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Case Report **Surgical Emergency**

Small Bowel Gist in a Patient with Neurofibromatosis Type 1: Revealed by Acute Abdominal Surgery: A Case Report

Dr. Maouni Iliass[1*], M. Gridda[1], Y. Ouhamou[1], M. Elabsi[1]

[1]Surgical Emergency Department, Ibn Sina University Hospital, Rabat, Morocco

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***Corresponding author:** Dr. Maouni Iliass
Surgical Emergency Department, Ibn Sina University Hospital, Rabat, Morocco

Abstract

We decided to report a case of gastrointestinal stromal tumor of the small intestine in a patient with neurofibromatosis type 1 because of the increase of its incidence as shown in the literature, also, by its revelation; its occurrence in a picture of acute peritonitis due to the rupture of the tumor. These tumors, commonly called by their English acronym GIST (Gastro-Intestinal Stromal Tumors), found in people with neurofibromatosis type 1 generally occur in the small intestine and are often multiple.

Keywords: GIST, small intestine, neurofibromatosis, peritonitis, tumor.

INTRODUCTION

Gastrointestinal stromal tumors (GISTs) occur mainly sporadically and represent about 1% of all neoplasia of the gastrointestinal tract. The origin of GISTs has a direct relationship with the interstitial cells of Cajal, which are part of the myenteric plexus of the digestive tract and are responsible for the control of intestinal motility (pacemaker). They can develop from all segments of the digestive tract, from the esophagus to the anus, or exceptionally the mesentery and peritoneum. They may project exophytically into the lumen or less frequently they dilate through the serosa of the organ [1]. The incidence is estimated at about 15 cases / million inhabitants/ year, the median age at diagnosis is about 60 years, and the sex ratio of about 1/1 [2]. Most mesenchymal tumors in the digestive tract have been considered as smooth muscle tumors (leiomyomas, leiomyosarcomas, etc. differential diagnosis) [3]. In 1983, Clark and Mazur introduced the term gastrointestinal stromal tumor (GIST) to describe a distinctive type of non-muscle smooth mesenchymal tumor [4]. Extensive study has shown that GISTs have a specific etiogenesis unrelated to the gastrointestinal smooth muscle at the expense of which, it is true, they develop. The most frequent clinical presentation of GIST is a digestive hemorrhage > 50% of the cases. This may be chronic with anemia or massive, requiring emergency treatment (40% of cases of hemorrhagic GIST). The majority of cases are localized, and less than 10% of patients present with metastatic disease [3]. We report a case of GIST on a neurofibromatosis background revealed late in an acute peritonitis picture.

MEDICAL OBSERVATION

The patient was 49 years old and single, with a pathological history of lower limb deformity and inability to walk since childhood, requiring the use of a wheelchair. She was referred to us with acute peritonitis. The interrogation with the patient on admission found a beginning of the symptomatology that goes back to 72h with epigastric abdominal pain that quickly generalized to the whole abdomen with vomiting. The clinical examination found a conscious patient, febrile at 39.5°, tachycardia at 112 beats/min, blood pressure at 110/56mmhg, and polygenic at 26 cycles/min, the abdominal examination found a generalized contracture the rest of the examination was characterized by the presence of skin lesions characteristic of Von Recklinghausen's disease ("Café au lait" spots) (Fig. 1). The biological workup showed a hyperleukocytosis of 27500 with 88% neutrophilic polynuclear, a hemoglobin level of 14.2 g/dl, a C-reactive protein of 361 mg/l, the hydroelectrolytic workup showed profound hypokalemia of 2.1 mEq/l, and a correct renal function. An abdomino-pelvic ultrasound was performed at the beginning showed a pelvic peritoneal effusion associated with an echogenic collection and containing air bubbles measuring 52mm

X 74 mm, then it was completed by abdomino-pelvic CT scan with contrast injection which objectified a peri-hepatic peritoneal effusion of medium abundance, peri-splenic and pelvic, and individualization of a pelvic mass of about 7 cm of great axis around which are agglutinated small intestines and the left colon with pneumoperitoneum bullae (Fig. 2); A standard X-ray of the pelvis showed a flattened and destroyed appearance of two femoral heads (Fig. 3). This fact confirms the bone involvement of his basic disease (neurofibromatosis type 1).

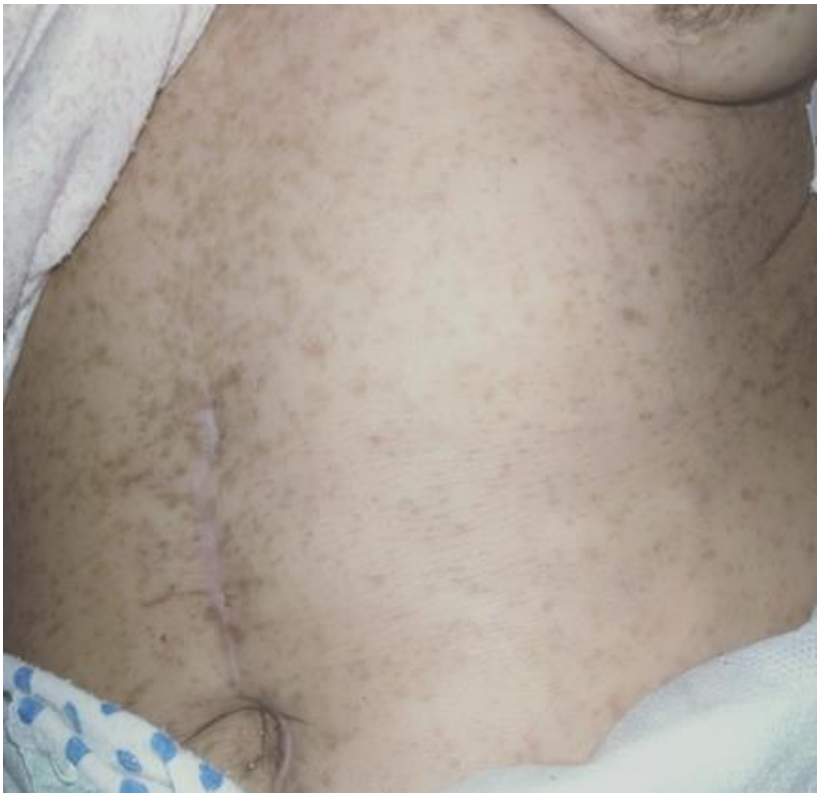


Figure 1: Café au lait skin stains

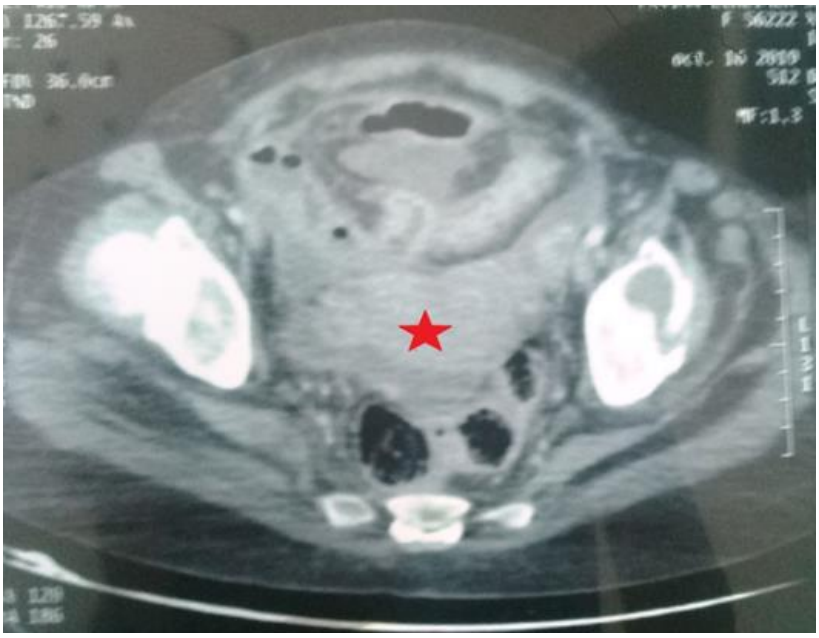


Figure 2: Axial section of abdominal CT scan showing a mass in contact with the sigmoid colon

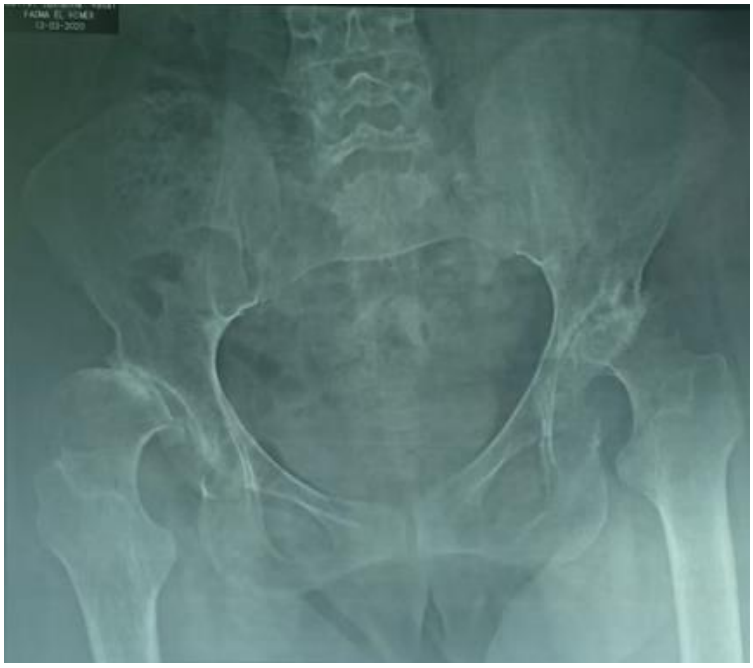
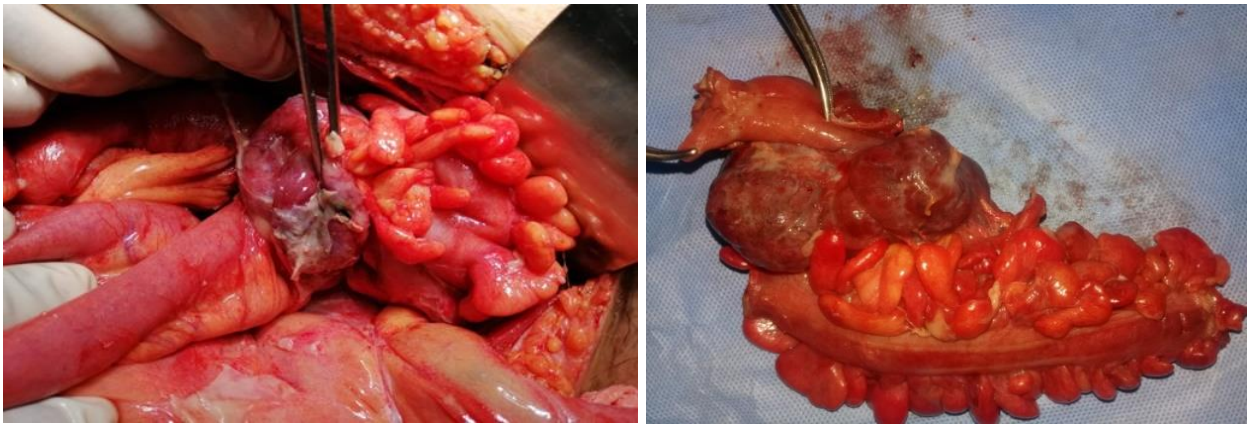


Figure 3: Bilateral femoral head necrosis

Faced with this clinical and radiological picture of peritonitis, the surgical indication was decided after a short resuscitation and especially a potassium recharge by central route. The surgical exploration revealed a generalized peritonitis, neglected with a purulent effusion and false membranes, after laborious liberation of the bowel, a mass was discovered originating at the level of the jejunum, 20 cm from the duodeno-jejunal angle, about 6 cm long and invading the sigmoid colon (fig. 4).



A B

Figure 4: An Intraoperative image showing a perforated mass taking the jejunum and sigmoid colon, B: Surgical specimen made of the sigmoid colon, jejunum and the tumor mass

This mass was perforated and made the two digestive segments communicate with the peritoneal cavity; we proceeded to a resection of the mass with a segment of jejunum and the sigmoid colon at a distance from the tumor. The continuity of the jejunum was re-established by a terminal anastomosis despite the risk of septicemia since the resection was made in the first loop, while the two ends of the left colon were abutted in a double stoma to the left according to Bouilly Volkmann. The postoperative course was simple with the patient being discharged at D+5. The anatomopathological and immunohistochemical study of the lesion showed that it was a tumor of size 6x5x5x4 cm, with spindle-shaped cells with eosinophilic cytoplasm and elongated or ovoid nuclei. The mitotic index was 1/25. These cells expressed CD117 and DOG1 positive (Fig. 5), concluding a gastrointestinal stromal tumor (GIST) at moderate risk of recurrence, for which adjuvant chemotherapy was decided at the multidisciplinary consultation meeting with imatinib 400 mg per day for 4 years. We subsequently referred the patient to a dermatologist for management of her cutaneous Von Recklinghausen's disease and proposed annual surveillance by CT enterography.

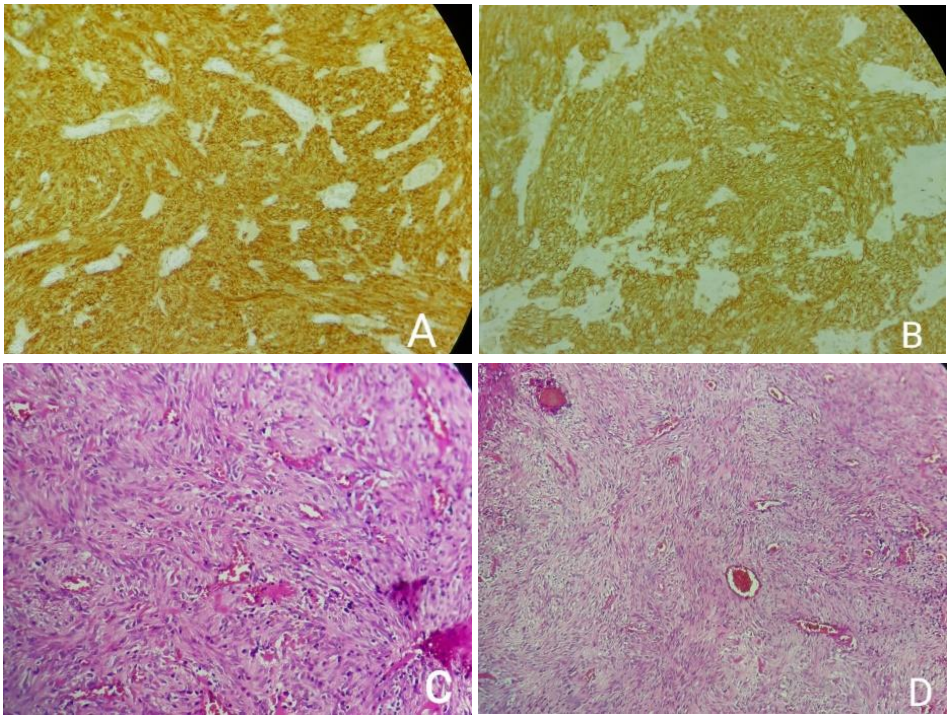


Figure 5: A,B): IHC study showing positive labelling by anti CD117 antibody (C,D): Spindle cell proliferations occupying the entire grape wall

DISCUSSION

GISTs are connective tissue tumors characterized by the expression of the CD117 marker (kit protein or c-kit). Only c-kit positive tumors are considered as GIST except in exceptional cases [2]. They are usually located in the stomach in 40% to 70%, in the small intestine in 20% to 40% and less than 10% in the esophagus, colon and rectum [5, 6]. Most GISTs are sporadic but there are a few cases of familial disease. GISTs can be seen in the context of Carney's triad or neurofibromatosis type 1. Our case had a strong suspicion of neurofibromatosis type 1, because of the very characteristic skin lesions and the bone defect in the pelvis. Recklinghausen's disease is an autosomal dominant inherited disease that involves an abnormality on chromosome 17. It is quite frequent with an incidence of 1/3500 births [7]. It progresses slowly and is characterized by the progressive appearance of pigment spots, skin tumors, tumors of the peripheral nerves (neurofibromas) or of the central nervous system (gliomas) and skeletal malformations. It may be accompanied by digestive manifestations such as stromal tumors, lesions of the intrinsic digestive nervous system or endocrine tumors of the duodenum [7]. These stromal tumors developed in the context of neurofibromatosis type 1 do not have mutations in the KIT and PDGFRA genes. They have no morphological features but are often multiple [8]. These tumors occur in 5% of patients with Von Recklinghausen disease [7]. The diagnostic circumstances of stromal GIST tumors are variable: incidental discovery, pain, mass syndrome, anemia, hemoperitoneum, and especially digestive hemorrhage which is the most frequent symptom [9]. Our patient presented with a picture of peritonitis, it is a rare mode of discovery, seven patients (7.6%) in the series of Magdy A *et al.*, had presented with rupture and peritonitis [10]. The stromal GIST tumor found in our patient is identical with the cases already reported in the course of Von Recklinghausen's disease: late discovery, after 49 years of age, in patients presenting the characteristic cutaneous signs of the disease, localization in the small intestine: this is the preferential site of occurrence of GIST associated with Von Recklinghausen's disease, followed by the stomach. Complete surgical resection is the only potentially curative treatment for GISTs [2]. Medical treatment is with imatinib, a pharmacological c-kit antagonist that inhibits tyrosine kinase function, which is recommended for advanced GIST, whether unresectable, metastatic or relapsed. Current data show that imatinib induces a 60-70% objective response rate, with 15-20% stable disease and 10-15% primary resistance. Secondary resistance (escape) is now reported in 10-30% of cases [2].

CONCLUSION

The benign or malignant character of these tumors is difficult to define; several prognostic factors have been proposed, but there are sometimes "borderline" tumors.

The only potentially curative treatment is the complete surgical removal of the lesion, but the use of Imatinib in recent years, a drug treatment that inhibits the KIT protein, has revolutionized their management.

Conflict of Interest: None.

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