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A Rare Association Between Gastrointestinal Stromal Tumors (GISTs) of the Small Intestine and Recklinghausen's Disease or Neurofibromatosis Type 1 (NF1): A Case Report

Aghali Ibrahim[1*], K. Camara[1], Salah Ben El Hend[1], Badr Slioui[1], Salah Bellasri[1], Redouane Roukhsi[1], Nabil Hammoune[1], Abdelilah Mouhsine[1]

[1]Radiology Department, Avicenne Military Training Hospital, Marrakech, Morocco

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***Corresponding author:** Aghali Ibrahim
Radiology Department, Avicenne Military Training Hospital, Marrakech, Morocco

Abstract **Case Report**

Neurofibromatosis type 1 (NF1) is a common neurocutaneous syndrome that predisposes to a wide variety of tumors. These tumors can be benign or malignant, such as gastrointestinal stromal tumors (GIST). However, the association of small bowel GIST and Recklinghausen's disease or neurofibromatosis type 1 (NF1) is rare. We report the case of a patient who underwent surgery for GIST.

Keywords: GIST, Recklinghausen's disease; neurofibromatosis type 1.

BACKGROUND

GISTs are malignant mesenchymal tumors that develop in the wall of the digestive tract. They most often occur in the stomach and small intestine, but localization in the rectum, colon, and esophagus is rarer [1]. They belong to the nosological entity of sarcomas and represent 18%, the most common form of sarcoma [2]. Genetically, neurofibromatosis type 1 (NF1), or Recklinghausen disease, is an autosomal dominant inherited disease characterized by multiple pigmented skin patches (café-au-lait spots) and neurofibroma [3]. Clinically, neurofibromatosis type 1 (NF1) is a common neurocutaneous syndrome, often associated with various tumors such as gastrointestinal stromal tumors [4,5]. These tumors can be benign or malignant and their search should be systematic when there are symptoms.

Individual cases should be presented and discussed in a multidisciplinary consultation meeting with a multidisciplinary team of specialists including a dermatologist, surgical oncologist, medical oncologist, pathologist, radiologist, gastroenterologist, and nuclear medicine specialists. The standard surgical treatment of GIST requires R0 resection (negative margins) when possible. Depending on the severity of GIST, preoperative treatment is necessary, especially for high- and intermediate-risk GISTs that require adjuvant treatments such as radiotherapy or chemotherapy [6]. In cases of metastatic disease, targeted therapies are available, but surgery remains an indication in some cases.

We report the case of a patient with NF1 who received surgical treatment for gastrointestinal stromal tumors (GISTs) of the small intestine.

CASE PRESENTATION

A 38-year-old patient with NF1 was referred to our hospital for an occlusive syndrome that had been evolving for three (03) days. A few days before this symptomatology, a diagnosis of Recklinghausen's disease (NF1) was made based on the histology of a cutaneous wart excised for multiple pigmented spots and a neurofibroma. The diagnostic hypothesis of an intestinal obstruction on NF1 was made and we carried out biological and radiological examinations which revealed a 40 mm diameter submucosal tumor of the small intestine, raising suspicion of a GIST.

A midline laparotomy for local excision of a small bowel gastrointestinal stromal tumor (GIST) was successfully performed with an R0 resection. Histology revealed that the small bowel tumor was an intermediate-risk GIST (positive immunostaining for KIT and CD34 microscopically).

After discharge, the patient was referred to the medical oncology department to continue treatment with possible adjuvant therapy.

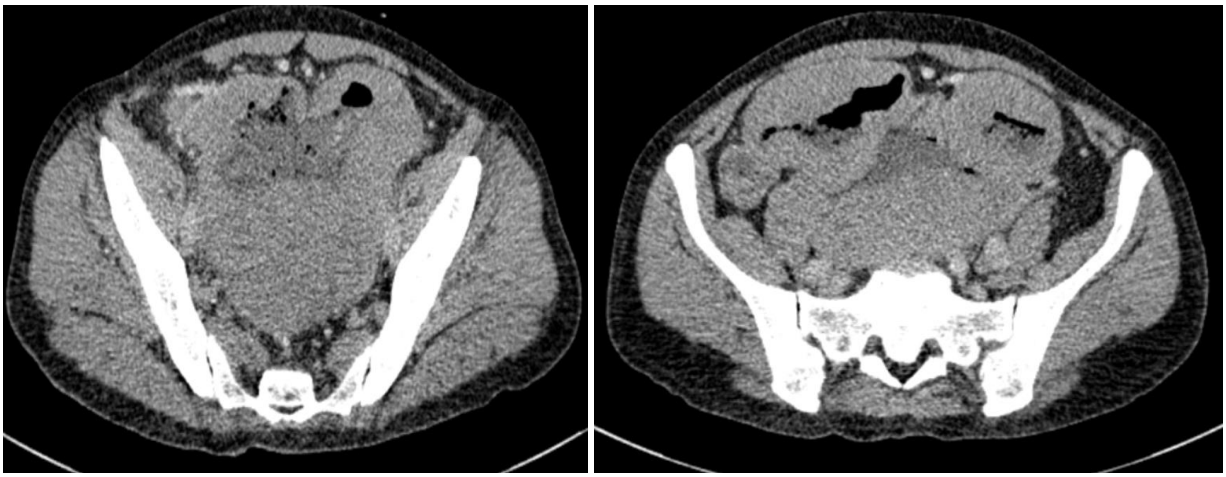

Figure 1 & 2: Axial CT scan: Small intestine GIST

DISCUSSION

We reported the case of a 38-year-old man with an association between small bowel GIST and Recklinghausen's disease (NF1). The incidence of GIST is estimated at approximately 15 cases/million inhabitants/year, or nearly 1,000 new cases per year in France [7,8]. Similarly, Martinbroto found an incidence of 1.1 cases/100,000 inhabitants/year [9]. Regarding age, GISTs develop mainly in the elderly, and the median age of diagnosis is 60-65 years [10]. In 2014, Karim Ibn Majdoud in Morocco reported the case of a 33-year-old man with small bowel GIST after NF1 lesions [11]. NF1 predisposes people to a wide variety of tumor types. These tumors can be benign or malignant such as gastrointestinal stromal tumors (GIST) with which it constitutes a syndromic entity [3,7,8,12]. In the typical picture of NF1-GIST association, GIST lesions are very often multiple, predominant in the small intestine, small in size and with a good prognosis [13]. Similarly, in our case, for ROSENBAUM, pilocytic lesions were found. Astrocytomas, gastrointestinal stromal tumors (GIST), pheochromocytomas and juvenile myelomonocytic leukemia are tumors frequently observed in patients with NF1 [4]. This is explained by genetic abnormalities of the NF1 gene, important in the development of tumors [4].

In our case, we performed local excision of the GIST as for Liand in Japan and Karim Ibn Majdoud in Morocco [5, 11]. The standard surgical treatment of GIST requires R0 resection (negative margins) when possible [10]. For Jean-Yves, localized GISTs are curable and surgery is their standard treatment [14].

After surgery, the patient was referred to oncology for possible adjuvant treatment. According to the recommendations of the American National Comprehensive Cancer Network (NCCN) in its clinical practice guidelines in oncology, high- and intermediate-risk GISTs require adjuvant treatment [6].

CONCLUSION

GISTs have been occasionally reported in the gastrointestinal tract of patients with syndromic NF1. Therefore, these patients should receive regular medical monitoring to detect these lesions early and act quickly to avoid complications and hope for a complete cure. This observation reports a rare case of association between Recklinghausen's disease (NF1) and a small bowel GIST in a 38-year-old young adult.

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