Rhabdomyosarcoma

Definition
Malignant tumour of skeletal muscle.

Epidemiology
Males greater than females.
Embryonal rhabdomyosarcoma is the commonest soft tissue tumour in children under 15.

Two-thirds embryonal	10-20
One-third pleomorphic	50-70

Classification
A. Pleomorphic or adult
B. Embryonal or childhood
   a. Alveolar
   b. Botryoid
   c. Spindle cell

Pathogenesis
No record of rhabdomyomas transforming into rhabdomyosarcomas.
There is a chromosomal translocation between chromosomes 2 and 13.
There is an association with low levels of p53 tumour suppressor gene.

Clinical
Mass in soft tissues.
In children the presence of an enlarging intramuscular mass is highly suggestive.
Embryonal rhabdo is more common in the head and neck than the extremities. It is also found in the genitourinary tract and retroperitoneally.
Pleomorphic rhabdo is more common in the buttock and thigh.

Natural history
Both forms are high-grade malignancies with rapid growth patterns.
The primary site of metastasis is the lung, but unlike other sarcomas lymphatic mets are not rare.

Radiology
Plain
Soft tissue swelling without bony change
Intralesional ossification and calcification is not seen

CT
Iso-dense with skeletal muscle hence poorly visualised on CT

MRI
Well visualised. Intramuscular location
Medium density on T1, high density on T2

Isotope scans
There is increased uptake in the early vascular phase (this is the case for virtually all high-grade soft tissue sarcomas).
There is no increased uptake in the late bone phase (there is increased uptake with liposarcomas).

Diagnosis
Gross pathology
Infiltration of surrounding tissues with no distinct capsule. Readily distinguishable from skeletal muscle.
Firm, grey to pink colour, bulges when cut.
Larger lesions may have extensive areas of haemorrhagic necrosis that may obscure the neoplastic tissue.

Histology
Largely homogeneous lesion with an infiltrative pseudocapsule.
1. Embryonal
   a. Vast majority of cells are small round blue cells clustered together in clumps with no particular pattern. The clusters of cells are separated by thin wisps of myxoid like matrix.
   b. Well-differentiated cells have an eosinophilic cytoplasm with cross striations.
   c. Poorly differentiated lesions are difficult to tell apart from other small round blue cell tumours and require EM, or positive immunohistochemical studies.
2. Alveolar
   a. This is an uncommon variant of embryonal rhabdo.
   b. Composed of well differentiated round cells arranged in an alveolar pattern separated by thick fibrous septae.
3. Botryoid
   a. This is another uncommon variant of embryonal rhabdo.
   b. Grape like clusters of round cells are massed beneath the

mucosal lining of hollow organs – particularly the bladder.

4. Pleomorphic
   a. Lesional cells are spindle shaped with marked variation in size and shape.
   b. Bizarre gigantic cells with huge hyperchromatic nuclei are scattered around. These cells can resemble a wristwatch with the watch representing the nucleus and the band representing the cytoplasm.
   c. Other configurations are tadpoles and belt buckles.

Electron microscopy
Cross fibrils

Immunohistochemistry
Myoglobin, desmin, actin, vimentin

Treatment
Extremities

1. Staging – MRI, CT, plain XR, CXR, CT chest, PET scan, bone scan
2. Neoadjuvant chemotherapy; this makes limb salvage practicable in most cases
3. Closely followed by wide surgical excision
4. If adequate surgical margins are not obtained postoperative radio-therapy is given
5. Postoperative chemotherapy is then continued for 2 years
6. Five year survival rate is 80% for stage I and II lesions

Radiotherapy
Definitive radiotherapy provides excellent palliation but is seldom curative.

Prognosis
Botryoid best; then embryonal, pleomorphic and alveolar.