Journal of Clinical Gastroenterology and Hepatology
ISSN: 2575-7733

Open access Short Communication

Understanding Rectal Gastrointestinal Stromal Tumors: Etiology, Diagnosis, and Treatment

Thomas Mark*

Department of Gastro-hepatology, Peking University, China

INTRODUCTION

Rectal gastrointestinal stromal tumors represent a subset of rare mesenchymal neoplasms arising from the gastrointestinal tract. Despite their infrequency, rectal pose unique diagnostic and therapeutic challenges due to their potential for local invasion, metastasis, and variable clinical behavior. This theory aims to elucidate the pathogenesis, clinical presentation, diagnostic modalities, and treatment approaches for rectal to enhance clinician awareness and optimize patient care. Rectal originate from the interstitial cells of or their precursors, which regulate gastrointestinal motility and peristalsis [1,2].

DESCRIPTION

Mutations in the proto-oncogene or platelet-derived growth factor receptor alpha gene are commonly implicated in the pathogenesis of leading to constitutive activation of tyrosine kinase signaling pathways and uncontrolled cellular proliferation. While the majority of rectal are sporadic, a subset may arise in the context of hereditary syndromes, such as neurofibromatosis and Carney triad. Rectal often present with nonspecific symptoms, including rectal bleeding, altered bowel habits, abdominal pain, and palpable mass on digital rectal examination. The clinical course of rectal varies widely, ranging from indolent, asymptomatic tumors to aggressive malignancies with metastatic spread. Large tumors or those located in close proximity to the anal sphincter may cause obstructive symptoms or urinary dysfunction, further complicating their management. The diagnosis of rectal relies on a combination of clinical evaluation, imaging studies, and histopathological examination. Endoscopic evaluation, including rectoscopy or colonoscopy, may reveal an intraluminal mass or mucosal irregularity, although deeper lesions may be missed. Cross-sectional imaging modalities, such as computed tomography and magnetic resonance imaging provide detailed anatomical information, delineating tumor size, extent of invasion, and presence of metastases. Definitive diagnosis requires histopathological examination of tissue specimens obtained via endoscopic biopsy or surgical resection, with immunohistochemical staining for serving as diagnostic markers for the management of rectal is guided by tumor size, location, mitotic index, and presence of metastases. Surgical resection remains the cornerstone of treatment for localized disease, aiming for complete tumor excision with negative margins while preserving sphincter function whenever feasible. In select cases, neoadjuvant or adjuvant imatinib therapy may be administered to downsize tumors, facilitate surgical resection, or reduce the risk of recurrence. Advanced or metastatic rectal may require systemic therapy with tyrosine kinase inhibitors such as imatinib, sunitinib, or regorafenib, to control tumor growth and improve progression-free survival. Rectal gastrointestinal stromal tumors represent a rare subset of mesenchymal neoplasms with diverse clinical presentations and variable prognoses. A multidisciplinary approach involving gastroenterologists, surgeons, radiologists, and oncologists is essential for accurate diagnosis, risk stratification, and individualized treatment planning [3,4].

CONCLUSION

Ongoing research efforts aimed at elucidating the molecular pathogenesis of and identifying novel therapeutic targets hold promise for further advancing the management of rectal and improving patient outcomes. Recent advances in the understanding of pathogenesis have spurred the development of novel therapeutic approaches aimed at overcoming resistance to conventional TKI therapy. Second-generation TKIs, including avapritinib and ripretinib, exhibit enhanced potency and specificity against resistant mutations, offering promising therapeutic alternatives for patients with refractory disease. Furthermore, combination strategies incorporating immunotherapy, radiotherapy, and locoregional interventions hold potential for synergistic antitumor effects and improved

Received:	29-May-2024	**Manuscript No:**	IPJCGH-24-20561
Editor assigned:	31-May-2024	**PreQC No:**	IPJCGH-24-20561 (PQ)
Reviewed:	14-June-2024	**QC No:**	IPJCGH-24-20561
Revised:	19-June-2024	**Manuscript No:**	IPJCGH-24-20561 (R)
Published:	26-June-2024	**DOI:**	10.36648/2575-7733.8.3.21

Corresponding author Thomas Mark, Department of Gastro-hepatology, Peking University, China, E-mail: mark@gmail.com

Citation Mark T (2024) Understanding Rectal Gastrointestinal Stromal Tumors: Etiology, Diagnosis, and Treatment. J Clin Gastroenterol Hepatol. 8:21.

treatment outcomes in select patient populations.

ACKNOWLEDGEMENT

None.

CONFLICT OF INTEREST

The authors declare that they have no conflict of interest.

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