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## Case Report

## Currarino triad: A case report of a 48-year-old patient with a neuroendocrine tumor

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## ABSTRACT

Currarino triad is a rare syndrome, with less than 250 cases reported, and it includes a combination of sacrococcygeal bony abnormalities, anorectal malformations and the presence of a presacral mass. Here we present a case of a 48-year-old male patient with history of severe chronic constipation, who was incidentally diagnosed with a presacral during imagiological investigation, which was histologically proven to be a neuroendocrine tumor.

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### Introduction

Currarino Triad, a syndrome first described by the pediatric radiologist Guido Currarino, includes an hemisacrum, a anorectal malformation, and presacral mass.

Etiopathogenesis and genotype-phenotype correlation are not yet clarified, and still the subject of scientific investigation.

Different imaging modalities can be used for the assessment of this condition.

With the widespread of antenatal ultrasound, some malformations can be detected before birth, enabling early detection, and appropriate patient management.

Multiplanar techniques, as computed tomography (CT) and magnetic resonance (MR), allow better characterization of the sacral congenital defects and presacral masses. Although the majority of the masses reported in Currarino triad are benign, a histologic sampling is mandatory to exclude malignancy, as it was the case of our patient.

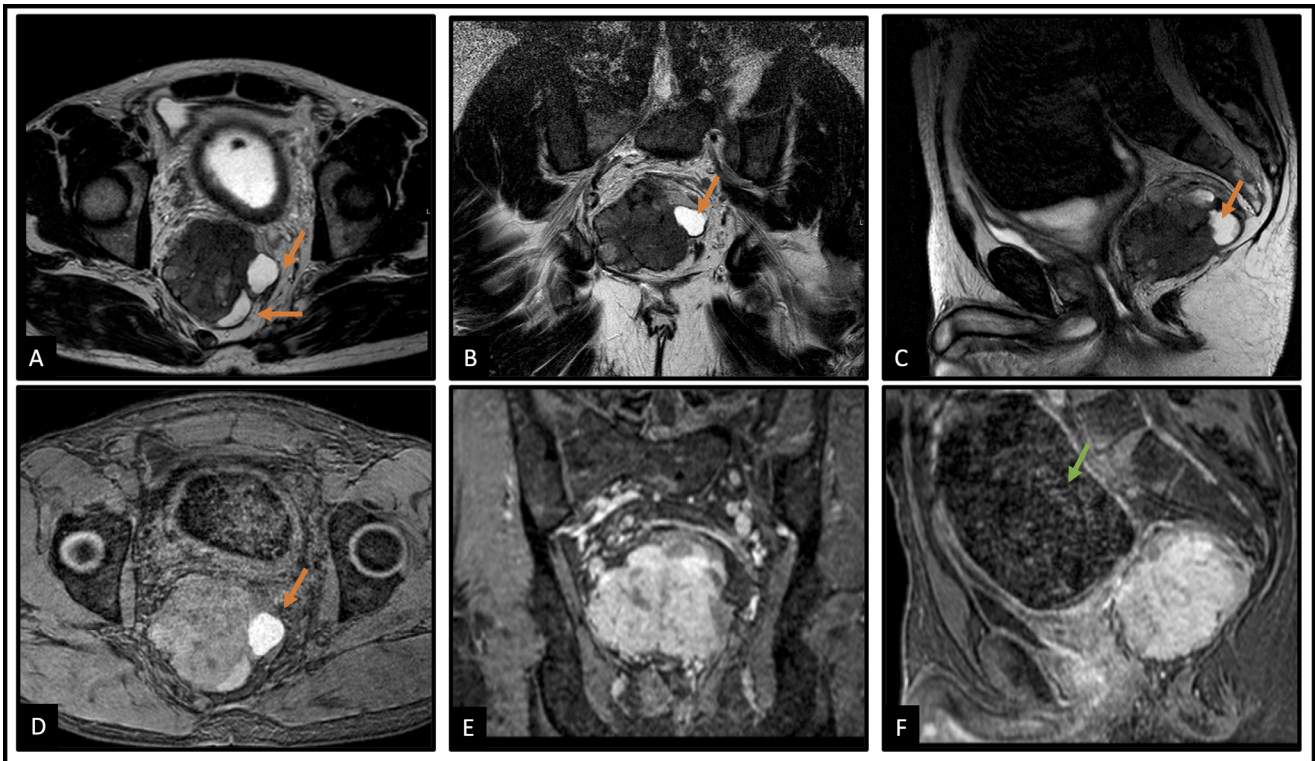
It is of utmost importance for clinicians and radiologists to be familiarized with the signs, symptoms and imagiological findings that constitute this triad, in order to provide prompt diagnosis and accurate patient management.

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**Fig. 1 – MRI of the pelvis. Axial (A), Coronal (B) and Sagittal (C) T2-weighted images. There is a pre sacral mass with lobulated contours exhibiting heterogenous signal with cystic areas (orange arrows) on the periphery. There seems to be cleavage planes with the surrounding structures. Axial T1-weighted images without contrast (D). The mass exhibits cystic areas with high signal (orange arrow) in relation with high density products (hemorrhagic or proteins). Coronal (E) and Sagittal (F) Axial T1-weighted images with contrast. The mass enhances avidly and heterogeneously, compatible with a hypervascular mass. There seems to be cleavage planes with the surrounding structures. Note on (F) a massive fecaloma on the rectum (green arrow). Of note, right partial sacral agenesis is partially seen on these images, better depicted on Fig. 5. (Color version available online.)**

## Case report

A 48-year-old male was sent to our institution to further characterize an incidental sacral mass detected on MR Defecography. At infancy he had surgery for anal imperforation, as did some of his siblings, according to the history provided by the patient. His daughter also had the same condition with an associated anterior myelomeningocele.

After anal imperforation correction surgery, he maintained chronic constipation, sometimes accompanied by fecal incontinence. In adulthood he developed colic inertia with severe constipation, that could only be managed with enemas and laxatives in doses far exceeding those recommended for the general population.

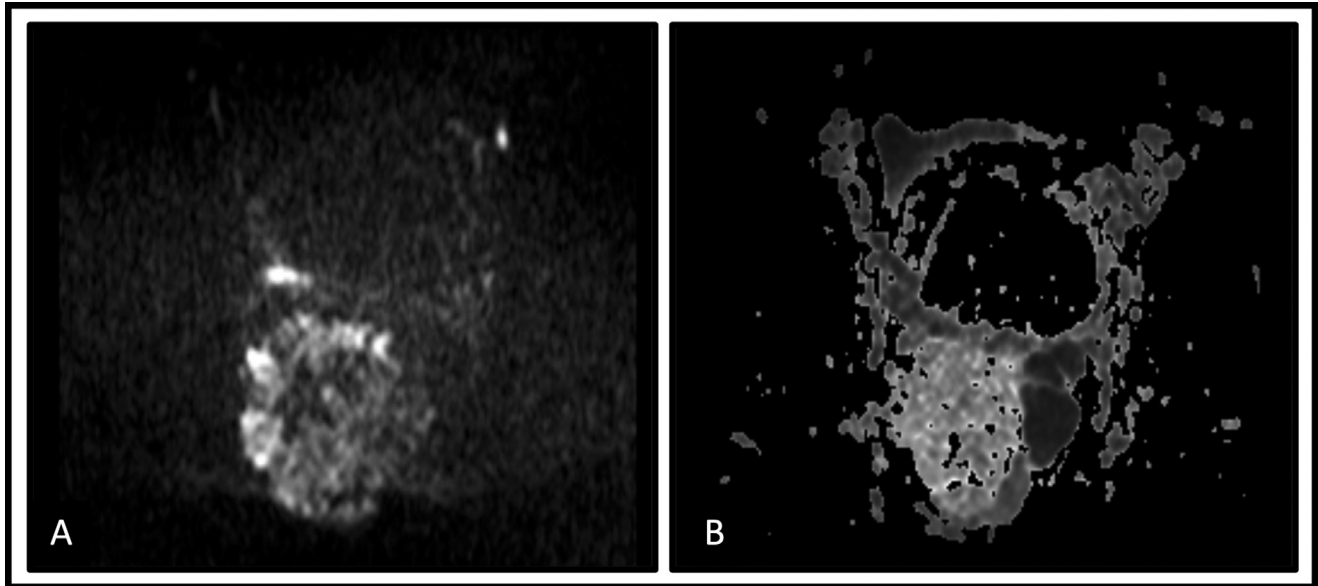
He was referred to a proctologic consultation and a MR Defecography was ordered. The results of this highly dynamic study were inconclusive, mainly because of a massive fecaloma prevented the patient from fully evacuating during the procedure. However, as incidental findings, not previously acknowledged, a presacral mass and partial sacral agenesis were depicted.

The mass was predominantly solid, with some cystic areas exhibiting high signal on T1-weighted images, in relation to hemorrhagic/high protein content (Fig. 1). It showed marked restriction diffusion, and there were also some enlarged pelvic lymph nodes, making the mass highly suspicious for malignancy (Figs. 2 and 3).

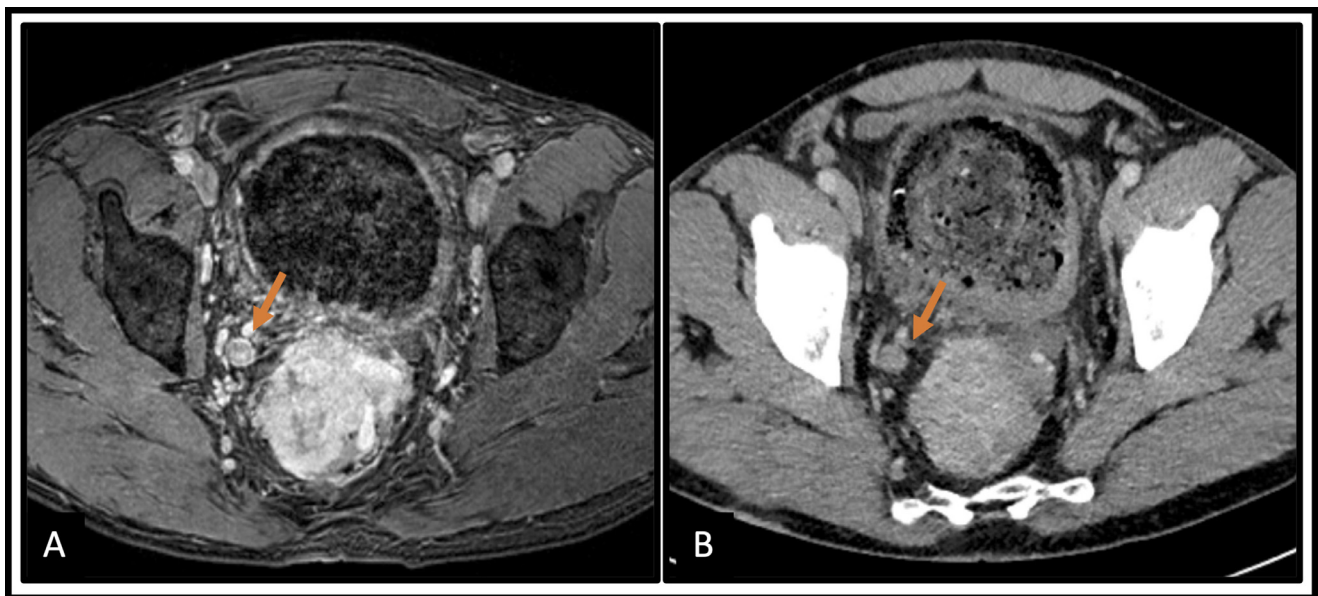
For staging purposes, a CT with IV contrast was performed. It confirmed the suspicious findings previously seen on MR, demonstrating a hypervascular and heterogenous mass with some gross calcifications within (Figs. 4 and 5), and suspicious pelvic lymph nodes. There were no signs of metastatic lesions elsewhere.

The case was presented on a multidisciplinary case conference, and the working diagnosis of Currarino Triad was proposed. A CT guided core biopsy was performed.

Microscopic examination of the biopsy fragments showed a well differentiated neuroendocrine tumor composed of nests of polygonal cells with abundant light eosinophilic granular cytoplasm and round nuclei with “salt and pepper” chromatin. No mitotic figures or necrosis were observed. Immunohistochemically the neoplastic cells were positive for CD56 and synaptophysin but negative for chromogranin.



**Fig. 2 – MRI of the pelvis. Diffusion-weighted images (A) and ADC MAP (B) demonstrating restriction diffusion, with low ADC values, raising suspicion of malignancy.**



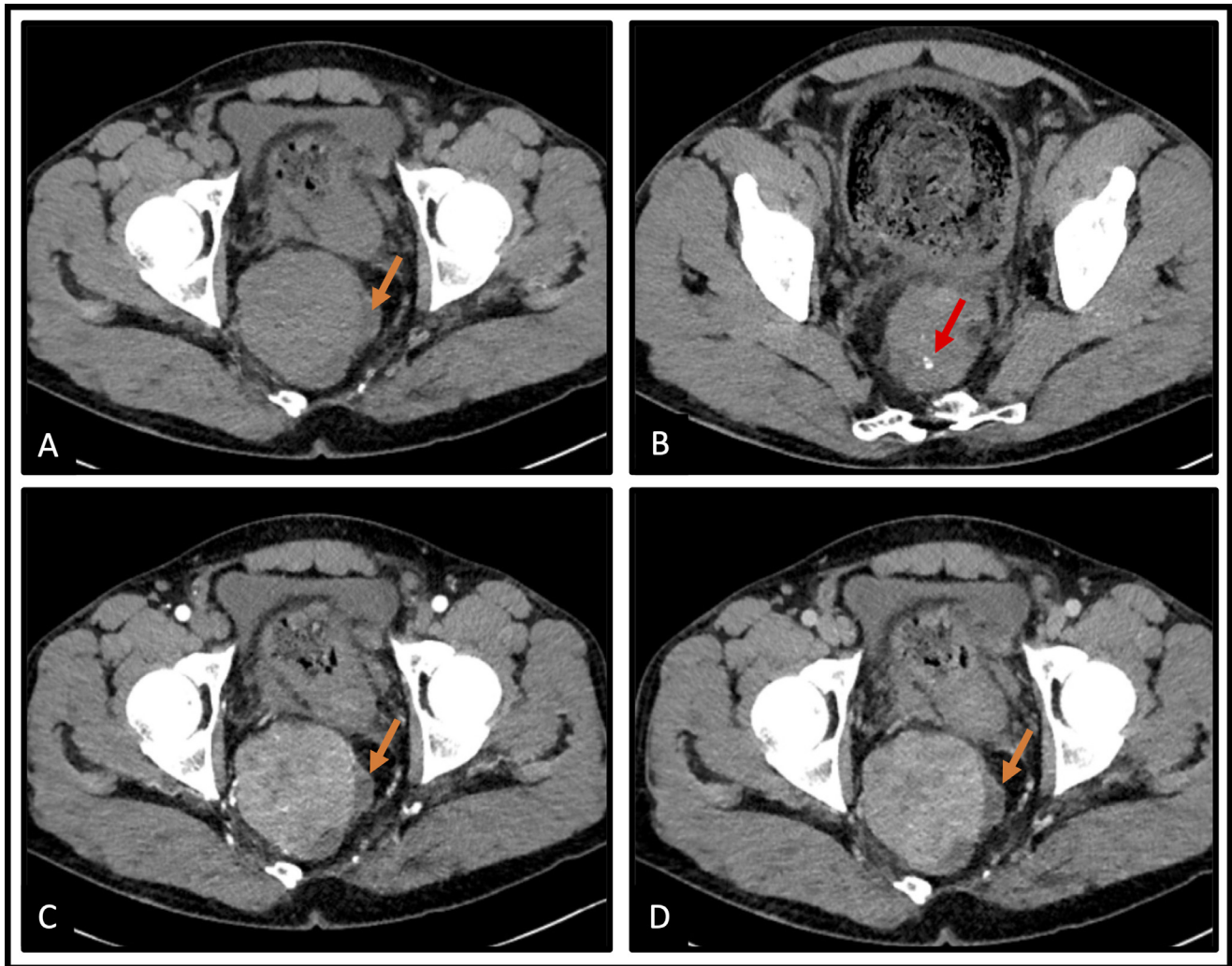
**Fig. 3 – MRI and CT of the pelvis. T1-weighted axial images post gadolinium (A) and CT axial images with iodated contrast (B) depicting a enhancing right iliac adenomegaly. Of note, right partial sacral agenesis is partially seen on these images, better depicted on Fig. 5.**

An abdominopelvic rectal amputation with a derivative colostomy were then performed.

The surgical specimen showed a mass with  $9.8 \times 8.7 \times 6.4$  cm localized in the retrorectal mesorectum with no apparent connection with the rectal wall (Fig. 6).

In our case the presacral mass was partially solid and cystic, the solid portion corresponded to a neuroendocrine tumor which was in close proximity to the cystic portion of

the lesion (Figs. 6 and 7), with focal areas of necrosis as well as lymphatic and perineural invasion. The cystic portion of the lesion had characteristics of a tailgut cyst (retrorectal cystic hamartoma)—multiloculated cyst lined by squamous and pseudostratified ciliated epithelium and disorganized bundles of smooth muscle in the cyst wall (Fig. 7). Although very rare, neuroendocrine tumors have been reported to arise in this type of lesion in Currarino syndrome [1–5].



**Fig. 4 – CT of the pelvis. Axial images without contrast (A and B), Axial Images with IV contrast on arterial phase (C) and venous phase (D). Hypervascular mass, with heterogenous contrast enhancement. As seen in MR there cystic lesions on the periphery that not enhance in relation with cystic contents (orange arrows). Some gross calcifications are also noted (red arrows in B). Of note, right partial sacral agenesis is partially seen on these images, better depicted on Fig. 5. (Color version available online.)**

The main differential diagnosis in this case would be a neuroendocrine tumor arising in presacral teratoma which has been described in the literature [6].

A teratoma is a germ cell tumor containing tissues derived from the 3 germ layers. After extensive sampling of the tumor it was not found any evidence of tissues derived from ectoderm (skin and skin appendages and neural tissue) or other tissues derived from mesoderm (bone, cartilage) which are commonly seen in this type of lesion [7]. Therefore our final diagnosis was of a well differentiated neuroendocrine arising in a tail gut cyst.

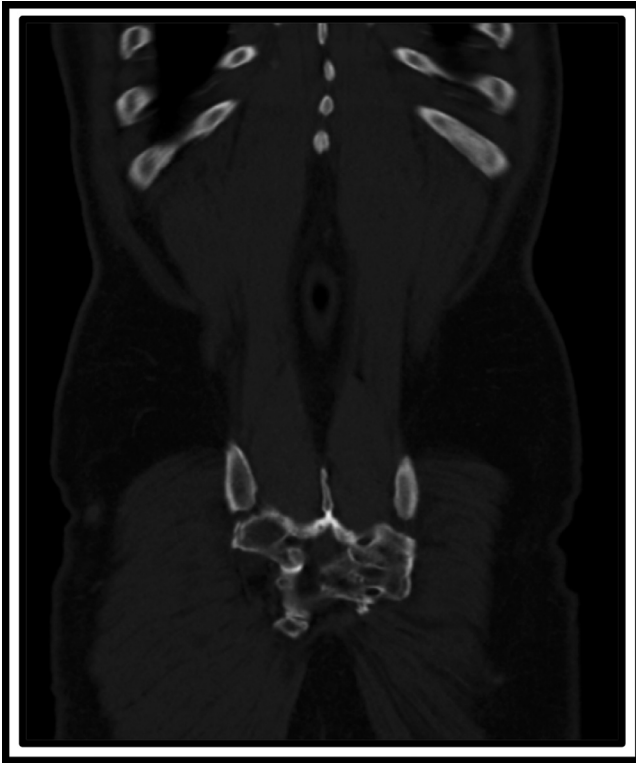
There were no major complications after surgery, and the patient went on a follow up surveillance plan, with no evidence of recurrence so far. About eighteen months after surgery, on a routine abdominal MR lesions suggestive of metastases were detected, affecting the liver, spleen and some bone structures. At the moment, the patient is on a chemotherapy palliative regimen.

## Discussion

The Currarino Triad was first described in 1981 by pediatric radiologist Guido Currarino and consists of sacroccygeal defect, presacral mass and anorectal malformation [8]. In the case presented, the patient presented the 3 characteristic features of the triad.

The most frequent sacroccygeal defect is hemisacrum or scimitar sacrum, but other segmental abnormalities may exist.

According to the literature, anterior meningocele is the most frequent type of presacral mass. Teratomas, enteric cysts, dermoid or epidermoid cysts, lipomas, hamartomas, or rectal duplications have also been reported. Malignant transformation of a presacral teratoma has been described but it is extremely rare [8].



**Fig. 5 – CT of the pelvis. Coronal images in a bone window reconstruction show partial right agenesis of the lower sacrum.**

The most common anorectal malformation is anorectal stenosis, followed by rectal stenosis or atresia, and anal ectopia, with or without fistula and intestinal duplication.

Although rare, malignancy, like neuroendocrine tumors, can arise in this type of lesion [9–11]. Other malignancies reported are malignant teratoma and leiomyosarcoma [12].

The actual incidence of this triad is unknown with less than 250 cases published in the literature. Due to intra and interfamilial phenotypic variability, it is believed that many cases may go unnoticed [13].

There is still an ongoing debate between the various theories of embryogenesis related to this disorder, but they all assume that it is caused by a caudal notochord malformation that leads to an aberrant secondary neurulation with incomplete separation of the ectodermal and endodermal layers in the developing embryo [8].

In its classical form, often described as the caudal regression syndrome, there are malformations of structures derived from the caudal region of the embryo, such as the urogenital system, the hindgut, caudal spine and spinal cord, and the lower limbs [14].

In 60% of cases of this syndrome there is a family history [6] and most are related to an autosomal-dominant trait. Mutations in the HLXB9 gene located in chromosome 7 (region 7q36) were found to be responsible for Currarino Syndrome.

As previously referred, our patient mentioned that some of his siblings also presented anorectal malformations, though unfortunately genetic testing was never performed to provide genetic correlation.

No obvious genotype-phenotype correlation has been identified. The phenotypic expression of this mutation is very variable and ranges from asymptomatic patients to patients with the complete triad [6]. There are incomplete forms of Currarino syndrome with absence of 1 or 2 characteristics, particularly in relatives of patients with Currarino syndrome, which is probably the case of the siblings and daughter of the patient in question [8].

The sacral bony defect is generally considered to be always present. There are also other anomalies which may be associated with the triad such as duplication of the urogenital tract, tethered cord, and different types of fistulas [15].

Better knowledge of the different clinical signs and symptoms of Currarino Triad at different stages of life enhances appropriate imaging and prompt diagnosis of this condition [8].

Currarino Triad may be diagnosed on prenatal ultrasound, and it is usually diagnosed in the first decade of life (more than 80% of cases). The incomplete form is usually diagnosed in adults [8]. Heterozygote patients can be asymptomatic and remain undiagnosed [8].

Clinically it presents as an imperforate anus at birth, intractable constipation from anorectal stenosis or extrinsic compression from a presacral mass, or as acute meningitis [8]. The chronic constipation from birth may simulate Hirschsprung disease and may require rectal biopsy for differentiation.

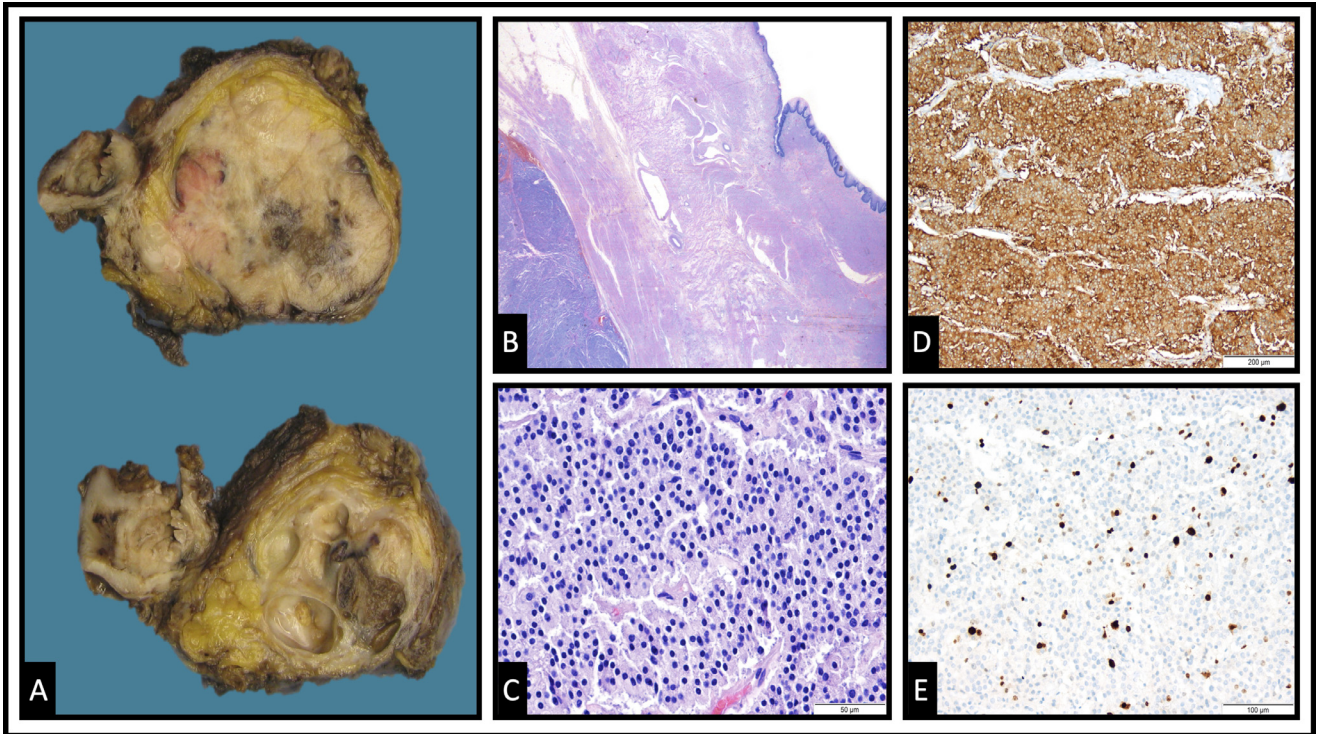
In pediatric patients, an adequate physical examination should be performed, to identify the type of ano-rectal malformation present. Digital examination of the rectum locates the distal rectal stenosis and, in some cases, the presacral tumor can be palpated [16].

In the terms of imaging spinal ultrasound is useful particularly in newborn and infants, and pelvic sonography is mandatory for all patients with Currarino Triad to exclude other associated urogenital anomalies [8].

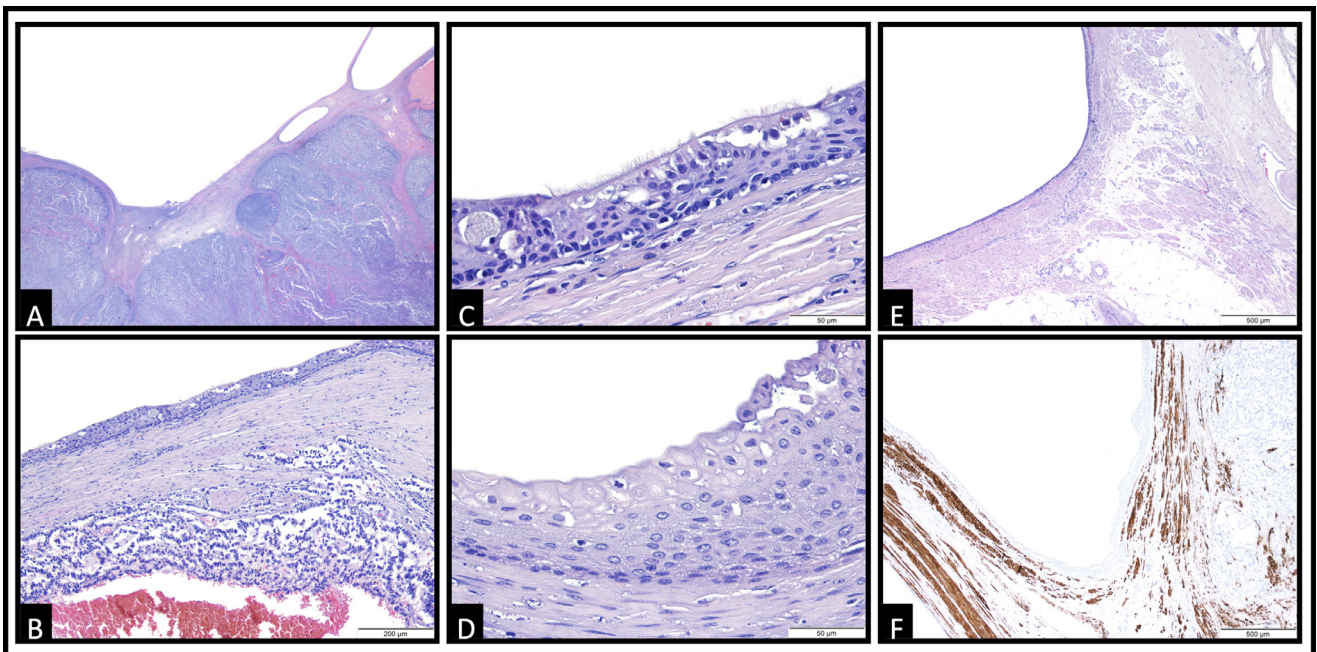
Sacral radiographs are useful for the diagnosis of sacral bone defects and the presence of the tumor, especially in newborns or infants.

CT and MRI are the imaging studies of choice to confirm the diagnosis and to better evaluate and exclude abnormalities of the spinal cord.

The surgical treatment of patients with Currarino Triad is focused on correcting the anorectal malformation and resecting the presacral tumor. There are some risks that should be taken into consideration, including nerve damage resulting in fecal incontinence and bladder dysfunction [11]. MRI is a very powerful tool, not only in the diagnosis, but also as a roadmap for planning the optimal surgical strategy in patients with myelomeningocele, syringomyelia, medullary cone or tethered spinal cord.



**Fig. 6 – Abdominopelvic rectal amputation with a multicystic and solid white mass localized in the retrorectal mesorectum (A) with no connection with the rectum (A, B). Histologically the solid portion of the lesion corresponded to a well differentiated neuroendocrine tumor (C). The neoplastic cells were positive for synaptophysin (D). The proliferative index measured by Ki-67 was 6% (E).**



**Fig. 7 – Multiloculated cystic spaces, in close proximity to the solid lesion (A, B), lined by squamous (D) and pseudostratified ciliated epithelium (C). Cyst wall with disorganized bundles of smooth muscle (E) which were immunohistochemically positive for desmin (F).**

## Conclusion

Currarino triad is a rare hereditary syndrome that includes anorectal malformation, sacrococcygeal defect and presacral mass. It usually manifests as persistent constipation in a child, but its clinical presentation is variable.

Early recognition of Currarino syndrome with adequate imaging assessment and surgical treatment may prevent serious complications such as meningitis, sepsis, urinary tract infection, and, rarely, malignant transformation of the presacral mass.

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