Case Report

Extragonadal dysgerminoma presenting as neck metastasis and masquerading as a thyroid swelling

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ABSTRACT

Germ cell tumors (GCTs) are common tumors of gonads. Their occurrence at extragonadal sites, either as primary tumors or as metastatic foci, is rare. Extragonadal GCTs have a prevalence of 1–2.5% of all GCTs with mediastinum being the most common site of involvement, followed by retroperitoneum, pineal gland, and sacrococcygeal region. Involvement of other extragonadal sites, including lymph nodes, is usually associated with metastatic disease. We report a case of a young female with an atypical presentation of a swelling in the left neck region creating a clinical and pathological dilemma.

Key words: Dilemma, extragonadal dysgerminoma, metastasis neck, thyroid

INTRODUCTION

Extragonadal germ cell tumors (EGGCTs) are defined as germ cell neoplasms displaying one of the histologies associated with gonadal origin but located outside the gonads.[1] They typically arise in the midline locations, and specific sites vary with age.[2] Histological, serological, and cytogenetic characteristics of EGGCTs are similar to those of gonadal GCT. However, major differences in clinical behavior suggest that gonadal and extragonadal tumors are biologically different. These EGGCTs are thought to arise from aberrant midline migration of primordial germ cells that tends to lodge at a site other than the gonads leading to aberrant locations.[3] We present this case not only because of its rarity but also because of its unusual location which created a diagnostic conundrum.

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CASE REPORT

A 20-year-old female came to the outpatient department with a left sided neck swelling since 2–3 months. The swelling was 5 cm × 3 cm in size, and it moved on deglutition [Figure 1a]. On palpation, the swelling was nontender and soft to firm in consistency. It was mobile, and the overlying skin was unremarkable. She also gave a history of loss of appetite and weight. Fine needle aspiration cytology (FNAC) smears of the swelling were highly cellular and showed large cells with rounded vesicular nuclei and irregular chromatin. The cytoplasm was eosinophilic to light blue, abundant, and fragile giving a tigroid background [Figure 1b]. Few lymphocytes and plasma cells were also seen. No lymphoid cells, lymphoglandular bodies or thyroid follicular cells could be identified. Based on these features, a diagnosis of a GCT possibly metastatic or a malignant epithelial tumor was entertained. On immunocytochemistry, these cells were positive for c-kit (CD-117), placental alkaline phosphatase (PLAP), and negative for cytokeratin (CK),

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leukocyte common antigen (LCA), thyroid transcription factor (TTF-1), CD-30, HMB-45, and S-100. Following this, a detail clinical workup was advised to find the primary. Her abdominal examination revealed an ill-defined mass in the left lumbar region. It was difficult to pinpoint its exact location. Thus, an ultrasonography (USG)/computed tomography (CT) abdomen was advised. USG showed a 10 cm × 5 cm mixed echogenic mass in the left lumbar region extending to the pelvis. CT abdomen showed a 10 cm × 8 cm sized mass in the retroperitoneal region compressing the surrounding structures [Figure 1c]. Ovaries were unremarkable.

Her routine hematological and biochemical parameters were within normal limits. Serum tumor markers showed high lactate dehydrogenase (4000 IU/L), CA-125 (100.6 μg/dL), carcinoembryonic antigen (4.2), β human chorionic gonadotropin (300 mIU/ml), and α-feto protein (2 μg/dL). USG-guided FNAC of the retroperitoneal mass exhibited features similar to the FNAC smears of the neck swelling. A diagnosis of retroperitoneal tumor metastasizing to the left side of the neck was made, and biopsy was advised. Excisional biopsy of the neck swelling and a USG-guided trucut biopsy of the retroperitoneal mass showed a similar histopathological picture. There were large round tumor cells with well-defined cell borders, large vesicular central nuclei, and watery clear cytoplasm. Few of the cells showed prominent nucleoli, the cells were arranged in nests separated by fibrous stroma infiltrated by lymphocytes. No lymphoid tissue could be identified. These cells on immunohistochemistry were positive for CD-117, PLAP and negative for CK, LCA, TTF-1, S-100, HMB-45, and CD-30 [Figure 2]. Based on the morphology, CD-117 and PLAP positivity, a final diagnosis of retroperitoneal dysgerminoma metastatic to the left side neck region was made. The patient underwent chemotherapy of three cycles of cisplatin, etoposide, bleomycin (PEB) followed by secondary surgery for removal of the residual retroperitoneal mass. Response to chemotherapy was extremely good, and follow-up is uneventful.

DISCUSSION

EGGCTs are either seminomatous or nonseminomatous in males and dysgerminomas or nondysgerminomas in females.[4] Extragonadal dysgerminomas are uncommon tumors. Their incidence varies from 1% to 5% of all GCTs.[3] They are most commonly located in the mediastinum where their incidence varies from 50% to 70% of all EGGCTs followed by retroperitoneum (30–40%). They can also arise in the pineal region, sacrococcygeal region, vagina, orbit, liver, and gastrointestinal tract.[2] An extensive search on PubMed and MEDLINE failed to reveal any information regarding such an atypical presentation. In fact, the existing information regarding EGGCTs is more than 6 years old and desperately needs an update. Hence, the current case becomes a relevant value addition to the world literature. Klinefelter syndrome is the only known risk factor for EGGCTs and usually causes mediastinal nonseminomatous GCTs.[5] Our patient was phenotypically and genotypically normal female with normal ovaries.

The tumor was located in the retroperitoneal region of the posterior abdominal wall. The retroperitoneal EGGCTs primarily manifest as abdominal pain (29%) and back pain (14%), followed by weight loss (9%), Fever (9%), vena caval or other thrombosis (8%), palpable abdominal abdominal mass (6%), cervical nodes (4%), and dysphagia (3%). However, EGGCTs presenting as metastasis in the soft tissue is a rare phenomenon and has never been reported in the literature. Our patient presented as the left sided neck swelling which moved on swallowing. Such an

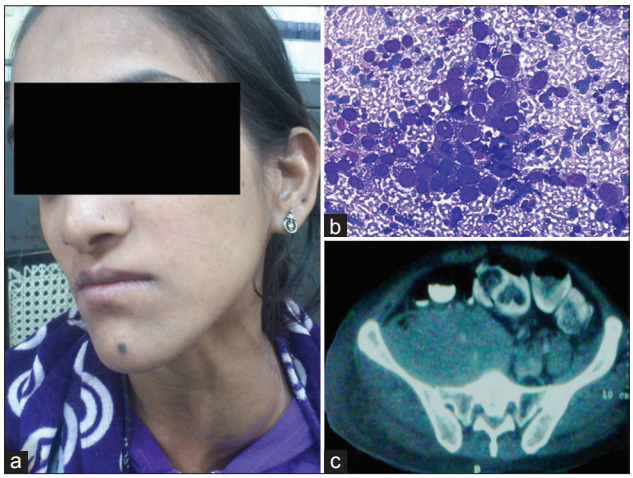

Figure 1: (a) Left side neck swelling which moved with deglutition (b) Fine needle aspiration cytology smears showing tumor cells against a tigroid background (Giemsa, ×40) (c) Retroperitoneal mass in the posterior abdominal wall seen on computed tomography abdomen

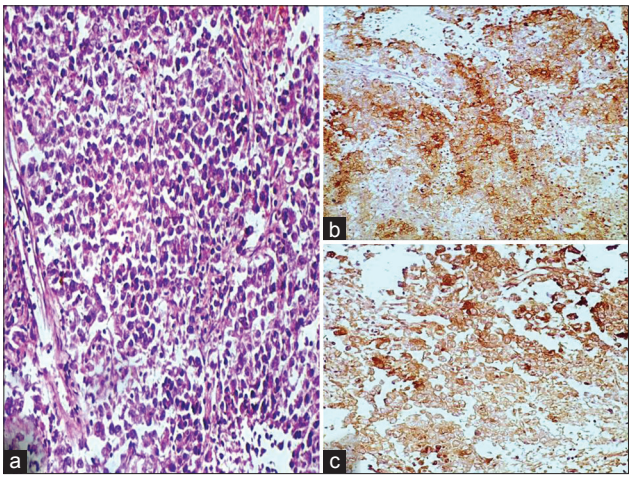

Figure 2: (a) Microphotograph showing tumor cells arranged in nests and separated by fibrous septa which is infiltrated by lymphocytes (H and E, ×40) (b) Tumor cells exhibiting PLAP positivity (immunohistochemistry, ×20) (c) CD-117 positivity of tumor cells (immunohistochemistry, ×20)

atypical presentation always causes erroneous diagnosis. It also possesses a great challenge to a pathologist. Moreover, since the approach to the diagnosis is through FNAC, it becomes much more difficult as the site is an important parameter for cytological diagnosis.

The question may also arise as to how the tumor metastasized to such an aberrant nonlymphoidal location. A possible hypothesis is that the tumor might have spread through hematogenous route. Though, the more common sites for its hematogenous spread are lung, liver, and bone.[6]

Tumor markers, though nonspecific, are helpful in the diagnosis and follow-up of EGGCTs. Serum lactic dehydrogenase is frequently elevated, but increased levels of serum alpha fetoprotein or human chorionic gonadotropin are generally not seen. If increased, they suggest that other germ cell elements are also present in the tumor (mixed GCT).[7] The panel of markers recommended are LCA, PLAP, CD-117, CD-30, CK, AFP, HMB-45, and EMA, which help to differentiate dysgerminomas from other tumors that mimic it, such as malignant mixed GCT, embryonal carcinoma, yolk sac tumor, clear cell carcinoma, lymphoma, and melanoma. Dysgerminomas show CD-117 and PLAP positivity.[8]

Radiology has its own limitations in EGGCTs especially for retroperitoneal tumors, as there are no definite radiological features distinguishing them from lymphoma, retroperitoneal metastasis, and retroperitoneal soft-tissue sarcoma.[3]

Regional lymph node metastasis is common to this tumor. However, it is the distant metastasis that results in poor prognosis. Therefore, an early diagnosis and treatment are essential to improve the prognosis. Standard cisplatin based chemotherapy plus additional secondary surgery is the treatment of choice. Though, the tumors are highly radiosensitive but it is usually used as a salvage treatment in cases of otherwise untreatable disease.[6]

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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