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SCIENTIFIC LETTERS

Tailgut cysts: Presentation of four cases☆,☆☆

Hamartoma quístico retrorrectal: presentación de 4 casos

Tailgut cysts are rare congenital tumours of the presacral space. The accepted origin is a developmental abnormality arising during the embryonic phase, developing from remnants of the primitive gut due to incorrect involution of the appendix or tail at the distal end of the caudal portion of the gut *(tailgut)*.[1]

This study looks at a series of 4 cases of tailgut cysts that have been surgically removed at our centre.

The clinical and pathological characteristics of these tailgut cysts are summarised in Table 1.

All patients were female, with a mean age of 47 years. The mean size of the lesions was 5.2 cm.

Two of the 4 patients had no local symptoms attributable to the lesion when first evaluated; one of these patients was referred after finding the lesion during a gynaecological check-up, and the other was being treated for an anal fistula. The other 2 patients reported experiencing pain for several months when sitting. In 3 of the cases, the rectal examination detected a well-defined, mobile retrorectal mass, while the other case had a soft, palpable, infracoccygeal mass.

All patients underwent magnetic resonance imaging (MRI), which showed lesions with well-defined borders, compression of the local rectal wall and no infiltration. The lesions were heterogeneous in appearance with multiple cystic components, showing areas of hypointensity or hyperintensity on T1-weighted sequences and hyperintense areas on T2-weighted sequences; only one lesion had a more complex morphology, with areas with solid and cystic components (partial hyperintensity on T1 and extreme hyperintensity on T2). In this case, an endorectal ultrasound was performed, with similar findings.

All patients were operated on by the same surgeon using a posterior approach with transsacral resection (Kraske technique); 3 patients also had a partial coccygectomy while 1 had a complete coccygectomy, with additional removal of the last sacral vertebra. Resection was always macroscopically complete, with no intraoperative injury to muscle fibres of the anal sphincter or significant blood loss. Primary closure was performed on all patients, and biological mesh was used in 2 cases, which was sutured to the levator ani muscle.

Histological examination revealed that the lesions had multiple cystic components; different subtypes were identified in the epithelium, along with sub-acute or chronic inflammatory changes in some samples. A solid component (coinciding with the complex solid-cystic lesion observed previously on MR images) was detected in one case, with the incidental finding of a neuroendocrine tumour (NET) measuring 1.5 cm, with Ki-67 labelling index of less than 2%; it was classified as grade 1.

The mean length of hospital stay was 4 days, with a mean follow-up of 8.5 months. No complications were detected during the immediate post-operative period or during the follow-up period, and there were no reports of disease recurrence.

Middledorpf first described a congenital presacral cyst in 1885,[2] but it was not until 1988 that Hjermstad and Helwig speculated that they originate from remnants of the primitive gut, providing the largest case series to date, with 53 cases.[1] More than 200 cases of tailgut cysts have been recorded in international literature. Most publications mention isolated cases, but some series are larger and from reference centres (Table 2).

Tailgut cysts are most common in middle-aged women, with a variable clinical presentation, and such cysts are diagnosed incidentally in 50% of cases.[1,3] In the event of a superinfection, symptoms of drainage or swelling compatible with anal fistula may appear, which is a common cause of initial delays in diagnosis. Physical examination usually reveals a well-defined retrorectal tumour that is compressing and displacing the posterior rectal wall without infiltrating it.[4]

It is important to perform an accurate differential diagnosis of all masses of the presacral space (including developmental cysts, teratomas, sacral chordomas, neural tube defects or neurogenic tumours) in order to prevent a higher rate of recurrence (up to 60% in some series) or other complications.[1,4]

The use of endorectal ultrasound may be useful for characterising involvement of the rectal wall, although the current trend is to perform more cost-effective imaging tests, primarily MRI.[5] Pre-operative biopsy is not currently recommended due to the possibility of local tumour spread; it also has no significant diagnostic value.[6]

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Table 1 Clinical data for cases of tailgut cysts.

Case number	1	2	3	4
Gender and age (years)	F 41	F 56	F 35	F 56
Symptoms	No	Pain when sitting	Pain when sitting	No
Physical examination	Retrorectal tumour	Infracoccygeal mass	Retrorectal tumour	Retrorectal tumour
Imaging test	MRI	MRI	MRI, transvaginal ultrasound	MRI, endorectal ultrasound
Initial diagnosis	Developmental cyst	Presacral lesion, undetermined nature	Developmental cyst/teratoma	Tailgut cyst
Size (cm)	$7 \times 7 \times 4$	$3.5 \times 3 \times 2$	$6 \times 5 \times 3$	$4.5 \times 2.5 \times 2$
Surgical technique	Posterior approach (Kraske)	Posterior approach (Kraske)	Posterior approach (Kraske)	Posterior approach (Kraske)
Follow-up (months)	15	6	6	7
Malignancy	No	No	No	Neuroendocrine
Recurrence	No	No	No	No

F: female; MRI: magnetic resonance imaging.

Table 2 Main published series that include tailgut cysts.

Series	Year	n	Mean age (years)	F	Technique	F/U (months)	Recurrence	Malignancy
Johnson AR et al.	1986	5	32	4	U	U	U	0
Hjermstad BM et al.	1988	53	35	41	Posterior approach (22) Transanal (9) Abdominoperineal resection (1) Unknown (21)	10	4	1
Hannon J et al.	1994	4	37.8	4	Transsacral (2) Combined abdominosacral (2)	36	1	0
Prasad AR et al.	2000	5	50.4	4	U	U	U	2
Singer MA et al.	2003	4	42.5	3	Parasacrococcygeal	U	U	0
Lev-Chelouche D et al.	2003	12	37	12	Posterior approach (9) Anterior approach (3)	54	0	0
Yang DM et al.	2005	5	43.6	5	U	U	U	0
Buchs N et al.	2007	10	38.4	10	Transperineal (6) Parasacrococcygeal (4)	60	1	0
Aflalo-Hazan V et al.	2008	11	40.8	8	Posterior approach (8) Combined abdominosacral (2) Abdominoperineal resection (1)	U	U	1
Woodfield JC et al.	2008	8	30	8	U	U	1	0
Grandjean JP et al.	2008	16	U	U	Posterior approach	U	1	0
Mathis KL et al.	2010	31	52	28	Posterior approach (20) Anterior approach (9) Combined abdominosacral (2)	U	1	4
Baek SW et al.	2011	8	40.4	8	Posterior approach	18	1	0
Rosa G et al.	2012	5	32	5	Transperineal (3) Transanorectal (2)	140	3	0
Chéreau N et al.	2013	28	U	U	U	U	U	6
Patsouras D et al.	2015	17	35	15	Posterior approach (16) Combined abdominosacral (1)	13	1	1

U: unknown; F: female; F/U: mean follow-up.

The treatment of choice for tailgut cysts is complete surgical excision with tumour-free margins, which is essential for a good outcome, especially in the event of possible superinfection or malignant transformation.[1,3] The chosen approach must be safe and effective. The posterior or transperineal approach is recommended in the case of accessible masses that do not extend beyond the third sacral vertebra, while bulky tumours in an anterior position or higher will require an abdominal or combined approach.[7] The Kraske technique allows good visualisation of the rectal wall and preservation of sphincter function, with low associated morbidity and mortality rates.[4,7]

The risk of malignant transformation reported is around 7%,[1,3] but may even reach up to 40%.[6] Tailgut cysts have been observed to transform into adenocarcinomas and NETs, which have a highly favourable prognosis compared to other malignancies. The presence of NET is unusual, and has been described in only 20 cases to date.[8] It is suggested that these tumours are possibly associated with hormones and oestrogen receptors as the potential therapeutic target, which is yet to be studied.[9]

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Chronic diarrhea: The first symptom of a metastatic medullary thyroid carcinoma☆

Diarrea crónica: el primer síntoma de un carcinoma medular de tiroides metastásico

Chronic diarrhoea (lasting for more than 4–6 weeks) is relatively common among the general population. The main causes according to pathogenesis are shown in Table 1. It is essential to take a detailed medical history and perform a thorough physical examination to adequately manage this disorder, since this makes it possible to screen for patients requiring more detailed tests and examinations.

This article looks at the case of a 36-year-old male with no notable medical history who started with diarrhoea with 3–4 stools a day, some during the night, without blood, mucus or pus. He had no abdominal pain, nausea, vomiting or fever. His symptoms were not related to meal times, and he showed no improvement, despite a lactose-free diet. On physical examination, his abdomen was soft, non-tender and with no palpable masses or organomegaly.

A full blood count and stool culture were ordered, both of which were within the normal ranges. Due to the persistence of symptoms for 4 months and the presence of nocturnal bowel movements, the patient was assessed by the digestive diseases department. A colonoscopy and endoscopy were ordered, revealing only the presence of erosive gastritis; the patient was found to be *Helicobacter pylori* positive. The patient took antibiotic treatment for *H. pylori* eradication, but symptoms persisted.

A broader study was ordered including an abdominal ultrasound. Numerous nodular hyperechoic liver lesions were observed that are compatible with metastasis. Therefore, a CT scan of the chest and abdomen was performed. The CT scan showed the same liver nodules, osteoblastic bone lesions and a thyroid nodule, all of which is indicative of a neuroendocrine tumour (Fig. 1).

More blood tests, including tumour marker tests, were ordered, showing very high calcitonin (38,653 ng/l, normal range 0–18.2) and carcinoembryonic antigen levels (1065 ng/ml, normal range 0–5.4). For better characterisation of the thyroid nodule, an ultrasound of the neck was performed which showed a lesion in the left thyroid lobe measuring 37 mm with ultrasound characteristics of malignancy. It also showed the presence of multiple swollen lymph nodes in the central, ipsilateral and contralateral compartments of the neck.

A biopsy of this nodule and one of the lymph nodes was compatible with medullary thyroid carcinoma (MTC) and, therefore, RET proto-oncogene mutation and urinary catecholamine and metanephrine tests were ordered due to the possibility of pheochromocytoma in the event of multiple endocrine neoplasia. The genetic study revealed a heterozygous RET mutation at pC618R. Urinary catecholamines and metanephrines were normal, which ruled out the concomitant presence of pheochromocytoma.

In view of these results, it was decided to perform a total thyroidectomy and removal of lymph nodes from the neck in order to prevent local symptoms and facilitate future

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