4.14.6. Orbit (VI): Neoplasia (IV): Rhabdomyosarcoma

- most common primary malignant orbital tumor of childhood
- origin
  - primitive undifferentiated pluripotential mesenchymal cells that differentiate toward skeletal muscle
  - not from the extraocular muscles
- clinical presentation
  - average age=7-8 years
  - sudden onset and rapid progression of unilateral proptosis

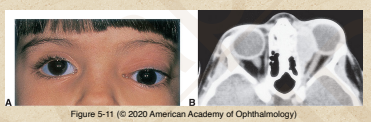
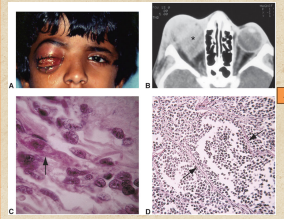


Figure 5-11 (© 2020 American Academy of Ophthalmology)

  - less dramatic course in patients in their early teens
    - gradually progressive proptosis lasting weeks to > 1 month
  - edema & reddish discoloration of eyelids
    - NOT accompanied by local heat or fever
  - ptosis
  - strabismus
  - ± palpable mass
    - particularly in the superonasal quadrant of the eyelid
    - may be retrobulbar
    - may involve any quadrant of the orbit
    - may rarely arise from the conjunctiva
  - unrelated history of trauma can lead to delay in diagnosis and treatment
- histologic types
  - 4 categories
    1. Embryonal
       - most common type
         - 80%
       - may develop in conjunctiva
         - grapelike submucosal clusters
         - botryoid variant
       - predilection for the superonasal quadrant of the orbit
       - loose fascicles of undifferentiated spindle cells
         - immature rhabdomyosarcomas
           - occasional cells show cross-striations (60%)
         - well-differentiated rhabdomyosarcoma
           - numerous cells with striking cross-striations
           - trichrome staining
       - immunohistochemistry
         - desmin
         - muscle-specific actin
         - vimentin
         - ± myogenin
       - electron microscopy
         - sarcomeric banding pattern
       - 94% 5-year survival



A, B, C: Embryonal; D: Alveolar

Figure 14-13 (© 2020 American Academy of Ophthalmology)

    2. Alveolar
       - 9%
       - predilection for the inferior orbit
       - poorly cohesive rhabdomyoblasts
         - separated by fibrous septa into alveoli
         - rounded rhabdomyoblasts either line up along the connective tissue strands or float freely in the alveolar spaces
       - most malignant form
         - 65% 5-year survival
    3. Pleomorphic
       - least common
       - straplike or rounded cells
       - most differentiated
         - cross-striations are easily visualized with trichrome stain
       - best prognosis
         - 97% 5-year survival
    4. Botryoid
       - rare variant of embryonal rhabdomyosarcoma
       - grapelike
       - not found in the orbit as a primary tumor
         - secondary invader from the paranasal sinuses or from the conjunctiva

- diagnostic workup
  - should proceed urgently
  - to define the location and extent of the tumor
    - CT and MRI
  - biopsy
    - anterior orbitotomy
      - often possible to completely remove a rhabdomyosarcoma if it has a pseudocapsule
        - the smaller the residual tumor, the more effective the combination of adjuvant radiation and chemotherapy in achieving a cure
      - in diffusely infiltrating tumors, a large biopsy specimen should be obtained so that adequate material is available for
        - frozen sections
        - permanent light-microscopy sections
        - electron microscopy
        - immunohistochemistry
      - more readily apparent on electron microscopy
      - cross-striations are often not visible on light microscopy
  - to evaluate for regional metastases
    - palpate the cervical and preauricular lymph nodes
  - chest radiography
  - under anesthesia at the time of the initial orbital biopsy
    - bone marrow aspiration and biopsy
    - lumbar puncture
- treatment
  - Intergroup Rhabdomyosarcoma Study Group guidelines
  - radiation therapy
    - 4500 to 6000 cGy
    - x 6 weeks
    - adverse effects of radiation
      - common in children
      - radiation dermatitis
      - bony hypoplasia
      - cataract
  - to eliminate microscopic cellular metastases
    - systemic chemotherapy
  - for recurrent cases
    - exenteration
- prognosis
  - survival rate >90%
    - if the orbital tumor has not invaded or extended beyond the bony orbital walls
    - orbital rhabdomyosarcoma better than extraorbital