CASE REPORT

Unusual presentation for a tailgut cyst

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SUMMARY

A 67-year-old man with a slow-growing, asymptomatic perianal lesion was presented to the colorectal clinic. Physical examination and radiological investigation were suggestive of an externally prolapsing colonic polyp. However, intraoperative findings of a cystic lesion containing a thick brown substance questioned the initial diagnosis. After complete surgical resection of the encapsulated lesion, histological examination revealed a tailgut cyst (cystic hamartoma). These uncommon lesions originate from an embryological remnant of the hindgut. They are generally benign but may have extremely rare malignant potential which warrants complete surgical resection. Throughout the literature, they are described to present with various symptoms and may be found in several anatomical locations, most commonly within the retrorectal space. In very rare instances, with only two other documented cases, they can be found externally at the anorectal junction. Although very rare, an anal tailgut cyst should be taken into account in differential diagnosis.

BACKGROUND

The rarity of perianal cystic hamartomas (tailgut cysts) means that they are often overlooked on the differential diagnosis list. Throughout the literature they have been described in numerous anatomical locations and with a variety of symptoms, which are similar to more common conditions. This makes correct diagnosis problematic. An externally protruding lesion which presented as a cosmetic concern is a unique presentation for these uncommon lesions.

In addition, this interesting case highlights how a seemingly straightforward procedure can reveal a more technically challenging operation than initially anticipated. As a surgeon, being prepared for such eventualities is integral to ensure optimal outcomes from surgery. In order to do this, the surgeon must have good awareness and broad knowledge of the possible differentials in order to amend the initial surgical plan. In this case, removal of the cystic capsule was required to prevent potential recurrence of the lesion while care was taken not to damage the integrity of the external anal sphincter.

Case presentation

A 67-year-old man was referred by his general practitioner to the colorectal clinic with an asymptomatic perianal lump that had been slowly growing in size for the past 20 years. He denied any pain, irritation or bleeding. Digital rectal examination revealed a 5 cm × 2 cm polypoid lesion at the 6

o'clock position at the anal verge. The rest of the physical examination was unremarkable. He had a medical history of hypertension and hypercholesterolaemia for which he took simvastatin and amlodipine. There was no family history of bowel cancer. He was an independent man who lived at home with his wife.

Investigations

The patient was investigated with routine laboratory blood tests, rigid and flexible sigmoidoscopy, proctoscopy and an MRI scan of the pelvis. Blood tests were all within normal limits (haemoglobin 155 g/L, white cell count $6.3 \times 10^9/L$, creatine 65 $\mu\text{mol/L}$, estimated glomerular filtration rate $>90 \text{ mL/min/1.73 m}^2$). A rigid sigmoidoscopy to 10 cm and proctoscopy did not reveal any abnormality except for an external anal polyp approximately 6 cm in size. Flexible sigmoidoscopy did not reveal any mucosal abnormalities. A pelvic MRI scan (figure 1) described a large smooth polyp measuring 6 cm in maximum diameter which appeared to be arising from a stalk attached to the 6 o'clock position of the lower anal canal. There was no other abnormality within the anal canal. The rectum and mesorectum were normal with no fatty stranding or adenopathy. Macroscopically it revealed an intact unilocular cyst measuring 42 mm × 25 mm × 15 mm (figure 2). The wall measured 2 mm thick and contained a thick dark brown fluid. Microscopically (figure 3), it showed a simple cyst, surrounded by a thin fibrous capsule and partly lined by stratified non-ciliated cuboidal and squamous epithelium with an innermost layer of apocrine-type cells (periodic acid-Schiff positive). The denuded areas showed florid foreign body-type granulomatous inflammation composed of scattered pigment-laden histiocytes and multinucleated giant cells. Immunohistochemistry showed positive staining of the epithelium with cytokeratin 7 and carcinoembryonic antigen. Cytokeratin 20 was negative. No obvious dysplasia or malignancy was seen.

Differential diagnosis

Initially the primary differential diagnosis for this lesion, based on clinical examination and imaging, was a protruding colonic polyp. However, during surgical resection the lesion was found to contain a thick brown substance within what appeared to be a well-defined membranous capsule. This finding made dermoid cyst, epidermoid cyst, anal gland cyst, sacrococcygeal teratoma, tailgut cyst (rectal cystic hamartoma) or thrombosed haemorrhoid other possibilities.

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Figure 1 MRI scan in the sagittal plane showing a large cystic lesion protruding from the anal canal.

Treatment

Examination under general anaesthesia and resection of anal lesion. The resected anal lesion (figure 2) was sent for histology.

Outcome and follow-up

The patient was followed up routinely in the colorectal clinic 8 weeks postoperatively. The patient was asymptomatic and the wound had almost completely healed.

DISCUSSION

Retrorectal cystic hamartoma (tailgut cyst) is a rare congenital lesion that derives from remnants of the embryological postanal gut.¹ They are more common in women with the female to male ratio at 5:1.^{2,3} The most common location for a tailgut cyst is the retrorectal space³ which is rich in structures and can be the source of a wide variety of congenital cysts, classically divided into enteric and neuroenteric cysts. The clinical presentation

Figure 2 Postoperative image of the resected specimen measuring 4.2 cm x 2.5 cm.

Figure 3 Microscopy of the resected anal lesion (H&E x200 magnification).

ranges from asymptomatic to a variety of non-specific symptoms due to the compression of surrounding structures.² It can also present as acute urinary retention, constipation or pelvic abscess.² They have an extremely rare potential of malignant transformation with adenocarcinoma or carcinoid being the most common according to the literature.^{1,4} They can have different cellular pathology and the diagnosis is usually made by histological confirmation.⁴ The presentation of a tailgut cyst in the anal region as a prolapsing tumour is very rare with only two documented cases in the literature.^{5,6} Diagnosis of a tailgut cyst requires a high index of suspicion. Due to the risk of malignant transformation, complete surgical resection is warranted and follow-up is compulsory due to risk of recurrence, formation of chronic abscesses and recurrent perianal fistulas.^{1,3}

Learning points

- ▶ Retrorectal cystic hamartomas (tailgut cyst) are rare causes of externally protruding anal lesions,
- ▶ They are believed to derive from the embryonic hindgut and have an extremely rare malignant potential.
- ▶ Planning and awareness for the unexpected is crucial to optimising surgical outcomes.
- ▶ Cystic lesions require removal of the capsule to reduce the risk of recurrence, which can be technically challenging in the perianal region.

Contributors DH was involved in planning and conception, performed the operation under supervision, wrote some parts of the case, performed an online research, design and revising. JW wrote some parts of this case report, performed an online research and was present during the surgery, involved in concept, design and revising. AC was the responsible consultant, he was involved in planning, conduct, conception and design and revising the article. FK was the histopathologist who provided us with the final diagnosis, involved in interpreting and diagnosing the histology slides, and also was involved in design and revising the article.

Competing interests None declared.

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