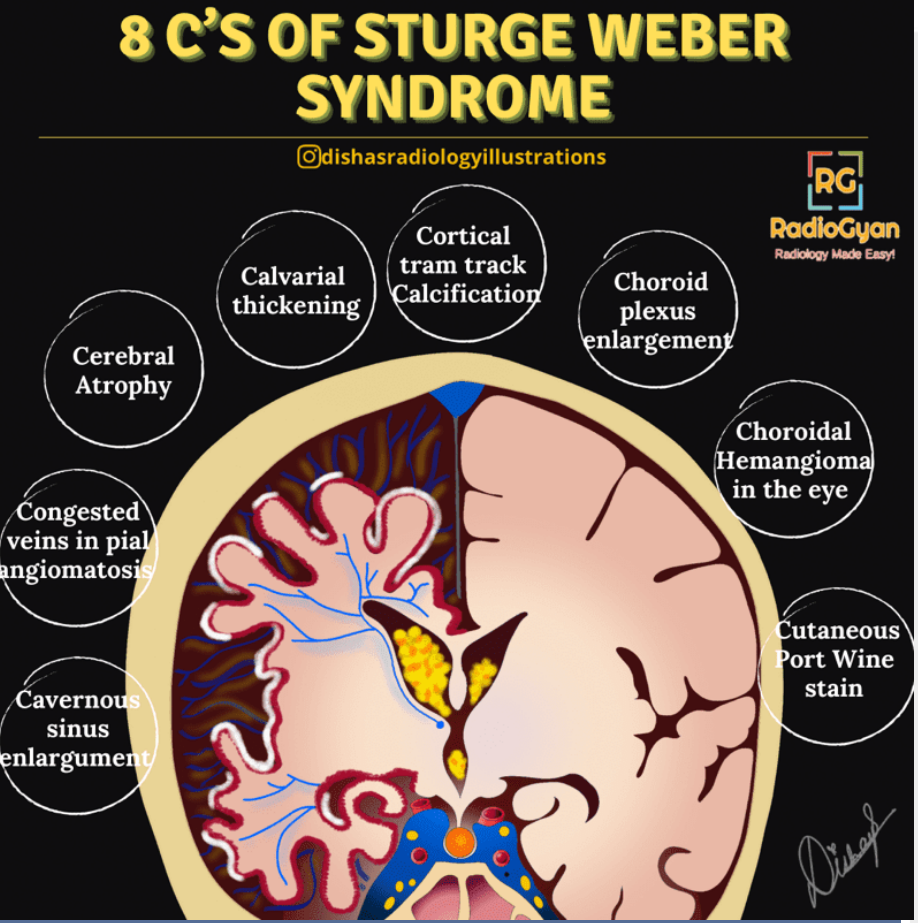
CASE 9











STURGE WEBER SYNDROME

- Congenital but sporadic, not inherited
- Postzygotic (i.e., somatic) mutation in *GNAQ*

Pathology

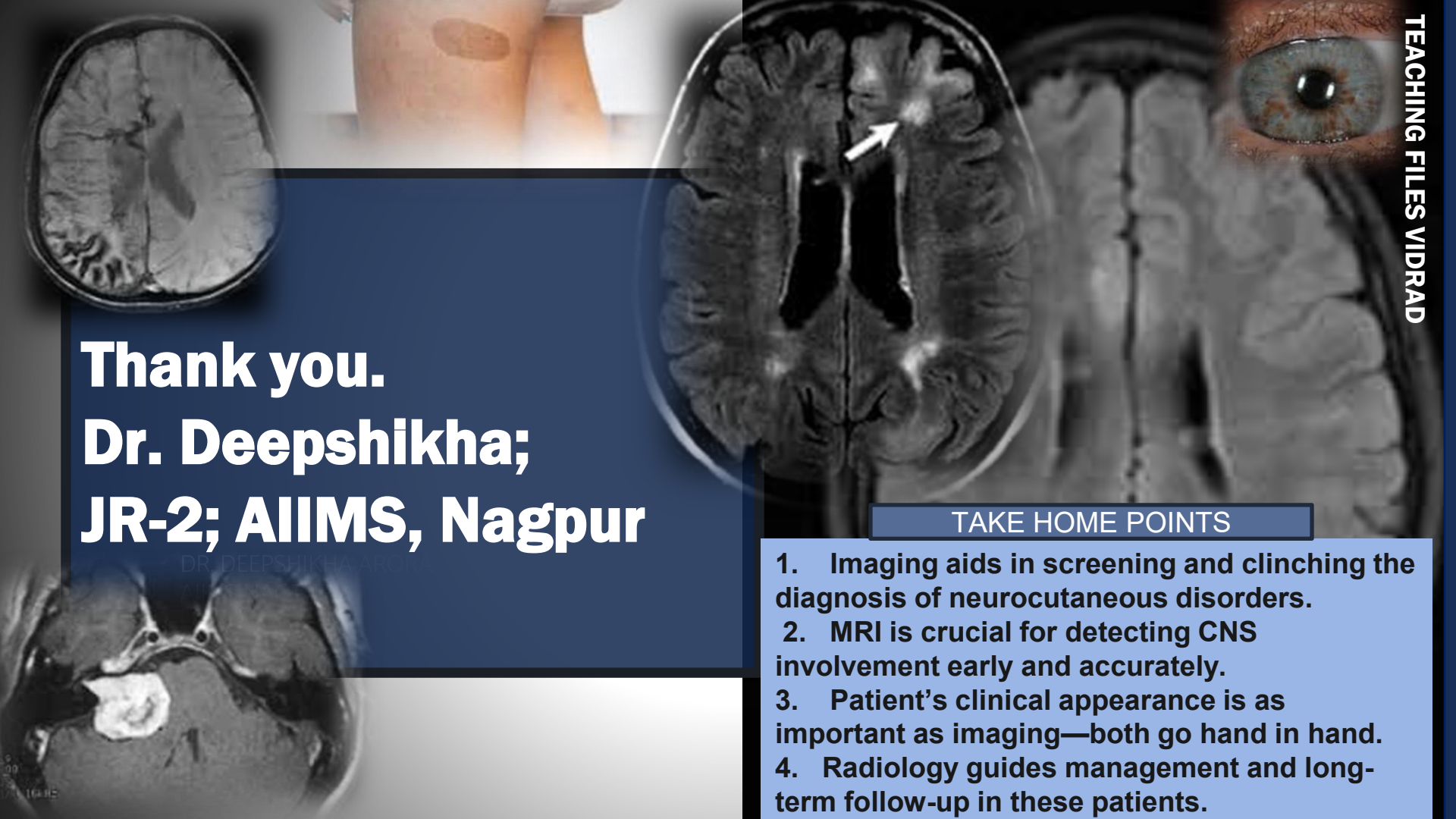
- Pial (leptomeningeal) angioma
- Cortical venous ischemia, atrophy
- Parietooccipital > frontal

Clinical Issues

- Unilateral facial CM
- Usual cutaneous distribution = CNV1, CNV2 > CNV3
  - Can be bilateral or even absent

Imaging

- CT
  - Atrophic cortex
  - Ipsilateral calvarium thick, sinuses enlarged
  - Cortical Ca^{++} (**not** in angioma!) increases with age
- MR
  - Cortical/subcortical hypointensity on T2
  - Ca^{++} "blooms" on T2*
  - Angioma enhances (unilateral 80%, bilateral 20%)
  - Ipsilateral choroid plexus enlarged
  - Enlarged medullary veins



TEACHING FILES VIDRAD

Thank you.
Dr. Deepshikha;
JR-2; AIIMS, Nagpur

TAKE HOME POINTS

1. Imaging aids in screening and clinching the diagnosis of neurocutaneous disorders.
2. MRI is crucial for detecting CNS involvement early and accurately.
3. Patient's clinical appearance is as important as imaging—both go hand in hand.
4. Radiology guides management and long-term follow-up in these patients.