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Case Report

Synchronous Gastrointestinal Stromal Tumor and Axillary Deep Fibromatosis: A Rare Association

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ABSTRACT

Gastrointestinal Stromal Tumor (GIST) is the most common mesenchymal tumor of the gastrointestinal tract and most commonly affects the stomach, while fibromatosis is a rare locally aggressive fibrous tissue neoplasm. There have been reports of GIST and fibromatosis occurring in same individual and in most of them fibromatosis occurs within the abdomen. In 75% of patients fibromatosis occurs after the diagnosis of GIST while in 20% it is seen synchronously. Here we report a case of axillary fibromatosis with synchronous GIST of the gastroesophageal junction in a 45-year-old male treated with surgery and now on adjuvant imatinib.

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Introduction

Gastrointestinal Stromal Tumor (GIST) arises from the interstitial cell of Cajal and is the most common mesenchymal tumor of the gastrointestinal tract. GISTs that arise in adults are characterized by the near universal expression of CD117 antigen, which is in contrast to other spindle cell tumors of the gastrointestinal tract that are typically CD117 negative. Gain of function mutations of the c-Kit gene which contribute to known molecular changes in the pathogenesis of GISTs, have been reported in about 80% of these patients.[1]

Those who lack c-Kit mutations may have mutations in the PDGFRA or BRAF or inactivating mutations in succinate dehydrogenase (SDH) genes. There are some tumors reportedly associated with GISTs including paraganglioma and pulmonary chondroma (as a part of the Carney triad).[2] There are also reports of coexistence of GIST with carcinoma of the stomach, breast, prostate, and kidney.[3] The coexistence of GIST and fibromatosis is extremely rare with only very few cases reported so far. Fibromatosis frequently occurs after the diagnosis of GIST within the abdomen.[4] Here we report a case of axillary fibromatosis with synchronous GIST detected incidentally in a 45-year-old man.

Case Report

A 45-year-old man was referred for pain and swelling of one month duration in the right axilla. On physical examination, an axillary mass measuring 7×6 cm was noted. MRI of the right shoulder showed a 6.5×6 cm sized mass of muscular plane infiltrating right latissimus dorsi and subscapularis muscles without infiltration to the chest wall (figure 1a). The patient underwent Contrast-enhanced computed tomography (CT) of the chest and abdomen which showed an enhancing soft tissue mass measuring 8.3×5.2×6 cm, involving muscular plane with irregular margins abutting the subscapularis and deltoid muscles along with axillary lymph nodes 23×9 mm in size. The chest scan was negative for lung metastasis. CT scan of the abdomen showed an exophytic mass 10×12×9

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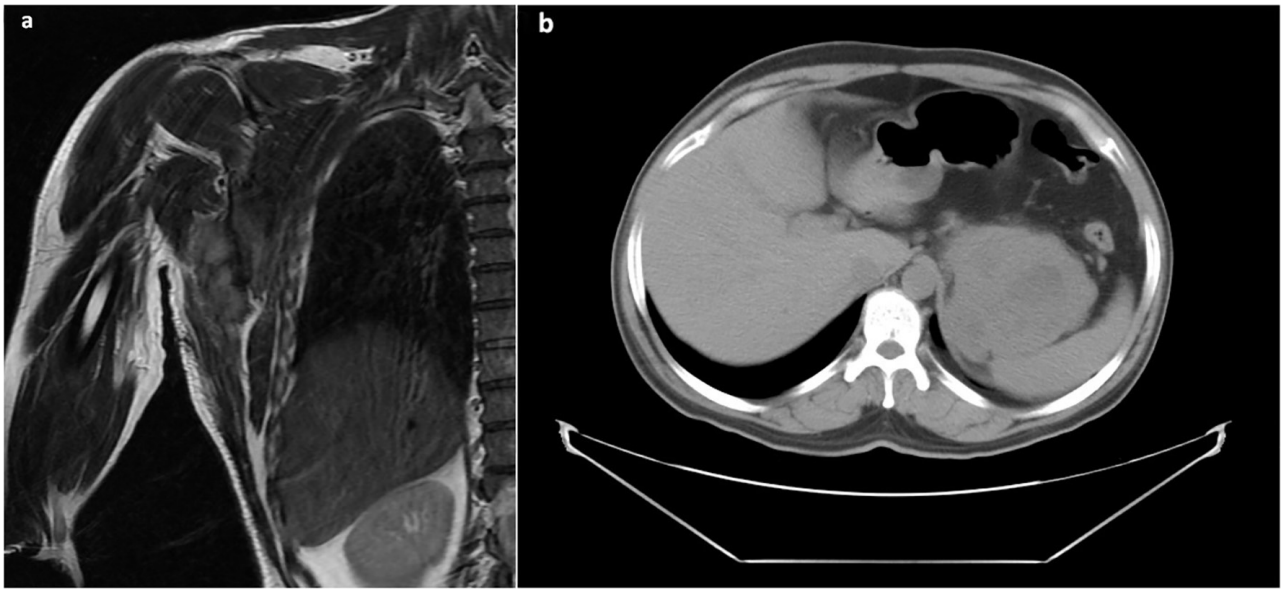


Figure 1: a) MRI of the right shoulder showed a 6.5×6 cm lesion involving muscular plane, infiltrating right latissimus dorsi and subscapularis. b) Computed tomography of the abdomen which showing an exophytic mass with central necrosis of size 8×7.2 cm in the gastro esophageal junction with loss of fat plane with left adrenal and spleen

cm in the gastroesophageal junction indenting the spleen and upper pole of the left kidney. Whole body [18F] fluoro-2-deoxy-D-glucose-positron emission tomography (FDG-PET) scan showed an abdominal mass of 8×7.2 cm with standardized uptake values (SUV) of 48.39 and an ill-defined enhancing mass in the right axilla with SUV of 5.5.

Upper gastrointestinal endoscopy showed edematous folds in the fundus of the stomach with no mucosal ulceration (figure 1b). Esophagus and the rest of the stomach was normal. The biopsy from the exophytic lesion in gastroesophageal junction was suggestive of spindle cell neoplasm. c-Kit and DOG1 staining was performed which were positive, suggestive of GIST showing brisk mitosis. Accordingly, sleeve gastrectomy with distal pancreatectomy and splenectomy was performed. There was no definite invasion of the spleen or pancreas, two perigastric and four peripancreatic lymph nodes were dissected which were not involved. All margins were free of tumor. In addition, the patient underwent lymph node dissection and partial scapulectomy for the axillary lesion where the pathology was suggestive of deep fibromatosis. Adjacent stroma showed foreign body giant cell reaction with deep margin of 0.7 cm. Three axillary lymph nodes were dissected which showed only reactive changes. The patient was planned to receive Imatinib mesylate 400 mg once daily for three years which was tolerated well by the patient. Informed written consent taken from the patient.

Discussion

Deep or aggressive fibromatosis also known as desmoid tumor is a rare tumor, that usually occurs between 20-40 years of age.[4] Desmoid tumors have been associated with familial adenomatosis polyposis as well as Gardner syndrome. The abdomen is the most common site of occurrence of desmoid tumors with highest frequency in surgical incision sites, a common presentation causing a diagnostic dilemma.[5] The WNT/b-catenin signalling is a significant pathway for development of desmoid tumors. Various treatment options for desmoid tumors (deep fibromatosis) include watchful waiting or surgical resection accompanied with systemic treatments such as tamoxifen, non steroidal anti inflammatory drugs or Imatinib mesylate which have been suggested mainly in inoperable cases.[6]

The simultaneous occurrence of fibromatosis with GIST is extremely rare. In about 75% of the patients, fibromatosis occurs after diagnosis of the GIST.[7] The age at presentation in patients with concurrent desmoid tumor and GIST is reported to be higher with a mean of 59 years. There are also reports of desmoid tumors developing at the excision site of the GISTs, making the diagnosis often more difficult.[8] Extra abdominal desmoid tumor, as in our patient coexistent with GIST is an extremely rare association.

The most common site of origin of GIST is in the stomach followed by the small intestine.[9] They are the most common mesenchymal tumors of the GI tract with median age of 60 years at diagnosis.[10] The treatment modality of choice is surgery followed by imatinib mesylate as neoadjuvant or adjuvant in inoperable tumors with sensitive mutations.[11] One of the proposed mechanisms for development of GIST and deep fibromatosis is activating mutations of Kit pathway; however, the largest series reported by Dumont et al. failed to show such mutations in any of the 28 patients who had GIST and fibromatosis.[7] Cross talk between the Kit and WNT signaling pathways have been suggested, but needs further studies.[12]

The synchronous occurrence of these two tumors is even much rarer when compared to metachronous appearance. Meanwhile, the occurrence of extra-abdominal fibromatosis concurrent with abdominal GIST is extremely rare with no similar cases reported so far. The treatment remains surgery for both primaries if operable. Preoperative imatinib may be an effective

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option for both tumors, but surgery remains the only potentially curative option and must be attempted in all patients.[13] Desmoid tumor must be kept in mind when GIST is diagnosed in a patient with atypical soft tissue masses either synchronously or at a later period in other parts of the body.

Conflict of Interest: None declared.

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