



Case Report

C-KIT NEGATIVE GASTRO INTESTINAL STROMAL TUMOUR WITH SUBCUTANEOUS TISSUE AND LYMPH NODE METASTASIS.

Md Mahiuddin Matubber¹, Syed Muhammad Baqui Billah²,
Subinoy Krishna Paul³, Jahirul Islam⁴, Roksana Afroj⁵

Abstract

We report a rare case of c-kit negative gastrointestinal stromal tumor with metastasis in left cervical lymph nodes and subcutaneous tissue. The patient presented to us with recurrent neck swelling, perforation of gas containing hollow viscous and multiple subcutaneous swellings. After confirmation of diagnosis we transferred the patient to medical oncologist. Patient expired two months after starting chemotherapy.

Introduction

Though most common among the mesenchymal tumors of gastrointestinal tract (GIT), gastrointestinal stromal tumors (GIST) are relatively rare form of cancer¹. Being positive for CD117 (c-Kit), the disease is primarily caused by KIT or platelet-derived growth factor receptor A (PDGFRA) activating mutation. Radiology, histology and immunohistochemistry have been the tool of diagnosis of this tumor. Different cancer experts have come up with the guidelines to diagnose and treat such tumors. National Comprehensive Cancer Network (NCCN) revised a guideline about the pathologic assessment and principles of surgery for GIST² while European Society for Medical Oncology (ESMO) formulated clinical guideline³ for diagnosis, treatment and follow-up at different stages of sarcoma. Researchers from Spain released the third version of GIST guidelines⁴ for diagnosis and treatment which speaks about the imatinib as first line of adjuvant treatment and sunitinib as second

line in high risk patients and surgery for localized disease. The common metastatic sites are liver, intraperitoneal cavity, liver, liver and intraperitoneal cavity, stomach, small intestine, omentum/mesentery, colorectal, bone and even lungs. Lymph node metastasis is very rare. We hereby report a young patient with supraclavicular lymph node metastasis of jejunal GIST who was c-Kit negative.

Case Report

A 21 year-old- male student non-icteric, non-diabetic and non- asthmatic was admitted to our hospital with intestinal perforation with a history of weight loss for the last 6 months. He had no history of fever, cough, voice change, bony pain, altered bowel habit, hematemesis, malena, or hemoptysis. He also suffering from recurrent painless small swelling over left side of neck about 2 years before. After 1 year of initial presentation of the swelling, the local doctor excised the swelling without any concrete diagnosis.

Author:

1. Associate Professor, Department of Surgery, Dhaka Medical Collage Hospital, Dhaka.
2. Technical Officer, Field epidemiology training program, IEDCR, Mohakhali, Dhaka.
3. Assistant professor, Department of Surgery, Dhaka Medical Collage, Dhaka.

4,5. Indoor Medical Officer, Department of Surgery, Dhaka Medical Collage, Dhaka.

Correspondence to: Dr. Md Mahiuddin Matubber, Associate Professor, Department of Surgery, Dhaka Medical Collage Hospital, Dhaka.

Mobile:01725176469

email:drmm1967@yahoo.com.

Nearly 1 year after excision, the patient again developed similar swellings in the same area (Fig1) which gradually increased in size and diagnosed as TB through Fine Needle Aspiration Cytology (FNAC). The patient received category 1 (CAT-1) anti Tb medication but there was no response to the treatment for the first one and a half months. Then he was sent for FNAC again and the diagnosis was proliferating trichilemmal tumor. In addition, another FNAC reported poorly differentiated metastatic carcinoma. He took 6 cycles of chemotherapy and 22 fractions of radiotherapy but there was no improvement. After 4 months of receiving chemotherapy and radiotherapy, he got admitted to surgery department with perforation of gas containing hollow viscus. Resection and anastomosis was done after laparotomy. Multiple para aortic and mesenteric enlarged lymph nodes were found with multiple lumps throughout the small intestine along the mesenteric border (Fig2). Histopathology of the resected intestine revealed GIST with lymph node metastasis though c-Kit was negative. After 12 days of operation, he developed multiple painless hard swelling over different parts of the body without any ulceration over the surface. Excision biopsy from the swelling was compatible with metastatic GIST. He was sent to oncology department for further treatment. The patient died two months after discharge from surgery department while receiving chemotherapy.



Fig-1: Metastasis in the cervical lymph nodes and subcutaneous tissue from GIST.



Fig-2: GIST in the small gut.

Discussion

With increasing knowledge of diagnostic evaluation through GIST pathogenesis, identification and differentiation of GIST from other submucosal tumors have become easier now-a-days. But because of its rarity, doctors sometimes miss the diagnosis at initial stage which turns into misdiagnosis leading to inept management at late diagnosis. Sometimes the patients come at such a stage when there is nothing much for the clinicians to do. Our case was so atypical that it was really hard to diagnose at primary level. Firstly we can think of the age of occurrence. Though the disease occur rarely in young age group and the outcome is favorable in young patients, our case was not like that⁵. Lymph node metastasis is rare in GIST but we had encountered such^{6,7}. Skin metastasis is another form of uncommon presentation⁸. Even other soft tissue metastasis is also rare, though skeletal muscle metastasis is documented⁹. Our case represents a unique atypical GIST where lymph node and subcutaneous metastasis was evident. The diagnostic heterogeneity at different levels of service

facilities needs to be readjusted for better management opportunities. Though different treatment are recommended by researchers¹⁰, we are still way behind those treatment modalities revealing limitations at treatment level as well as at diagnosis.

Conclusion

We presented an atypical case of c-Kit negative GIST at early age with lymph node and subcutaneous metastasis, the prognosis of which was not favorable probably due to diagnostic dilemma at initial stage. From our experience and search we can advocate for any neck swelling to be assessed meticulously at every stage of encounter.

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