

# Gardner syndrome complicated with hydronephrosis.

## A case report.

Paolo Cabassa<sup>1\*</sup>, Mario Morone<sup>1</sup>, Enza Gatti<sup>1</sup>, Mariapaola Narbone<sup>1</sup>, Roberto Maroldi<sup>1</sup>

*1. Department of Radiology Spedali Civili, Brescia University, Brescia, Italy*

\* **Correspondence:** Paolo Cabassa, Department of Radiology Spedali Civili, Brescia University,  
1 viale Europa, Brescia 25123, Italy  
 [paolocab@libero.it](mailto:paolocab@libero.it)

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### ABSTRACT

Gardner syndrome is an autosomal dominant disease characterized by the presence of colonic polyposis, osteomas and soft tissue tumors. We present a case of a man who was admitted for a relapse of adenocarcinoma of the rectum. CT-staging showed multiple locations of desmoid tumors and osteomas, with final diagnosis of Gardner syndrome. The follow-up CT, after surgery and chemotherapy, showed a relapse of the lesions with hydronephrosis due to ureteral compression.

### CASE REPORT

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A 38 year-old man was admitted for a relapse of adenocarcinoma of the rectum. In a hospitalization, one year previously, the patient had been submitted to anterior resection of the rectum for a moderately differentiated adenocarcinoma. In that occasion, a computed tomography (CT) staging showed perirectal and iliac lymph-node metastases and no other findings. At physical examination a mass was palpable in the paraumbilical region; the mass was firm, non-tender, and not painful. Colonoscopy revealed numerous polyps covering the whole colon and rectum. CT staging was performed to evaluate the extension of the abdominal mass and stage the disease. CT demonstrated an 11 x 6 x 19 cm solid mass with inhomogeneous enhancement in the anterior abdominal wall involving the rectus abdominis muscle; another mass was visible in right iliac fossa. Other solid nodules with similar features were recognizable in the right external oblique muscle and paravertebral muscles. Another one, involving the mesenteric fat tissue, was poorly margined and associated with some prominence of the adjacent mesenteric vasculature (Figure 1). Furthermore CT depicted a lentiform ossified lesion in the frontal bone and multiple exostoses in the skull (Figure 2). CT did not show lymph-node or metastases.

The patient underwent ultrasonography-guided biopsy and histopathologic analysis revealed that the tumour consisted of spindle-shaped cells embedded in a collagenous stroma

(Figure 3). No mitoses were found. The final pathologic diagnosis was desmoid tumor. The patient was transferred for surgical resection of masses and polypectomy; at surgery a fixed retroperitoneal mass involving the mesentery and vessels was found. Subsequently this mass was not removed and the patient received adjuvant chemotherapy.

Genetic tests showed a mutation of adenomatous polyposis coli (APC) gene in the patient and confirmed the suspicion of Gardner syndrome (GS).

A further CT staging was performed ten months later, to evaluate response to chemotherapy, demonstrated a relapse of masses in the abdominal wall and an increase in size of the mesenteric mass compromising the right ureter causing hydronephrosis, with concomitant reduced renal cortical enhancement (Figure 4). The patient started new cycles of chemotherapy and is continuing his follow-up.

#### DISCUSSION

Gardner syndrome (GS) is a rare (1:1000000 population in USA) autosomal dominant inherited disorder due to germline mutations in the APC tumor suppressor gene on chromosome 5 with complete penetrance and variable expressivity (1). GS was first reported by Gardner and his

colleague in the early 1950s (2). GS is considered a variant of familial adenomatous polyposis (FAP), it was reported that GS is caused by truncating mutations of the APC gene (codons 1403 and 1578) differing from classic FAP (codons 169-1600) (1). GS represents a multisystem disease characterized by the triad of colonic polyposis, multiple osteomas and mesenchymal tumors of the skin and soft tissues. FAP is characterized by the presence of hundreds or thousands of colorectal tubular adenomas. Intestinal polyps become malignant in virtually 100% of patients if not treated. Hence, prophylactic colectomy is indicated.

Desmoid tumors are rare, locally aggressive, non-encapsulated masses resulting from a benign proliferation of fibrous tissue. Abdominal desmoid can occur sporadically and develop anywhere in the abdomen, including the musculature of the abdominal wall or cavity (especially the mesentery), the retroperitoneum and the pelvis. Common locations are the incision sites. However, desmoid forming in the mesentery are especially common in patients with adenomatous polyposis or GS, occurring in 9%-18% of cases (3).

Although they do not metastasise and are histologically benign, they are locally aggressive with a propensity to invade local structures and recur after resection. The infiltrative nature of mesenteric desmoid tumors can lead to bowel loop or ureter obstruction or damage to blood vessels, causing life-threatening complications (4).

The incidence of desmoid is only 2-4 per million per year; 0.03 % of all tumors. They are typically seen in the third and fourth decades of life. Women are reported to be affected more commonly than men (5-6). Prior abdominal surgery is an important risk factor for the development of mesenteric fibromatosis in patients with GS (3.5-5.7% of patients and usually appear within 3 years following surgery although they can appear at anytime). Desmoid tumors may be also associated with trauma and estrogen therapy. Mesenteric desmoid tumors are a major cause of morbidity and mortality in patients with GS, who have undergone prophylactic colectomy.

At microscopic analysis, mesenteric fibromatosis is composed of fibroblasts in an uninflamed, abundant collagenous stroma. The fibroblasts are elongated, spindle-shaped cells with regular uniform nuclei and scant cytoplasm.

The clinical presentation of GS is variable and its diagnosis is often delayed, despite the presence of clues for a significant amount of time (6). Most patients with desmoid tumors are clinically asymptomatic, and the tumors cause little or no focal symptoms until late in their course. Patients may present to a physician because of a palpable mass, abdominal pain, or gastrointestinal bleeding (5). Desmoid can be locally aggressive and may invade contiguous structures. Some complications that have been reported include small-bowel obstruction and hydronephrosis (4).

The diagnosis of FAP and GS can be made by genetic testing for gene mutations (APC), by the demonstration of multiple colonic polyps during colonoscopy and histopathologic analysis. The occurrence of a lesion in the

rectus abdominis muscle and the adjacent musculoaponeurosis should alert the radiologist to the possibility of a desmoid tumor. The diagnosis of a desmoid tumor should be strongly considered in this case because of the patient's age, sex, history of previous surgery, and imaging findings but final diagnosis is only by biopsy (7). A definitive diagnosis based solely on imaging features is not possible. Ultrasonography is useful in the initial evaluation of patients with soft-tissue masses and could be a guide for biopsy. Desmoid tumor appears as well-defined lesions with varying echogenicity (8).

CT and magnetic resonance (MR) imaging are useful techniques in the evaluation of the size, site, extent, and relationship between the lesion and surrounding structures, so they are useful in planning surgical resection and predicting prognosis. CT and MR typically demonstrate an irregular, infiltrating mass. On CT, most mesenteric desmoid are isodense relative to muscle, although large lesions may display areas of low attenuation caused by necrosis. Lesions, at MR imaging, show low or intermediate signal intensity on T1-weighted images and have heterogeneously intermediate or high signal intensity on T2-weighted images. However, CT and MR appearance of desmoid tumor varies, depending on their composition. Early stage lesions are more cellular, hypodense on CT and hyperintense on T2-weighted MR images. As desmoid tumors evolve, collagen deposition increases with a resultant decrease in signal intensity on T2-weighted image and homogeneous, soft-tissue attenuation on CT scan (9-12). Desmoid tumors may show homogeneous, inhomogeneous or no significant enhancement on CT and MR scans enhance after injection of contrast material.

There are no specific imaging features on MR or CT to distinguish desmoid tumors from other solid masses. On the basis of imaging findings alone, mesenteric fibromatosis is difficult to differentiate from malignant neoplasm of the mesentery, such as lymphoma, metastatic disease, Gastrointestinal Stromal Tumors and soft-tissue sarcomas. The differential diagnosis for rectus abdominis lesions includes acute hematoma and other soft-tissue tumours, such as fibrosarcoma, lymphoma, rhabdomyosarcoma, neurofibroma, benign fibrous tumour, and primitive neuroectodermal tumours (8-10).

Since GS may involve different organs, it is usually very difficult to treat it, and the therapeutical effect is also uncertain.

The therapeutic management of desmoid tumors is controversial. Numerous treatments have been tried, but none has proven effective (13). Most authors recommend avoiding surgery because it carries a high mortality rate and often requires sacrifice of considerable lengths of small bowel. Even if excision is successful, recurrence occurs in up to 88% of cases and so operation is recommended only for symptomatic patients (14).

TEACHING POINT

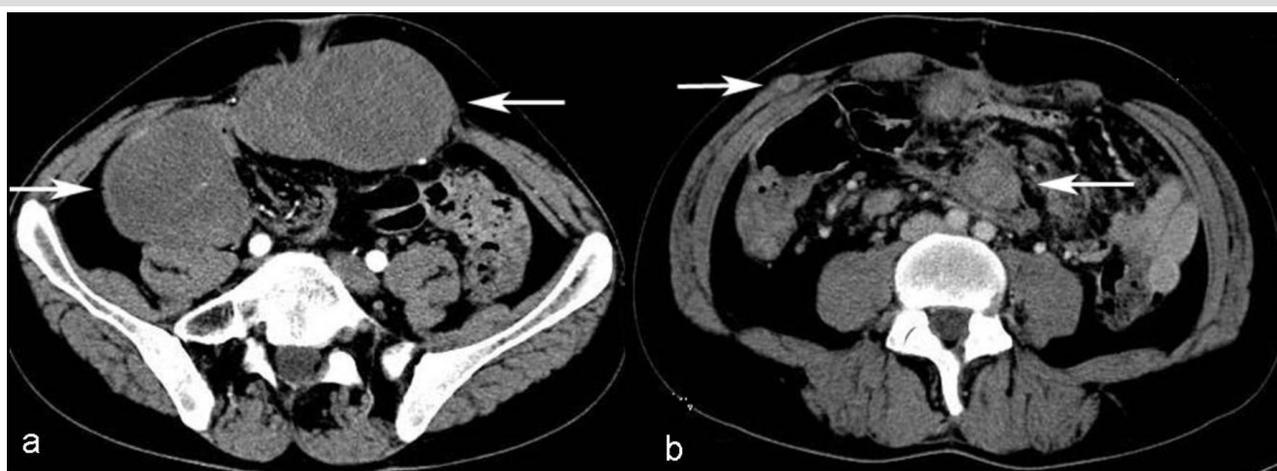
The detection of multiple osteomas and desmoid tumors in a patient with history of abdominal surgery (particularly for rectal adenocarcinoma) could help in suspecting the diagnosis of Gardner syndrome. Desmoid tumors don't metastasise, recur after resection and are locally aggressive; their infiltrative nature can lead to bowel loop or ureter obstruction, or damage to blood vessels, causing life-threatening complications.

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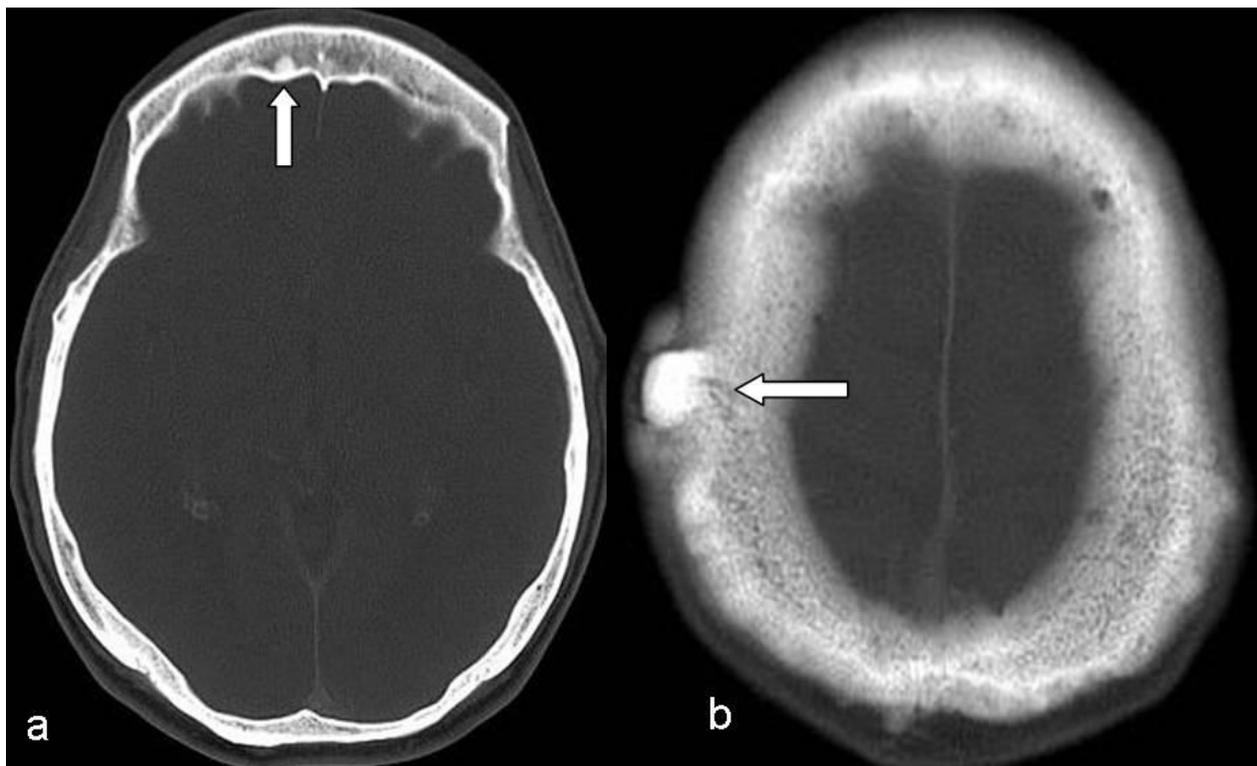
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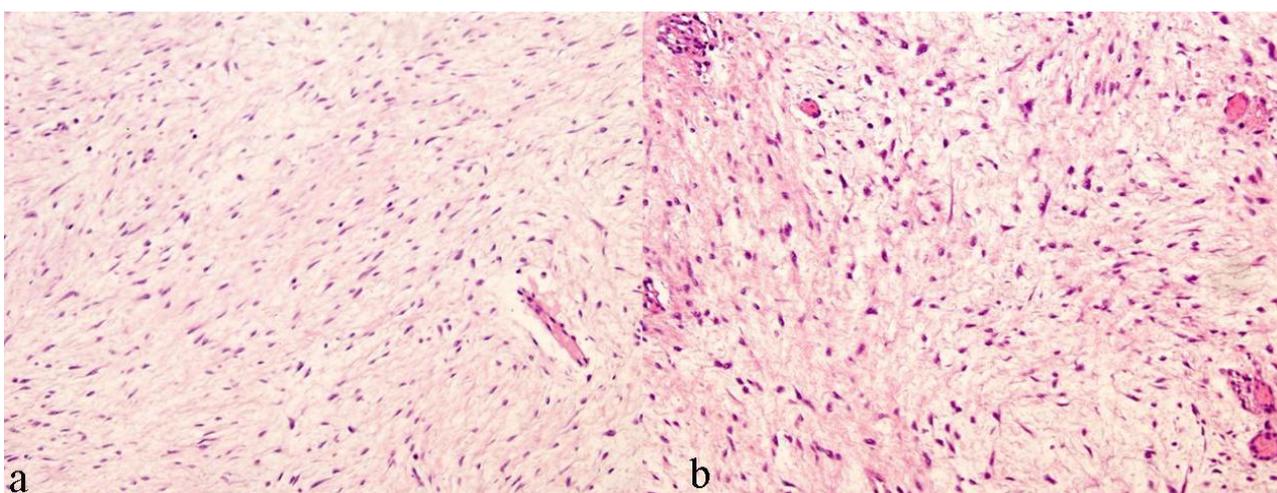
FIGURES



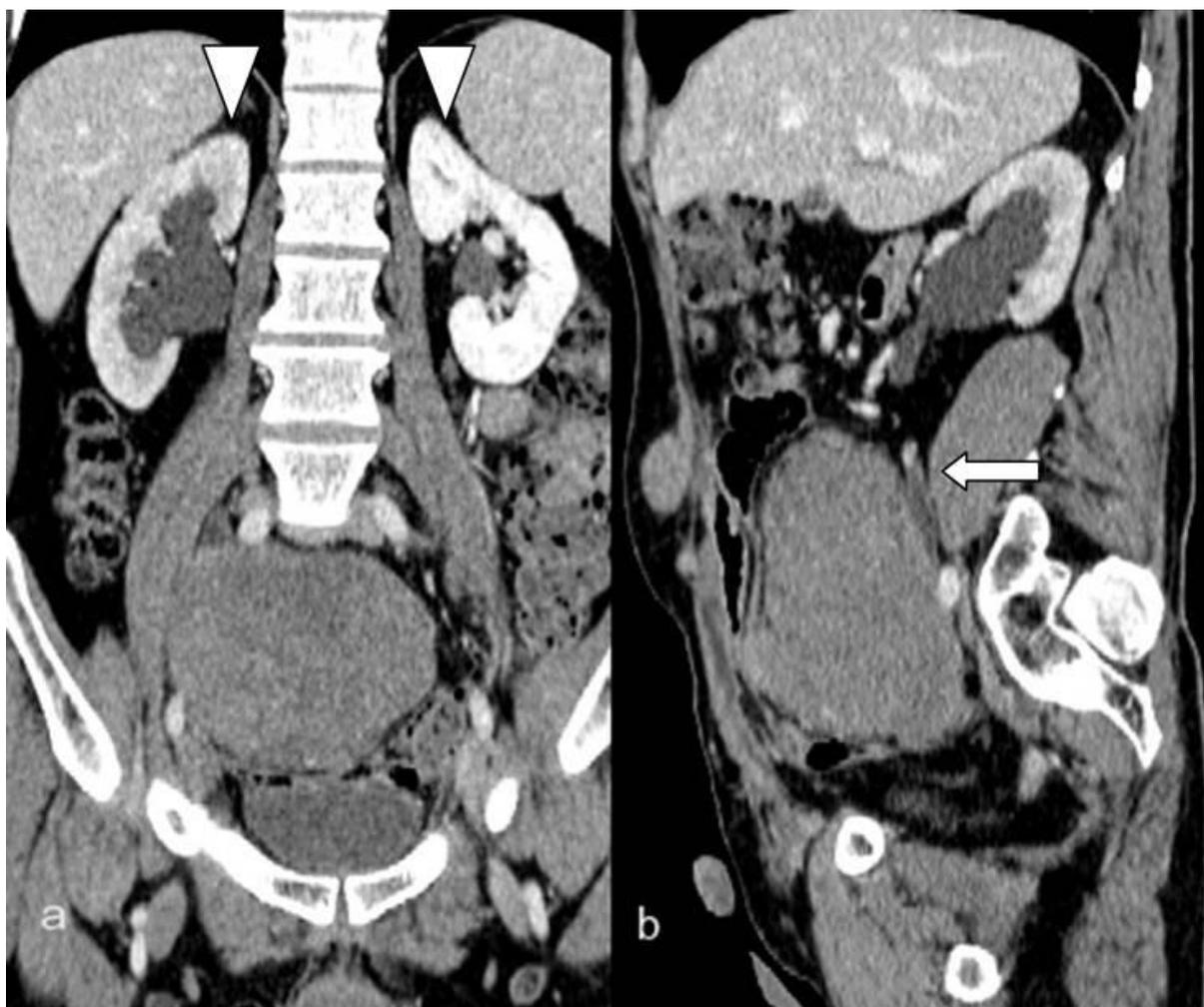
**Figure 1:** A 38 year-old man with abdominal masses. Axial multidetector CT scan of the abdomen during arterial phase (figure a) demonstrates a 11 x 6 x 19 cm solid mass with inhomogeneous enhancement in the anterior abdominal wall (right arrow in figure a) involving the rectus abdominis muscle; another mass was visible in right iliac fossa (left arrow in figure a). Axial multidetector CT scan of the abdomen during portal phase (figure b) also showed solid nodules in the right external oblique muscle (left arrow in figure b) and in mesenteric fat tissue in the left side that was poorly margined and associated with some prominence of the adjacent mesenteric vasculature (right arrow in figure b). (Imaging parameters: 5 mm slice thickness; 120kv; variable mA with automatic exposure control, this level 130 mA)



**Figure 2:** Axial multidetector CT scan of the head (bone window) depicts a lentiform ossified lesion in the right frontal bone (arrow in figure a) and multiple exostoses in the skull (arrow in figure b). (Imaging parameters: 6 mm slice thickness; 120kv; variable mA with automatic exposure control, this level 300 mA)



**Figure 3:** These high-power views (a: 10x magnification; b: 100x magnification) of a needle-core ultrasonography-guided biopsy that was taken during fine-needle aspiration show the orderly arrangement of uniform fibroblasts embedded in a collagenous stroma (hematoxylin and eosin stain). No mitoses were found.



**Figure 4:** Coronal (a) and sagittal (b) multiplanar reconstruction (MPR) multidetector CT staging to evaluate response to chemotherapy demonstrated, during portal phase, increase in size of the mesenteric mass compromising the right ureter ( arrow in figure b) causing a hydronephrosis with reduced renal cortical enhancement of the right kidney (arrowheads in figure a). (Imaging parameters: 5 mm slice thickness; 120kv; variable mA with automatic exposure control, this level 160 mA)

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**ABBREVIATIONS**

- GS = Gardner Syndrome
- APC = adenomatous polyposis coli
- FAP = familial adenomatous polyposis
- CT = computed tomography
- MR = magnetic resonance

**KEYWORDS**

Gardner; desmoid tumors; computed tomography

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