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Association of right breast cancer and ileal gastrointestinal stromal tumor in a patient with type I neurofibromatosis: Case report and review of the literature

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ABSTRACT

INTRODUCTION: The Neurofibromatosis type I (NF1) is an autosomal dominant syndrome that affects 1/3000–1/4000 individuals. Patients with this condition are predisposed to different tumors, like neurofibromas, optic nerve gliomas, gastrointestinal stromal tumors (GIST) and breast cancers.
PRESENTATION OF CASE: A 78-year-old female patient affected by NF1 in May 2018 during follow-up for a carcinoma of the right breast had persistent anemia requiring regular blood transfusions. She presented with NF 1 with disseminated cutaneous neurofibromas, asthma, hypothyroidism, arterial hypertension and uterine prolapse.

She had performed gastroscopy and colonoscopy both negative for neoplastic lesions. She was subjected to chest and abdomen CT which revealed the presence of an ileal lesion of 6.5 × 4 cm suspected of GIST.

The patient underwent laparoscopic ileal resection in 120 min and was discharged on the sixth postoperative day.
DISCUSSION: NF1 is caused by biallelic loss of a tumor suppressor gene. Most GISTs are localized in the stomach and small intestine. Surgery is the first line of treatment for localized disease. The main goal of surgery is complete excision with negative margins. The association between breast cancer and intestinal GIST in NF1 is reported only from two previous studies.
CONCLUSION: It is a rare case of association of breast cancer and ileal GIST in NF1. Laparoscopic resection of intestinal GIST has shown in some studies to have oncological outcomes comparable to laparotomy. Furthermore, laparoscopy is associated with better perioperative outcomes and shorter hospital stays. Further studies with a higher level of evidence are needed.

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1. Introduction

NF1 is an autosomal dominant syndrome that affects 1/3000–1/4000 individuals [1]. It is clinically diagnosed with the diagnostic criteria of the National Institute of Health [2] as following:

- 6 or more café au lait macules (> 0.5 cm in children or > 1.5 cm in adults);
- 2 or more cutaneous/subcutaneous neurofibromas or one plexiform neurofibroma.
- Axillary or groin freckling.
- Optic pathway glioma.
- 2 or more Lisch nodules.
- Bony dysplasia (sphenoid wing dysplasia, bowing of long bone not pseudarthrosis);
- First degree relative with NF1.

Patients with NF1 are predisposed to both benign and malignant tumors, including neurofibroma, malignant peripheral nerve sheath tumors, neuroblastomas, gastrointestinal stromal tumor (GIST) and breast cancer [1].

In this case report we describe a patient NF1 with a rare association of breast cancer and ileal GIST subjected to laparoscopic

Abbreviations: NF1, neurofibromatosis type I; GIST, gastrointestinal stromal tumors; CT, computed tomography; HPF, high power field; ER, oestrogen receptor; PgR, progesterone receptor.

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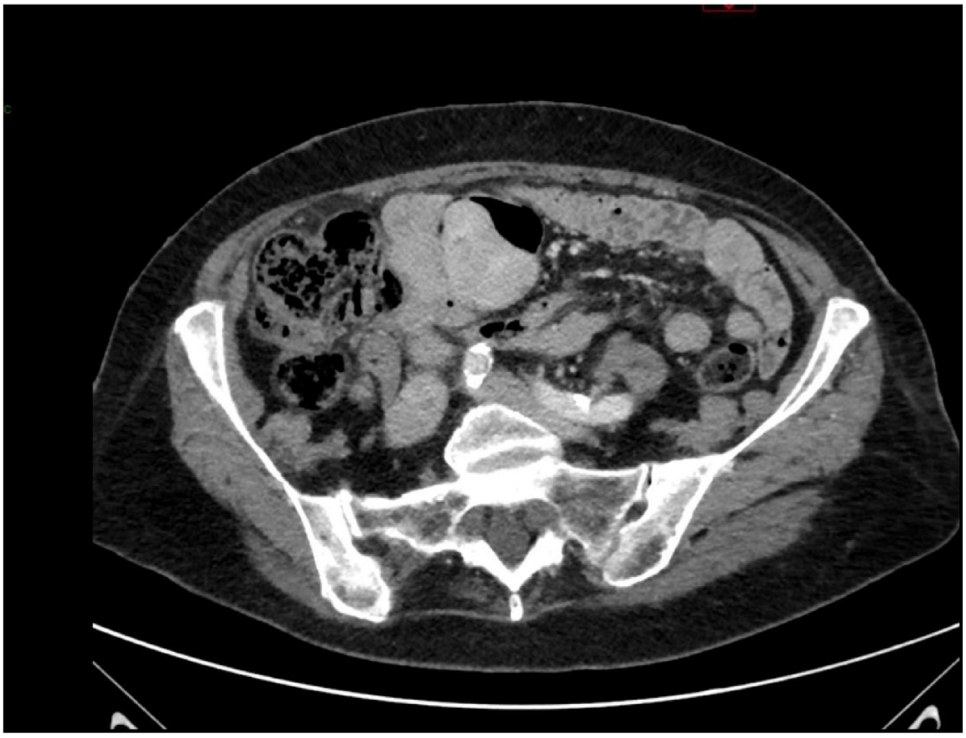

Fig. 1. CT with contrast enhancement which the presence of an ileal loop in the meso-hypogastric site with a lesion with a predominantly endoluminal development of 6.5 × 4 cm with colliquate areas inside, highly suspected for GIST.

resection. This paper has been reported in line with the SCARE criteria [3].

2. Presentation of case

A 78-year-old female patient affected by NF1 in May 2018 during follow-up at the breast clinic treated six years previously for right breast cancer had persistent anemia (hemoglobin 6.5 g/dl) requiring regular blood transfusions. In September 2012 she was subjected to right tumorectomy with axillary dissection. The histological examination was mixed type infiltrating carcinoma of the breast pT2pN1a G2 50 mm in diameter. The margins were negative. ER 98%, PgR 95%, Ki67 11%, C-erb negative. Exclusive hormone therapy with Anastrozole for 5 years.

Gastroscopy and colonoscopy were performed as well as endoscopic video capsule all negative for neoplastic lesions. The chest and abdomen CT revealed a lesion of 6.5 × 4 cm with colliquate areas inside in an ileal loop in the meso-hypogastric site, highly suspicious of GIST (Fig. 1).

After CO2 insufflation of the abdomen to 12 mmHg, ports were positioned in supra-umbilical site for the camera: other three work ports (5 mm of diameter) in the right side, left side, and hypogastrium. We discovered the presence of an ileal mass of about 7 cm of diameter positioned in the middle ileal location. We performed the resection of the ileal meso, through Thunderbeat (Olympus Deutschland, GmbH), preparing the loops of the small bowel. A Mc Burney's incision was made and the neoformation was extracted. An anisoperistaltic laterolateral anastomosis was performed using 60 mm GIA. Laparoscopic ileal resection was completed in 120 min.

The patient was discharged on the sixth postoperative day. Final histological examination revealed GIST at high risk (dimensions greater than 5 cm and mitosis greater than 5 × 10 HPF) according to Joensuu. Surgical resection margins were negative.

Regular oncological follow-up without breast or abdominal recurrences until March 2020.

3. Discussion

GISTs are the most common mesenchymal tumors of the gastrointestinal tract [4]. Most of them are in the stomach (50–70%) and small intestine (35%) [5]. Surgery is the only potentially curative therapy for patients with resectable GISTs. Complete resection of the tumor without lymphadenectomy is recommended [6].

The association between breast cancer and intestinal GIST in NF1 is extremely rare, reported only from two previous studies [7,8]. Invernizzi [7] described a 60-year-old woman affected by NF1 who was coincidentally diagnosed with a gastrointestinal stromal tumour, a breast carcinoma and a peripheral nervous system tumour.

Takeuchi [8] was remarkably similar to our case because reported a case of a 76-year-old woman with a history of NF-1 who right modified mastectomy for lobular carcinoma and laparoscopic tumour resection combined with small intestine surgery for GIST.

The incidence of GIST in NF1 patients is between 3.9 and 25%. The characteristics and genesis of tumors in GISTs in NF1 is different from that of sporadic GISTs.

In GISTs NF1: main site (small intestine), multiple lesions in many cases, better prognosis for the same size and stage, no c-Kit mutation but loss of heterozygosity of NF1.

In Sporadic GISTs: mainly single site lesions (stomach), worse prognosis, c-Kit mutation [7,8].

A lot of clinical studies and meta-analysis have demonstrated that the laparoscopic resection for GISTs of the stomach had associated with lesser pain, shorter hospital stay, faster postoperative recovery, and similar recurrence rates compared to open approach [9,10]. However there is a scarcity of studies on the outcomes of laparoscopic small bowel GIST resections. Currently there is only one meta-analysis that compares laparoscopic versus open approach in GIST of the small bowel and demonstrated 3-years disease free survival and minor complications in laparoscopic group [11].

Another study of 5096 patients [12] revealed no significant differences in 90-day mortality (adjusted for confounding factors) between laparoscopic and open resection.

These results support that laparoscopy may be a more suitable and more effective approach than the OPEN technique in line with our case report with breast cancer and ileal GIST treated with laparoscopic approach.

4. Conclusion

It is a rare case of association of breast cancer and ileal GIST in NF1(third case report in literature). Laparoscopic resection of intestinal GIST has shown in some studies to have oncological outcomes comparable to laparotomy. Furthermore, laparoscopy is associated with better perioperative outcomes and shorter hospital stay. Further studies with a higher level of evidence are needed.

Declaration of Competing Interest

None.

Funding

None.

Ethical approval

None.

Consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Author's contribution

Dario Bono: Conceptualization, Methodology, Software, Data curation, Writing - Original draft preparation, *Alberto Musso*, *Manuela Scamuzzi*, *Francesco Tomaselli*, *Rinaldo Caponi and Roberto Saracco*: Writing - Review and editing, Supervision and Project administration.

Registration of research studies

1. Name of the registry: Researchregistry.com.
2. Unique identifying number or registration ID: 6094.
3. Hyperlink to your specific registration (must be publicly accessible and will be checked): https://www.researchregistry.com/browse-the-registry#home/registrationdetails/5f7f4cdada4b8200167b368b/.

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