

## Caso clínico

# Peritoneal mucinous carcinomatosis in a patient with a history of Hirschsprung's disease. The role of home parenteral nutrition

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### Abstract

A case of peritoneal mucinous carcinomatosis in a patient who suffered a Hirschsprung disease 30 year before is presented. TH present condition caused an irreversible intestinal obstruction and the patients received home parenteral nutrition without unremarkable complications longer than two years.

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Key words: *Hirschsprung disease. Intestinal obstruction. Peritoneal carcinomatosis. Home parenteral nutrition.*

Hirschsprung's disease (HD) results from an absence of ganglionar cells in the mucosa and muscular layers of the colon. There is a failure of neural crest cells to migrate to the mesodermal layers, possibly mediated by abnormalities in end-organ cell surface receptors or local deficiency of nitric oxide synthesis.

The disease occurs in 1 of 5,000 births, and accounts for 15-20% of cases of neonatal intestinal obstruction. When the symptoms appear later they present as alternative episodes of constipation and diarrhoea. Hypochromic anaemia, hypoproteinemia and failure to thrive are common. Treatment is surgical; resection of the aganglionic segment with the ganglionated proximal bowel pulled through seromuscular cuff of the distal rectum (pull-through procedure). Complications following surgery include chronic constipation, strictures, enterocolitis and bowel obstruction.

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### CARCINOMATOSIS MUCINOSA PERITONEAL EN UN PACIENTE CON ANTECEDENTE DE ENFERMEDAD DE HIRSCHSPRUNG. EL PAPEL DE LA NUTRICIÓN PARENTERAL

### Resumen

Presentamos el caso de un paciente en quien se diagnosticó una carcinomatosis mucinosa peritoneal treinta años después de haber padecido una enfermedad de Hirschsprung. El cuadro clínico actual cursó con una obstrucción intestinal no resoluble quirúrgicamente y fue tratado con nutrición parenteral domiciliaria durante más de dos años sin complicaciones.

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Palabras clave: *Enfermedad de Hirschsprung. Obstrucción intestinal. Carcinomatosis peritoneal. Nutrición parenteral domiciliaria.*

We present a patient with a history of HD who developed 30 years later anal fistulae that resulted to be a mucinous signet cell adenocarcinoma.

### Case report

The patient is a 46 year-old man who underwent a partial colectomy and ileoanal pull-through procedure because of HD at age 9. He did well until his 30's when he presented pain in the lower abdomen irradiated to the sacral area. A presacral abscess was diagnosed, refractory to systemic and prolonged antibiotic therapy. Later on he developed an enteric fistula (fig. 1) initially treated in a conservative manner. Recurrent episodes of enteric fistula in the pelvis led to perform an abdominoperineal resection of the colon and the creation of an end ileostomy. Despite the surgical procedure, new fistulae appeared to the sacral area with involvement of soft tissue, muscle and bone. Computed tomography and magnetic resonance imaging demonstrated a large multilobular cystic process in the presacral space (fig. 2). Mucous material was observed through the fistulae. Histological examination showed abundant mucous with a moderate number of epithelial

cells with moderate atypia, consistent with the diagnosis of mucinous signet cell adenocarcinoma. Although he received radiotherapy as well as systemic chemotherapy with 5-fluorouracil and leucovorin the disease progressed to a complete intestinal obstruction. For the following two years he was managed conservatively with home parenteral nutrition, intravenous rehydration at home and a decompression gastrostomy with no major incidences. The patient subsequently died of carcinoma after a short deterioration of his basal status.

## Discussion

Colorectal mucinous adenocarcinoma constitutes up to 10-20% of the colorectal cancers<sup>1</sup>. This type of colorectal cancer is considered to have a worse prognosis than typical adenocarcinoma. The presence of signet-ring cells is a non-significant indicator of a poor prognosis.

It has been reported a higher incidence of colorectal mucinous adenocarcinoma in patients with inflammatory bowel disease. There are a few reports in the literature of a carcinoma arising after use of a double-stapled ileal pouch-anal anastomotic technique<sup>2</sup> or in long-standing multiple perianal and presacral fistula<sup>3</sup>. There is only one reported case of mucinous adenocarcinoma arising from a stricture after a Soave procedure<sup>4</sup>. We present another patient who developed an adenocarcinoma years after

surgical treatment of HD. Deterioration of general health began long before diagnosis. The main complains our patient presented were abdominal pain and distension.

Because of the inflammatory changes associated with peritoneal tumour implants, fistula formation and adhesions are common, they may finally lead to a chronic partial or total bowel obstruction. Our patient eventually developed an irreversible bowel obstruction and required home parenteral nutrition as well as intravenous hydration to maintain his body weight and hydration status.

Although radiological investigations can be helpful, they are not diagnostic and a laparotomy is often required to reach a diagnosis. None of the surgical procedures performed in our patient for 15 years allowed the diagnosis. Our explanation is that the condition probably was not initially present but developed as a consequence of chronic inflammation related to recurrent colonic and rectal fistulae inducing dysplasia in the course of time.

So far, an association between HD and mucinous adenocarcinoma has not been identified. HD is linked to medullary thyroid carcinoma and other endocrinopathies included in the multiple endocrine neoplasia type 2 (MEN 2) syndrome 5. RET protooncogen germ line mutations are associated with MEN 2 as well as with familial and sporadic HD. Other cancers in HD are exceptional.

The ileo-anal Soave pull-through is the most common surgical procedure performed in patients with total colonic aganglionosis. The technique presents a low number of complications, such as fecal incontinence, strictures or episodes of intestinal obstruction.

Advanced abdominal disease causes intestinal obstruction that accounts for a great burden for patients. Within this scenario, we consider that home parenteral nutrition represents an acceptable therapeutic regimen to prolong survival in patients with locally advanced cancer and a good quality of life.

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Fig. 1.—Barium enema. Rectum appears narrow and irregular. Mass effect in the pre-sacral area. Long fistulous tract to left buttock.



Fig. 2.—Axial T1-weighted MR. Irregular and heterogeneous mass involving rectum, invading bone and reaching subcutaneous soft tissues.