Gastrointestinal Stromal Tumor presenting in a neurofibromatosis patient

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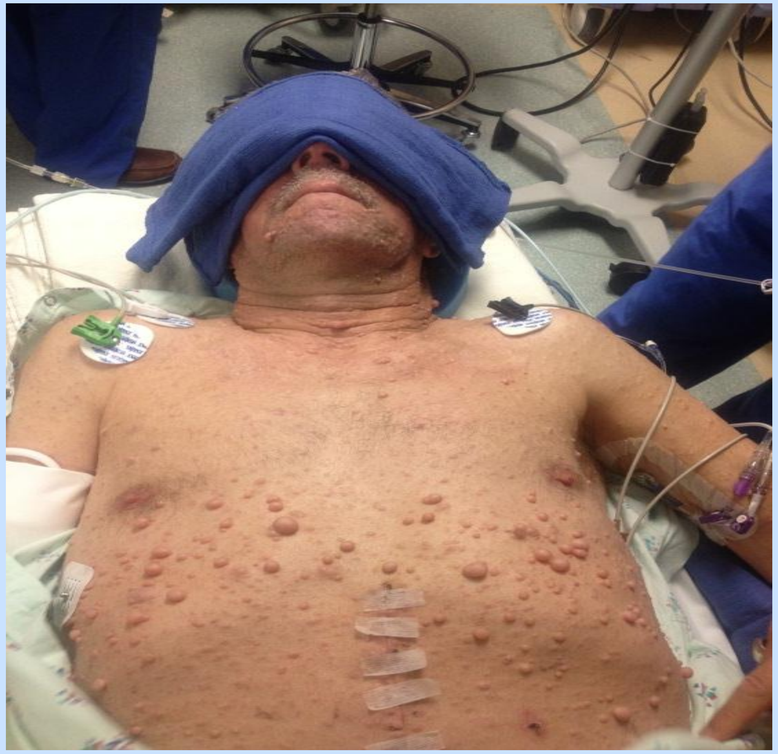
Introduction

Neurofibromatosis is one of the most common autosomal dominant conditions occurring in approximately 1/3000 births, has long been associated with multiple GI tumors (2-25% of patients) including Neurofibroma's and leiomyomas. GIST is a less common tumor in these particular patients. These are usually present in the stomach and small intestines



case

A 59-year-old, Caucasian male with a history of neurofibromatosis type 1 started complaining of increased acid reflux that was not controlled on PPI's. Patient denied any abdominal pain, nausea, vomiting, abnormal bowel habits or bloody stools. His abdomen was soft, with multiple neurofibroma's covering his trunk and face. CT abdomen revealed an exophytic mass in the distal jejunum measuring 4.2x3.5x3.2 cm. PET CT showed two foci of abnormal uptake in the abdomen, one in left upper abdomen which could represent a small bowel lesion with another lesion near the duodeno-jejunal junction. He underwent CT guided biopsy which confirmed the presence of metastatic tumors. He underwent exploratory laparoscopy with small bowel resection and end to side duodenojejunostomy. Pathology later confirmed the diagnosis of gastrointestinal stromal tumor in both sites. PCR and sequencing was negative for c-KIT and PDGFRA. CD117 was strongly positive. Later on, he presented again with nausea and vomiting for one week. CT abdomen showed distended stomach suggesting gastric outlet obstruction without a leak. He underwent subsequent EGD with dilatation of the anastomosis site.

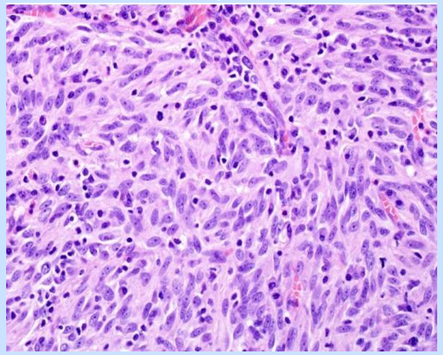


Figure (1)
Small bowel GIST: spindle shaped cells with scattered mitotic features

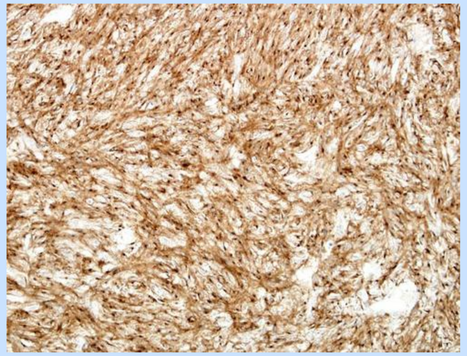


Figure (2)
Small bowel GIST: +CD 117 (c-Kit)

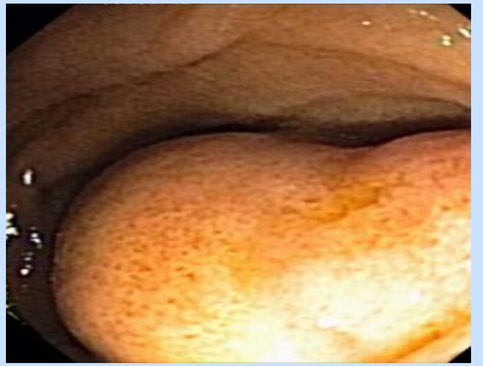


Figure (3)
Endoscopy shows small bowel intraluminal GIST

Conclusion

GIST's are the most common mesenchymal neoplasms of the Gastrointestinal tract. Most GIST occur sporadically and multiplicity is very rare. NF associated GIST are phenotypically and genotypically distinct from sporadic GIST. Though GIST 's display strong positivity for c-KIT, mutation studies for KIT and PDGFRA are typically negative in NF1 associated GIST. It has predilection to the stomach and small intestines. Clinical manifestations depend on its size and location. Gastrointestinal tract bleeding (50%) is the most common, followed by abdominal pain (20-50%) and obstruction (20%). Although uncommon, rupture and peritonitis have been reported.

References

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* Ku M.C., Tsai C.M., Tyan Y.S., Multiple gastrointestinal tumors in a patient with type 1 Neurofibromatosis presenting with tumor rupture and peritonitis. Clinical Imaging 34(2010) 57-59