Case 1338

Eurorad

Desmoid tumours complicating familial adenomatous polyposis

Published on 17.12.2001

DOI: 10.1594/EURORAD/CASE.1338
ISSN: 1563-4086
Section: Abdominal imaging
Imaging Technique: CT
Imaging Technique: MR
Imaging Technique: CT
Imaging Technique: MR
Case Type: Clinical Cases
Authors: L. Manganaro, C.Carrozza, M.L. Angeli, L.Bertini, L.Ballesio
Patient: 22 years, male

Clinical History:

The patient, who had undergone a total colectomy 2 years previously as a result of familial adenomatous polyposis, presented with two big masses in the lower inferior quadrant of the abdomen.
Imaging Findings:

The patient presented with intense pain of the lower inferior quadrant of the abdomen. He had undergone a total colectomy 2 years previously as a result of familial adenomatous polyposis (FAP).
Abdominal ultrasound demonstrated a conspicuous hydronephrosis. A CT scan of urinary tract was performed with oral and intravenous contrast material showing hydronephrosis and also two masses. One mass, of around 6 x 7 cm, was located on the left side; the other one, of around 10 x 8 cm, was located on the right side under the umbilical area. Both masses were very adherent to the psoas muscle and to the intestinal loops. Both masses appeared with high vascularisation and were apparently capsulated.

These findings were confirmed by an MRI examination that showed the masses to have a high signal intensity onT2-weighted images with marked and homogeneous enhancement after contrast administration.

In the face of these findings a surgical biopsy was performed and revealed that the two masses were desmoid tumours. The patient was treated with chemoterapy.

A CT scan after treatment demonstrated that the mass located on the left side was reduced by 50% and the mass on the right was highly necrotic.

Discussion:

Familial adenomatous polyposis (FAP) is an autosomal dominant condition caused by germline mutation of the APC tumour suppressor gene, which occurs with a frequency of approximately 1:7500 of the population; about 15% of cases result from a new, rather than an inherited, mutation.
Desmoid tumours are one of the most important and intriguing extracolonic manifestation of FAP and theycontribute

significantly to the morbidity and mortality rate.

Desmoid tumours are often multiple and occur most frequently in the abdomen; they appear with locally invasive proliferation of myofibroblast cells and occur in about 10% of patients with FAP. They normally present early in the forth decade of life, the majority following surgery by a little over 2 years. Trauma (surgical), oestrogens and the presence of a particular range of APC mutations have been implicated in the aetiology of desmoids, although their natural history remains obscure.

A variety of treatments, including surgical excision, non-steroidal anti-inflammatory drugs (NSAIDs), anti-oestrogens and cytotoxic chemotherapy have been advocated, but good evidence for the efficacy of most of these is lacking, because only small numbers of patients with FAP and associated desmoids have been studied.

Differential Diagnosis List: Desmoid tumors associated with familial adenomatous polyposis.

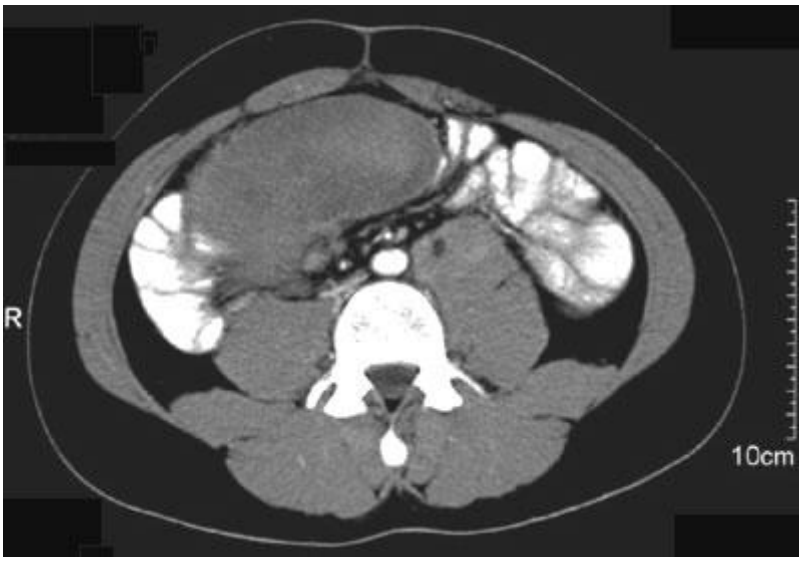
Final Diagnosis: Desmoid tumors associated with familial adenomatous polyposis.

References:

Clarke SK, Neale KS, Landgrebe GC,Phillips RKS. Desmoid tumors complicating Familial Adenomatous Polyposis. British Journal of Surgery. 1999 Sep;86(9) :1185-9. (PMID: 10504375)

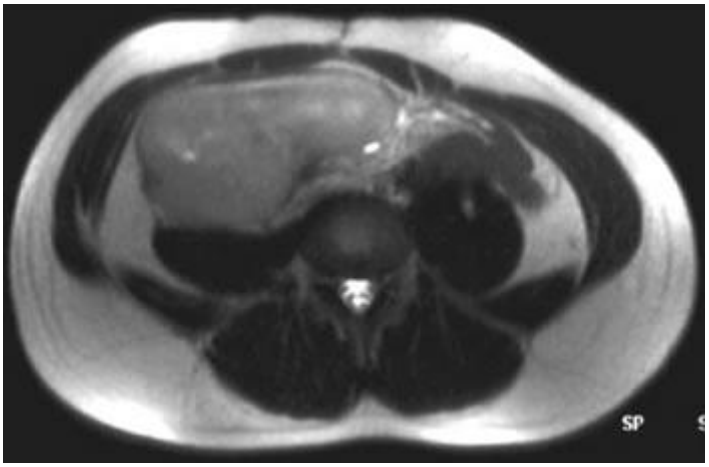
Figure 1

a



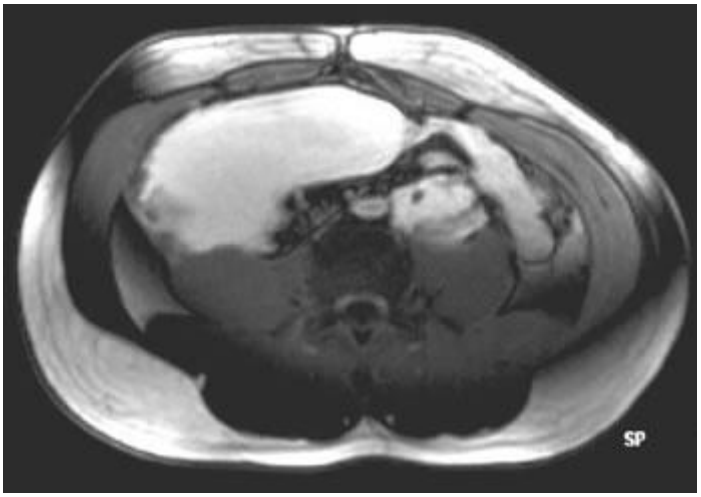
Description: CT scan demonstrating an abdominal mass adherent to intestinal loops and another one embracing the left ureter. **Origin:**

b



Description: T2-weighted image of the masses. **Origin:**

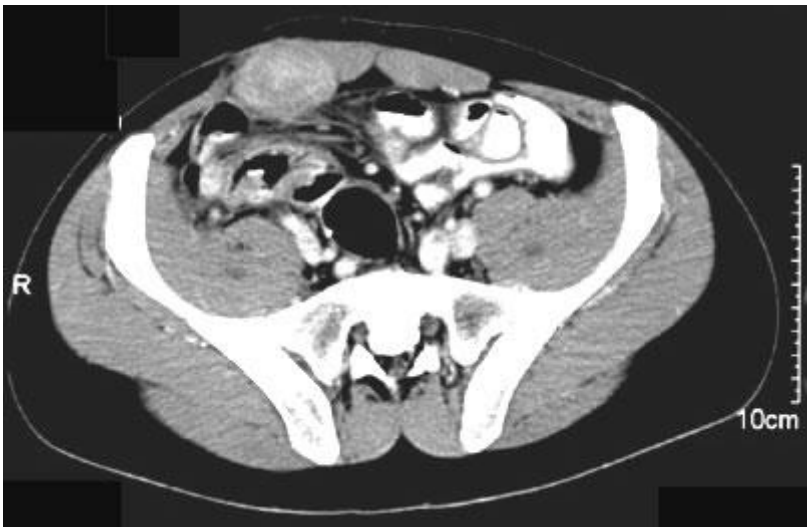
c



Description: After paramagnetic contrast administration the masses appear with a high signal intensity.
Origin:

Figure 2

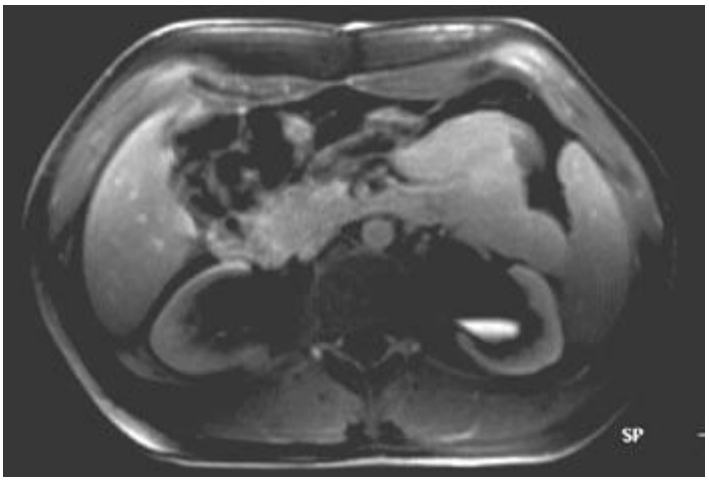
a



Description: CT scan of a mass infiltrating the abdominal wall. **Origin:**

Figure 3

a



Description: MR T2-weighted image demonstrating the left kidney hydronephrosis. **Origin:**