

CASE REPORT

Silent Retroperitoneal Seminoma Arising from an Occult Undescended Testis: Diagnostic Challenges and the Role of Ultrasound-Guided FNAC

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ABSTRACT

Retroperitoneal masses in adult males present a significant diagnostic dilemma due to their deep location, varied etiologies, and nonspecific clinical manifestations.¹ Germ cell tumors, particularly seminoma, may rarely present primarily as retroperitoneal disease, especially in association with cryptorchidism, leading to delayed or erroneous diagnosis.^{2,3} We report the case of a 42-year-old male who presented with prolonged constitutional symptoms including back pain, abdominal distension, anorexia, intermittent fever, and generalized weakness for over one year. Radiological evaluation revealed a large retroperitoneal pre- and paravertebral mass suggestive of lymphadenopathy.² Initial ultrasound-guided fine needle aspiration cytology (USG-FNAC) was interpreted as metastatic adenocarcinoma. Repeat FNAC demonstrated dispersed medium-to-large atypical cells with vesicular nuclei, prominent nucleoli, pale cytoplasm, and a characteristic tigroid background. Immunocytochemistry showed strong positivity for placental alkaline phosphatase (PLAP) and negativity for AFP, cytokeratin, leukocyte common antigen, and carcinoembryonic antigen, confirming the diagnosis of seminoma.⁵ Subsequent clinical examination revealed absence of the left testis in the scrotum, consistent with an undescended testis, explaining the unusual presentation.³ The patient showed marked response to radiotherapy. This case highlights the critical role of USG-FNAC combined with immunocytochemistry in diagnosing seminoma at an unusual site, emphasizes the importance of careful clinical correlation, and underscores the need to consider germ cell tumors in the differential diagnosis of retroperitoneal masses in adult males.^{1,6}

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KEYWORDS

• Seminoma • Retroperitoneal mass • Cryptorchidism • Immunocytochemistry • FNAC

INTRODUCTION

Retroperitoneal masses encompass a wide spectrum of benign and malignant lesions, including lymphomas, sarcomas, metastatic carcinomas, and germ cell tumors.¹ Accurate diagnosis is often challenging due to the deep anatomical location and nonspecific clinical presentation. Ultrasound-guided fine needle aspiration cytology (USG-FNAC) has become a valuable diagnostic modality for evaluating such lesions, offering a minimally invasive, rapid, and cost-effective method for tissue diagnosis.^{6,7}

Testicular germ cell tumors (TGCTs) are the most common malignancies in young and middle-aged men, with seminoma representing a major histological subtype.² Cryptorchidism is the strongest known risk factor for TGCTs, increasing the risk by several folds, particularly when the testis is intra-abdominal.³ Seminoma arising in an undescended testis may present with retroperitoneal metastasis without obvious scrotal findings, leading to diagnostic confusion.^{4,5}

We report a case of retroperitoneal seminoma diagnosed on FNAC, initially misinterpreted as metastatic adenocarcinoma, emphasizing the diagnostic pitfalls and the pivotal role of cytology and immunocytochemistry.

Case Report

A 42-year-old male presented to the surgical unit with complaints of generalized weakness, back pain, abdominal bloating with postprandial distension, anorexia, neck pain, and occasional evening rise of temperature for 17 months. There was no history of urinary symptoms or scrotal swelling. Past history was significant for malaria and jaundice 6–7 years earlier, and he had completed a 6-month course of antitubercular therapy. There was no significant family history. The patient had stopped smoking and alcohol consumption several years earlier.

On examination, the patient appeared pale and debilitated. Bilateral paravertebral swellings were noted at the T9 level. Abdominal examination revealed fullness without organomegaly. Testicular examination

was initially not emphasized; however, on re-evaluation, the left testis was found to be absent from the scrotum.

Investigations

Chest radiograph showed non-homogeneous opacities in bilateral upper lung zones with fibrotic bands in the left upper zone. Ultrasonography revealed a retroperitoneal mass, following which USG-guided FNAC was performed.

Computed tomography of the thoracolumbar region demonstrated a large lobulated pre and paravertebral soft tissue density lesion suggestive of retroperitoneal lymphadenopathy.²

Laboratory investigations revealed hemoglobin of 7.2 g/dL, elevated serum alkaline phosphatase (380 U/L), and serum creatinine of 2.2 mg/dL. Liver function tests and serum bilirubin were within normal limits.

Cytological Findings

The initial FNAC showed clumps of malignant epithelial-appearing cells with moderate nuclear pleomorphism and occasional cytoplasmic vacuolation, leading to a provisional diagnosis of metastatic adenocarcinoma.

Repeat FNAC smears were moderately cellular and showed predominantly dispersed medium-to-large round cells with vesicular nuclei, prominent nucleoli, and pale finely granular cytoplasm. The background showed a characteristic tigroid appearance with scattered lymphocytes. No gland formation, mucin, or necrosis was identified. These features were suggestive of a germ cell tumor, particularly seminoma.⁵

Immunocytochemistry

Immunocytochemical studies on cell block material showed:

- **PLAP:** Strongly positive
- **AFP:** Negative
- **Cytokeratin:** Negative
- **LCA:** Negative

- **CEA: Negative**

The immunoprofile confirmed the diagnosis of seminoma.⁵

Treatment and Outcome

The patient was treated with cobalt tele-radiotherapy. Following completion of radiotherapy, the retroperitoneal mass showed marked regression, consistent with the known radiosensitivity of seminoma.^{2,3}

DISCUSSION

Diagnostic Challenges of Retroperitoneal Seminoma

Primary or secondary retroperitoneal seminoma represents a diagnostic challenge due to its deep location, absence of scrotal symptoms, and frequent radiologic resemblance to lymphadenopathy, lymphoma, or metastatic carcinoma.^{1,2} In patients with occult cryptorchidism, the diagnosis is often delayed because testicular examination may be overlooked, and constitutional symptoms predominate over localizing signs.³ As demonstrated in the present case, prolonged nonspecific symptoms led initially to misdiagnosis and inappropriate differential considerations such as tuberculosis and metastatic adenocarcinoma.

Cytomorphological Pitfalls and Role of Repeat FNAC

Seminoma may show variable cytologic patterns depending on sampling, tumor degeneration, or crush artifact, leading

to interpretive errors on FNAC.^{5,6} Initial aspirates may appear cohesive with epithelial-like clusters, mimicking adenocarcinoma, particularly in paucicellular or hemorrhagic smears. Repeat FNAC with better cellular yield often reveals classical features such as dispersed large cells with vesicular nuclei, prominent nucleoli, pale cytoplasm, and a tigroid background, as seen in the present case.⁵ This highlights the importance of repeat sampling when cytology and clinical findings are discordant.

Immunocytochemistry as a Decisive Tool

Immunocytochemistry (ICC) significantly enhances diagnostic accuracy in FNAC samples from retroperitoneal masses. Seminoma shows characteristic positivity for PLAP and negativity for epithelial, lymphoid, and yolk sac tumor markers.⁵ In the current case, the ICC profile conclusively excluded adenocarcinoma and lymphoma, underscoring the indispensable role of ancillary techniques in cytology-based diagnosis.^{6,7}

Comparative Analysis of Reported Cases

A review of published cases demonstrates that USG-guided FNAC, when combined with ICC, has consistently enabled early and accurate diagnosis of retroperitoneal seminoma, even in the absence of a detectable scrotal primary.^{1,5-7} Most cases share common features: adult male patients, large retroperitoneal masses, initial diagnostic confusion, and excellent therapeutic response following correct diagnosis.

Table 1: Role of USG-Guided FNAC in Retroperitoneal Seminoma

Author & Year	Age (Years)	Clinical Presentation	FNAC Interpretation	ICC Markers	Final Diagnosis	Outcome	Reference
Paudel <i>et al.</i> , 2022	38	Abdominal pain, weight loss	Suggestive of GCT	PLAP +	Seminoma from undescended testis	Good response to therapy	[1]
Bedi <i>et al.</i> , 2015	45	Back pain, retroperitoneal mass	Seminoma on FNAC	PLAP +, AFP –	Primary retroperitoneal seminoma	Complete remission	[5]
Chakrabarti <i>et al.</i> , 2014	40	Abdominal mass	Malignant round cell tumor	ICC diagnostic	Seminoma	Favorable	[6]
Sain <i>et al.</i> , 2024	40	large retroperitoneal mass without scrotal symptoms	seminomatous germ cell tumor from retroperitoneal mass	PLAP aided	Primary retroperitoneal seminoma	Good outcome	[7]
Present case	42	Constitutional symptoms, back pain	Initially adenocarcinoma; repeat FNAC diagnostic	PLAP +, AFP –, CK –	Seminoma from occult cryptorchid testis	Marked regression	–

Clinical Significance

This comparative analysis reinforces that **USG-guided FNAC is a reliable, minimally invasive, and cost-effective first-line diagnostic modality** for retroperitoneal masses, particularly in resource-limited settings.^{6,7} When supplemented with ICC and careful clinical examination—including assessment for cryptorchidism—it can prevent misdiagnosis, unnecessary surgical procedures, and treatment delays.^{3,5}

Implications for Practice

All adult males presenting with retroperitoneal masses should undergo mandatory testicular examination and consideration of germ cell tumors, irrespective of age or absence of scrotal complaints.²⁻⁴ FNAC with ICC should be strongly advocated as part of the diagnostic algorithm for deep-seated retroperitoneal lesions.

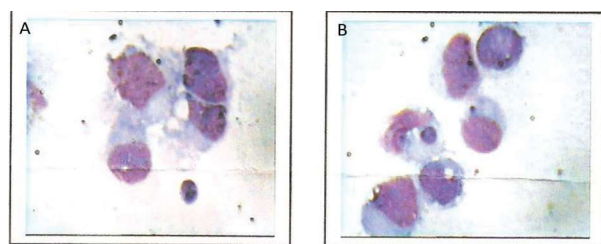


Figure 1: Ultrasound-guided fine needle aspiration cytology (USG-FNAC) smears from the retroperitoneal mass. A,B: Smears show predominantly dispersed medium-to-large pleomorphic malignant cells with vesicular nuclei, and pale finely granular cytoplasm with few of the cells showing cytoplasmic vacuolation. (MGG, X40)

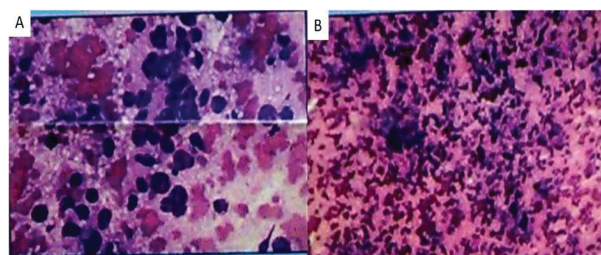


Figure 1A: High-power view showing large round tumor cells with vesicular nuclei, prominent nucleoli, and pale cytoplasm consistent with seminoma (MGG stain, X20). **B:** Low-power view showing moderately cellular FNAC smear with dispersed tumor cells in a tigroid background (MGG stain, X10).

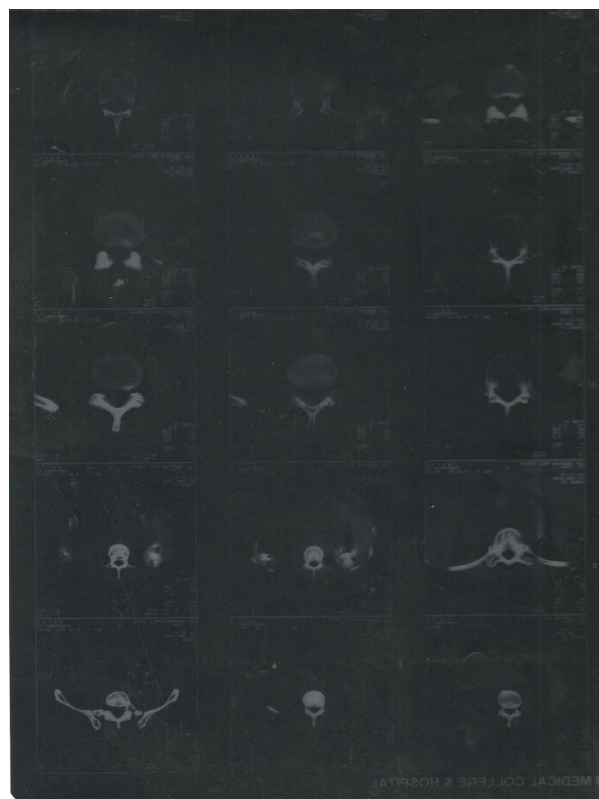


Figure 3: CT scan showing lobulated retroperitoneal pre and paravertebral soft tissue mass suggestive of lymphadenopathy.

CONCLUSION

Seminoma should be considered in adult males presenting with retroperitoneal masses, particularly in the presence of cryptorchidism. Ultrasound-guided FNAC with immunocytochemistry plays a decisive role in achieving an accurate diagnosis and ensuring timely, effective treatment.

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