Small Bowel Leiomyoma in a Patient with Neurofibromatosis - A Case Report and Review of Literature

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Abstract:
Neurofibromatosis, an autosomal dominantly inherited disease occurs 1 in 3000 live births. The disease has varied manifestations in the skin, nervous tissue, bone and soft tissues. Gastrointestinal tract may also be involved with multiple Neurofibromas, giving rise to obscure gastrointestinal bleeding and obstruction. There is an increased association between benign small bowel Leiomyomas and Neurofibromatosis but this has been documented in very few instances. Due to its rarity, a high degree of suspicion for gastrointestinal tumor in patients with Neurofibromatosis is needed for timely diagnosis. Present report describes an interesting case of a small intestinal Leiomyoma in a patient with cutaneous Neurofibromatosis.

Keyword: Small Intestinal Leiomyoma, Neurofibromatosis.

Introduction:
Leiomyomas of the gastrointestinal tract, although rare, are the most common benign non epithelial tumors of the small intestine. The incidence of small bowel Leiomyoma is comparatively high in patients with neurofibromatosis compared to the general population because there is an increased association between the two but this has been documented in very few instances.

Case Report:
A 45 yr old male was admitted at our hospital with a history of right sided lower abdominal pain of 5 days duration. There was no history of fever, vomiting, hematemesis, melena or altered bowel habits. Since early puberty, he had developed multiple soft painless skin lesions over his face and body. There was no significant past medical history of note and no family history of similar skin lesion. On physical examination, the patient was alert, and oriented. He had generalized cutaneous fibromatosis.
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(Fig 1) There was no pallor. His abdomen was tender over the right lumbar and iliac region with guarding. Bowel sounds were normal. Routine blood investigations were within normal limits. X-ray of the chest and abdomen were normal. Ultrasound abdomen detected an ill defined mass in the right iliac region. CT Abdomen plain and contrast revealed an irregular soft tissue mass of size 7x6 cm arising from the small intestine. With the above findings the patient was planned for surgery and exploratory laparotomy was done. Intraoperatively a smooth, bosselated mass of size 9x7 cms (Fig 2) arising from the anti-mesentric border of ileum 20 cm proximal to the ileocaecal junction was identified. The mass was firm in consistency and appeared necrotic on gross examination. Proximal bowel loops appeared normal and there was no evidence of obstruction. Other intra abdominal organs appeared normal and there was no gross lymphadenopathy. Ileal resection of the affected segment with wide margin of clearance was done and end to end anastomosis was carried out. Post operatively the patient had an uneventful recovery. Microscopic examination of the specimen revealed well differentiated smooth muscle cells, suggestive of leiomyoma arising from the subserosal layer of a normal ileum (Fig 3).

**DISCUSSION:**
Tumors of the small intestine are rare. Even though the small bowel accounts for 80% of the length and 90% of the mucosal surface of the gastrointestinal tract, only 3% to 6% of gastrointestinal tumors and 1% of gastrointestinal malignancies arise from the small bowel. Benign tumors of the small bowel occur in 0.1% of the surgical specimens. Of these, adenomas, leiomyomas and lipomas are the three frequent tumors. Many benign tumors remain asymptomatic and are discovered incidentally at surgery or post mortem examination. Clinically, leiomyoma is probably the most important symptomatic small bowel tumor. It is found at all level of small intestine but most commonly in the subserosa and submucosal surface. It is usually diagnosed in the fifth decade of life, although it may occur at any age. It has no sex predilection. Grossly, leiomyomas are white gray lesions. Microscopically, they contain well differentiated smooth muscle cells and no mitoses, which differentiates them from their malignant counterpart.

Patients may present with abdominal pain, gastrointestinal bleeding (due to central necrosis or ulceration of tumor mucosa), intestinal obstruction or volvulus. In a study of 1399 benign tumors of the small intestine, only 14 cases were associated with neurofibromatosis. The frequency of involvement of the gastrointestinal tract in neurofibromatosis varies from 12-25% but only a small number of patients have symptoms. Hochberg, however found 5 cases of intestinal leiomyomas in 39 cases of neurofibromatosis with gastrointestinal involvement in a survey of literature. This suggest an increased frequency of leiomyomas in patients with neurofibromatosis than previously recognised.

A high degree of suspicion for gastrointestinal tumor in patients with neurofibromatosis is needed for timely diagnosis. Gastrointestinal involvement in neurofibromatosis is often asymptomatic and there is delay in diagnosis. The average interval from onset of GI symptoms to diagnosis with GI neoplasm was 2.8 years in patients reviewed in a study. Diagnosis may be difficult due to nonspecific symptoms, as well the predominantly small bowel location making radiographic visualization
difficult. Explorative laparotomy may be essential for removing any doubts regarding diagnosis. Leiomyomas in patients with neurofibromatosis tend to be multiple and preferentially involve the proximal small bowel. The usual mode of presentation is as a result of haemorrhage but ulceration, intussusception, perforation and obstruction can occur.

It is a well known clinical observation that small bowel tumors are rarer than tumors of stomach and colon. Many speculations have been offered: the rapid transit time, the fluid content of small intestine reduces exposure to any potential carcinogens and the relative sterility of the small bowel as compared with the large bowel. Lowenfels hypothesized that an immune surveillance system of the small bowel militates against the development of tumors. The increased incidence of primary and metastatic small bowel tumors in immune suppressed states, supports this hypothesis. The occurrence of multiple small intestinal leiomyomas in patients with neurofibromatosis suggest a defective or deficient immune surveillance, thereby predisposing to neoplastic transformation. Due to the high incidence of malignancy and the possibility of malignant degeneration in the more frequently occurring benign neoplasms, all neoplasms should be resected with the adequate margins, since differentiation from leiomyosarcoma is difficult even on pathological examination. Lymph node resection is not routinely performed because leiomyosarcoma does not spread via lymphatics. Recurrence and metastasis are rare.

CONCLUSION:
Small intestinal leiomyoma should be considered in a patient with neurofibromatosis presenting with gastrointestinal symptoms due to an increased association between the two conditions. Hence a high index of suspicion is needed for timely diagnosis and management.

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