

Scrotal Involvement in Gist: A Case Report of Testicular Metastasis and Literature Review

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ABSTRACT

Gastrointestinal stromal tumors (GISTs) are rare mesenchymal neoplasms, commonly metastasizing to the liver and peritoneum. Scrotal involvement is extremely rare. We report a 63-year-old male with ileal GIST and peritoneal carcinomatosis, initially treated with surgery and Imatinib. After disease progression, he presented with abdominal and scrotal swelling. Imaging revealed extensive metastases, and orchiectomy confirmed metastatic GIST without direct testicular invasion. Despite systemic therapy escalation, liver metastases progressed. Scrotal metastases of GIST are uncommon, possibly spreading via a persistent peritoneo-vaginalis duct. Primary extra-gastrointestinal stromal tumors (E-GISTs) of the scrotum must also be considered. This case underscores the importance of recognizing atypical metastatic sites. Diagnosis relies on immunohistochemistry (CD117, CD34), while treatment remains challenging due to tyrosine kinase inhibitor resistance.

Keywords: Gastrointestinal Stromal Tumor, Scrotal Metastasis, Peritoneo-Vaginalis Duct, Extra-Gastrointestinal Stromal Tumor

Introduction

Gastrointestinal stromal tumors (GISTs) are mesenchymal neoplasms originating from the interstitial cells of Cajal in the digestive tract. Mutations in the KIT, PDGFRA, and occasionally BRAF genes are the primary drivers of oncogenesis [1]. The stomach is the most common site of origin, followed by the small intestine. Metastatic GISTs predominantly affect the liver and peritoneum. Scrotal involvement, however, remains exceedingly rare.

Case Presentation

A 63-year-old male was diagnosed in 2012 with ileal GIST and peritoneal carcinomatosis. He underwent ileal resection and received Imatinib (400 mg daily) until 2018. Upon disease progression in 2020, Imatinib was reintroduced at the same dose, resulting in a complete response. However, the patient was lost to follow-up until 2022, when he presented with abdominal and scrotal swelling.

Scrotal ultrasound showing a large 25 cm mass occupying the right hemi-scrotum, with non-visualization of the right testis [Figure 1A] and a left testis displaced superiorly and laterally, measuring 35 × 15 × 33 mm [Figure 1B]. A CT scan showed extensive intraperitoneal masses and multiple liver nodules [Figure 2]. A right orchiectomy was performed. Histological examination revealed a malignant spindle cell mesenchymal tumor with sarcomatous involvement of the testicular parenchyma without direct invasion [Fig 3A, 3B]. Postoperatively, the patient resumed Imatinib (400 mg daily), which was later increased to 800 mg due to disease progression in the liver and peritoneum in July 2023. By January 2024, further progression in the liver led to the initiation of Sunitinib. No additional scrotal metastases were noted.

Discussion

Scrotal and Testicular Metastases of GIST: The first case of testicular metastasis from a GIST was reported by Dorić et al. in 2007 [2]. The patient presented with right testicular enlargement seven months after resection of a jejunal GIST. Orchiectomy revealed tumor nodules within the tunica vaginalis, with histology confirming metastatic GIST tissue. The testicular

parenchyma was not involved. Similar cases were described by Perrone et al. in 2008 and Thomas et al. in 2015, both proposing metastases occurred via a persistent peritoneo-vaginalis duct [3,4].

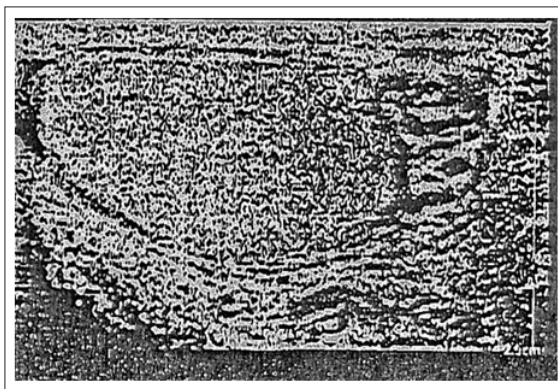
The peritoneo-vaginalis duct is an embryonic structure that connects the peritoneal cavity to the inguinal region, aiding in testicular descent. It typically closes after birth; however, its persistence may result in conditions such as indirect inguinal hernia or hydrocele [5]. In the context of malignancies originating from the gastrointestinal or genitourinary tract, a patent peritoneo-vaginalis duct may facilitate metastatic spread. This hypothesis is further supported by a case reported by Ahmad et al. in 2015, where metastatic GIST tissue was found in the epididymis [6]. Recently, Rota et al. (2024) described an unusual case of GIST metastasizing to the scrotum, penis, and myocardium [7].

E-GIST Involving the Scrotum

Extra-gastrointestinal stromal tumors (E-GISTs) account for approximately 5% of all GISTs. Primary scrotal E-GISTs are exceptionally rare. In 2004, Froehner et al. reported a case of a scrotal mass extending from the abdomen along the inguinal canal, associated with large abdominal masses [8]. A CT-guided biopsy confirmed GIST histology, but the primary site could not be determined. In 2007, Kang et al. described a scrotal E-GIST with no evidence of an alternative primary site, a finding further corroborated by Ali et al. in 2020 [9,10]. These cases highlight the need for thorough histological evaluation to distinguish E-GISTs from other differential diagnoses, such as germ cell tumors.

Conclusion

GISTs can metastasize to unusual locations, including the testis and scrotum. Diagnosis is confirmed using immunohistochemical markers such as CD117 and CD34. For localized primary GISTs, surgical resection remains the cornerstone of treatment, followed by adjuvant Imatinib based on recurrence risk. However, metastatic GISTs carry a poor prognosis due to resistance to tyrosine kinase inhibitors (TKIs), rendering the management of rare metastatic sites particularly challenging.



Figures 1A: Figure 1A: Scrotal ultrasound showing a large 25 cm mass occupying the right hemi-scrotum, with non-visualization of the right testis.

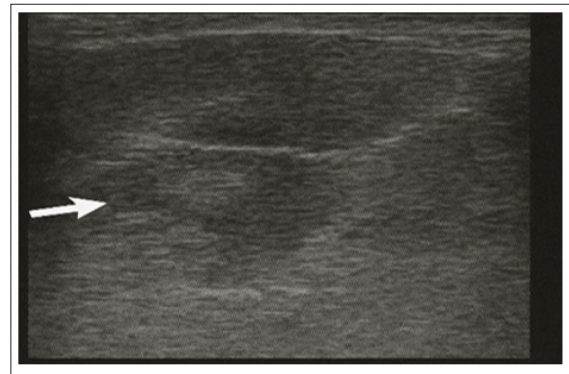
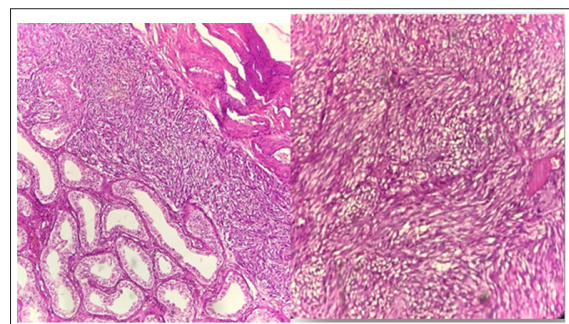


Figure 1B: left testis displaced superiorly and laterally, measuring 35 × 15 × 33 mm.



Figure 2: CT scan showing extensive intraperitoneal masses and multiple liver nodules



Figures 3A,3B: Testicular GIST metastasis showing spindle cells arranged in intersecting fascicles with paranuclear vacuoles and eosinophilic cytoplasm.

Patient Consent: Written informed consent was obtained from the patient for publication of this case report and accompanying images.

Conflict of Interest: The authors declare no conflicts of interest.

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