

Pancreatic Extragastrointestinal Stromal Tumor in a Patient with Neurofibromas Presenting with Iron Deficiency Anemia

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Abstract

Background: Extragastrointestinal stromal tumors are unusual, with those arising in the pancreas even more so. Because extragastrointestinal stroma tumors involving the pancreas may present nonspecifically as an upper gastrointestinal bleed, it is important to adequately work up the patient to rule out malignancy to optimize the outcome for the patient. While surgery with complete resection of the tumor is the treatment of choice, it is important to consider the role of tumor markers in targeted therapy for a nonresectable disease or nonsurgical patients.

Case presentation: We describe a 53-year old man with a history of neurofibromas and irritable bowel syndrome who presented with abdominal pain and persistent iron deficiency anemia. He was found to have an extragastrointestinal stromal tumor of the pancreas, which stained positive for CD117 and DOG1. The treatment plan for this patient is targeted therapy with Imatinib, a tyrosine kinase inhibitor effective in the treatment of CD117+ cancer cells, followed by possible surgical resection.

Conclusions: The pancreatic extragastrointestinal stromal tumor is a rare malignancy that can present with acute, nonspecific findings of a gastrointestinal bleed including anemia, fatigue, and melanotic bowel movements. With adequate history and physical examination, we can appropriately work up a patient with this rare disease and develop an appropriate treatment plan for the patient.

Keywords: pancreatic extragastrointestinal stromal tumor; neurofibromas; anemia, melena; CD117; Imatinib

List of Abbreviations: eGIST – extragastrointestinal stromal tumor EUS – endoscopic ultrasound; FNA – fine needle aspiration; GIST – gastrointestinal stromal tumor GIT – gastrointestinal tract; NF1 – neurofibromatosis 1; PRBCs – packed red blood cells UGI – upper gastrointestinal

Background

Gastrointestinal stromal tumors (GISTs) tend to occur as primary mesenchymal tumors in the gastrointestinal tract (GIT). In descending order of frequency, occurrences in the stomach, small bowel, large bowel, and esophagus make up over 85% of GISTs [1]. Extragastric stromal tumors (eGISTs) make up 5-10% of GISTs, with the disease of the pancreas accounting for 5% of these cases [2]. Immunohistochemical staining is useful for diagnosis, as positivity for CD117 confirms the diagnosis of GIST [1]. CD117 is also known as c-kit, which is a receptor tyrosine kinase. Increased expression of this protein leads to upregulated cell proliferation, which results in the tumorigenesis of GISTs. Other tumor markers include PDGFR- α , CD34, SMA, desmin, and DOG1 [3,4]. GISTs have also been known to occur in patients with a history of neurofibromatosis I (NF1). Neurofibromatosis I is due to a defect in the NF1 gene which encodes neurofibromin 1, a regulator of RAS proteins [5]. RAS proteins are key regulators in signaling cascades of cellular processes [6]. Treatment for GISTs is surgical resection, with the use of targeted antineoplastic agents for nonresectable disease or nonsurgical candidates.

Case Presentation

A 53-year old man with a history of neurofibromas diagnosed by the presence of skin lesions but never genetically confirmed to have NF1 presented with diffuse periumbilical pain and a one-day history of multiple bouts of melanic loose stools. He had been hospitalized one month prior with low hemoglobin (5.6g/dL, reference range 13.8-17.2g/dL) and iron deficiency anemia on laboratory findings. Additionally, he was found to have a mass measuring 2.3 x 1.5cm at the pancreaticoduodenal groove on a CT scan of the abdomen. He was discharged and in the process of being worked up as outpatient with endoscopic ultrasound (EUS) and biopsy of duodenal and pancreatic abnormalities one week prior to return to the hospital with abdominal pain and dark stools. The possibility of dark stools occurring secondary to treatment of patient's iron deficiency anemia with iron supplements was considered on the differential diagnosis. Associated symptoms included occasional dizziness and palpitations. The patient denied fevers, chills, vomiting, chest pain, shortness of breath, or dysuria. During this admission, the patient was found to have iron deficiency anemia again with a hemoglobin of 8.3, hematocrit 26, ferritin 57, iron 32, TIBC 299, and % saturation of 11. Patient's laboratory also showed mild leukocytosis to 11.1. The patient received 1 unit of packed red blood cells (PRBCs). The following day, the patient's anemia had not improved, with hemoglobin falling further to 7.2. The second unit of PRBCs was administered to the patient and efforts

were made to obtain biopsy results from his recent outpatient EUS work up. A fine-needle aspiration (FNA) biopsy of the pancreatic mass was consistent with a gastrointestinal tumor; cytopathology report showed spindle cells with fibrillar cytoplasm in a background of rare stromal components and blood and immunohistochemistry of the biopsy tested positive for CD117 and DOG1. The patient was diagnosed with pancreatic eGIST. He underwent further workup to rule out the additional disease with colonoscopy. The colonoscopy showed diverticulosis and a small polyp, which was resected. Before surgical resection of the pancreatic eGIST, the patient will further undergo a capsule endoscopy to rule out the possibility of additional disease in the upper gastrointestinal tract. The treatment plan was to proceed with targeted therapy with Imatinib for the CD117+ pancreatic eGIST.

Discussion and Conclusions

This case illustrates the interesting constellation of findings in a patient with a rare pancreatic eGIST and history of neurofibromas. The findings may mimic symptoms of an upper gastrointestinal bleed (UGI) such as persistent anemia, vague abdominal pain, fatigue, and dark stools. Work up of a pancreatic mass on imaging in a patient presenting with symptoms of a UGI bleed should include the possibility of an eGIST. Definitive diagnosis can be obtained with EUS and FNA biopsy of the suspicious mass. The treatment of choice for gastrointestinal stromal tumors is surgery with complete surgical resection of GIST with the goal of obtaining negative microscopic margins. Targeted antineoplastic agents may be used to shrink the tumor for greater ease of resection. Imatinib (Gleevec) is an inhibitor of receptor tyrosine kinase, which makes it effective in treating GIST tumors caused by activating mutations of the *c-kit* gene.

Authors' contributions

IY gathered the patient's history and physical exam findings, performed the research for the topic, and wrote the manuscript.

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