46th ANNUAL DIAGNOSTIC SLIDE SESSION, 2005
REFERENCES AND DIAGNOSES

MODERATOR: E. Tessa Hedley-Whyte, M.D.

EDITOR: Leroy R. Sharer, M.D.

Case 2005-4

Submitted by: Angelica Oviedo, M.D., Kaiser Hospital, Oakland, CA 94611

Diagnosis: Atypical teratoid rhabdoid tumor (ATRT)

Comment: Tumor cells were positive on ICC for epithelial membrane antigen (EMA) and negative for smooth muscle antigen (SMA). The Ki-67 labeling index was high. Fluorescence in situ hybridization (FISH) revealed monosomy of chromosome 22 in 73/100 cells, and the tumor cells were negative for INI1 by ICC. Members of the audience recognized the contributions of Dr. Lucy Rorke to the definition of this entity.

From the presenter: ATRT has a much worse prognosis than medulloblastoma. Immunohistochemistry for EMA is frequently positive in ATRT. Loss of all or part of chromosome 22 is seen in most ATRT's. Nuclear staining for INI1 (which corresponds with the chromosome 22 loss) is negative in ATRT and positive in medulloblastoma, by immunohistochemistry.

References:

Burger P, Yu I, Tihan T, Friedman H, et al: Atypical teratoid/rhabdoid tumor of the central nervous system: a highly malignant tumor of infancy and childhood frequently mistaken for medulloblastoma. Am J Surg Pathol 1998; 22:1083-1092.

Judkins A, Mauger J, Rorke L, Biegel J: Immunohistochemical analysis of hSNF5/INI1 in pediatric CNS neoplasms. Am J Surg Pathol 2004; 28:644-650.