Grand Rounds Presentation

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April 17, 2014

History and Examination

CC: “My gold weight is too heavy and I want it removed”

HPI: 27 y.o. black female, s/p right upper eyelid gold weight placement for CN 7 palsy following right CN 7 schwannoma resection (2012) states that for the past 2 months her right eyelid has begun to droop more and more, and that she has to tilt her head back to see. She also complains of double vision which she had before but is now a bit worse.

History and Examination

- PMH: as per HPI, whole brain XRT x 2 without improvement (2012)
- PSH: Right hemicraniectomy for right CN 7 schwannoma, right static facial sling by ENT (2013)
- POH: none
- Meds: none
- Gtt: none
- Allergies: none
- FH: neg for glaucoma, blindness, strabismus, cancer
- SH: occasional ETOH, denies smoking, illicit drug use

History and Examination

- BCVA: OD: 20/20-2, OS: 20/20
- Pupils: anisocoria OD 3 mm-2 mm min reactive, +APD OD; OS 4 mm-2 mm brisk reactive
- Color vision: 8/8 OU
- EOM: see photo
- CVF: constricted OD, full OS
- Tapp: 10/13 at 9AM
- SLE:

LLA: see photos AC: D/Q

C/S: W/Q OU I/P: round, reactive OS

K: few inf PEE OD; cl OS L: clear OU

History and Examination

Funduscopic Exam:

- Vitreous: clear OU
- C/D: 0.3 s/p OU
- Mac: flat OU
- V/P: WNL OU

Previous Clinical Exam (2012)

- gold weight placement by ENT for exposure keratopathy OD (2012);
- migration of weight (shown above)
- repositioned by Dr. Shinder (2012)

Present Clinical Examination

Question

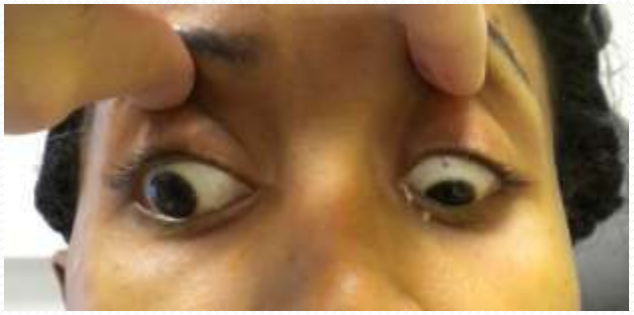
Will you remove or replace the gold weight?

Motility Examination

diplopia

diplopia

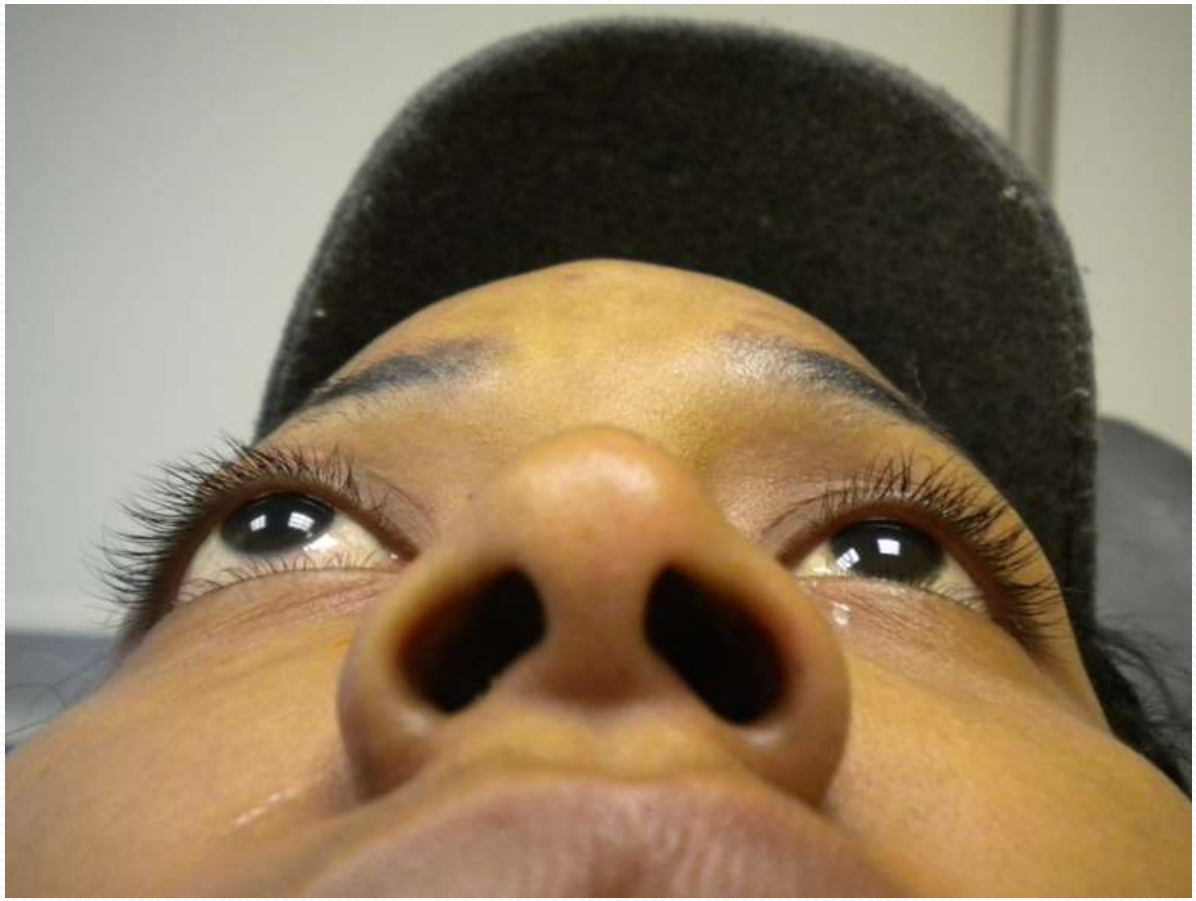
diplopia



diplopia

Patient Care

Clinical Examination



Clinical Examination

Clinical Examination

- MRD1: <1 mm OD; 4 mm OS
- Levator function: right: 11 mm left: 18 mm
- Lagophthalmos: 1 mm OD

Review of Cranial Nerve Exam

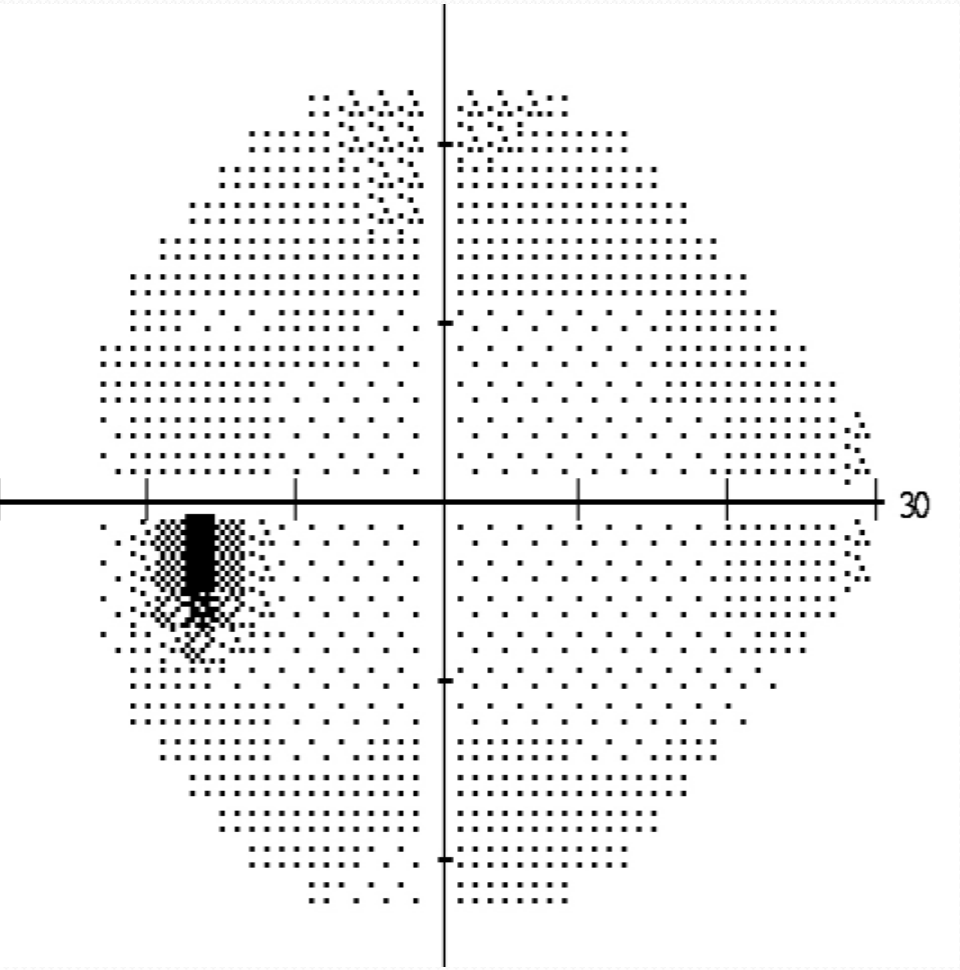
- CN 1: intact
- CN 2: impaired (+APD, visual field deficits)
- CN 3: somatomotor- impaired OD, levator and sup/med/inf recti impaired
  parasympathetic- impaired accommodation
- CN 4: intact (no hypo/hypertropias,torsion or head tilt)
- CN 5: impaired sensation in right V1, V2, V3 distribution
- CN 6: trace deficit OD

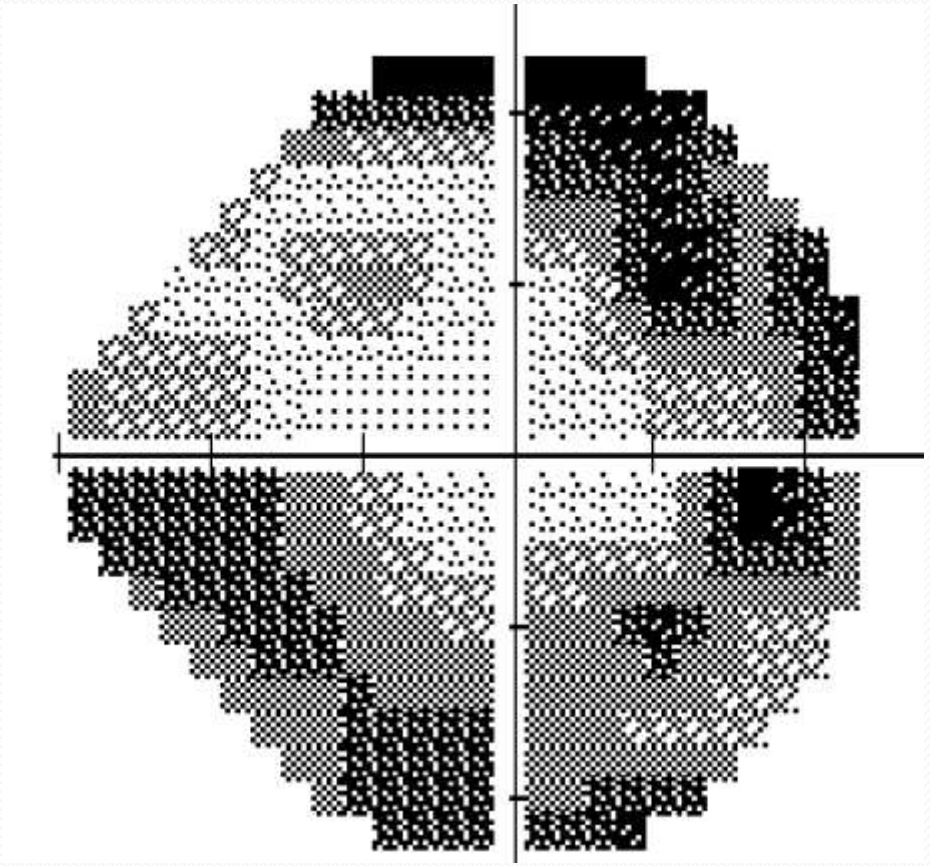
Review of Cranial Nerve Exam

- CN 7: impaired right side (LMN facial palsy, lagophthalmos)
- CN 8: impaired hearing right ear
- CN 9: intact
- CN 10: intact
- CN 11: intact
- CN 12: impaired- deviation of tongue to right side

- Sympathetic motor: right ptosis (mild)

HVF 24-2





Relevant Exam Findings

- Right proptosis
- Mild Right ptosis
- Afferent pupillary defect, right eye, with dec. VF
- Anisocoria with right miosis and decreased EOMs
- No convergence response of right eye

Differential Diagnosis

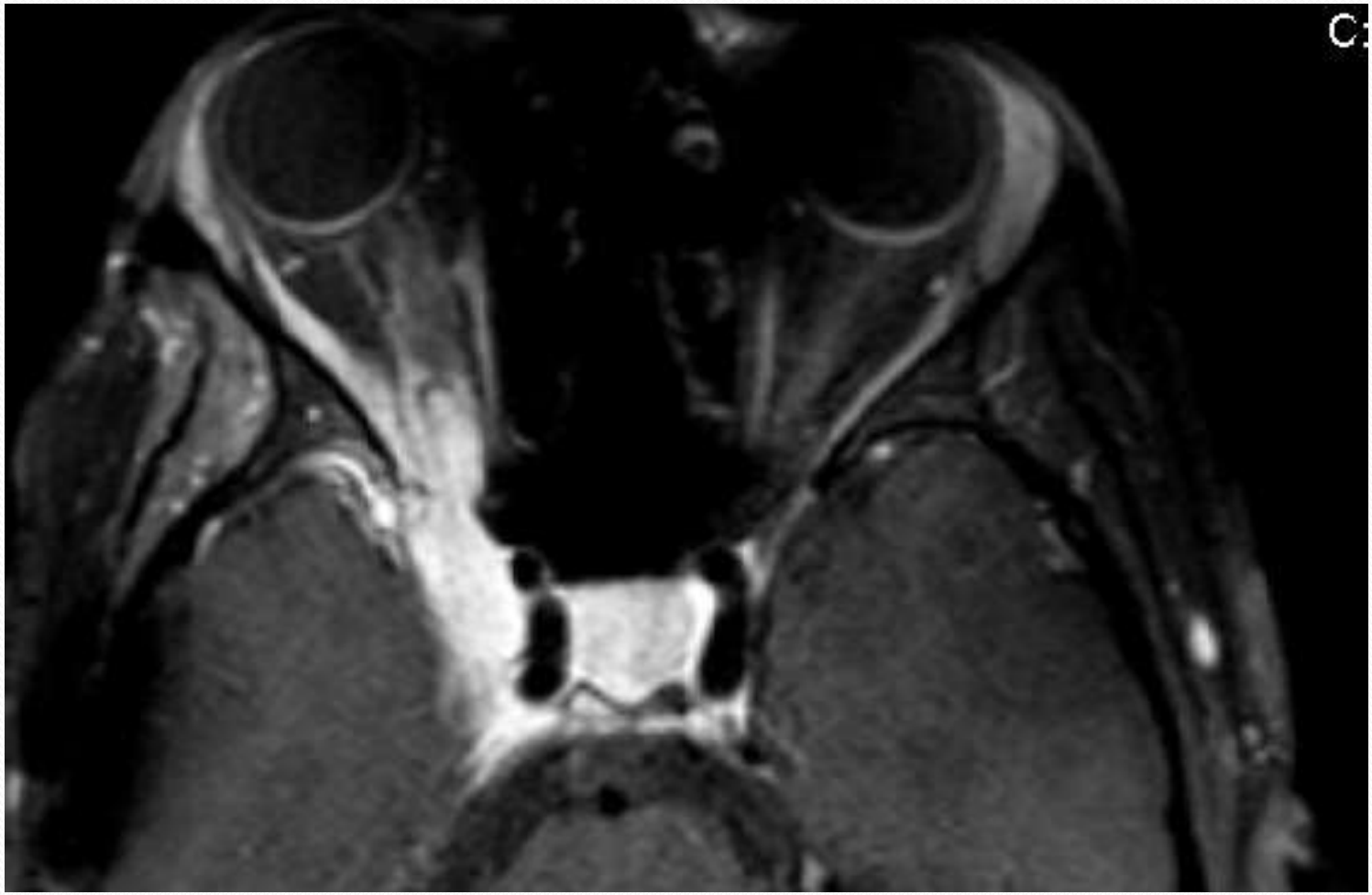
a) *neoplastic*: cavernous sinus/intraconal/apex mass- meningioma, optic nerve glioma, schwannoma

b) *vascular*: cavernous hemangioma, intracranial aneurysm, carotid-cavernous or dural-sinus fistula

c) *inflammatory*: thyroid eye disease, sarcoidosis, inflammatory orbital pseudotumor

d) *miscellaneous*: radiation neuronitis

Imaging



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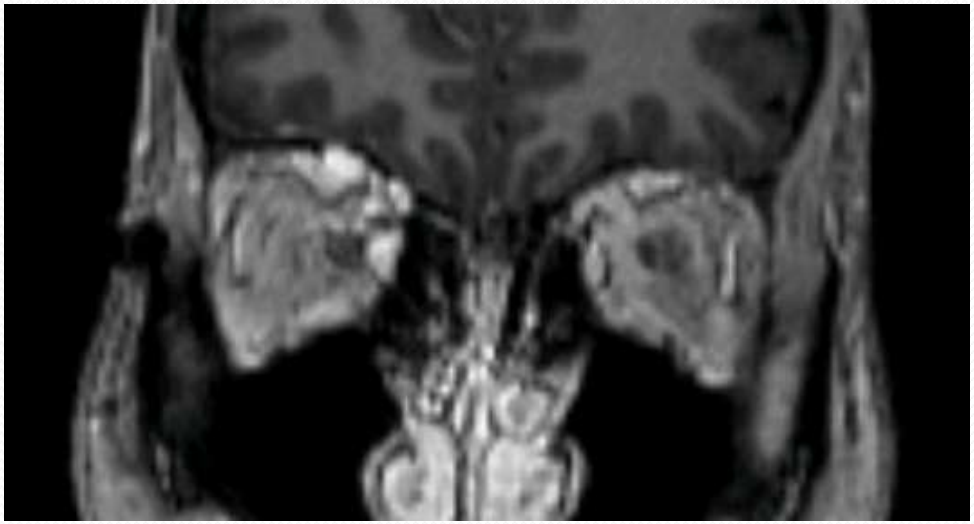
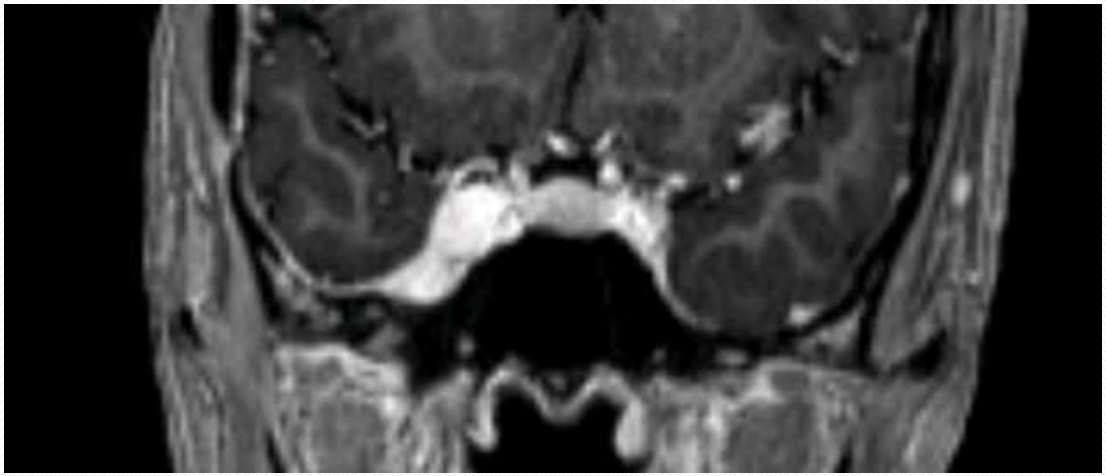




Diagnosis: Neurogenic Ptosis

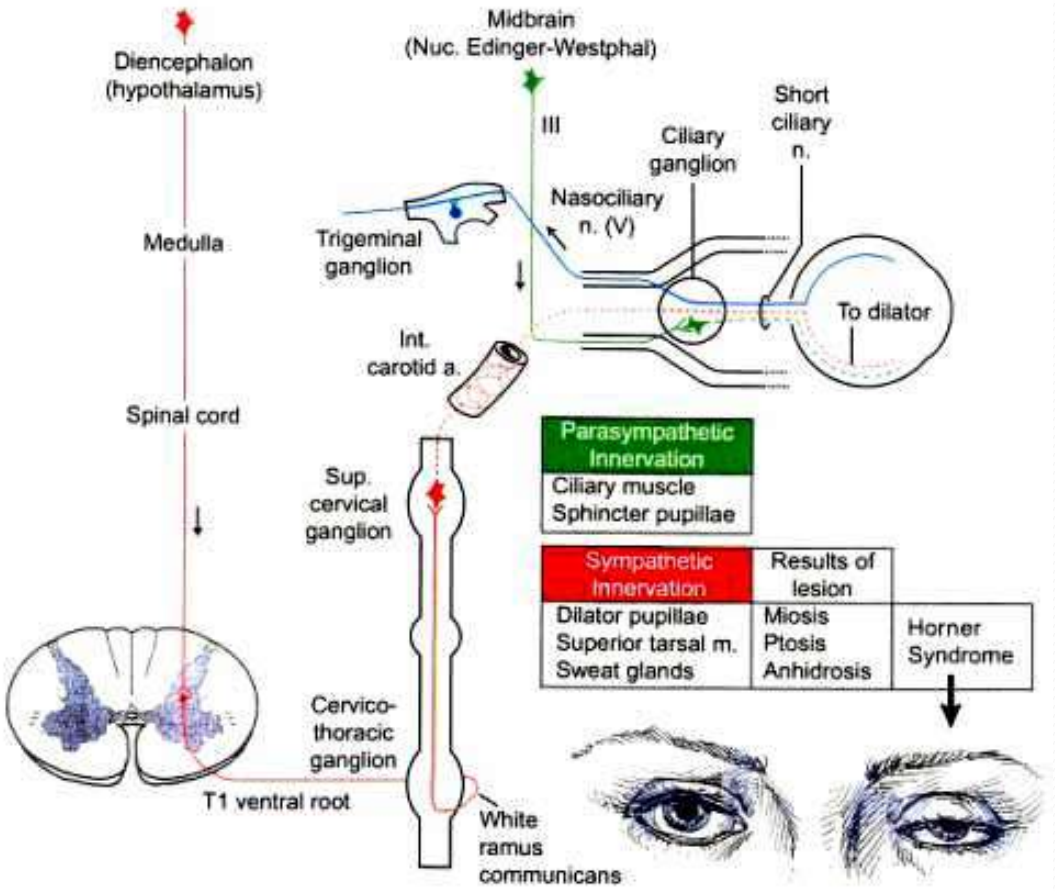
- Horner's syndrome due to expansive tumor involvement of cavernous sinus and orbital apex; likely schwannoma given history of multiple tumors
- Pupil-involving CN 3 palsy

Patient Care, Medical Knowledge

Horner's syndrome

- Results in a neurogenic ptosis due to loss of innervation of the muller muscle
- Classic triad consists of ptosis, miosis, and anhidrosis
- Localizing tests may be performed to determine if the lesion is in a 1st, 2nd or 3rd order neuron

Horner's Syndrome-Anatomy



http://www.dartmouth.edu/~humananatomy/figures/chapter_45/45-7.HTM

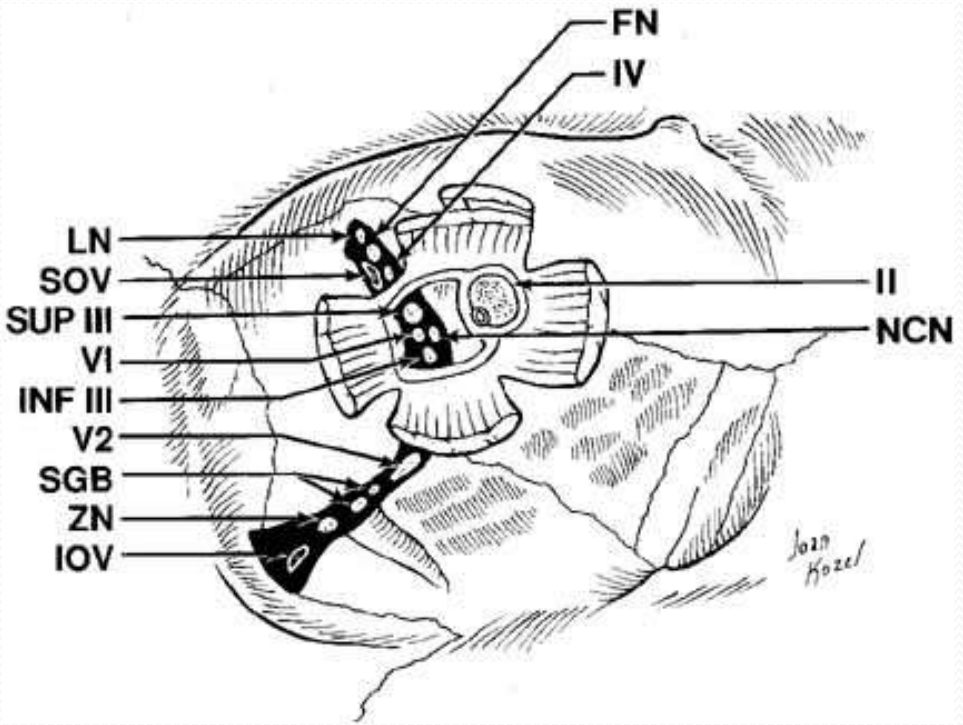
Medical Knowledge

Horner's Syndrome- Localization

- In presence of ataxia, diplopia, vertigo, lateralized weakness: brainstem
- In presence of b/l or ipsilateral weakness, sensory level, bowel and bladder impairment: cervicothoracic
- Arm pain or hand weakness (brachial plexus involvement): lung apex
- Ipsilateral extraocular pareses (esp CN6 palsy): cavernous sinus
- If isolated and accompanied by head and neck pain: ICA dissection

Signs of Orbital Apex Lesions

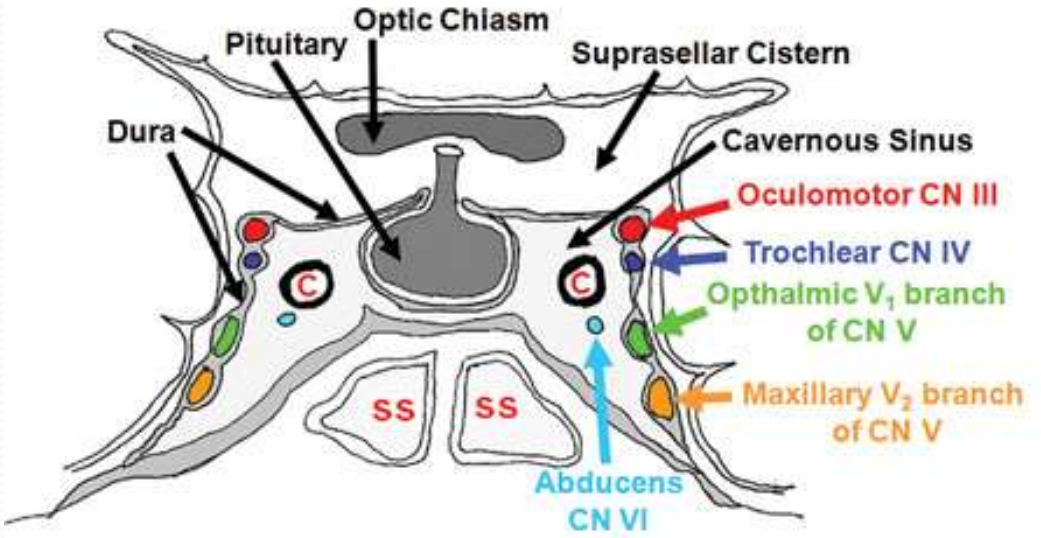
- Ophthalmoplegia (CN 3, 4, 6)
- Ptosis (CN 3- parasympathetics)
- Decreased corneal sensation (CN V1)
- Visual loss (CN 2 involvement)
- Proptosis
- Swelling/chemosis of ocular surface membranes



http://www.oculist.net/downaton502/prof/ebook/duanes/pages/v2/v2c021.html

Signs of Cavernous Sinus Lesions

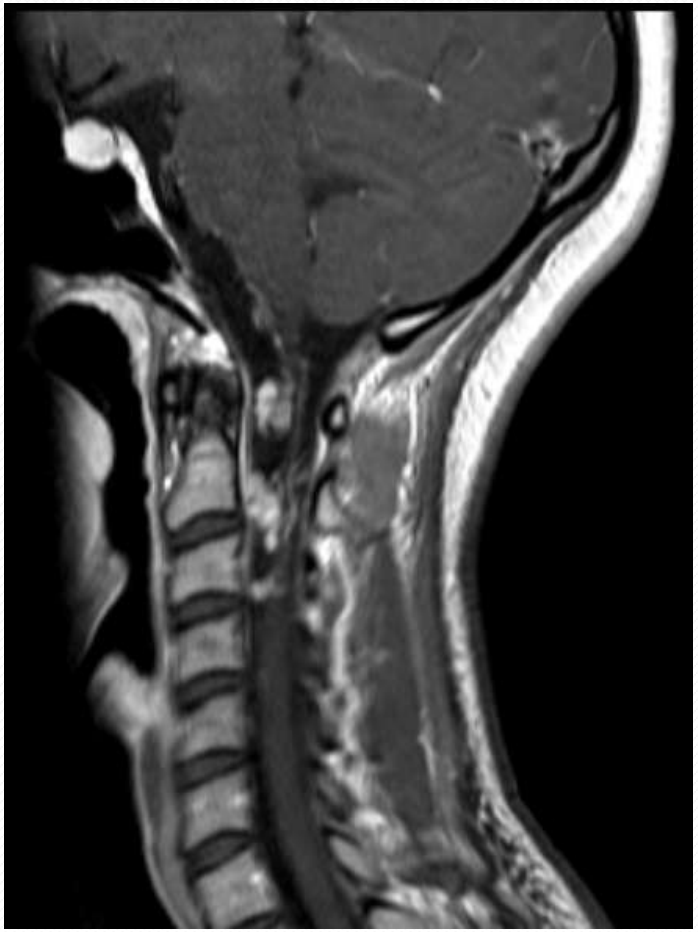
- Unilateral and isolated/combined 3rd, 4th or 6th CN palsies
- Painful ophthalmoplegia
- Proptosis
- Ocular hypertension



Review of Previous Imaging

- Dumbbell neuromas at C2 and C4

- S/p resection of right CN 7 schwannoma (imaging not available)



NF-2 Diagnostic Criteria

- Confirmed/definite: Bilateral vestibular schwannomas
- Presumptive/probable: Unilateral vestibular schwannoma or any two (meningioma, glioma, schwnannoma, juvenile posterior subcapsular lens opacity, juvenile cortical cataract)+ family history
- Monitor patients with:

yearly MRI until age 50, yearly ophthalmic exams, annual hearing screens with brainstem auditory evoked response

Schwannomatosis

- Is the third major form of neurofibromatosis
- Incidence: 0.58 per million persons
- No known gender or race predilection; average age of symptom onset is 25 to 30 y.o.
- Mostly sporadic, 20% familial
- Mutations in SMARCB1 tumor suppressor gene on chr. 22 found in up to 50% of families with schwannomatosis and 10% of sporadic cases

Schwannomatosis

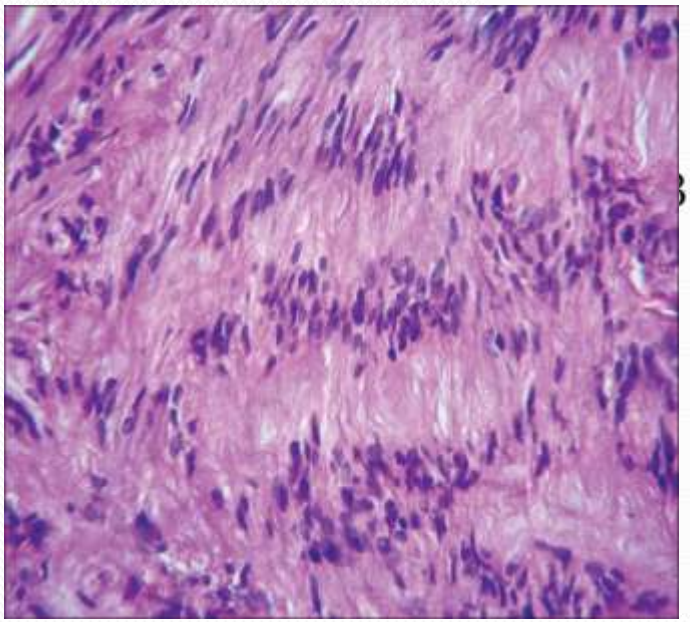
- Typical locations of tumors:
- Peripheral nerves (cranial nerves, arms and legs)
- Spinal nerve roots (in 75% of patients, usually dorsal roots)
- Subcutaneous (20-30% of patients)- skin moves freely

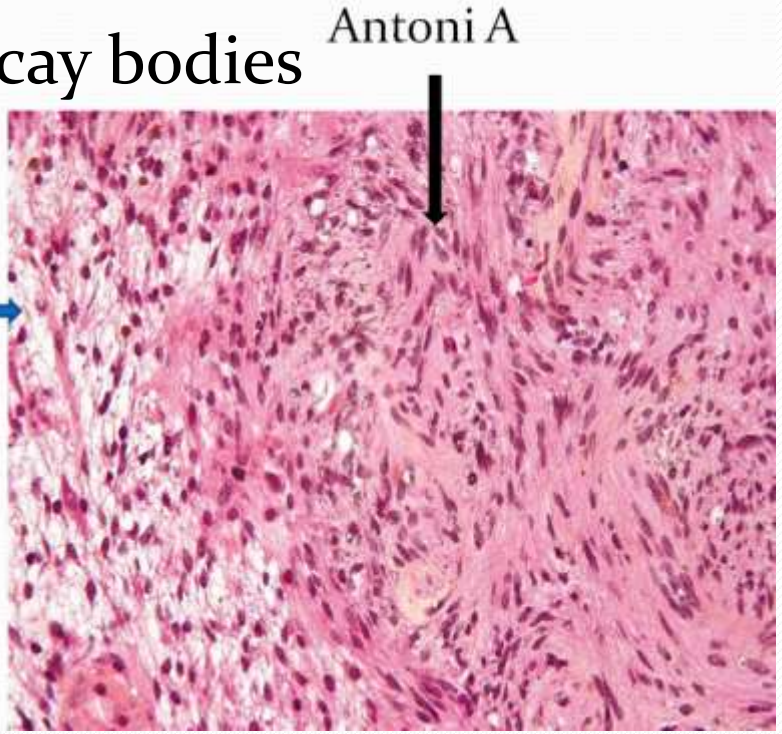
Diagnosis

- Age <30 yrs old AND:
- 2 or more non-intradermal schwannomas, at least one with histologic confirmation
- No evidence of vestibular schwannoma on high-resolution brain MRI with cuts through auditory canal
- No cutaneous or ocular manifestations of NF-1
- No first-degree relative with NF-2, no known NF-2 mutation
- Unmanageable pain is often a key symptom

Schwannomas

- benign, slowly-growing, encapsulated nerve peripheral nerve tumors
- Antoni A, Antoni B, and Verocay bodies





Genetic Testing

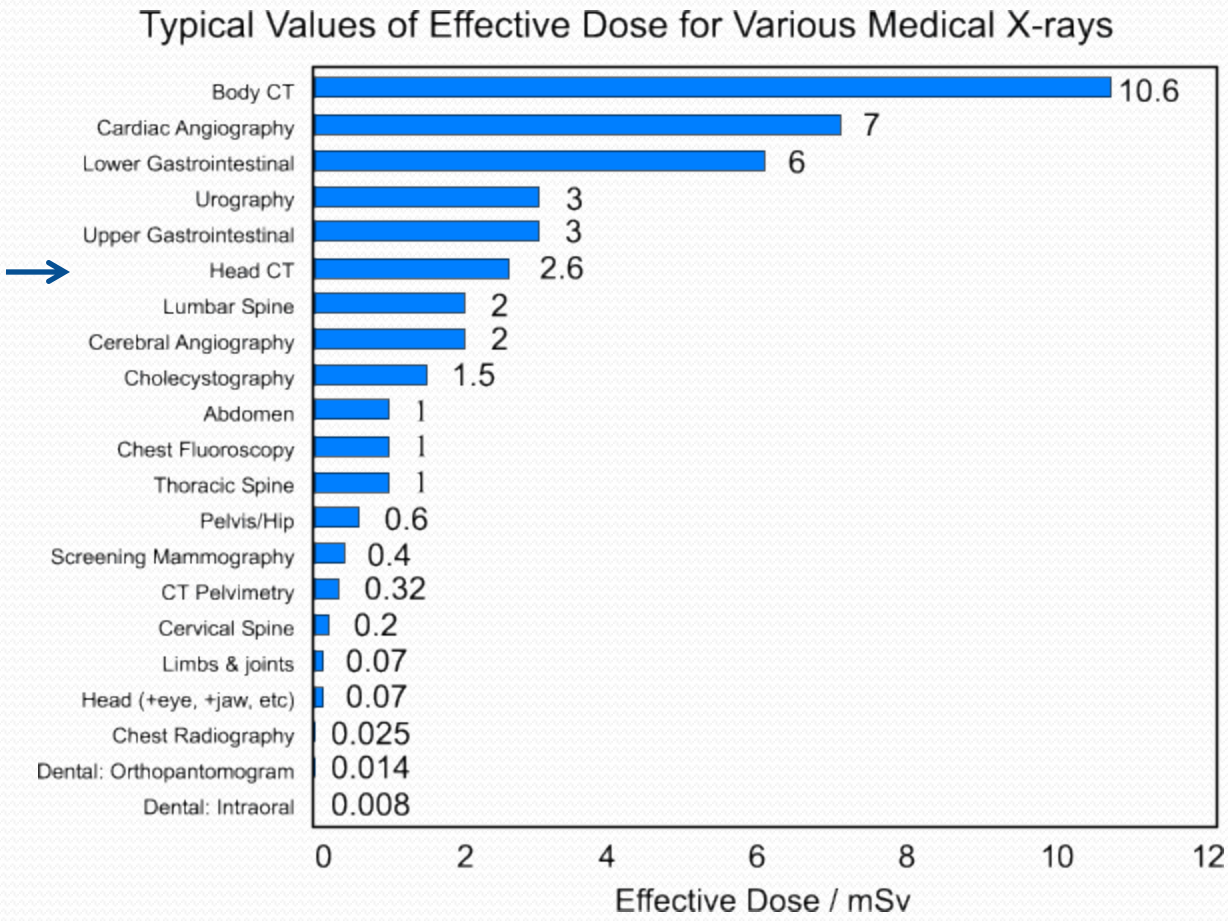
- A molecular diagnosis of NF-2 can rule out schwannomatosis and make counseling more straightforward
- Results of testing generally have no impact on surgical decision-making
- The genetics of schwannomatosis have not been fully elucidated; the possibility of germ-line mutations exists

Schwannomatosis- Treatment

- Most schwannomas may be resected by experienced surgeons
- Intraoperative neurophysiological monitoring may be helpful
- If asymptomatic, yearly MRI scans may be useful for monitoring
- Patients should be educated about early signs/symptoms of a symptomatic schwannoma
- If malignancy is suspected, CT-guided biopsy may be used
- Chemotherapy is generally ineffective

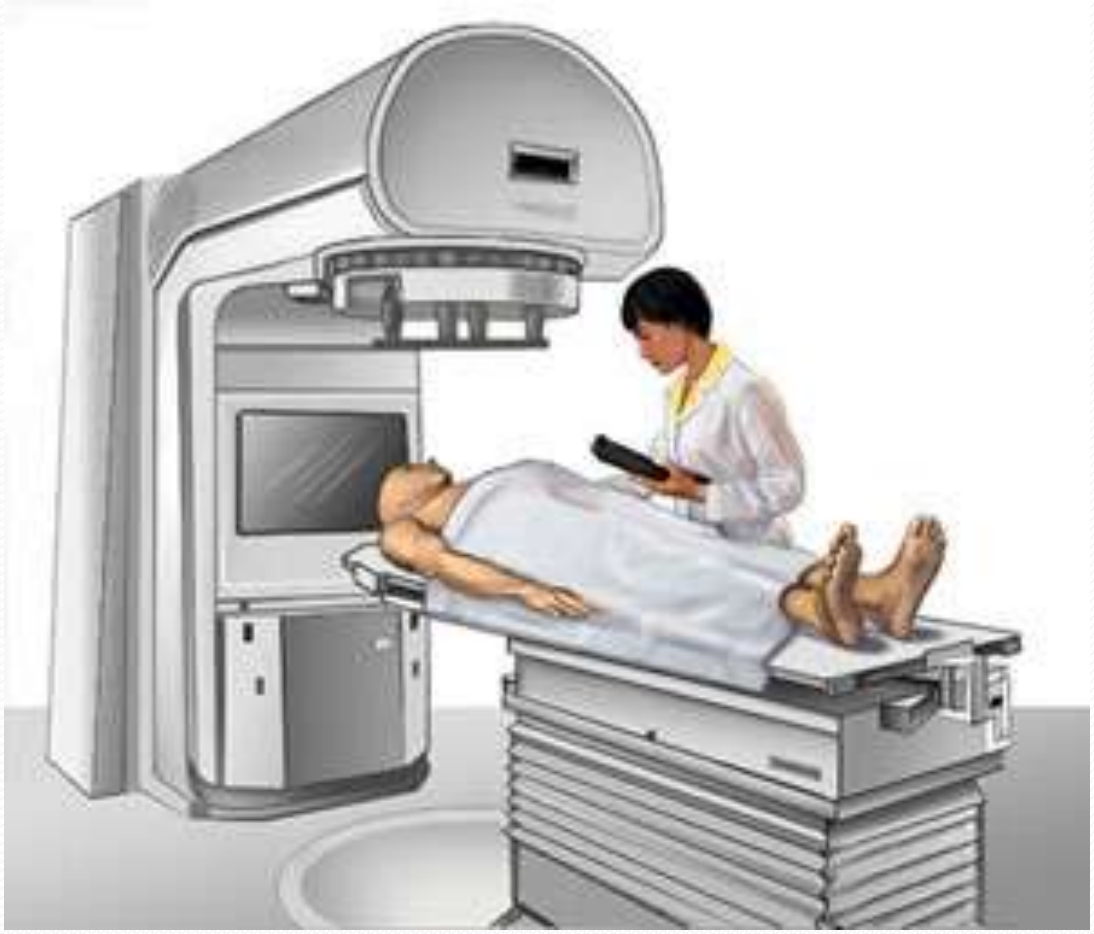
Medical Knowledge, Practice-Based Learning and Improvement

Radiation Exposure- CT Scans



http://www.arpansa.gov.au/radiationprotection/basics/xrays.cfm

External Beam Radiation Therapy

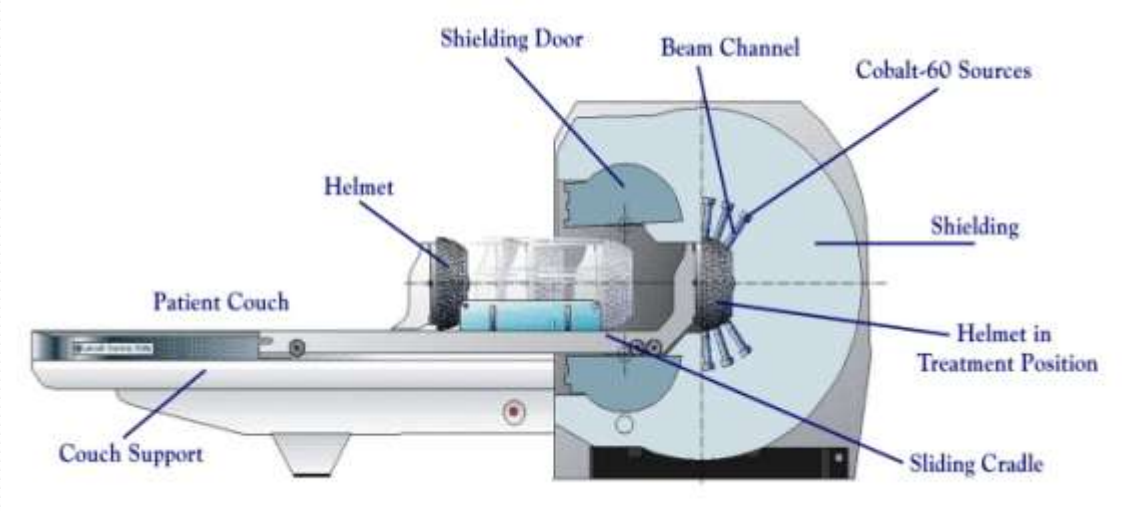


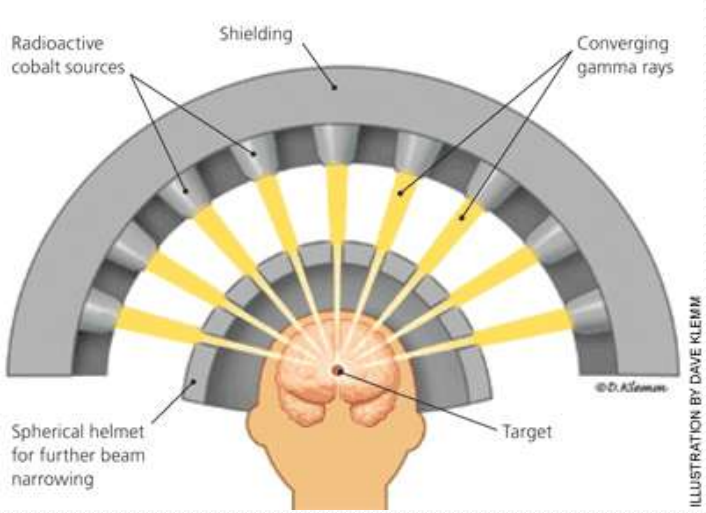
Medical Knowledge

Stereotactic Radiosurgery

- Narrow beams of radiation from different angles deliver precise doses of radiation
- Often used for:
- Acoustic neuromas
- Gliomas/glioblastomas
- Metastatic brain tumors
- Meningiomas
- Pituitary tumors
- Fractionated radiosurgery allows for a larger cumulative dose of radiation

Stereotactic Radiosurgery





www.nejm.org

Medical Knowledge

Side effects of Brain RT

- Heachaches
- Cerebral edema
- Hair loss
- Nausea/vomiting
- Fatigue
- Hearing loss
- Memory/speech problems
- Seizures

Patient Update

- Discussed the case with hematology/oncology and radiation oncology; patient to have radiosurgery performed at Bellevue

Practice-based Learning and Improvement, Professionalism, Interpersonal and Communication Skills, Systems-based Practice

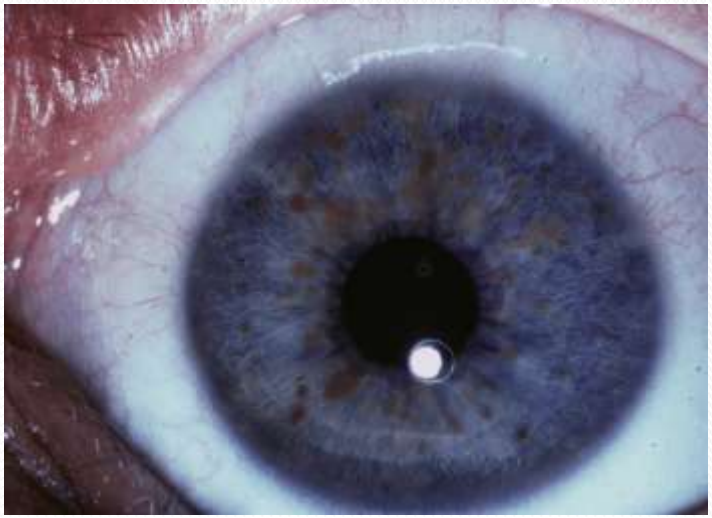
Key Points

- A patient presenting with ptosis must receive careful evaluation with examination for MRD1, levator function, motility, orbital signs
- Cranial nerve exam may aid in localizing orbital apex and cavernous sinus lesions
- NF1 and NF2 both have typical ophthalmic manifestations, whereas schwannomatosis does not

Question 1

- You examine a patient with the following clinical exam. The ophthalmic finding shown below is found in what percentage of patients above age 6?

A) 20%
B) 40%
C) 60%
D) 80%
E) 90%



Medical Knowledge

Question 2

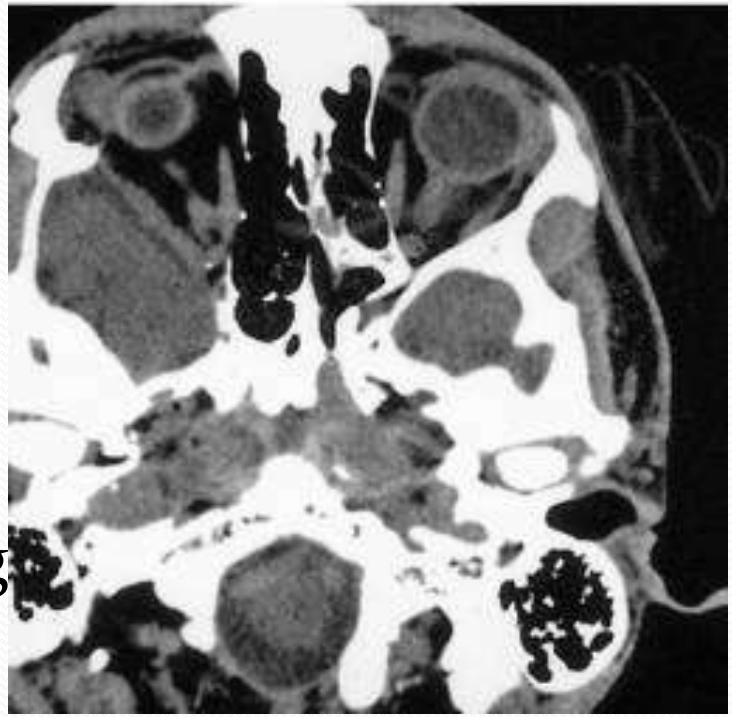
- You are shown the CT scan of a patient with no history of trauma. Which of the following is most likely?

A) The visual acuity of the right eye is normal

B) There is a family history of the disease

C) The patient has intractable pain

D) The patient has difficulty hearing from both ears



Reflective Practice

- This case allowed me to participate in the care of a patient with a new-onset neurogenic ptosis in the setting of multiple cranial nerve tumors. Consideration of the differential diagnosis after a careful clinical exam and communication with radiation-oncology, hematology-oncology, and neurosurgery allowed for appropriate and timely care of the patient.

Core Competencies

- Patient Care: The case involved thorough patient care and careful attention to the patient's past medical history. The patient received workup and appropriate medical management by ophthalmology, hematology-oncology, ENT and neurology services.
- Medical Knowledge: This presentation allowed me to review the presentation, differential diagnosis, proper evaluation/work up for neurogenic ptosis, as well as management of neurofibromatosis.
- Practice-Based Learning and Improvement: This presentation included a literature search of third nerve palsy, its differential diagnoses, workup and treatment.
- Interpersonal and Communication Skills: The patient was treated with respect and every effort was made to communicate with the patient in a logical, understandable manner.
- Professionalism: The patient's workup and treatment was initiated in a timely manner. She was informed of her diagnosis and explained the reasoning behind our workup.
- Systems-Based Practice: The patient was managed in a coordinated manner by the neurology, ophthalmology, hematology-oncology, and ENT services.

Selected References

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- Widemann BC, Acosta MT et al. CTF meeting 2012: Translation of the basic understanding of the biology and genetics of NF1, NF2, and schwannomatosis toward the development of effective therapies. Am J Med Genet A. 2014; 164A(3):563-78.

Thank You

- Our Patient
- Asher Neren
- Dr. Shinder