

Mesenteric cyst and recurrent abdominal pain in a patient with Gorlin-Goltz syndrome: a case report

L. MONACO, F. GUIDA, G. D'ONOFRIO, N. DI MARTINO

SUMMARY: Mesenteric cyst and recurrent abdominal pain in a patient with Gorlin-Goltz syndrome: a case report.

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Gorlin-Goltz syndrome (GGS) is an infrequent autosomal do-

minant multisystemic disease with complete penetrance and variable expressivity. It is estimated to have an incidence of 1:50,000 - 1:150,000 cases with a M/F = 1:1. This report describes a case of recurrent abdominal pain due to a large mesenteric cyst in a 38-year-old female patient affected by a rare disease: Gorlin-Goltz syndrome.

KEY WORDS: Abdominal pain - Mesenteric cyst - Gorlin-Goltz.

Introduction

Gorlin-Goltz syndrome (GGS) is an infrequent autosomal dominant multisystemic disease with complete penetrance and variable expressivity.

The pathogenesis of the syndrome is attributed to abnormalities in the chromosome 9 (q22.3-q31) and loss or mutations of human patched gene (PTCH1 gene).

This syndrome has been termed with several names such as, basal cell nevus syndrome, GGS, nevoid basal cell carcinoma syndrome (NBCCS), multiple basal cell carcinoma (BCC) syndrome and it is estimated to have an incidence of 1:50,000 - 1:150,000 cases with a M/F = 1:1 (1-3).

This article describes a case of recurrent abdominal pain due to a large mesenteric cyst in a 38-year-old female patient affected by a rare disease: Gorlin-Goltz syndrome.

Case report

A 38-year-old female patient was referred to our surgical department for the presence of recurrent and inguavescent abdominal pain in the last month.

Her medical history was positive for the presence of Gorlin-Goltz syndrome diagnosed ten years before. Clinical examination revealed a typical appearance with frontal bossing, prognatism and multiple skin incision for the removal of BCC.

Clinical examination of the abdomen revealed a mass in the right hypocondrium that was of elastic consistence and not painful. In addition, the mass moved with the variation of the patient's position.

Laboratory testings were unremarkable and also beta-HCG were negative. An eco- and CT- scan of the abdomen were performed.

Abdomen CT-scan revealed the presence of a 12-cm cystic lesion of the proximal mesentery with fluid content, very close posteriorly to the right renal hilum and inferiorly to the aorto-iliac carrefour (Figure 1).

A laparoscopic approach was performed with 3 - trocars technique: two 10-mm trocars (intraumbilical and epigastric) and a 5-mm trocar in the right flank. The exploration of abdominal cavity showed the lesion strictly adherent to mesentery and not dissociable from the proximal ileum.

General and Laparoscopic Unit, Villa Esther - Pineta Grande Hospital, Avellino, Italy

Corresponding author: Francesco Guida, e-mail: francescoguida83@alice.it

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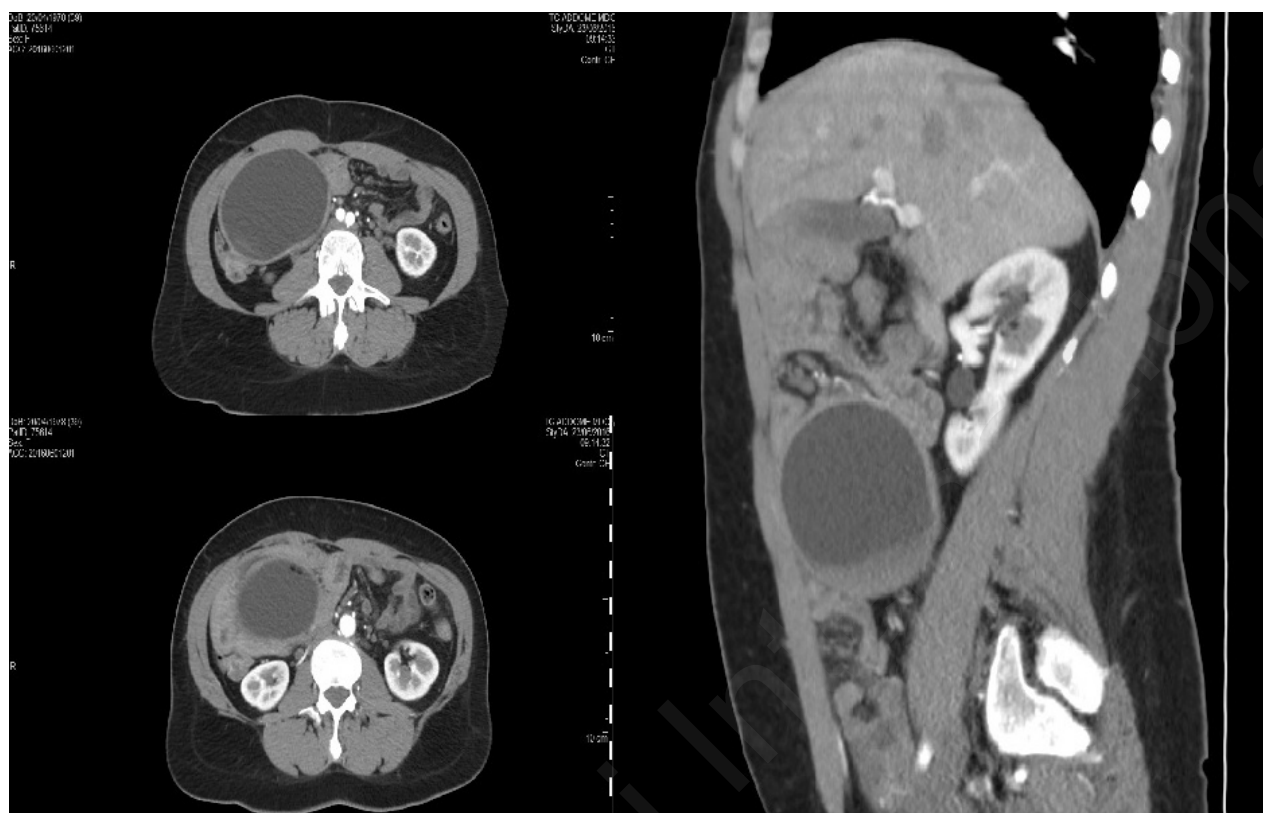


Figure 1 - Abdomen CT-scan showing a cystic lesion of the proximal mesentery very close to the right renal hilum and inferiorly to the aorto-iliac carrefour.

Also, due the great size of the formation, the laparoscopic mobilization was not possible. For these reasons, a supraumbilical incision was made and a laparotomic en-bloc resection of the cyst and of the ileum was performed. Digestive continuity was restored with an end-to-end ileal anastomosis with absorbable sutures. A drain was placed near the anastomosis. Post-operative course was uneventful and bowel function restarted in 3rd postoperative day. Oral intake was administered in 6th postoperative day and the day after the drain was removed and the patient was discharged.

Pathologic examination showed a 10-cm mesenteric cystic formation adherent to the resected ileum with fluid, yellowish and dense content (sebum-like). The formation had a thickened wall with infiltration of inflammatory cells. No abnormalities were found in the ileal mucosa (Figure 2).

Discussion

Nevoid basal cell carcinoma syndrome (NBCCS), also known as Gorlin syndrome, is a rare autosomal dominant disorder caused by mutations in the Patched

(PTCH) gene on chromosome 9q22 (3, 4).

The PTCH protein acts in a negative feedback pathway as a receptor for different hedgehog proteins. Hedgehog signalling is involved in cell growth and differentiation in a wide variety of tissues and organs. The PTCH germ line mutations in NBCCS patients are either inherited from an affected parent or due to *de novo* events. Tumorigenesis starts if inactivation of the second, normal PTCH allele by either loss-of-heterozygosity or point mutation occurs by chance in a single cell (two-hit mutagenesis). Tumorigenesis results in reactivation of the hedgehog/PTCH pathway and not only causes basal cell carcinoma, but also contributes to the formation of tumors such as medulloblastoma and rhabdomyosarcoma (5). Furthermore, several recent studies found evidence for an involvement of hedgehog signalling and PTCH in tumorigenesis of the GI- tract (5-11).

NBCCS patients often have a coarse facial appearance with macrocephaly, frontal bossing and prognathism. Falx calcification is frequently found in affected individuals and skeletal anomalies such as bifid ribs, wedge-shaped or fused vertebra and thumb deformities are common. Multiple keratocysts of the

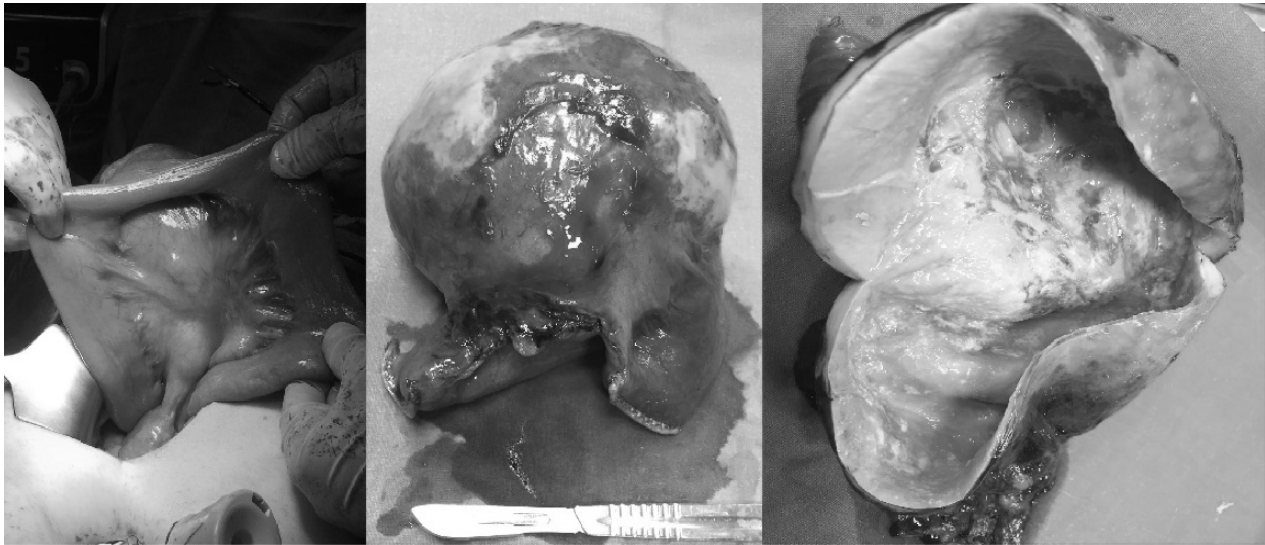


Figure 2 - Surgical specimen.

jaw can develop between childhood and young adulthood, and most patients get their first basal cell carcinoma (BCC) in their early 20s (12).

In the case we have described the patient presented with mesenteric cyst whose occurrence is less than 5% of individuals with Gorlin syndrome (13).

The rarity of the two conditions strongly argues against an independent occurrence of both: mesenteric

cyst and NBCCS in our patient.

Maybe a causative relation between the mesenchymal proliferation in cystic wall and the PTCH germ line mutation exists and it could be an entirely independent pathogenetic chain of events.

At pathological examination, cystic wall had inflammatory infiltration and we know that chronic inflammation often leads to displasia and tumors.

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