PEComas (tumors with perivascular epithelioid cell differentiation) are a family of related benign and malignant mesenchymal neoplasms which include angiomyolipoma (AML), clear cell “sugar” tumor of the lung (CCST), lymphangioleiomyomatosis (LAM), and a group of rare histopathologically similar lesions arising in other anatomic sites. The presence of a distinctive cell type, the perivascular epithelioid cell, is characteristic of this group of tumors. In this study, cytogenetic analysis of a malignant PEComa arising in the retroperitoneum of a 54 year old female revealed the following clonal karyotypic abnormalities: 31,X,+X,der(5)(5;5)(q13;p15.1),+der(5)(5;5)(p15.1;q13),+9,+12,+16,+19,+20[9]/62,XXX,-X,-1,-2,+3,-4,der(5)(5;5)(q13;p15.1)x2,der(5)(5;5)(p15.1;q13),+der(5)(5;5)(p15.1;q13),-7,-8,+9,-10,-11,+12,-13,-14,-15,+16,-17,-18,+19,+20,-21,-22[7]. Cytogenetic studies of PEComas are few. Most are of AML and have revealed trisomy 7, a finding not substantiated by subsequent FISH studies. To the best of our knowledge, this is the first case of a clonally aberrant malignant PEComa.

Introduction

PEComa is a family of related tumors with a marked female predominance characterized by nests and sheets of epithelioid to spindled cells with ample clear to eosinophilic cytoplasm and focal association with blood vessel walls.

PEComas with malignant potential are further characterized by infiltrative growth, marked hypercellularity, nuclear enlargement and pleomorphism, high mitotic activity, atypical mitotic figures and coagulative necrosis.

Immunohistochemical studies show coexpression of melanocytic and muscle markers.

The following genetic abnormalities have been observed in related members of the PEComa family:

- Trisomy 7 has been reported in angiomyolipoma, however, the significance of these findings are unclear as trisomy 7 has also been observed in normal tissue. (Cancer Genet Cytogenet 99:42-45, 1997. Cancer Genet Cytogenet 29:123-134, 1997. Cancer Genet Cytogenet 106:182, 1998).

Case Summary

A 54 year old female presented with abdominal fullness. Radiographically, a 12 x 12 cm retroperitoneal mass was identified.

Histopathologically, the tumor was composed of nests of epithelioid cells arranged around thin-walled blood vessels. Individual cells exhibited abundant clear to granular, eosinophilic cytoplasm.

The neoplastic cells were immunoreactive for melanocyte marker melan-A.

Features supporting the designation of malignant PEComa in this case included areas of poorly differentiated spindle-shaped cells, focal necrosis and increased mitotic activity (6-14 mitotic figures per 10 high power fields) where atypical mitotic figures could be appreciated.

A representative portion of the retroperitoneal mass was cultured in-situ in MF media for 6-7 days.

Cytogenetic analysis revealed the following abnormal karyotype:

- 31,X,+X,der(5)(5;5)(q13;p15.1),+der(5)(5;5)(p15.1;q13),+9,+12,+16,+19,+20.

Conclusions

- Near-haploidy is rare in human malignancy but has been seen in a subset of childhood acute lymphocytic leukemia with poor prognosis, however, the mechanism, role, and significance of near-haploidy is unknown.
- Structural rearrangements in conjunction with near-haploidy is more often seen in solid tumors than in leukemias.
- Abnormal cytogenetic studies and the near-haploidy modal count is supportive of the pathologic diagnosis of a malignant neoplasm.
- This case shows the importance of adjunct cytogenetic studies in the diagnosis of solid tumors.
- Further studies are necessary to ascertain the significance of these findings.