Case 2

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Sunset
History

- 24 year old male presented with a 3 day history of right flank pain, sharp in nature.
- Denies fever, chills, hematuria or dysuria.
- Past history: left side kidney stone.
- PE: enlarged left supraclavicular lymph node.
CT SCAN

- Multiple masses in the pelvis measuring up to 10 cm in maximum dimension, along the aorta and inferior vena cava
- Differential diagnosis: lymphoma vs. testicular neoplasm
- A 3.5 cm left supraclavicular lymph node was biopsied and submitted to rule out lymphoma
LEFT SUPRACLAVICULAR LYMPH NODE
Cytokeratin
Summary of immunologic findings

Positive: Pankeratin, EMA, Vimentin,
Focally positive: desmin, WT1, CD99, MSA
Negative: LCA, Tdt, chromogranin,
synaptophysin, S100, TTF-1, CK7 and CK20
Differential diagnosis

- Ewing’s sarcoma/primitive neuroectodermal tumor group
- Embryonal rhabdomyosarcoma
- Alveolar rhabdomyosarcoma
- Neuroblastoma
- Malignant lymphoma
- Site specific neoplasms: Merkel cell carcinoma for scalp, small cell undifferentiated ca & solid variant of adenoid cystic ca for head and neck region (parotid)
Molecular findings

Molecular cytogenetic analysis of the paraffin block using fluorescent in situ hybridization (F.I.S.H) is positive for 22q12 translocation - highly characteristic marker for Ewing’s sarcoma or desmoplastic small round cell tumor.
Desmoplastic small round cell tumor

- Highly aggressive neoplasm described by Gerald and Rosai (1991)


- Predilection for young males (15-41 yrs)
- Male to female ratio: 4:1
- Case reports of this tumor in very young children, in elderly male and in females have been reported

  *The expanding clinical spectrum of desmoplastic small round cell tumor: a report of two cases with molecular confirmation. Hum Pathol. 1999; 30:430-435*
Clinical presentation

- Most common clinical presenting symptoms and signs: Peritoneal cavity abdomen, abdominal pain, abdominal distension, palpable abdominal mass.

- Other primary sites: paratesticular region pleura, posterior cranial fossa, soft tissue and bone, ovary and parotid gland.
Desmoplastic small round cell tumor

- Arise from an undifferentiated cell with potential for multiple lines of differentiation
- *Gerald et al.* suggested that DSRCT can be a primitive tumor histologically related to mesothelium (*mesothelioblastoma*)
  1. prevalence of its main location (peritoneal and pleural surfaces)
  2. repeated demonstration of chromosomal translocation involving **WT1** gene, also expressed in primitive mesothelium

- Subsequent paper described tumor involving parotid glands, brain, scalp and ethmoid sinuses
Desmoplastic small round cell tumor

- Neoplastic masses are solid, firm, multilobulated with gray tan cut surfaces
- Tumors consist of solid sheets, large nests, small clusters and cords of cohesive small, round, cells lying in a hypocellular, desmoplastic, collagenous stroma
- Tumor cells small with hyperchromatic nuclei, clumped chromatin, inconspicuous nucleoli and ill defined light eosinophilic cytoplasm
- Mitotic figures are numerous
- Necrosis, dystrophic calcification and rosette like structures may be seen
Desmoplastic small round cell tumor

- Specific immunohistochemical staining profile trilineage coexpression (epithelial, muscular and neural markers)

- Immunohistochemical staining profile:
  - Cytokeratin 88%
  - CAM 5.2 87%
  - Desmin 81% (dot like)
  - Vimentin 88%
  - NSE 84%
  - CD99 23%
  - Actin 3%
  - WT1 91%

Desmoplastic small round cell tumor

- Reciprocal translocation
  \[ t(11;22)(p13;q11 \text{ or } q12) \]
- Fusion of exon 7 of the Ewings sarcoma gene EWS on chromosome 22 with exon 8 of the Wilm’s tumor suppressor gene WT1 on chromosome 11
- The chimeric transcript corresponding to the fusion gene product can be detected by reverse transcriptase-PCR
Treatment and prognosis

- Chemotherapy: cyclophosphamide, doxorubicin, vincristine, ifosfamide and etoposide
- Recommendation: adjuvant abdominopelvic radiotherapy followed by surgery and chemotherapy
- DSRCT: highly aggressive clinical course with multiple local recurrences
- The average survival is <2 to 3 years
Desmoplastic small round cell tumor

- It is important to consider DSRCT as a possible diagnosis when a young male presents with non specific abdominal symptoms and radiologic evidence of a disseminated, intraabdominal malignancy
- Small round cells in a fibrous stroma
- Trilineage immunohistochemical expression
- EWS/WT1 chimeric transcript: t(11;22)(p13;q12)
Desmoplastic small round cell tumor - References