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CASE REPORT

OLGU SUNUMU

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# Jejunal GIST: A Case Report of Incidentally Detected in A Patient with Breast Cancer

## Meme Kanserli Bir Hastada Tesadüfen Saptanan Jejunal GIST: Olgu Sunumu

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**ABSTRACT**

Gastrointestinal stromal tumors (GISTs) represent the most prevalent mesenchymal neoplasms of the gastrointestinal (GI) tract, typically originating in the stomach (60%) and small intestine (30%). Among the segments of the small intestine, the jejunum is a relatively uncommon site for GIST occurrence. The coexistence of GISTs in patients with a prior history of breast cancer is exceedingly rare, and poses a unique diagnostic and therapeutic challenge due to the possibility of metastatic disease versus a second primary tumor. We present the case of a 55-year-old female with a prior diagnosis of breast carcinoma, who was incidentally found to have a jejunal mass during routine oncologic surveillance. Radiological and endoscopic evaluations raised suspicion for a submucosal tumor, which was subsequently confirmed as a jejunal GIST via histopathological and immunohistochemical analysis following surgical resection. The patient underwent curative segmental jejunal resection with negative margins and had an uneventful postoperative recovery. Risk assessment based on size and mitotic index placed the tumor in a low-risk category, and no adjuvant therapy was deemed necessary. This case highlights the importance of considering GISTs in the differential diagnosis of incidental small bowel masses, especially in patients with a history of malignancy, and underscores the role of complete surgical excision in achieving curative outcomes.

Keywords: GIST, Jejunum, Breast cancer

**ÖZ**

Gastrointestinal stromal tümörler (GIST'ler), gastrointestinal (GI) sistemin en yaygın mezenkimal neoplazmlarını temsil eder ve genellikle mide (%60) ve ince bağırsağın (%30) bölgelerinde ortaya çıkar. İnce bağırsağın segmentleri arasında, jejunum GIST gelişimi açısından nispeten daha az rastlanan bir bölgedir. GIST'lerin, daha önce meme kanseri öyküsü olan hastalarda görülmesi oldukça nadirdir ve metastatik hastalık ile ikinci bir primer tümör ayrımının yapılmasını gerektirdiği için tanı ve tedavi açısından benzersiz zorluklar oluşturur. Bu yazıda, daha önce meme kanseri tanısı almış olan 55 yaşında bir kadın hastada, rutin onkolojik tarama sırasında tesadüfen saptanan jejunal kitle olgusu sunulmaktadır. Radyolojik ve endoskopik değerlendirmeler submukozal bir tümörden şüphelenilmesine yol açmış ve cerrahi rezeksiyon sonrasında yapılan histopatolojik ve immünohistokimyasal analizler ile bu kitlenin jejunal GIST olduğu doğrulanmıştır. Hasta, negatif cerrahi sınırlarla küratif segmental jejunal rezeksiyon geçirmiş ve ameliyat sonrası dönemi sorunsuz tamamlamıştır. Tümörün boyutu ve mitotik indeksi esas alınarak yapılan risk değerlendirmesi, tümörün düşük risk grubunda olduğunu göstermiş ve adjuvan tedaviye gerek duyulmamıştır. Bu olgu, küçük bağırsakta tesadüfi olarak saptanan kitlelerin ayırıcı tanısında GIST'lerin de göz önünde bulundurulması gerektiğini vurgulamakta ve kür sağlayıcı cerrahi rezeksiyonun önemine dikkat çekmektedir.

Anahtar Kelimeler: GIST, Jejunum, Meme kanseri

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## Introduction

Gastrointestinal stromal tumors (GISTs) are the most common mesenchymal neoplasms of the gastrointestinal (GI) tract, accounting for approximately 1–2% of all gastrointestinal malignancies. These tumors are believed to originate from the interstitial cells of Cajal, which serve as pacemaker cells regulating GI motility. GISTs most frequently occur in the stomach (about 60% of cases) and the small intestine (approximately 30%), with the jejunum representing a relatively uncommon site of origin.

Clinically, GISTs may present with nonspecific symptoms such as abdominal pain, gastrointestinal bleeding, or anemia, and in many cases, they may remain asymptomatic and be discovered incidentally during imaging or endoscopic procedures. The diagnosis is confirmed through histopathological examination and immunohistochemical staining, with most GISTs expressing CD117 (c-KIT) and DOG1, which are critical markers for differential diagnosis.

The occurrence of GISTs in patients with a history of another primary malignancy, such as breast cancer, is notably rare. In such cases, distinguishing between metastatic disease and a second primary tumor becomes essential for appropriate management. There is limited literature on the coexistence of breast cancer and GISTs, and it remains unclear whether this association is purely coincidental or may reflect underlying genetic or environmental predispositions.

In this report, we present a rare case of a 55-year-old woman with a prior history of breast carcinoma who was incidentally diagnosed with a jejunal GIST during routine oncologic follow-up. The lesion was successfully managed with surgical resection, and the patient achieved complete recovery. This case adds to the growing body of literature on multiple primary malignancies and highlights the importance of a thorough diagnostic approach in patients with a previous history of cancer.

## Case Presentation

The patient, who was 55 years old, was diagnosed with breast cancer 8 years prior and was under stable follow-up after total mastectomy and adjuvant treatment, applied to us after which a suspicious mass was detected in the small intestine during routine control scans at an external center. The patient's physical examination result was unremarkable. Laboratory results were normal except for a slight decrease in hemoglobin value. When the patient's tomography scans obtained at our center were evaluated, a 2 cm hypervascular mass was observed in the proximal jejunal loop which was not an exophytic extension and did not cause bowel obstruction (Figure 1). Apart from this, no other findings or additional features were found to suggest breast cancer recurrence. It was thought that the described mass could be a GIST or carcinoid tumor. No findings were observed in the patient who underwent upper or lower gastrointestinal endoscopy, except for a small duodenal diverticulum.

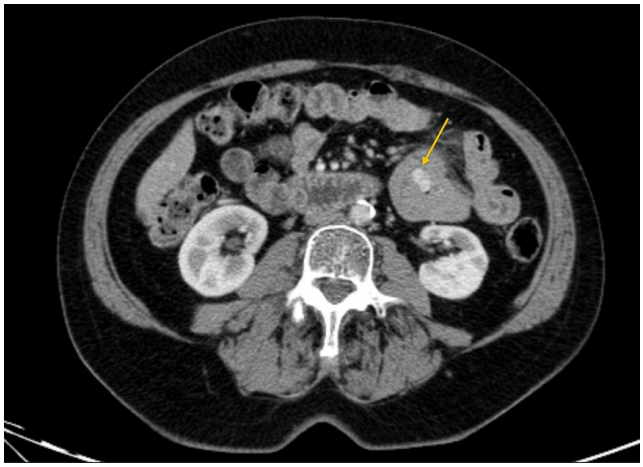

Figure 1: Axial CT scan shows a hypervascular lesion on jejenum.

Additionally, the Ga-68 DOTATATE PET-CT examination did not reveal any conclusive evidence of a carcinoid tumor. The patient was informed that a diagnostic laparoscopic examination would be performed and that if the mass could be observed on laparoscopic examination, primary resection would be performed, but if not found, laparotomy would be performed. At the patient's request for laparotomy, all possible risks were explained to the patient and the decision for surgical resection was made.

In the exploration performed with a median incision above the umbilicus, a non-obstructive mass of approximately 1.5 cm in size was observed 25 cm from Treitz, located in the small bowel serosa, close to the mesenteric surface (Figure 2). The

proximal and distal parts of the mass appeared normal. No pathologic finding was observed in other intestinal loops and solid organs. Following the decision to resect the small bowel loop where the mass was located, the loop was resected with a stapler from five cm proximal and distal parts, and a side-by-side anastomosis was performed. The patient was allowed to consume oral food on the fifth postoperative day. He was discharged with full recovery on the eighth day.

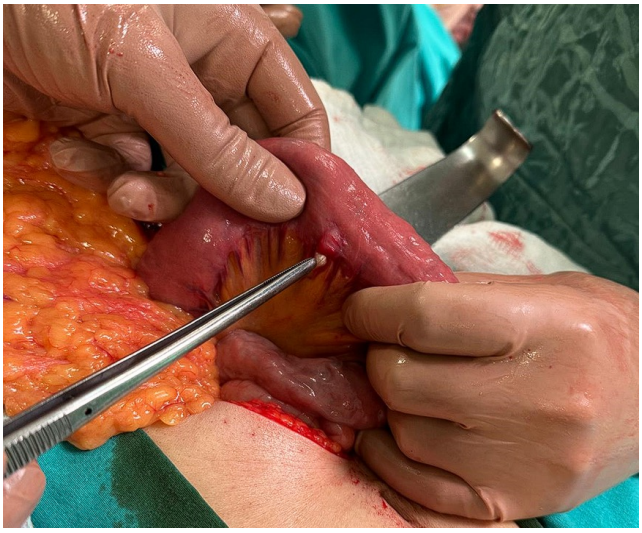

Figure 2: Intraoperative examination, lesion found on the mesenteric side of jejenum.

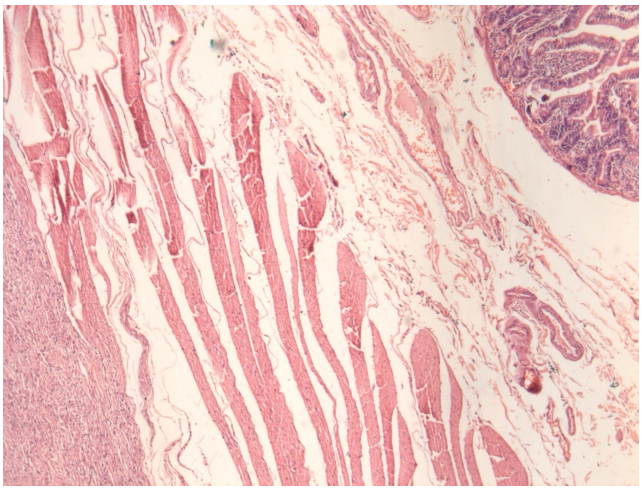

Figure 3: The relationship between the tumor and the intestinal wall is observed (H&E, x5).

Pathological evaluation revealed a mixed cell type with spindle and epithelioid morphology. The tumor was limited to the muscularis propria, there was no bleeding, necrosis, or ulceration, and the surgical margin was negative (Figure 3). Neoplastic cells were found to be positive for CD117, DOG1, and CD34, and negative for Desmin, S100, Caldesmon, and HMB45 (Figure 4). Ki67 index was between %5-10.

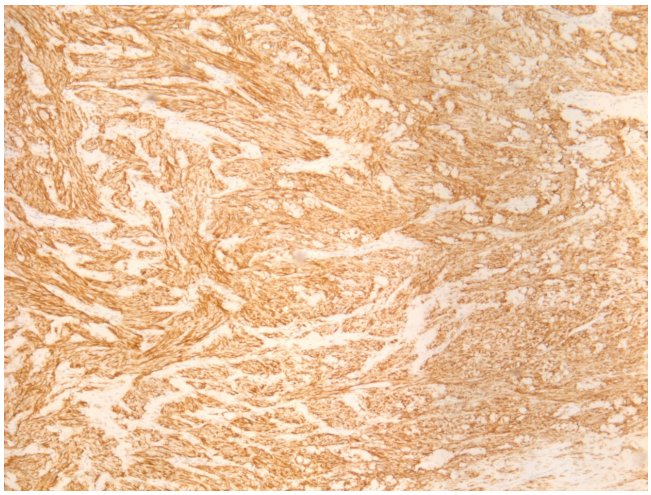

Figure 4: DOG1 staining of tumor (x5)

## Discussion

GISTs are defined as neoplasms with KIT-positive mesenchymal spindle cells or epithelioid features that are commonly found in the gastrointestinal tract. Ligand-independent activation of KIT plays an important role in the molecular pathogenesis of these tumors [1]. Data in the literature reveal that the vast majority of GISTs (75–85%) carry mutations in the KIT gene or, less commonly, in the PDGFRA gene (5%) [2, 3]. Only 10–15% of adult GIST cases and 85% of pediatric cases lack these mutations; these cases are referred to as "wild-type" GISTs [1]. Most GISTs are sporadic, with only 5–10% showing hereditary characteristics. Hereditary GIST cases are frequently associated with Carney triad, Carney-Stratakis syndrome, neurofibromatosis type 1 (NF1), and familial GIST [4].

In particular, familial GIST cases with germline KIT mutations have been reported to be accompanied by hereditary breast and ovarian cancer syndrome, albeit rarely [5]. However, when a large single-center case series of sporadic GIST cases was examined, it was found that a primary cancer developed metachronously or synchronously in approximately 18% of these cases. The most common of these primary cancers are gastrointestinal, genitourinary, gynecological, and breast cancers [6]. The coexistence of GISTs and other malignancies raises important questions

about potential shared molecular pathways or environmental risk factors, although definitive causal mechanisms remain to be clarified.

There is a history of breast cancer in our case, and under current conditions, a genetic study on GIST could not be performed. Therefore, it was not possible to obtain genetic information about the case. However, since there is no history of a hereditary disease in the patient's family and according to the available data, it is thought that the case was a sporadically developing GIST and that this tumor was accompanied by breast cancer metachronously. When genetic studies are available, it will be possible to clarify this relationship more clearly. From a clinical perspective, such patients warrant careful long-term follow-up to monitor for both GIST recurrence and the development of other malignancies.

The clinical presentation of GISTs can be quite variable. The symptoms that occur in patients may vary depending on the location and size of the tumor. The most common symptoms generally include gastrointestinal bleeding, abdominal pain, a palpable mass, or intestinal obstruction findings. These symptoms may vary depending on the size of the tumor and the pressure it exerts on the surrounding tissues. Smaller tumors may often remain asymptomatic, while larger tumors, particularly those arising from the stomach, may cause more obvious symptoms such as a palpable mass. However, rarer and life-threatening clinical presentations of GISTs are also encountered in the literature. In particular, atypical conditions such as massive gastrointestinal bleeding and intussusception leading to intestinal obstruction have been reported [7, 8].

In our case, the small size of the tumor probably caused the absence of symptoms. Such asymptomatic cases are usually detected incidentally during imaging performed for another reason, as in our case. The diagnosis of such GIST cases, which do not present any clinical symptoms, is more challenging and often relies on high-resolution cross-sectional imaging modalities or endoscopic evaluation. Endoscopic ultrasound (EUS) may assist in characterizing submucosal lesions, particularly when located in the stomach or duodenum. However, small intestinal GISTs, such as those in the jejunum, are often beyond the reach of conventional endoscopy, which can delay diagnosis.

Treatment approaches in GIST are shaped by factors such as tumor stage, localization, size, molecular features, and metastatic status. As frequently emphasized in the literature, the primary treatment option in localized jejunal GIST cases is complete resection of the tumor with negative surgical margins, which forms the basis of curative treatment [9]. In our case, complete resection was achieved with surgical intervention, and no additional adjuvant treatment was required.

The risk assessment in this case is best interpreted within the framework of the AFIP (Armed Forces Institute of Pathology) classification, as detailed in Table 1, which considers tumor size, mitotic activity, and anatomical location [10]. According to this system, the tumor in this case—measuring 1 cm in diameter and exhibiting 3 mitoses per 50 high-power fields (HPF)—was classified as having a 0% risk of progressive disease (recurrence or metastasis). The tumor was limited to the muscularis propria without mucosal or serosal invasion, and surgical margins were free of tumor infiltration. These findings further support a favorable prognosis and negate the need for adjuvant therapy in the absence of high-risk features.

Table 1. AFIP Risk Classification for GIST (Gastrointestinal Stromal Tumors)

| Tumor Size (cm) | Mitotic Rate (per 50 HPF) | Tumor Location | Risk of Progressive Disease |
|---|---|---|---|
| ≤2 | ≤5 | Any | Very Low |
| >2 – ≤5 | ≤5 | Gastric | Low |
| >2 – ≤5 | ≤5 | Non-gastric | Intermediate |
| >5 – ≤10 | ≤5 | Gastric | Intermediate |
| >5 – ≤10 | ≤5 | Non-gastric | High |
| >10 | Any | Any | High |
| Any size | >5 | Any | High |

Nevertheless, accurate risk stratification is crucial for postoperative management and surveillance planning. Patients classified as low-risk often require less intensive follow-up, whereas those in intermediate or high-risk categories benefit from closer surveillance and consideration of adjuvant tyrosine kinase inhibitor therapy.

In cases of GIST that cannot be surgically removed, have relapsed, or present with metastasis at diagnosis, tyrosine kinase inhibitors (TKIs) play an essential role in the treatment process. Imatinib is used as a first-line treatment by suppressing kinase activity, especially in tumors carrying KIT and PDGFRA mutations [10]. In cases of resistance to imatinib or progression after initial response, sunitinib and regorafenib are used as second- and third-line treatments, respectively [11]. Recent advancements have also introduced novel agents such as ripretinib for later lines of therapy, expanding the therapeutic arsenal against advanced GIST.

## Conclusion

Follow-up imaging in breast cancer patients is very important for early detection of recurrence and evaluation of possible metastatic spread. However, during routine follow-up imaging studies, new tumors can be detected incidentally in different localizations that can directly affect the patient's survival. In particular, rare tumors of the gastrointestinal system, such as jejunal GIST, which can be asymptomatic, can be noticed incidentally by imaging methods. This can positively affect the prognosis by providing early diagnosis and therefore rapid implementation of appropriate treatment approaches. Therefore, long-term follow-up of breast cancer patients has a critical role not only for the control of the primary disease but also for the early diagnosis of other malignancies that may develop incidentally.

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