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Gastrointestinal Stromal Tumors (GIST) of the Small Intestine in A Patient with Recklinghausen Disease Or Neurofibromatosis Type 1 (NF1): A Case Report

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Abstract:
Neurofibromatosis type 1 (NF1) is a frequent neurocutaneous syndrome that predisposes for various benign and malignant tumorssuch as gastrointestinal stromal tumors (GISTs). We report a NF1 patient who received surgical treatment for small intestine gastrointestinal stromal tumors (GISTs).

Keywords: GISTs, Recklinghausen Disease; Neurofibromatosis type 1

Background:
GISTs are malignant mesenchymal tumors that develop in the wall of the digestive tract, most often in the stomach and hail, more rarely in the rectum, colon, esophagus [1]. They belong to the nosological entity of sarcomas and represent 18%, the most common form of sarcoma [2]. Neurofibromatosis type 1 (NF1) or Recklinghausen disease, is an autosomal dominant inherited disorder characterized by multiple pigmented skin spots (café-au-lait spots) and neurofibroma[3].Neurofibromatosis type 1 (NF1) is a frequent neurocutaneous syndrome that predisposes for various benign and malignant tumorssuch as gastrointestinal stromal tumors [4,5].
The treatment strategy should be defined by a multidisciplinary team experienced in soft tissue sarcomas, comprising an oncological surgeon, medical oncologist, pathologist, radiologist, gastroenterologist, and nuclear medicine specialists. Surgical treatment with R0 resection (negative margins), if possible, remains the mainstay of GIST management.In some cases, preoperative therapy may be introduced. High- and intermediate-risk GISTs require adjuvant therapy [6]. For metastatic disease, targeted therapies are available, but surgery may also be used in some cases.
We report a NF1 patient who received surgical treatment for small intestine gastrointestinal stromal tumors (GISTs).

Case presentation:
A 38-year-old male patient with NF1 (figure 1) was referred to our hospital for occlusive syndrome made of cessation of materials and gases, abdominal pain in the umbilical region, vomiting and

abdominal distension lasting for 72 hours. In fact, some days before this symptomatology,he was diagnosed Recklinghausen disease (NF1)according to the histology of an excised skin wart done for multiple pigmented skin spots and neurofibroma.The diagnostic hypothesis of an intestinal occlusion on NF1 was posed and we carried out biological and radiological examinations.

A submucosal tumor 40 mm in diameter was detected in the small intestine by the abdominal scanner.

Midline Laparotomy for local excision of the small intestine GIST were performed successfully. The small intestine tumor was found to be intermediate-risk GIST by the histology(positive immunostaining of KIT and CD34 microscopically).

After his discharge, the patient was referred to the oncology department for continued treatment with an adjuvant therapy.

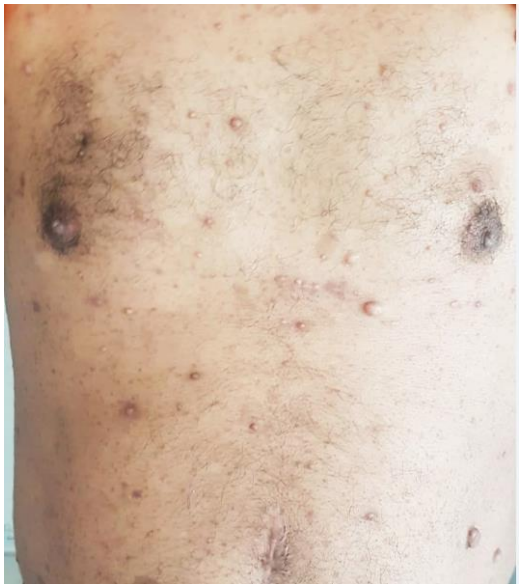

Figure 1: skin lesion of NF1

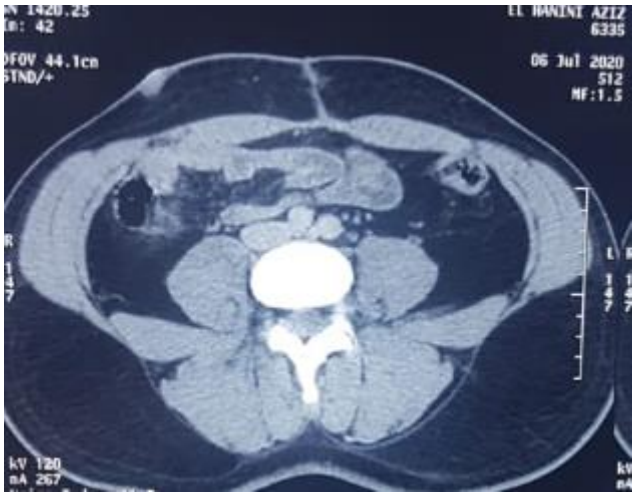

Figure 2: tumor of the small intestine

Discussion:

We reported the case of a 38 years old man with an association between small intestine GIST and Recklinghausen disease (NF1). The incidence of GISTs is estimated at about 15 cases/million

inhabitants/year or nearly 1000 new cases per year in France [7,8]. For Martin-broto found an incidence of 1.1 cases/100,000 inhabitants/year [9].GISTs mainly develop in older adults, and the median age of diagnosis is 60–65 years [10]. In 2014, Karim Ibn Majdoud in Morocco, reported a case of 33 years old man with small intestine GIST after NF1 lesions [11]. NF1 is associated with a wide variety of benign or malignant tumors including the GISTs with which it constitutes a syndromic entity [3,7,8,12]. In this case, GISTs are very often multiple, predominant in the small intestine, small in size and good prognosis; typical table of associated NF1 [13]. For ROSENBAUM, some neoplasiasfrequently observed in NF1 patients are pilocytic astrocytomas, gastrointestinal stromal tumors, pheochromocytomas and juvenile myelomonocytic leukemia [4]. In fact,the development of NF1-associated tumors is largely explained by the underlying defect of the NF1 gene which results in activation of the RAS proto-oncogene- a key mechanism of tumorigenesis [4].

In our case, we did the surgery for local excision of the GIST as well as for Liand in Japon and Karim Ibn Majdoudin Morocco [5, 11]. Surgical treatment with R0 resection (negative margins), if possible, remains the mainstay of GIST management [10].For Jean-Yves, localized GISTs are curable, and surgery is their standard treatment [14].

After surgery, the patient was sent to oncology for adjuvant therapy. In fact, for the American National Comprehensive Cancer Network (NCCN) Clinical Practice Guidelines in Oncology, High- and intermediate-risk GISTs require adjuvant therapy [6].

Conclusion:

GISTs have been reported to occasionally occur in the digestive tract of the patients with NF1. Therefore, these patients should undergo regular physical examinations so that early detection and early treatment can be achieved.We present a rare case of a Recklinghausen disease (NF1) patient with GIST of small intestine.

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