EXTRASKELETAL CHONDROSARCOMA WITH A ALLELE LOSS IN CHROMOSOME-22 AND CHROMOSOME-10

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ANGIOSARCOMA OF THE SCALP WITH A COMPLEX HYPO-TETRAPLOID KARYOTYPE. A Molina, CD Bangs and T Donlon. Stanford University Medical Center, Departments of Medicine (Oncology) and Pathology (Clinical Cytogenetics), Stanford, Ca 94305

Angiosarcoma is a rare (less than 2% of all soft tissue sarcomas), aggressive malignant neoplasm of vascular endothelial cell origin which can arise in any tissue of the body. Cutaneous angiosarcoma almost exclusively affects the elderly and primarily involves the head and neck region in the area of the scalp. We performed a cytogenetic study of a resected cutaneous angiosarcoma in a 72 year old man. The patient underwent wide surgical excision of several erythematous ulcerated scalp nodules present in the dermis and subcutis. Histological examination revealed diffuse infiltrative cells with pleomorphic hyperchromatic nuclei and vascular neoplasms. Numerous mitotic figures were seen. Tumor tissue was disaggregated with collagenase and cultured. Chromosomes were analyzed by the G-band method. Twenty cells were examined, revealing a hypo-tetraploid cell line with a chromosome number ranging from 85-91 and a modal number of 87. A representative karyotype was identified: 86, XY, +X, +Y, +1, +2, +2q?, +2q?, -3, -3, +13(p), +13(p), +del(5)(p21.3), +del(5)(p21.3), +6, +7, +7, +8, +del(8)(11q33)11.2, +9, +9, +9, +10, +10, +11, +12, +13, +13p, +14, +14, +15, +16, +16, +17, +18, +19, +19, +20, +20, +21, +22, +22, +22, +5mar.

Consistent structural abnormalities were two chromosome #2% with abnormal long arms, two short arm deleted copies of #3, an abnormal #6 derived from a translocation with #1, a #13 with extra material on the short arm and multiple marker chromosomes. Numeric variation between cells was seen. To our knowledge this is the first cytogenetic study on angiosarcoma of the scalp. Despite surgical resection and adjuvant radiation therapy, the patient developed widespread metastatic disease. Cytogenetic analyses performed at an earlier stage in the evolution of this malignancy might offer insights into the mechanisms underlying tumor progression and the generation of aneuploidy.


Extraskeletal Myxoid Chondrosarcoma is a rare tumor of the adulthood. Non-random reciprocal translocation between chromosome 9 and 22 has been reported associated with this tumor. We have performed cytogenetic and molecular genetic investigation of a myxoid chondrosarcoma from the right frontal-temporal region in a 27 year old female. A portion of this tumor was snap-frozen for DNA analysis and the other portion was mechanically dissociated and put in the tissue culture medium. Twenty-five analyzable metaphases from a 10 day short term culture of the tumor revealed 46 XX karyotype. When the tumor DNA was compared with lymphocyte DNA from the same person with the probe D22S9 (mapped to chromosome 22 q11 region), control DNA was homozygous for this probe (5.8Kb), tumor DNA has extra lower band at 5.2Kb in addition to the 5.8Kb band. This finding is suggestive of tumor DNA had possible small internal deletion of one allele in the region of 22q11. Analysis of MBC DNA revealed homozygosity for the D22S1 probe which is mapped to chromosome 22q11 - q13. Markedly decreased hybridization signal was noted in the tumor DNA indicating loss of chromosome 22 sequence in the tumor cells. The tumor DNA and control DNA were also compared with 9q RFLP probes including ASSP3, Aldo B, no difference were observed. Hox I, NFE-B and DIOS1 loci were not informative. Allele loss was noted in the tumor DNA with DIOS4 probe. This study reveals a defect of extraskeletal chondrosarcoma may reside in the region of 22q11 region which is consistent with the previous cytogenetic observation. In our case, no translocation was observed, but submicroscopic deletion may be present. Allele loss in chromosome 10 warrants further study.