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Cytogenetic Analysis of a Rare Primary Undifferentiated Testicular Sarcoma

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1. Introduction

Testicular tumors are the most common solid malignancies in young adult males. The vast majority (>95%) arise from germ cells, with seminoma being the most frequent subtype, followed by non-seminomatous tumors such as embryonal carcinoma, yolk sac tumor, teratoma, and choriocarcinoma.

Primary testicular sarcomas, however, are extremely uncommon. Only a small number of cases have been reported in medical literature. A diagnostic challenge arises because germ cell tumors may sometimes contain sarcomatous components, making it difficult to distinguish between a true primary sarcoma and a germ cell tumor that has undergone sarcomatous transformation.

A key cytogenetic marker in germ cell tumors is isochromosome 12p (i(12p)) or other forms of 12p amplification, which are present in most adult testicular germ cell tumors regardless of histologic subtype. Therefore, detecting or excluding 12p abnormalities using fluorescence in situ hybridization (FISH) is valuable in determining tumor origin. This report describes an exceptionally rare case of primary undifferentiated testicular sarcoma with cytogenetic evaluation for 12p amplification.

2. Case Presentation

A 36-year-old man presented with a one-year history of painless enlargement of the right testis. He had no urinary symptoms, systemic complaints, or significant past medical history. Physical examination did not reveal lymph node enlargement or other abnormalities.

Scrotal ultrasonography and computed tomography identified a large multiloculated mass in the right testis measuring approximately 9.4 cm in diameter. No metastatic lesions were detected at that time. Serum tumor markers, including alpha-fetoprotein (AFP) and beta-human chorionic gonadotropin (β-hCG), were within normal limits.

The patient was lost to follow-up but returned two years later with marked disease progression. Imaging showed significant tumor enlargement to approximately 21 cm, along with widespread metastases involving the lungs and multiple bones, including the skull base, spine, pelvis, and femur.

A radical orchiectomy was subsequently performed. Chemotherapy was initiated immediately without waiting for final pathology, using a standard germ cell tumor regimen consisting of etoposide and cisplatin for four cycles. In addition, palliative radiotherapy was administered to bone metastatic sites.

The patient showed a partial response to treatment, with complete regression of pulmonary metastases and partial improvement in bone lesions. Despite aggressive management, his condition deteriorated, and he died 12 months after surgery.

3. Pathological and Cytogenetic Findings

Microscopic examination of the orchiectomy specimen revealed a highly undifferentiated malignant tumor consistent with sarcoma. No components suggestive of germ cell tumor differentiation were identified.

mmunohistochemical studies were performed using a broad panel of markers, including cytokeratins, desmin, vimentin, S-100, CD30, AFP, β-hCG, placental alkaline phosphatase (PLAP), and other lineage-specific markers. The tumor did not show consistent expression of epithelial, muscular, neural, or germ cell markers, supporting an undifferentiated sarcomatous phenotype.

To further clarify the tumor's origin, fluorescence in situ hybridization (FISH) analysis was performed on formalin-fixed, paraffin-embedded tissue samples to assess for amplification of chromosome arm 12p. This genetic abnormality is strongly associated with germ cell tumors.

The FISH study demonstrated no evidence of 12p amplification or isochromosome 12p formation, suggesting that the tumor was not derived from a germ cell lineage. This finding supported the diagnosis of a primary undifferentiated testicular sarcoma rather than a germ cell tumor with sarcomatous transformation.

4. Discussion

Primary testicular sarcomas are extremely rare, and their diagnosis is challenging due to their overlapping features with germ cell tumors that contain sarcomatous components. Correct classification is important because treatment strategies and prognosis differ significantly between these entities.

Isochromosome 12p is a well-established cytogenetic hallmark of germ cell tumors and is present in most adult cases across all histological subtypes. As a result, detection of 12p amplification

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by fish is a useful tool for confirming germ cell origin in poorly differentiated tumors.

In the present case, the absence of 12p amplification strongly indicated that the tumor was not derived from a germ cell tumor, supporting a diagnosis of primary testicular sarcoma. This makes the case particularly valuable, as it provides molecular evidence distinguishing primary sarcoma from germ cell tumor–associated sarcomatous transformation.

Clinically, the tumor demonstrated aggressive behavior with rapid growth and widespread metastasis. Despite multimodal treatment including surgery, chemotherapy, and radiotherapy, the patient had only a temporary partial response and ultimately succumbed to the disease within one year.

This case emphasizes the importance of early diagnosis and follow-up in testicular masses, as delayed treatment may allow progression to advanced metastatic disease. It also highlights the importance of integrating histopathology, immunohistochemistry, and cytogenetic testing in evaluating rare and poorly differentiated testicular tumors.

6. Conclusion

Primary undifferentiated testicular sarcoma is a rare and highly aggressive tumor that can mimic germ cell tumors clinically and histologically. Cytogenetic evaluation, particularly analysis of chromosome 12p, is essential for accurate diagnosis.

In this case, the absence of 12p amplification confirmed that the tumor was not germ cell–derived, supporting a diagnosis of primary testicular sarcoma. This report underscores the importance of combining morphological, immunohistochemical, and molecular techniques to ensure accurate classification of rare testicular neoplasms.

References

1. Khan O, Protheroe A. Testis cancer. Postgrad Med J. 2007;83(984):624-632.
2. Guo CC, Punar M, Contreras AL, Tu SM, Pisters L, Tamboli P, Czerniak B. Testicular germ cell tumors with sarcomatous components: an analysis of 33 cases. Am J Surg Pathol. 2009;33(8):1173-1178.
3. Fuzesi L, Rixen H, Kirschner-Hermanns R. Cytogenetic findings in a metastasizing primary testicular chondrosarcoma. Am J Surg Pathol. 1993;17(7):738-742.
4. Rodriguez E, Mathew S, Reuter V, Ilson DH, Bosl GJ, Chaganti RS. Cytogenetic analysis of 124 prospectively ascertained male germ cell tumors. Cancer Res. 1992;52(8):2285-2291.
5. Bosl GJ, Ilson DH, Rodriguez E, Motzer RJ, Reuter VE, Chaganti RS. Clinical relevance of the i(12p) marker chromosome in germ cell tumors. J Natl Cancer Inst. 1994;86(5):349-355