# CYTOGENETIC AND MOLECULAR CYTOGENETIC ANALYSES OF UNTREATED TESTICULAR GERM CELL TUMORS

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Abstract

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OF UNTREATED TESTICULAR GERM CELL TUMORS

An isochromosome of the short arm of chromosome 12 or i(12p) has been reported in 80-90% of testicular germ cell tumors (TGCTs). In the current study of 46 primary tumors or lymph node metastases, acquired chromosomal abnormalities were identified in 29 specimens, of which 20 were i(12p)-positive, and the remaining 9 were i(12p)-negative.

Chromosomal microdissection and sequence-independent polymerase chain reaction amplification were used to develop a 12p-specific paint probe, which was used with a whole chromosome 12 paint probe, for two-color fluorescence in situ hybridization analysis of the abnormalities of chromosome 12 in 27 of the tumors. An i(12p) was confirmed in 18 tumors, 8 of which had additional 12p abnormalities. The remaining 9 tumors had other structural rearrangements of chromosome 12. A breakpoint in chromosome band 12p13 was identified in all but one of the i(12p)-negative tumors. Overrepresentation of sequences from the short arm of chromosome 12 was present in all 27 tumors.

In addition to abnormalities of chromosome 12, other numerical and structural abnormalities were analyzed. Numerical abnormalities included overrepresentation of apparently normal copies of chromosomes X, 7, 8, 21, 12, 3, and 17 compared to chromosomes 4, 10, 11, 13, and 18. In nonseminomas chromosomes 7, 8, X, 12, and 17 were overrepresented relative to chromosomes 18 and 13, while in

seminomas, chromosomes 21 and 14 were overrepresented with respect to chromosomes 11, 18, and 10. Chromosomal breakpoints in the centromeric and heterochromatic regions of chromosomes 1 and 9, the short arm of chromosome 9, and the centromeric and stalk regions of chromosomes 13, 14, and 15 occurred in both primary tumors and lymph node metastases.

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