Poorly differentiated colonic adenocarcinoma showing rhabdoid feature: An extremely unusual anatomopathological diagnosis☆

Adenocarcinoma de colon pobremente diferenciado con diferenciación rabdoide: un diagnóstico anatomopatológico extremadamente infrecuente

Rhabdoid tumours were first identified in renal tumours at the end of the twentieth century as rare variants of Wilms' tumour.[1] Since then, tumours with this type of differentiation have been described at various locations, each characterised by greater aggressiveness of the lesions and poor patient prognosis.[2] The rarest site of extrarenal tumours with rhabdoid differentiation is the colon.

This case study concerns an 80-year-old patient with ASA II who attended the clinic for a screening colonoscopy finding of a tumour at the hepatic flexure of the colon.

The pathological diagnosis of the biopsy of the lesion taken during the colonoscopy was poorly differentiated adenocarcinoma, with additional immunohistochemical (IHC) study results indicating diffuse positivity for CK7 and negativity for CK20. As these findings were not typical of colorectal carcinoma, a definitive diagnosis of primary colorectal adenocarcinoma could not be confirmed.

CEA levels at the time of the study were 2.6 ng/ml.

After ruling out distant disease by thoracic, abdominal, and pelvic CT scan, a laparoscopic right hemicolectomy was performed, with no complications arising during the procedure or the post-operative period.

The pathological study of the specimen confirmed the diagnosis of poorly differentiated adenocarcinoma of the colon with rhabdoid differentiation (component comprising 70% of the lesion), which infiltrated the pericolic fatty tissue without reaching the serosa. However, angiolymphatic and perineural invasion was observed. Three of the 12 lymph nodes isolated were positive for metastatic carcinoma. End stage pT3N1 Mx.

The immunohistochemistry study of the differentiated component was positive for pan-CK AE1-AE3, CK20, EMA and CDX2 and negative for vimentin. Furthermore, the dedifferentiated rhabdoid component was positive for focal pan-CK AE1-AE3 and CK7, β-catenin and vimentin, and negative for CK20, EMA and CDX2. Both neoplastic components were negative for other markers like actin HHF35, desmin, CD56 and TTF1. p53 overexpression was seen in both components. No microsatellite instability was observed (Fig. 1).

The multidisciplinary committee decided to commence eight cycles of adjuvant chemotherapy (CT) with capecitabine in monotherapy. Eight months after the diagnosis and surgical treatment, the patient was in good general condition, with good tolerance of the adjuvant therapy and with no signs of disease progression.

Since it was defined as a pleomorphic giant cell carcinoma, fewer than 100 cases of undifferentiated carcinomas with rhabdoid component in the gastrointestinal tract have been published in the literature, with just 20 cases in colon resection specimens having been published, making the colon the most unusual site in the whole body[3] (Table 1). These tumours tend to appear in patients over the age of 60, irrespective of gender. The existence of this tumour as an independent pathological entity continues to be disputed as some experts consider it to be a phenotypic variation of a 'not otherwise specified' (NOS) adenocarcinoma. There are two types of tumour with rhabdoid component: pure, in which only rhabdoid cells are differentiated without another epithelial element, and composite, in

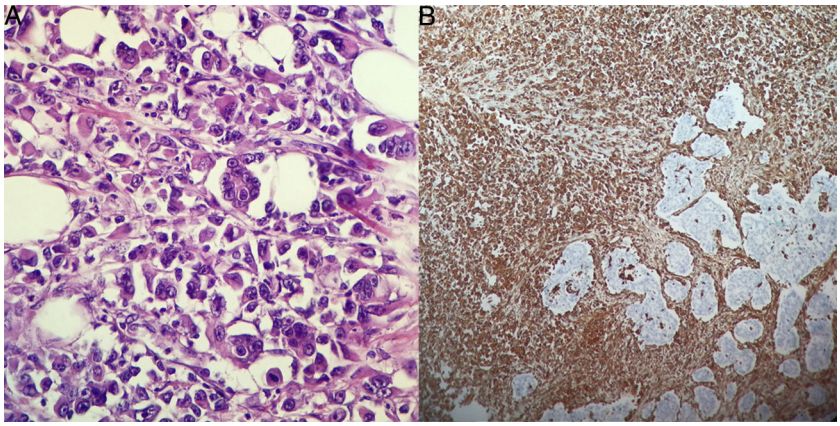

Figure 1 (A) Magnification (×40) of the haematoxylin and eosin stain of neoplastic cells with rhabdoid phenotype: marked pleomorphism and visible nucleoli, frequent mitotic figures, large eosinophilic cytoplasms. (B) Staining with vimentin (×20), positive in the mesenchymal component with rhabdoid phenotype and negative in the more differentiated epithelial component (adenocarcinoma).

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Table 1 Colon tumours with rhabdoid differentiation previously reported in the literature (arranged by date of publication).

Author	Age and gender	Location	Histology	Reference
Chetty et al.	72 M	Caecum	Composite	Chetty R, Bhathal P. Caecal adenocarcinoma with rhabdoid phenotype: an immunohistochemical and ultrastructural analysis. Virchows Arch A Pathol Anat Histopathol. 1993;422:179–182
Yang et al.	75 M	Transverse colon	Pure	Yang A, Chen W, Chiang H. Malignant rhabdoid tumour of colon. Histopathology. 1994;24:89–91
Macak et al.	50 M	Rectum	Composite	Macak J, Kodet R. Rectal adenocarcinoma with rhabdoid phenotype. Pathologica. 1995;87:696–699
Marcus et al.	84 F	Transverse colon	Composite	Marcus V, Viloria J, Owen D, Tsao M. Malignant rhabdoid tumour of the colon. Dis Colon Rectum. 1996;39:1322–1326
Nakamura et al.	76 M	Caecum	Pure	Nakamura I, Nakano K, Nakayama K, Ishii Y, Ohta K, Takahashi M, et al. Malignant rhabdoid tumour of the colon: Report of a case. Surg Today. 1999;29:1083–1087
Kono, et al.	66 M	Caecum	Composite	Kono T, Imai Y, Imura J, Ono Y, Hagiwara S, Taira K, et al. Cecal Adenocarcinoma with prominent rhabdoid feature: report of a case with immunohistochemical, ultrastructural, and molecular analyses. Int J Surg Pathol. 2007;15:414–420
Mastoraki et al.	62 F	Descending colon	Pure	Mastoraki A, Kotsilianou O, Papanikolaou I, Foukas P, Sakorafas G, Safioleas M. Malignant rhabdoid tumour of the large intestine. Int J Colorectal Dis. 2009;24:1357–1358
Han et al.	23 F	Rectum	Pure	Han S, Li J, Liu Z, Cheng J, Guo S, Wu X. Malignant rhabdoid tumour of rectum: report of a case. Tech Coloproctol. 2010;14:199–200
Pancione et al.	71 F	Ascending colon	Pure	Pancione M, di Blasi A, Sabatino L, Fucci A, Dalena A, Palombi N, et al. A novel case of rhabdoid colon carcinoma associated with a positive CpG island methylator phenotype and BRAF mutation. Hum Pathol. 2011;42:1047–1052
Remo et al.	73 F	Ascending colon	Composite	Remo A, Zanella C, Molinari E, Talamini A, Tollini F, Piacentini P, et al. Rhabdoid carcinoma of the colon: a distinct entity with a very aggressive behaviour. Int J Surg Pathol. 2011;20:183–188
Lee et al.	62 M 83 F	Sigmoid Rectum	Composite Composite	Lee S, Seol H, Kim W, Lim S, Kim W, Hwang T, et al. Rhabdoid colorectal carcinomas: reports of two cases. Korean J Pathol. 2013;47:372
Samalavicius et al.	49 M	Rectum	Pure	Samalavicius N, Stulpinas R, Gasilionis V, Baltruskeviciene E, Aleknavicius E, Mickys U. Rhabdoid carcinoma of the rectum. Ann Coloproctol. 2013;29:252

Table 1 (*Continued*)

Author	Age and gender	Location	Histology	Reference
Romera Barba et al.	77 M	Descending colon	Pure	Romera Barba E, Sánchez Pérez A, Duque Pérez C, García Marcilla J, Vázquez Rojas J. Malignant rhabdoid tumour of the colon: a case report. Cir Esp. 2014;92:638–640
Baba et al.	45 F		Composite	Baba Y, Uchiyama T, Hamada K, Ishihara Y, Tanaka H, Isono Y, et al. A case report of undifferentiated carcinoma of the sigmoid colon with rhabdoid features. Nihon Shokakibyo Gakkai Zasshi. 2014;111:1384–1390.
Agaimy et al.	79 M	Caecum	Pure	Agaimy A, Rau T, Hartmann A, Stoehr R. SMARCB1 (INI1)-negative rhabdoid carcinomas of the gastrointestinal tract. Am J Surg Pathol. 2014;38:910–920
Moussaly et al.	87 F	Transverse colon	Composite	Moussaly E, Atallah J. A rare case of undifferentiated carcinoma of the colon with rhabdoid features: a case report and review of the literature. Case Rep Oncol Med. 2015;2015:1–5
Cho et al.	73 M	Caecum	Composite	Cho I, Kim S, Min Y, Noh M, Hong R. Poorly differentiated cecal adenocarcinoma showing prominent rhabdoid feature combined with appendiceal mucinous cystadenoma: a case report and review of the literature. Oncol Lett. 2015;9:1527–1530
Kalyan et al.	31 F	Caecum	Composite	Kalyan A, Pasricha G, Monga D, Singhi A, Bahary N. Case report of rhabdoid colon cancer and review of literature. Clin Colorectal Cancer. 2015;14:e5-e8
D'Amico et al.	65 M	Ascending colon	Pure	D'Amico F, Bertacco A, Cesari M, Mescoli C, Caturegli G, Gondolesi G, et al. Extraordinary disease-free survival in a rare malignant extrarenal rhabdoid tumour: a case report and review of the literature. J Med Case Rep. 2018;12:39

which rhabdoid phenotype cells mix together with the adenocarcinoma.[4] In these cases, rhabdoid phenotype cells are believed to derive from malignant epithelial cells in which a clone suffers sarcomatoid dedifferentiation.[3]

They are histologically characterised by the presence of pleomorphic mesenchymal cells with specific immunohistochemical findings co-existing with a conventional epithelial tumour. Loss of INI1 nuclear expression due to deletions, translocations or other types of mutation that involve the tumour suppressor gene SMARCB1/INI1 at locus 22q11.2 has been seen in some cases.[5]

These tumours are extremely unusual and very aggressive, with significantly lower survival rates than other histological forms of colon cancer.

The rhabdoid variant is also very rare, and even more so at certain sites, like the gastrointestinal tract. The malignant neoplasms may be completely dedifferentiated towards this phenotype or, as more often occurs, this rhabdoid phenotype may be seen as an area in a typical epithelial neoplasm as a whole.

The finding of this histological characteristic in neoplasms implies greater dedifferentiation and therefore a worse prognostic course than for other neoplasms.

References

1. Marcus V, Viloria J, Owen D, Tsao M. Malignant rhabdoid tumor of the colon. Report of a case with molecular analysis. Dis Colon Rectum. 1996;39:1322–6.
2. Pancione M, di Blasi A, Sabatino L, Fucci A, Dalena A, Palombi N, et al. A novel case of rhabdoid colon carcinoma associated with a positive CpG island methylator phenotype and BRAF mutation. Hum Pathol. 2011;42:1047–52.
3. D'Amico F, Bertacco A, Cesari M, Mescoli C, Caturegli G, Gondolesi G, et al. Extraordinary disease-free survival in a rare malignant extrarenal rhabdoid tumor: a case report and review of the literature. J Med Case Rep. 2018;12:39.
4. Moussaly E, Atallah J. A rare case of undifferentiated carcinoma of the colon with rhabdoid features: a case report and review of the literature. Case Rep Oncol Med. 2015;2015, 5313485313485.

5. Agaimy A, Daum O, Märkl B, Lichtmannegger I, Michal M, Hartmann A. SWI/SNF complex-deficient undifferentiated/rhabdoid carcinomas of the gastrointestinal tract: a series of 13 cases highlighting mutually exclusive loss of SMARCA4 and SMARCA2 and frequent co-inactivation of SMARCB1 and SMARCA2. 2016;40:544–53.

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Duodenal obstruction due to intramural haematoma as a complication of endoscopic biopsy☆

Obstrucción duodenal por hematoma intramural como complicación de la biopsia endoscópica

Bowel obstruction secondary to an intramural haematoma is a rare complication and generally arises following abdominal trauma, anticoagulant therapy or haematological diseases.[1] Endoscopic biopsy of the duodenum is a very rare cause of intramural haematoma, with only isolated cases published in the literature, mostly in children.[2,3] In a series of almost 27,000 procedures conducted over a 12-year period, the incidence of this complication was 1/1922 procedures (2015).

We present the case of a 27-year-old female patient with coeliac disease diagnosed in 2013 due to her family history, HLA-DQ2-positive, anti-tissue transglutaminase antibody levels of 30 IU/ml and duodenal biopsies with mild villous atrophy (Marsh 3A). The patient was not following a strict gluten-free diet or undergoing clinical monitoring when she attended the clinic due to asthenia and intermittent diarrhoea, with a BMI of 19. The gastroscopy showed normal duodenal mucosa. Biopsies were obtained using standard forceps. The patient experienced no bleeding during the examination and was discharged asymptomatic. 12 h after the examination, she experienced nausea, vomiting and epigastric pain. Upon re-examination, she was found to be afebrile with BP 108/60 mmHg, HR 65 bpm and abdomen painful upon palpation of the mesogastrium, but without signs of peritoneal irritation. The abdominal X-ray revealed no pneumoperitoneum; Blood test: leukocytes $10.8 \times 10^3/\mu l$, Hb 11.8 g/dl, platelets $222 \times 10^3/\mu l$, prothrombin time 98%, APTT 29.1 s, CRP 1.3 mg/dl, GPT 19 U/l, GGT 43 U/l, AP 55 U/l, total bilirubin 0.6 mg/dl and amylasaemia 130 U/l. Due to the persistent signs of obstruction and intense pain, despite nasogastric suction and analgesia, an abdominal CT scan was performed, which showed a large accumulation measuring $4 \times 8 \times 10$ cm, AP × LL × cc, in contact with the duodenum, with displacement to the left of the head of pancreas and superior mesenteric artery and vein. The content of the accumulation was heterogeneous, consistent with a duodenal haematoma. Because only a portal-phase CT scan was performed, active bleeding could not be ruled out due to its hyperdense content that could correspond to extravasation of the IV contrast. In light of the above, an exploratory laparotomy was performed.

The examination revealed a duodenal haematoma with involvement from the pylorus and which dissected the first portion of the duodenum and initial part of the second, with no evidence of transmural perforation. The haematoma was drained with evacuation of clots, leaving the gastrostomy tube in place. After the operation, total parenteral nutrition was started together with IV antibiotic therapy. The patient recovered without complications and was discharged 28 days after the procedure on an oral diet.

The histological study of the duodenal biopsies showed increased intraepithelial lymphocytes without villous atrophy (Marsh 1).

The coagulation study was normal (platelet count, prothrombin time and APTT); more targeted studies were not performed (Fig. 1).

Bowel obstruction secondary to duodenal haematoma is a very rare endoscopy-related complication, with only isolated cases published in the literature, mostly in healthy children.[2,3] Endoscopic biopsies may entail a greater risk for patients with haematological disorders. Although there is very little information available,[4] the indication for biopsy must be thoroughly justified in these patients.

Our patient did not exhibit any coagulation disorder that could have contributed to the development of a haematoma. Despite a normal platelet count, prothrombin time and APTT, this patient may nonetheless have been susceptible to a greater risk of bleeding secondary to platelet dysfunction or another coagulation disorder associated with the underlying disease. However, no platelet aggregation studies were performed or fibrinogen or D-dimer levels assessed that could be of use. The only procedure undertaken was the mucosal biopsies obtained with standard forceps. As such, the haematoma appears to have been

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