Rectal Gastrointestinal Stromal Tumor: A Case Report

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CASE REPORT

A 55- years old diabetic hypertensive male presented with chief complaints of bleeding per rectum on and off and altered bowel habits for 2 years. He had negative family history for any abdominal malignancy. Digital rectal examination revealed an ulceroproliferative growth on anterior and lateral walls of rectum 4 cm from anal verge, firm to hard in consistency involving ¼ circumference. Upper margin of growth could not be felt and on withdrawal finger was stained with blood.

Transanal proctoscopic incisional biopsy was done from the rectal growth and it was found to be a case of rectal GIST. The diagnosis was confirmed by immunohistochemistry showing immunoreactivity (score 3+ i.e. immunoreactive in 51-75% cells) to CD117. Colonoscopy confirmed the presence of irregular rectal growth of ulceroproliferative nature of size 6 x 4 cm, starting 4 cm from anal verge. Rest of the colonic examination negated the presence of any synchronous tumour.

Abdominopelvic CECT showed gross circumferential thickening of rectum of size 3.1 x 6 cm with heterogenous enhancement and loss of layers of differentiation suggestive of rectal mass abutting the prostate [figure - 1]. No regional lymphadenopathy or distant metastases were found.

Serum CEA was within normal limits. Apart from microcytic/hypochromic anaemia, other hematological parameters were normal. After proper consent, patient underwent abdomino-perineal excision (APE) with total mesorectal excision (TME) and permanent colostomy. Postoperative period was uneventful except for seroma in the perineal wound which was managed conservatively. Patient was discharged on 10th postoperative day.

Gross observation of the resected specimen showed 35 cm long colon and rectum with their mesentery. Cut open specimen revealed a grey brown mass of size 7.2 x 6.4 x 5.1 cm in rectum. Cut section showed grey white mass involving full thickness of rectal wall. Histopathological examination of the APE specimen confirmed it as rectal GIST (spindle cell type) involving muscular layer up to serosal layer with mitotic count > 10/50 HPF. Proximal, distal and circumferen-

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tial radial margins were uninvolved by the tumor. Specimen lymph nodes were also uninvolved by the tumour (figure - 2, figure - 3).

Following the histopathological confirmation and risk assessment, patient was started on adjuvant therapy with Imatinib mesylate 400 mg OD, to decrease the risk of recurrence. After one year of follow up and patient on continued Imatinib, no signs of disease progression have been found.

DISCUSSION

Rectal GIST constitutes 5% of all GISTs and 0.1% of all tumors originating in the rectum. Common sites of metastasis for GIST include liver, peritoneum and omentum; lymph node and extra-abdominal metastases are rare.6

In the past decade the understanding and treatment of GIST has reached remarkable advances due to (a) the identification of constitutively active signals (due to mutation of c-kit and PDGFRA genes encoding receptor tyrosine kinases) and (b) the development of therapeutic agents that suppress tumor growth by specifically targeting and inhibiting these signals. As the incidence of rectal GIST is much lower than that of GIST in the stomach or small intestine, the clinicopathological profiles of rectal GIST have not yet been accurately characterised, and therefore it is the tendency to validate the same prognostic factors for the latter as for such tumours at other sites, particularly gastric GIST. The three established prognostic factors for GIST are tumor size, mitotic index and tumor site of origin, with mitotic count the most important (Table 1).7,8 Individuals with rectal GIST have higher risk of progression than those with small bowel or gastric GISTs of comparable size and mitotic count. Degrees of cellularity and atypia have also been suggested as useful criteria, but their reproducibility is more problematic. The epithelioid phenotype, which seems to lead to a worse outcome, together with symptoms lasting for at least a year, might be considered as further prognostic factors.

It is generally agreed that complete surgical resection with negative tumour margins is the principal curative procedure for primary and non-metastatic tumours, particularly for those at a low risk.9 For rectal GIST, various surgical procedures may be considered, including local excision, anterior resection of the rectum and abdomino-perineal resection. The choice of procedure depends on tumour size and location.

Neoadjuvant imatinib may enhance the resectability of inoperable malignant GIST and may allow for optimal surgical timing. Therapy with imatinib is also used in the adjuvant post-operative treatment of tumours at a high risk or in cases of incomplete surgical resection. A randomized trial has demonstrated prolonged recurrence-free survival in patients assigned to one year of imatinib therapy compared with

<table>
<thead>
<tr>
<th>Size</th>
<th>Mitotic Count</th>
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<tr>
<td>Very low risk</td>
<td>&lt; 2 cm</td>
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<tr>
<td>Low risk</td>
<td>&lt; 5 cm</td>
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<tr>
<td>Intermediate</td>
<td>6–10/50 HPF</td>
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<td>Intermediate</td>
<td>5–10 cm</td>
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<tr>
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<td>&gt; 5 cm</td>
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<tr>
<td>High risk</td>
<td>&gt; 10 cm</td>
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<tr>
<td>Any size</td>
<td>&gt; 10/50 HPF</td>
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</tbody>
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Table-1: Defining risk of aggressive behavior in GIST7

Figure-1: CECT of pelvis with I.V. and per-rectal contrast, axial section showing gross circumferential thickening of rectum with heterogenous enhancement and loss of layers differentiation suggestive of rectal mass abutting the prostate

Figure-2: Typical spindle cell gastrointestinal tumor composed of interlacing fascicles of cells with cigar-shaped nuclei (HE x 100)

Figure-3: Shows few mitotic figures (HE x 400)
those assigned with placebo. So it is accepted that imatinib is a valid treatment for advanced or metastatic tumours as well as localized GIST post surgery, but further evidence for the efficiency of this drug is needed in the case of high risk tumours and for the neoadjuvant therapy.

CONCLUSION

Although rectal GIST is extremely rare, it should be included in differential diagnosis when a tumour in the rectum is detected. The diagnostic workup of rectal GIST is essentially the same as that advised for any other type of rectal neoplasia. Preoperative biopsy of the tumour is important, since it can reach a certain preoperative diagnosis by means of the immunohistochemical characterization of CD34 and CD117 and management can be tailored accordingly especially with regards to neo adjuvant therapy. Because of small body of evidence especially in rectal GIST, it is difficult to assess the necessary extent of surgical resection and lymphadenectomy, indication for treatment with imatinib and optimal length of the adjuvant therapy. Further studies with large series of patients and long term follow up of ongoing trials are necessary to establish the most effective treatment strategy for patients with rectal GIST.

REFERENCES