A 50-year-old patient presented with marked dilation of the second portion of the duodenum on abdomino-pelvic computed tomography (CT), discovered after an examination due to dyspepsia (Fig. 1). She had a family history of megaduodenum on her father’s side. Surgical intervention was decided, performing a subtotal duodenectomy with pyloric and duodenal papilla preservation with Y de Roux reconstruction and cholecystectomy. Re-intervention was performed on day 4 due to biliary peritonitis secondary to punctiform perforation of the first duodenal portion, adjacent to duodeno-jejunal anastomosis. An antrectomy with preservation of duodenal papilla and a new gastro-jejunal anastomosis were performed (1,2).

Discussion
The so-called familial megaduodenum, first reported in 1938 by Weiss, consists of an uncommon hereditary myopathic degeneration with an autosomal dominant character, which generally presents before the age of 20 years (1-3). The clinical presentation features a history of recurrent intestinal pseudo-obstruction, along with weight loss, due to bacterial overgrowth. Given the non-specificity of the symptoms, patients usually experience a delay in diagnosis (1-3).
The main radiologic finding is dilatation of the duodenum without a mechanical cause. In our case, more frequent mechanical causes such as annular pancreas, Wilkie syndrome, adhesions and tumors, among others, were ruled out, as was a paraneoplastic neurological origin. This added to the presence of family history, and histological examination would confirm the diagnosis. Although in our case, no significant abnormality in the duodenal wall was demonstrated and the myenteric plexus was normal (1-4).

In less severe cases, treatment is based on a hypercaloric and low-fiber diet, as well as antibiotherapy for bacterial overgrowth. In the most severe cases, enteral nutrition by jejunostomy or parental nutrition is contemplated (1-3).

Multiple surgical techniques for its repair, most of them aggressive, have been described. The bulk of the available literature is based on cases, which precludes establishing a “gold standard”. The main objectives of intervention are to relieve or avoid obstruction, improve emptying and restore the continuity of the gastrointestinal tract, paying special attention to the duodenal papilla (5).

In conclusion, familial megaduodenum is an uncommon disease, with few cases described in the literature, whose diagnosis is based on evidence of a duodenal dilation of functional cause together with the existence of family history. Conservative treatments are rarely sufficient, with surgical treatment, in the absence of disorders of intestinal motility, effective in achieving symptomatic relief in selected cases.

References
Fig. 1. Coronal cut of abdomino-pelvic computed tomography (CT) scan with oral contrast evidencing marked dilatation of the second portion of the duodenum (star), without evidence of apparent obstructive cause.