OPHTHALMOLOGY GRAND ROUNDS

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June 20th, 2013

HPI

40 yo old male presents with complaints of dysphonia, facial pain, nasal congestion, epistaxis, and left sided epiphora.

History

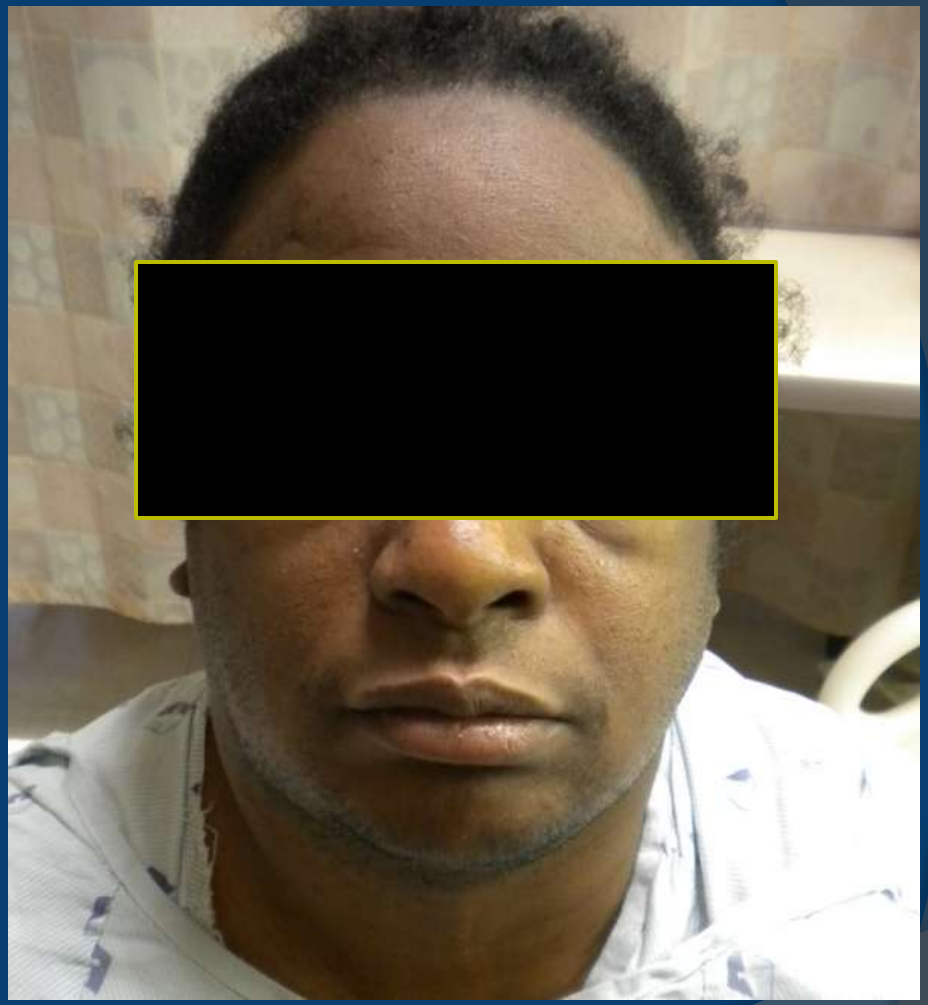
- POH: none
- Gtt: none
- PMH: DM II
- All: nkda
- FH: no known eye disease

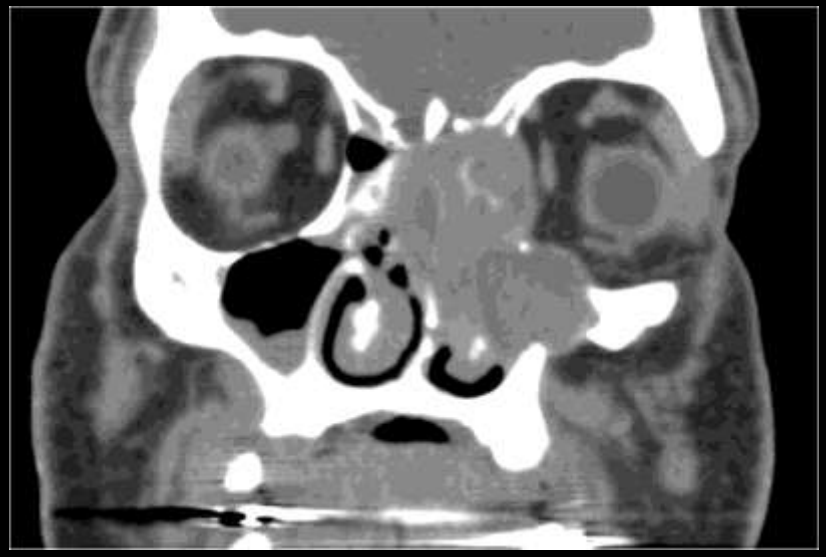
EXAM

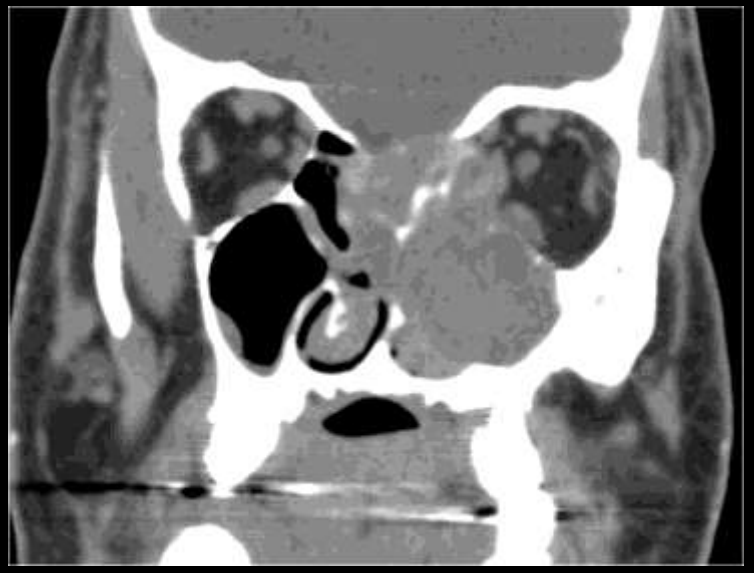
- nVAsc: 20/20, 20/20
- p: r and r, no apd
- EOM: full ou, no pain, diplopia in extreme upgaze and right gaze
- CVF: full ou
- Ttono: 14/14

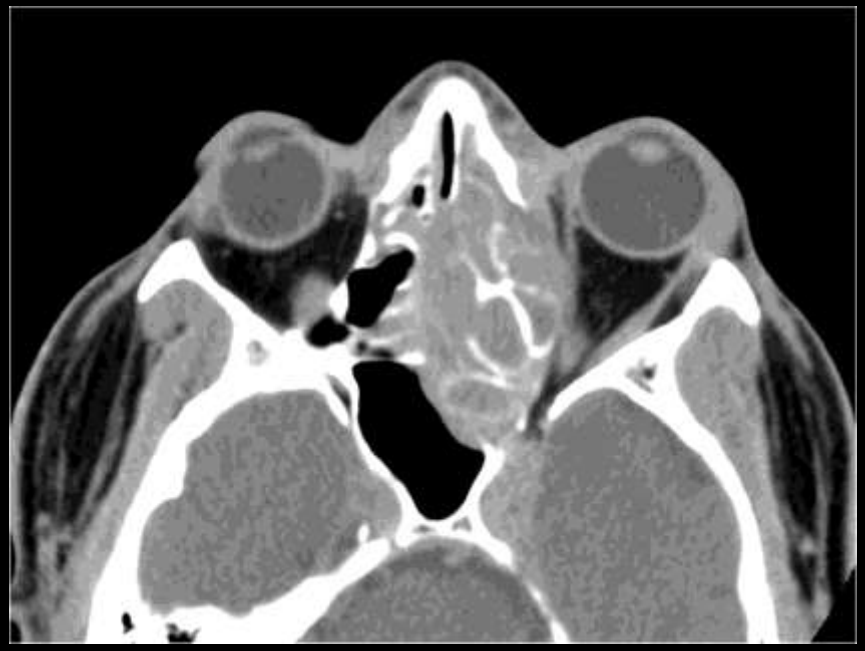
- PLE: wnl ou
- DFE: wnl ou, 0.3 s/p ou

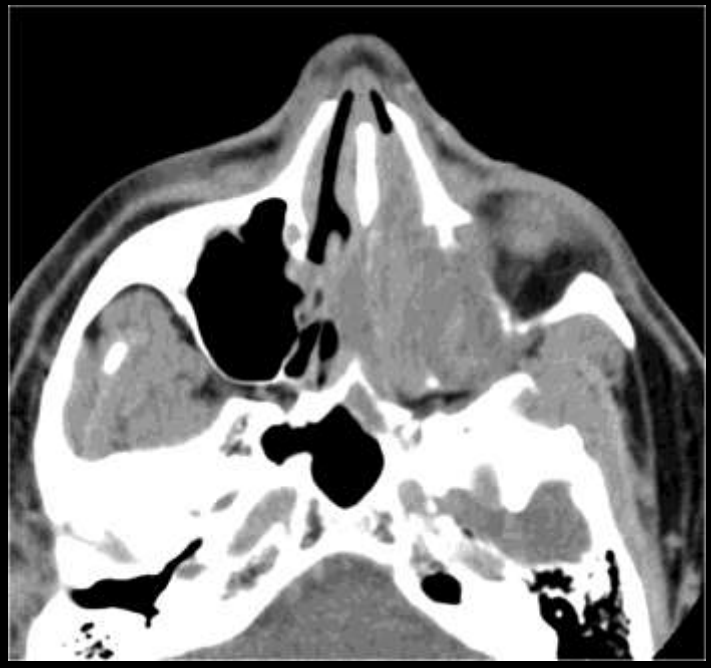
HEENT:
Bilateral submandibular and cervical lymphadenopathy









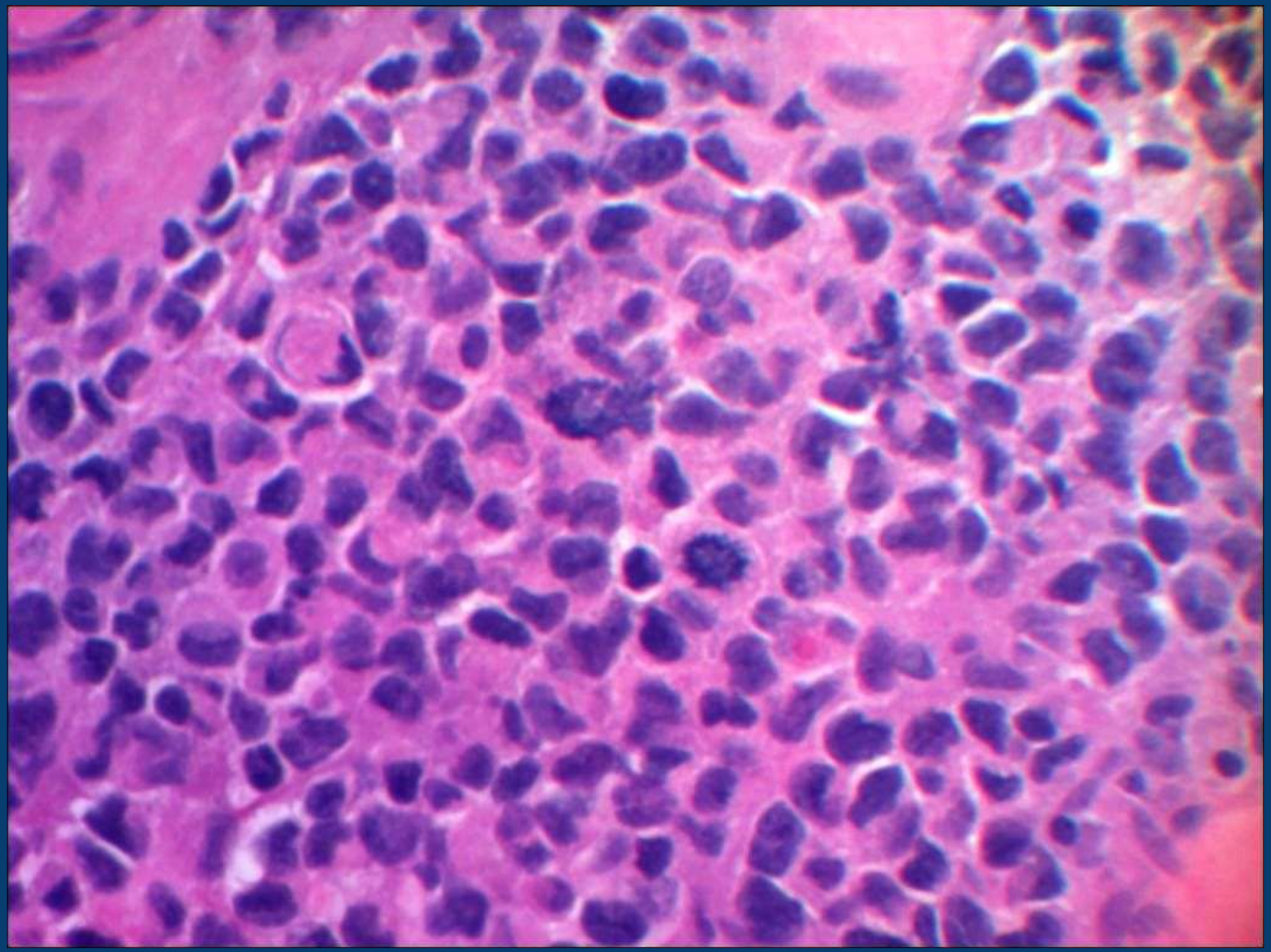


Differential Diagnosis

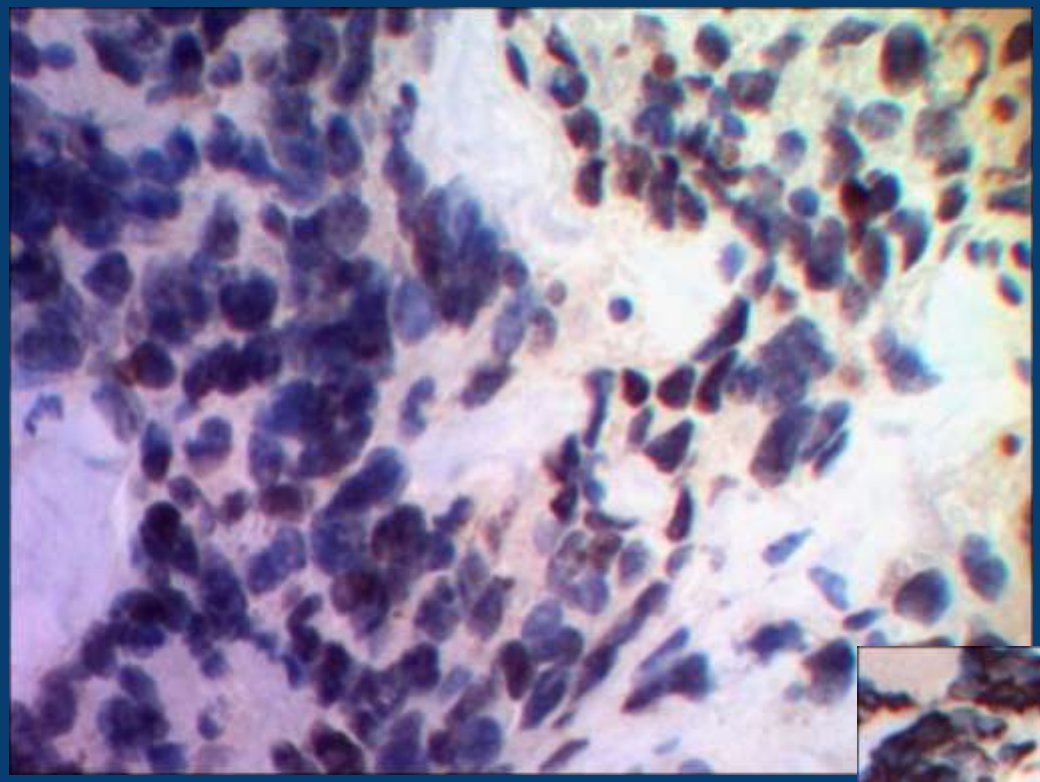
Differential Diagnosis

- Maxillary sinus Squamous Cell CA
- Lymphoma
- Mesenchymal tumor
- Metastasis
- Rhabdomyosarcoma

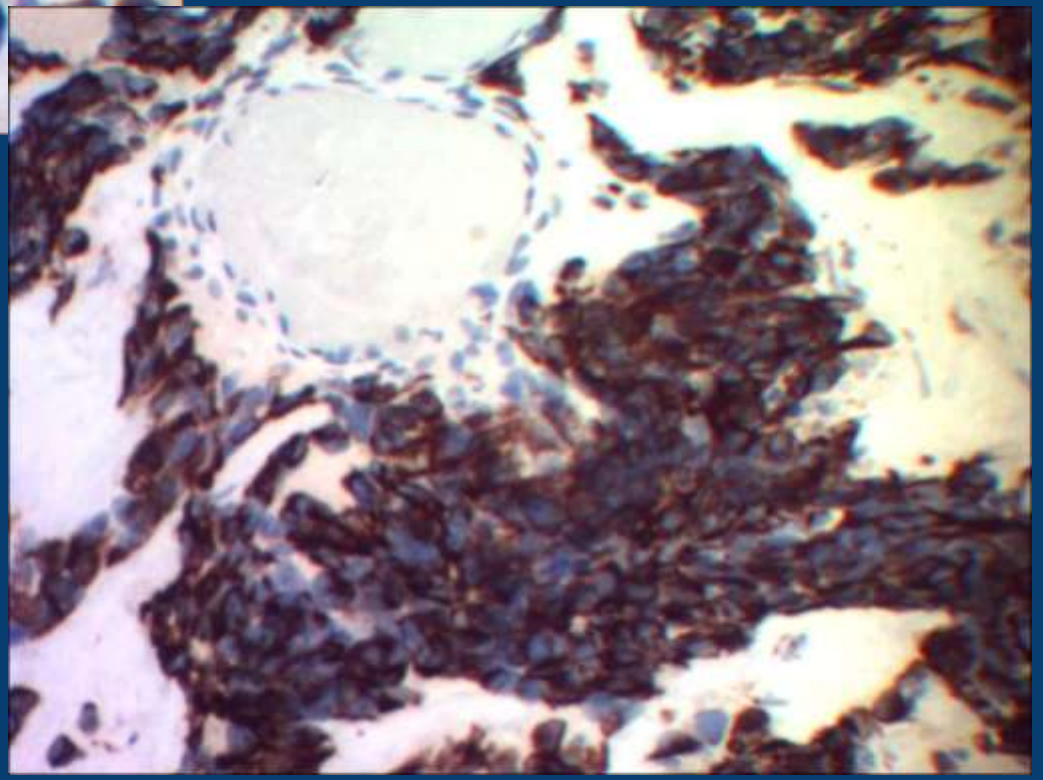
What do you want to do?



Patient Care, Interpersonal and Communication Skills



<= Myogenin positive



Desmin positive ➔

Histiopathology Results

- The tumor is composed of small round blue cells with numerous mitotic figures, necrosis and individual cell apoptosis. The tumor cells stain w Myogenin, Desmin and CD56 supporting the diagnosis for rhabdomyosarcoma, adult-type.

LICH: Small round blue cell tumor, favor adult-type embryonal rhabdomyosarcoma.

MSK: High grade Rhabdomyosarcoma, favor solid alveolar type.

- Composed of cells with histopathologic features of striated muscle in various stages of embryogenesis.
  - Develops from undifferentiated mesenchymal cells that possess the capacity to differentiate into striated muscle
- Most occur in soft tissues of the orbit but they can rarely occur in the other ocular adnexal structures and even within the eye
- Head and neck rhabdomyosarcomas usually appear in the first decade of life.
- Slight predilection of males
- No major predilection for race

- 5% of all childhood cancers

- MOST COMMON primary orbital malignancy in children
- MOST COMMON soft tissue malignancy in children
- MOST COMMON mesenchymal tumor of orbit
- MOST COMMON met is to chest

- DOES NOT originate from extraocular muscles

Adult Rhabdomyosarcoma

- There are only *25 reported cases* of adult RMS of the maxillary sinus from 1950-2000 with two-thirds of these being patients under the age of 30, and less than 20 cases of adult orbital RMS from 1965-2012

Adult Rhabdomyosarcoma

- Common presenting symptoms of adult maxillary RMS include nasal congestion, nasal discharge, epistaxis, facial pain, and proptosis and visual disturbance if RMS invades the orbit
- Tend to be aggressive and commonly spread to the cervical lymph nodes, lungs, and bones.
- The alveolar subtype, the second most common type of adult RMS (20-30%), is mostly undifferentiated, rapidly develops distant metastasis, and has the worst prognosis

History

1854

Dr. Weber is generally credited with providing the first acceptable description of rhabdomyosarcoma, which occurred in the tongue of a 21-year-old man

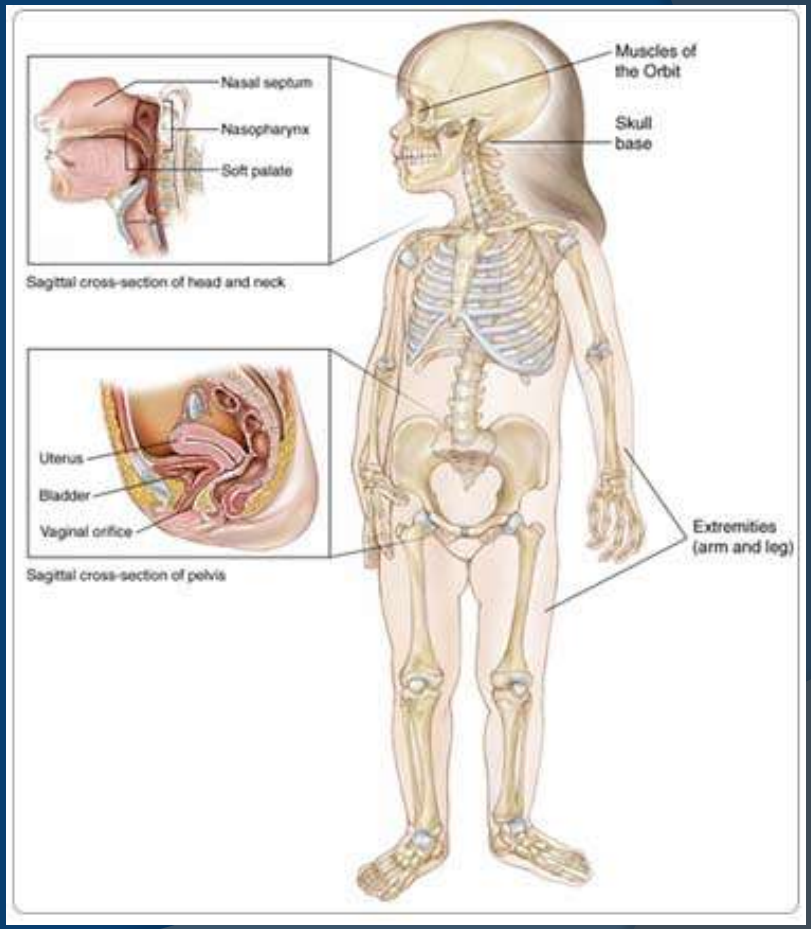
1882

Dr. Bayer first published description of an orbital rhabdomyosarcoma

Where does it occur?

- Head and neck area (45%)
- Trunk (40%)
- Extremities (15%)

*About 25%–35% of head and neck rhabdomyosarcomas arise in the orbit



3 Main Histopathology Types

Pleomorphic

- Usually occurs in adults
- Least common in the orbit
- Best prognosis

Embryonal

- Majority of Orbital Rhabdomyosarcomas
- Good Prognosis
- Botryoid is subtype

Alveolar

- 2nd most common in the orbit
- Worst Prognosis
- Usually in extremities

Primary Ocular Rhabdomyosarcoma

1. Orbit

2. Eyelid

- Rhabdomyosarcoma confined to the eyelid is rare. However, orbital rhabdomyosarcoma frequently presents as a visible and palpable subcutaneous mass deep in the eyelid. Management same as orbital

3. Conjunctival

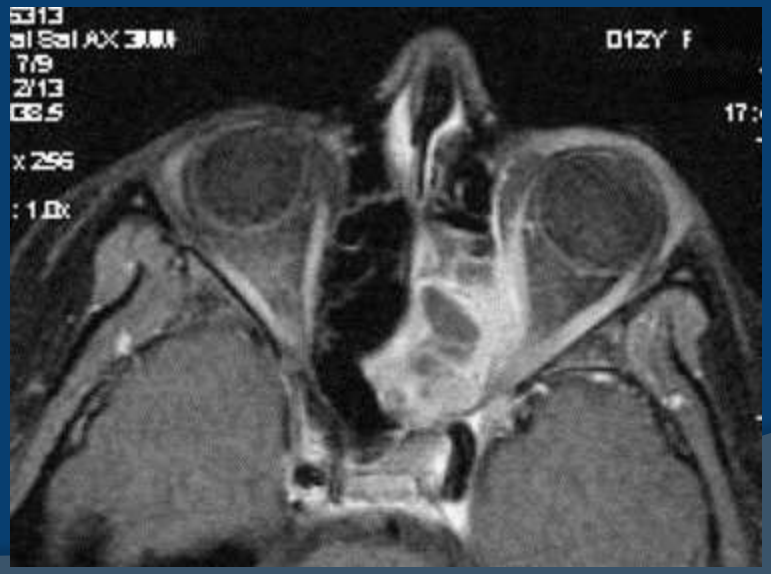
- Rhabdomyosarcoma confined to the conjunctiva is rare. Usually appears as a fleshy pink mass in the forniceal conjunctiva, most often in the superior fornix

4. Anterior Uvea (Iris and Ciliary Body)

- It generally presents in an infant or young child as a solitary, fleshy iris mass that shows slow growth.

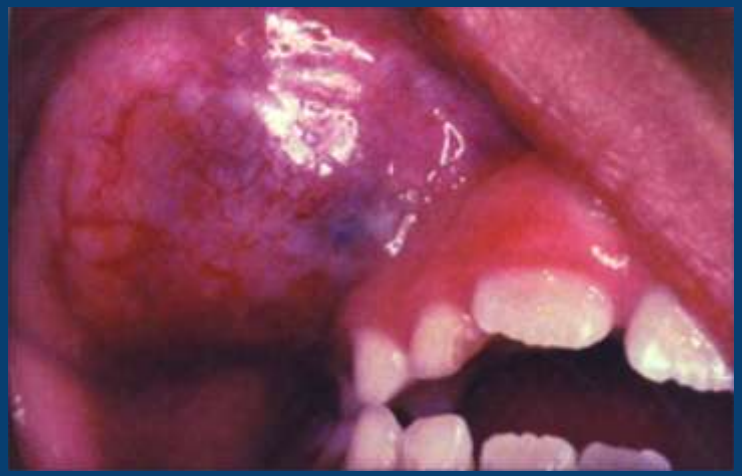
Secondary Orbital Rhabdomyosarcoma

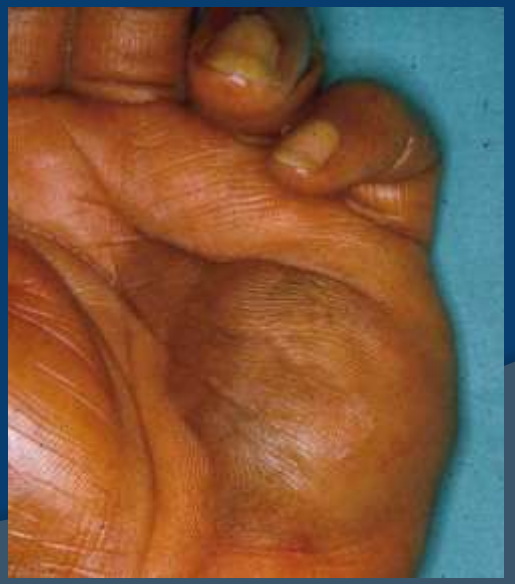
- Direct extension from paranasal sinuses or nasopharynx.
- The patient with sinus or nasopharyngeal rhabdomyosarcoma often presents with signs of sinusitis, nasal congestion, or epistaxis
- Nasopharyngeal rhabdomyosarcoma can be more difficult to diagnose and affected patients can sometimes present with rapid, bilateral visual loss due to invasion of the orbital apices, leading to optic nerve compression.



Orbital metastasis from extraocular rhabdomyosarcoma

- Can be the site of metastasis from a primary neoplasm in other parts of the body without direct communication to the orbit.





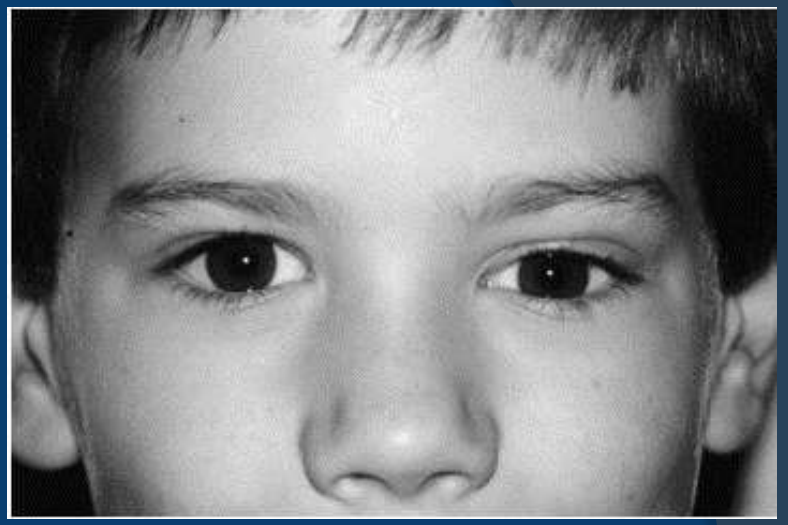
Primary Orbital Rhabdomyosarcoma

25–35% of head and neck rhabdomyosarcomas

10–20% of all rhabdomyosarcomas

Mean age at diagnosis of 8 years

Clinical Features



- proptosis (80–100%)
- globe displacement (80%)
- conjunctival and eyelid swelling (60%)
- blepharoptosis (30–50%)*
- palpable mass (25%)
- pain (10%)

- Others: NLDO, choroidal folds, retinal venous tortuosity, and optic disk edema

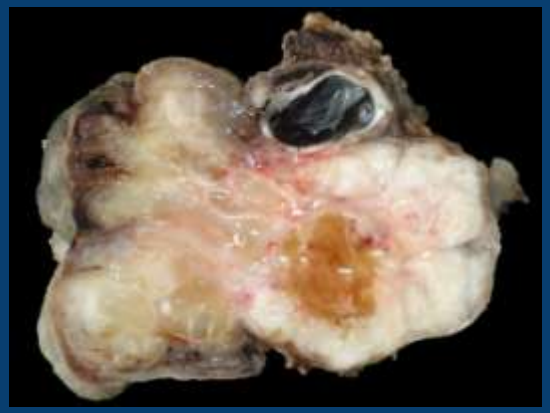
Where can it go?

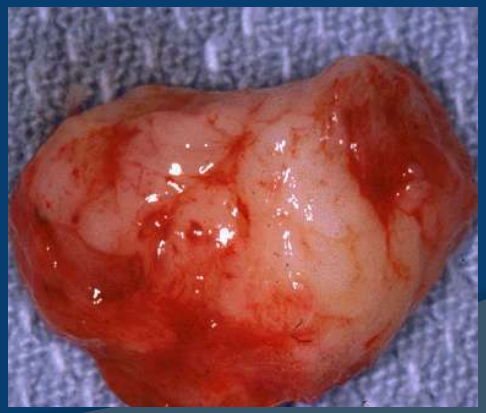
- Orbital bone and into the cranial cavity.
- Mets to lung and bone.
  - Metastasis generally occurs via hematogenous dissemination.
- Regional lymph node metastasis is rare
- Tumors that extend from the orbit anteriorly into the conjunctiva or eyelid can gain access to lymphatic channels

Gross Pathology

- Well circumscribed tumor in the early stages.
- Larger, more aggressive tumors have an irregular border as a result of tumor invasion though the pseudocapsule.

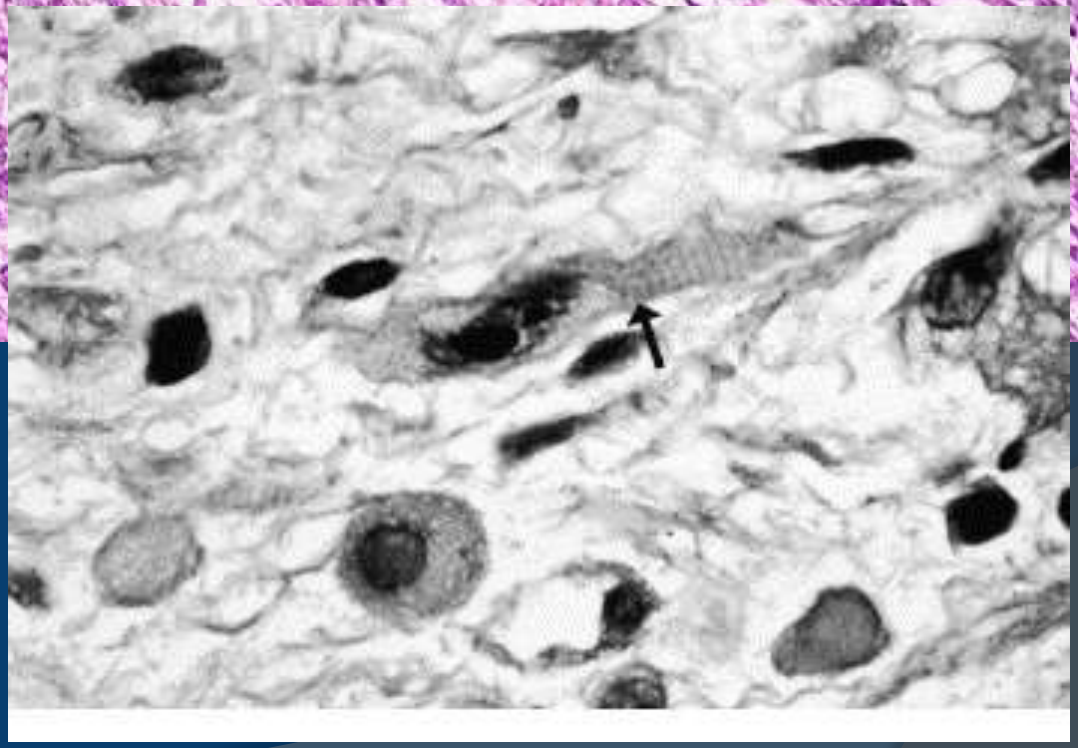
- In fresh sections, the tumor has a light gray to pink color and may show areas of hemorrhage and cyst formation.





Pathology

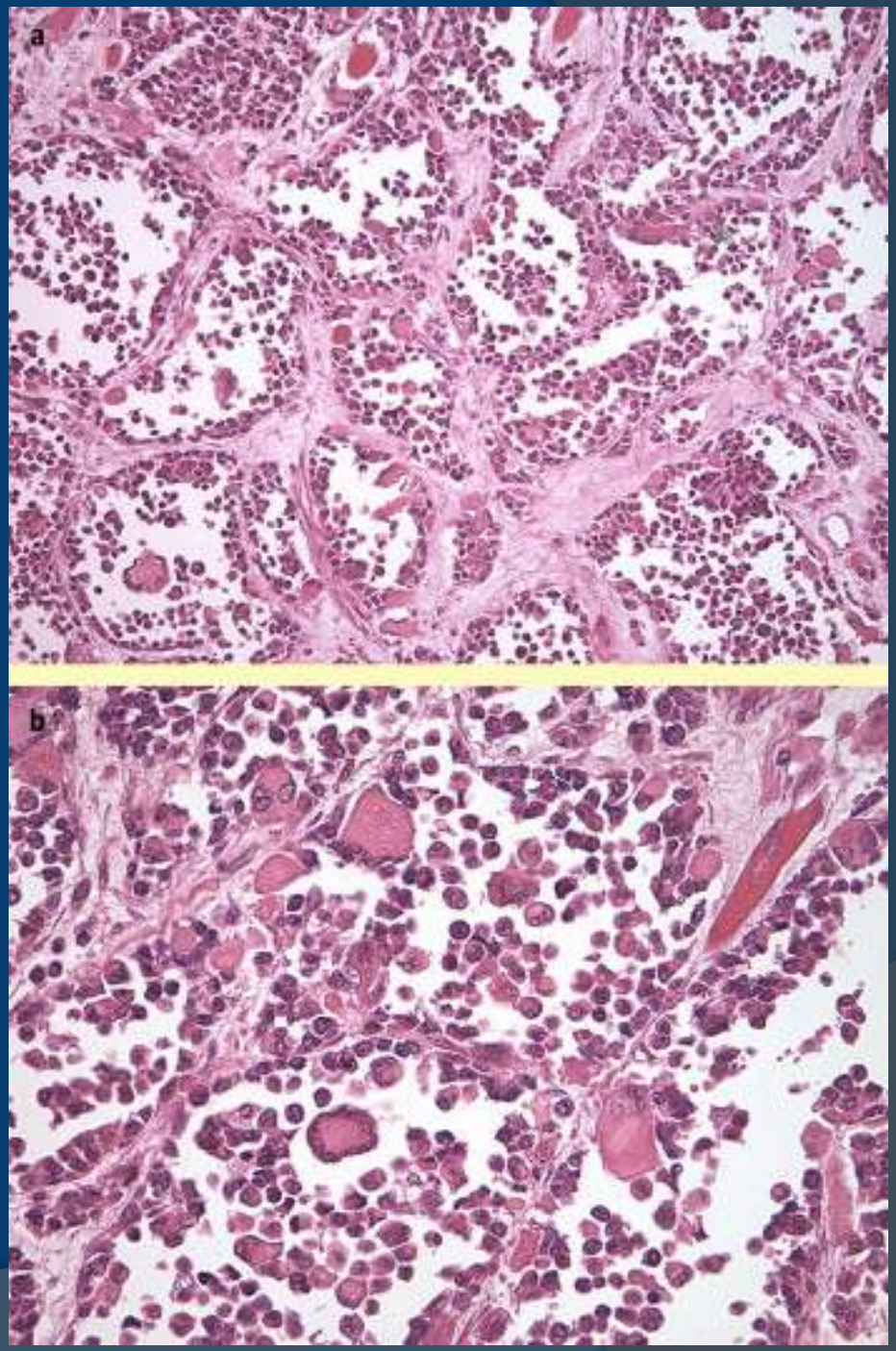
- Embryonal characterized histopathologically by elongated spindle cells with prominent cytoplasm. May show features characteristic of skeletal muscle in various stages of embryogenesis.



- Cross striations can sometimes be identified

Pathology

- The alveolar type appears as loosely arranged, malignant cells with septae that are reminiscent of the alveoli of the lung

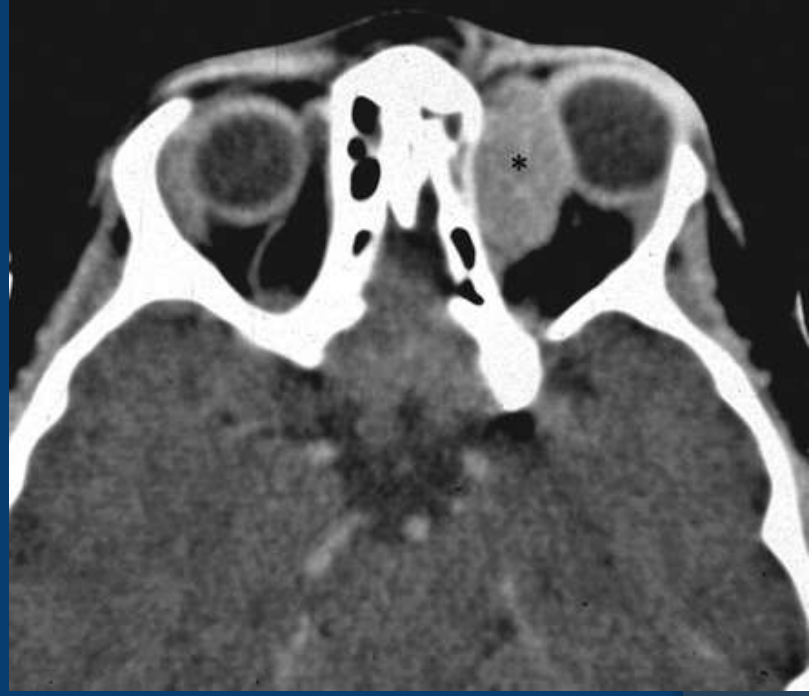


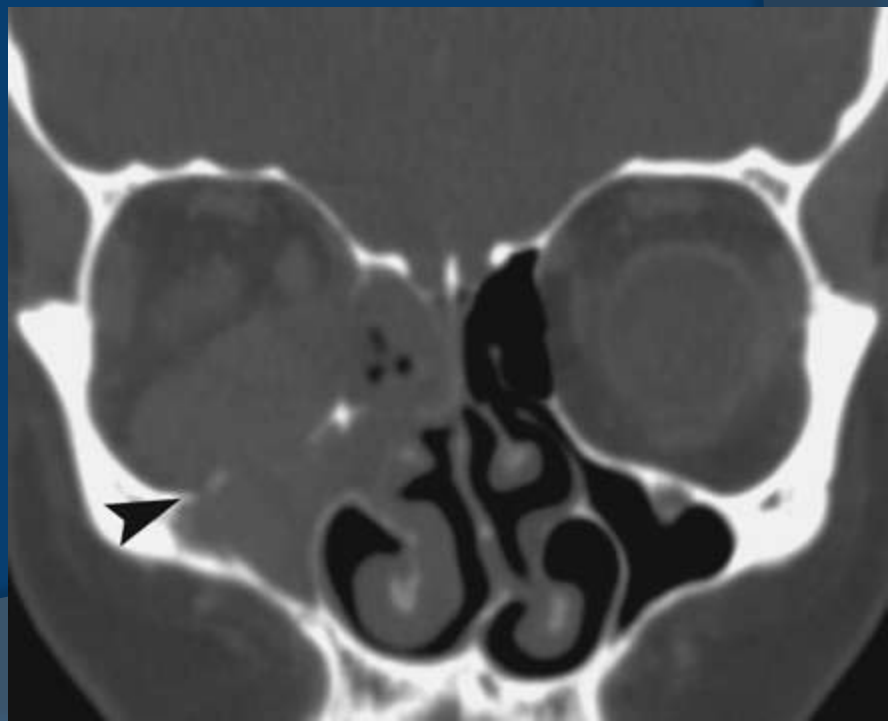
Round Cell and Spindle Tumor Differential

- Neuroblastoma
- Ewing's sarcoma
- Lymphoma
- Fibrosarcoma
- Fibrous histiocytoma
- Leiomyosarcoma
- Malignant Schwannoma
- Alveolar soft-part sarcoma
- Hemangiopericytoma
- Granulocytic sarcoma
- Osteogenic sarcoma
- Nodular fasciitis
- Rhabdoid tumor`

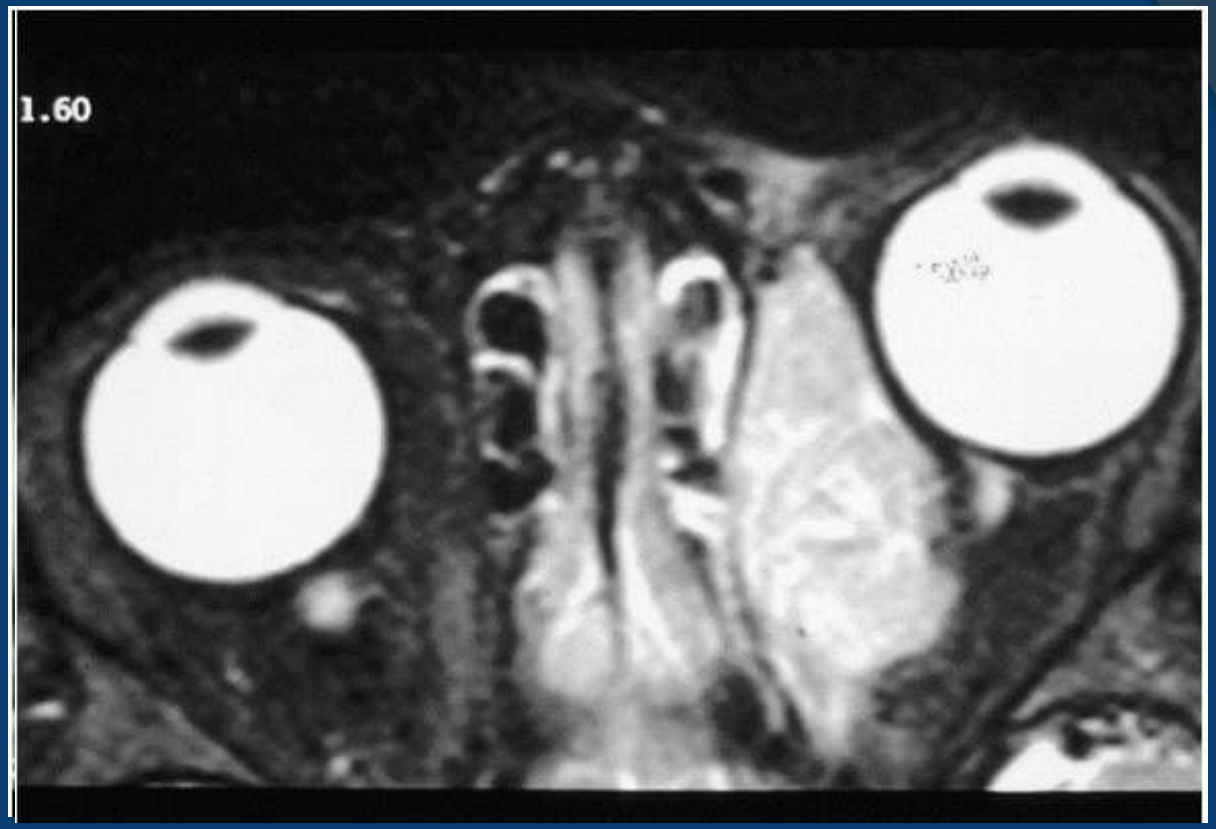
CT

- Early stages as a well-circumscribed, homogeneous, round to ovoid mass that is isodense to muscle
- Usually extraconal and superonasal
- Larger tumors tend to erode bone and occasionally extend to the sinuses or nasopharynx.
- Orbital rhabdomyosarcoma shows moderate to marked enhancement with contrast agents





MRI



- T1-weighted image, the tumor appears in the early stages as a round to ovoid mass that is usually located in the orbital soft tissue superonasally. It usually has a hypointense signal with respect to orbital fat, but is isointense with respect to extraocular muscles
- It generally shows moderate to marked enhancement with gadolinium and is best delineated with fat suppression techniques.
- On T2-weighted image, the lesion is hyperintense to extraocular muscles and orbital fat.

Staging

Table 2. Staging of Rhabdomyosarcoma by the Intergroup Rhabdomyosarcoma Study Group Staging Classification

Group	Description
I	Completely resected localized disease implying both gross impression resection and microscopic confirmation of complete resection and absence of regional lymph node involvement
Ia	Confirmed to muscle or organ of origin
Ib	Contiguous involvement outside the muscle or organ of origin
II	Residual disease and/or regional lymph node involvement
IIa	Grossly resected localized tumor with microscopic residual disease and no evidence of gross residual tumor or regional lymph node involvement
IIb	Completely resected regional disease with no microscopic residual tumor[a]
IIc	Grossly resected regional disease with microscopic residual tumor[a]
III	Incomplete resection with biopsy or gross residual disease
IV	Distant metastatic disease present at onset

[a]Regional disease implies involvement of the regional lymph nodes.

Treatment

1. Surgery
   - Biopsy vs. debulking
   - Keep periosteum intact and do not invade pseudocapsule

2. Irradiation
   - (4,000 to 5,000 cGy) but often with ocular complications

3. Chemotherapy
   - Agents employed: vincristine , actinomycin D, ifosfamide and etoposide.
   - Have greatly increased survival

Prognosis for Pediatrics

- 1970's: 70% mortality…..now >90% Survival

- Better when it's in the orbit because:
  - Get symptoms sooner
  - More favorable morphology
  - Favorable anatomic location
  - Few lymphatics in the orbit

Long-term Survivors of Adult Rhabdomyosarcoma of Maxillary Sinus Following Multimodal Therapy: Case Reports and Literature Reviews

Tsung-Han Wu, MD; Jen-Seng Huang, MD; Hung-Ming Wang[1], MD; Cheng-Hsu Wang, MD; Kun-Yun Yeh, MD

Chang Gung Med J Vol. 33 No. 4
July-August 2010

Age and gender

Patient ages ranged from 15 to 77 years and the median age was 29.2 years. More than two-thirds of patients were younger than 30 years of age at diagnosis, with a rapid drop-off in reported cases thereafter. Fourteen patients were female (60.9%) and nine patients were male (39.1%) in the current analysis. The sex distribution of this study showed a modest preponderance of females to males (1.5:1) overall.

Adult maxillary RMS also tends to have a poor prognosis because of extensive local disease found at diagnosis, resulting in difficult or impossible surgical excision

Head and neck cases account for 18 percent of adult RMS with a 5-year survival rate of 8 percent or less

Our patient

- Systemic workup with PET was positive for bilateral cervical lymph node metastasis
- Currently the patient is undergoing chemotherapy with vincristine, dactinomycin, and cyclophosphamide, an extrapolation of the standard protocol from the pediatric literature. Following induction chemotherapy, the patient will undergo a repeat PET for assessment of any residual disease and will be evaluated for possible surgical resection and post-operative radiation.

Reflective Practice

Orbital rhabdomyosarcoma is one of the few life threatening diseases seen initially by ophthalmologists and prompt diagnosis and treatment can save the life of the affected patient.

Therefore, eye care physicians should be aware of this tumor, recognize its clinical features, and refer the patient for prompt treatment.

Core Competences

- Patient Care: The case involved thorough patient care ability to explain findings and need for treatment to patient. Once diagnosed, the patient received proper management and care.
- Medical Knowledge This presentation allowed us to review the presentations, proper evaluation/work up, and previous cases
- Practice-Based Learning and Improvement: This presentation included a current literature search of developing associations and current treatment modalities.
- Interpersonal and Communication Skills: The patient was treated with respect and every effort was made to communicate with the patient and treat regarding his wishes
- Professionalism: The patient was treated in the proper manner.
- Systems-Based Practice: The patient was discussed with colleagues and treated appropriately

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Thank you

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