Interesting Case

Los Angeles Society of Pathologists
June 9, 2009

Miriam Peckham, PSF
Loma Linda University Pathology Department
Clinical Presentation

• 28-year-old male
• Left epigastric pain
Gross Examination

• 15 cm mass in greater omentum

• Firm, gray-tan heterogeneous surface
Immunohistochemistry Profile

Positive for:
- Cytokeratin Cocktail
- Vimentin
- EMA
- NSE
- CD99

Negative for:
- Chromogranin
- GFAP
- S-100
- Desmin
Diagnosis

Desmoplastic Small Round Cell Tumor (DSRCT)
DSRCT

- First described 20 years ago
- Rare
- Young males (15-35)
- Peritoneal based
  - abdomen and pelvis
  - often multicentric or multinodular
DSRCT

• Lacks definite visceral origin
  • Mesothelial cells?
• Small, round, nested blue cells within a desmoplastic stroma
“Your results may vary”

• Occasionally spindled, rhabdoid, signet-ring or pleomorphic cells
Polyphenotypic

• Shows neural, mesenchymal, and epithelial markers

  • Majority:
    • EMA and cytokeratins
    • Vimentin and desmin
    • Neuron-specific enolase

  • Uncommon:
    • Chromogranin and synaptophysin

• Expresses WT1 - Wilms Tumor Gene
Genetic Characteristics

Reciprocal translocation of chromosomes 11 and 22

\[ t(11;22)(p13;q12) \]
t(11;22) Translocation

• Fusion of WT1 gene (chromosome 11) and EWS gene (chromosome 22)
• Different fusion product than \( t(11;22) \) of Ewing’s sarcoma
WT1

• EWS-WT1 activates IGF-1
  • Promotes cellular proliferation

• Overexpression
  • Dual expression of mesenchymal and epithelial markers
Differential Diagnoses

• Important to differentiate from other small blue cell tumors
  • Small cell carcinoma
  • Merkel cell carcinoma
  • Poorly-differentiated carcinoma
  • Malignant mesothelioma
  • Lymphoma
  • Rhabdomyosarcoma
  • Ewing’s sarcoma/PNET
  • Wilms tumor
  • Neuroblastoma
Differential Diagnoses

• Diagnostic consideration based upon:
  • Age group - Pediatric to young adult?
  • Location - Intra or extra-abdominal?
  • Staining patterns - Polyphenotypic?

• Definitive diagnosis:
  • Presence of \( t(11;22)(p13;q12) \)
Behavior and Prognosis

• Very aggressive (<2 year survival)
• Often multifocal at presentation
Treatment

• Surgical excision

• Adjuvant chemo and radiotherapy
  • Lal & Sue, et al, showed 55%, 3-year survival with combination therapy
DSRCT—Summary

• Intra-abdominal small round cell tumor of childhood, often multicentric

• Polyphenotypic marker profile
  • Desmin, cytokeratin, NSE

• Characteristic 11;22 translocation

• Aggressive tumor, high mortality
References


Acknowledgments

- Dr. Don Chase
- Dr. Craig Zuppan
- Dr. Jun Wang
- Dr. Katie Wilkinson
- Pedro Chase

Accepted for publication by the California Tumor Tissue Registry online journal, July, 2010.
http://www.cttr.org