Case #7
Gladwyn Leiman, MD

22 year old male
FNA biopsy of an irregular abdominal “nodularity”
Case history

- 22 year old sportsman
- Vague abdominal discomfort
- One month delay in seeking medical care
- Irregular abdominal nodularity
- Multiple omental masses on CT scan
- Clinical diagnosis: Likely lymphoma
On site evaluation

INTRA-ABDOMINAL DESMOPLASTIC SMALL ROUND CELL TUMOR OF YOUNG MALES

DIFFERENTIAL DIAGNOSES

- All small round blue cell tumors of childhood
- Lymphoma and leukemia excluded
- Small cell sarcomas eg synovial sarcoma
- Metastatic melanoma
- Peritoneal mesothelioma
Intra-abdominal desmoplastic small round tumor of young males
Desmoplastic small round cell tumor of young males
Small round cell tumor of young males
Small round cell tumor
Tumor

"The destruction of an entity", described by Gerald and Rosai in 1989, 1991
DSRCT - 1

- **Age**: Mean 20.8 years
- **Gender**: M:F 2:1......5:1
- **Location**: Abdomen, pelvis, scrotum, thorax head and neck
- **Symptoms**: Vague mass, pain, distension
- **Location**: Serosal masses, nodules, seeding
- **No attachment to any organ**

DSRCT - 2

- **Unifying pathologic features**: SRBC nests separated by abundant hyaline
- **Divergent differentiation**: Epithelial, mesenchymal, myogenic, neural
- **EM** – intermediate filaments, lipid, glycogen. **NO** microvilli or neural granules

DSRCT - 3

- **Immune Profile**: Keratin, EMA, VIM, DES, WT1, CD99 [Not LCA, SYN, CGA,]
- **Genetics**: Characteristic reciprocal translocation, t(11;22) (p13;q12) [differs from Ewings, PNET, WT]
- **Prognosis**: Aggressive, abysmal, inevitably fatal
Desmoplastic round cell tumor (with divergent differentiation) is a distinct entity.

- It has a unique chromosomal translocation
- It probably has an oncogenic relationship to Wilms' tumor and Ewing's sarcoma
- Cell of origin possibly primitive mesothelium, submesothelial mesenchyme

What happened to this patient?

- Tumor reduction surgery x2 a year apart
- Multiple courses chemotherapy
- Never played sport again
- Internet Support Group
- Serious depression and inanition
- Died two years later at Christmas

Why it was important

- On-site presence at FNAs = 50% diagnosis
- Correct diagnosis possible on FNA with no prior experience of entity
- Diagnosis was new to clinicians
- Behind a ‘homerun’ may be a patient with a terrible disease, years of physical and emotional trauma, and a helpless anguished family
Desmoplastic Small Round Cell Tumor

Twenty years ago, in 1989, Gerald and Rosai first drew attention to this tumor entity which seemed to involve young male patients in their teens and twenties, affecting predominantly the peritoneal cavity, without organ involvement. Similar tumors have, however, subsequently been seen in female patients, in older adults up to the age of approximately 50, and have been found to involve a wide variety of sites of sites of origin such as pelvic peritoneum, tunica vaginalis, pleura, lung, and even head and neck sites such as scalp, ethmoid and posterior cranial fossa. In the intervening two decades, less than 200 cases have been reported. The probability of the individual cytopathologist seeing a primary tumor of this type is thus very low, and the likelihood of developing expertise, negligible. It would seem that correct cytodiagnosis would be difficult in the context of primary tumor recognition in FNA.

Symptomatology varies according to the site involved. In abdominal cases, which still predominate in the literature, the initial symptoms may be vague and misleading, involving dyspepsia or abdominal distension. Most abdominal cases are very advanced at the time of presentation, involving lobulated nodularity and masses of the peritoneum and omentum, but conspicuously not arising in any underlying organ. Histologic appearances are classic, consisting of well-defined masses of small round blue tumor cells separated by very abundant desmoplastic stroma. Divergent differentiation is a feature of this tumor, complicating the histologic appearance. Tumor cells themselves are small, with very high nuclear to cytoplasmic ratios, minimal cytoplasm, apoptosis, molding, necrosis and mitotic figures. The cell of origin is thought to be primitive mesothelium or submesothelial mesenchyme. Differential diagnosis includes all the small round blue cell tumors of childhood. However, the histopathology is fairly distinctive and a limited panel of immunochemistry usually suffices to exclude other small round cell tumors, and confirm this entity. These tumors are almost always positive for epithelial markers, Vimentin, WT1 and CD99. Many are positive for desmin, and neural markers such as CD56. They are, however, negative for chromogranin and synaptophysin as well as for leukocyte common antigen. If necessary, genetic studies can be done; this tumor harbors a specific karyotypic abnormality, namely t(11;22) (p13;q12), a fusion of Ewing’s sarcoma gene and Wilm’s tumor suppressor gene.

Prognosis of this small cell tumor is extremely poor; it is more aggressive than many of the other small round-celled tumors of young patients. Despite combinations of surgery, radiation and chemotherapy, it is regarded as inevitably fatal; a handful of patients have been reported to survive with tumor for more than five years. It is important to distinguish desmoplastic small round cell tumor from other entities such as Ewing’s, PNET, small cell carcinoma, lymphoma, neuroblastoma, and Wilm’s tumor.

- This is a rare tumor, mainly of young males, predominantly in the abdominal cavity, with many extraneous sites and many divergent forms of differentiation described.
- Symptomatology is very vague and diagnosis usually delayed.
- Cytopathologic diagnosis is possible in the context of the clinical picture in abdominal cases.
- Histopathologic appearances are characteristic and the cytogenetics is unique.
- This tumor is aggressive and inevitably fatal with current chemo-radiation and surgery.
Limited bibliography:


- Chang, F. Desmoplastic small round cell tumors. Arch Pathol Lab Med 2006;130:728-732