CASE REPORT

An elusive cause of perianal pain in a patient with Hirschsprung's disease

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SUMMARY

A 23-year-old woman presented to the outpatients department with symptoms of acute onset perianal pain of 2 weeks' duration and constipation. She was a known case of Hirschsprung's disease and was operated on in infancy. Rectal examination ruled out acute fissure or perianal sepsis, and subsequent colonoscopy was normal. Because of persistent symptoms, she was evaluated further by imaging which revealed a presacral lesion closely abutting the nerve roots, suggesting the possibility of a nerve sheath tumour. The tumour was excised by the perineal route, and surprisingly the histopathology report revealed an epidermoid (sebaceous) cyst. Presacral tumours are very rare lesions which most colorectal surgeons never come across in their careers. Tumours range from indolent, to benign, to more aggressive malignant lesions. Usually the symptoms are non-specific and surgical excision remains the best treatment. Multidisciplinary team work is needed for most of these cases.

BACKGROUND

The presacral (retrorectal) space is a potential space between the mesorectum anteriorly and the sacrum posteriorly, and is a common site of embryological remnants.¹ Tumours arising in this space are very rare and can range from simple benign cysts to infiltrating malignancies. Developmental cysts are the most common retrorectal lesions.² Symptoms are variable, depending on the type of tumour, size, mass effect and associated complications.² Clinical examination may miss the tumour if it is very soft. Surgical excision is the ideal treatment prior to which thorough evaluation is needed. The surgical approach depends on the location of the tumour and invasion of the surrounding structures.

We present a rare case of presacral epidermoid cyst in a young woman diagnosed with Hirschsprung's disease and treated in childhood, who presented with symptoms mimicking acute fissure in ano without any palpable mass.

CASE PRESENTATION

A 23-year-old woman presented to the outpatients department with complaints of sudden onset pain in the perianal region and constipation of 2 weeks' duration. Pain was pricking in nature with severe intensity, and was aggravated by passing stools. The patient was a known case of Hirschsprung's disease, diagnosed at birth for which she had undergone diversion sigmoid loop colostomy.

The Duhamel procedure was performed at 6 months of age. The patient developed adhesive intestinal obstruction post surgery for which a diversion transverse colostomy was done, which was reversed at 1 year of age. There was no other significant history. The patient was slimly built and abdominal examination revealed a right hypochondrium and left iliac fossa scar. Rectal examination revealed a normal sphincter tone with the rectum loaded with faeces. There was no fissure in ano or signs of perianal sepsis, or any palpable mass.

INVESTIGATIONS

X-ray of the abdomen showed no evidence of intestinal obstruction. Colonoscopy was normal. As the patient did not improve with conservative management, contrast enhanced CT of the abdomen and pelvis was undertaken which showed a well defined hypodense lesion, 4.2 cm×4.9 cm×4.7 cm, in the presacral region on the left side in close relation to the lower sacral foramina S4 and S5 with mild scalloping of the adjacent sacrum (figure 1). MRI of the pelvis was done to further characterise the lesion, which demonstrated a hypointense lesion in T1 and heterogeneously hyperintense lesion on T2 images (figure 2). The lesion was displacing the anal canal anteriorly and to the right side. Fat planes were preserved between the lesion and the rectum anteriorly while posteriorly the lesion was closely abutting the left sacral nerve roots (figure 3).

DIFFERENTIAL DIAGNOSIS

The preoperative diagnosis was of a nerve sheath tumour causing her symptoms.

Figure 1 CT scan of the pelvis showing a hypodense lesion in the presacral region on the left side.

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Figure 2 MRI of the pelvis (T2 weighted), showing a heterogeneously hyperintense lesion with preserved fat plane between the lesion and the rectum.

TREATMENT

The case was discussed in a multidisciplinary team meeting involving neurosurgery, orthopaedics and medical oncology, and it was decided to proceed with surgery. The lesion was reached by a posterior approach, placing a left perianal incision and entering the presacral space. A soft cystic mass was felt extending from the 5 to 10 o'clock position. By gentle dissection, the cyst was removed in toto after ligating the pedicle. Interestingly, a cut section showed pultaceous material and HPE proved it to be an epidermoid cyst (figures 4, 5).

OUTCOME AND FOLLOW-UP

The patient's symptoms subsided postoperatively and she was followed-up regularly for more than a year without any recurrence.

DISCUSSION

The presacral space is bounded by the mesorectum anteriorly, the presacral fascia posteriorly, the endopelvic fascia laterally, the peritoneal reflection superiorly and Waldeyer's fascia

Figure 4 Cut specimen with the cyst wall and its contents.

inferiorly.³ It contains totipotent cells from which various tumours develop.¹ The normal contents include loose connective tissue, the median sacral artery, the superior rectal vessels and branches of the sympathetic and parasympathetic nervous system.¹ The true incidence of tumours is unknown, while that reported in the literature is between 1.4 and 6.3 cases/year.⁴ Young women are most commonly affected, with a sex ratio of 5:1. Benign tumours are most common in women while malignant tumours have a slight male preponderance.⁴

Uhlig and Johnson⁵ first classified the lesions into congenital, neurogenic, osseous, inflammatory and miscellaneous. Dozois *et al*⁶ subcategorised them into benign and malignant within this broad group. Congenital tumours are the most common, accounting for 55–70% of lesions, and include developmental cysts, chordomas and anterior sacral meningoceles. Developmental cysts comprise 60% of the lesions and can originate from all three embryonic germ layers. Developmental cysts are divided into epidermoid cyst, dermoid cyst, enterogenic cyst, tail gut cysts and teratoma, based on the cell layer of origin.⁶

Epidermoid and dermoid cysts result from abnormal closure of the ectodermal tube. Dermoid cysts can be differentiated from the former by the presence of skin appendages, such as sweat glands, hair follicles and sebaceous cysts.⁷ Post anal dimple or sinus may be present if the cysts communicate with the skin. Enterogenous (duplication) cysts are in continuity with

Figure 3 MRI of the pelvis (T2 weighted) showing the close relation of the lesion to the sacral foramina and nerve roots.

Figure 5 Photomicrograph of the cyst wall confirming the epidermoid cyst with absence of skin appendages.

the rectum. They are lined by mucosa and have a definite muscular wall with a myenteric plexus. Malignant degeneration has been reported in these cysts.⁷ Tail gut cysts (cystic hamartomas) are circumscribed, unencapsulated and multilocular, and are lined by transitional epithelium in addition to squamous epithelium. Teratomas arise from totipotent cells and are most common in infancy. They may be solid or cystic and have malignant potential.⁷

Currarino triad has been described, which includes anorectal malformation, sacral agenesis and presacral tumours, and this condition can be associated with Hirschsprung's disease.⁸ Following an extensive literature search, we were unable to find out a case report of presacral epidermoid cyst in a patient with Hirschsprung's disease. We think this is likely the first report of the two conditions occurring in the same patient.

Chordomas are the most common malignant presacral tumours which arise from the notochord. Anterior sacral meningocele form due to herniation of the dural sac through a defect in the sacrum, and can present with headache, recurrent meningitis, constipation, urinary symptoms and low back pain. Inadvertent biopsy will lead to life threatening meningitis.⁹

Neurogenic tumours originate from the peripheral nerves and are the second most common presacral tumours (10%). The majority (85%) are benign, and are slow growing and asymptomatic. Osseous tumours (5–10%) include a spectrum of tumours arising from bone, cartilage, fibrous tissue or marrow, and are rapidly growing. Inflammatory lesions, including foreign body granuloma (barium leak, surgical sutures) and infective processes originating from the abdomen (perforative diverticulitis) or perianal region are no longer considered presacral tumours as they do not originate in the presacral region. Miscellaneous tumours account for 10–25% and includes metastasis, lymphoma, fibrosarcoma, liposarcoma, malignant fibrous histiocytoma, lymphangioma, lipoma, leiomyoma and haemangioma.¹⁰

Symptoms are variable and depend on the volume of the cyst, location, mass effect, presence or absence of infection, and benign or malignant nature of the lesion. More than 80% of benign lesions are asymptomatic whereas only <10% of malignant lesions are symptom free.¹¹ Symptoms range from constipation, rectal fullness and tenesmus, to rectoragia, hypogastric pain, urinary frequency, dysuria and sciatic-type pain. However, rectal bleeding is common in duplication cysts.² The most common complications of congenital cysts are infection, which is chronic in half of cases, abscess and fistula formation. Malignant degeneration is noted (7%) with adeno and squamous cell carcinoma being the most common histological type. More than 95% of tumours can be felt on digital rectal examination.⁴ However, in our patient the cyst was not palpable on digital rectal examination.

Plain X-ray may demonstrate sacral destruction (chordoma), calcification in the mass (teratoma) or pathognomonic signs such as 'scimitar sign' (anterior sacral meningocele). Contrast enhancing CT or MRI of the pelvis is widely used and can differentiate solid from cystic lesions, vascular, adjacent organ involvement and bony destruction. In our patient, imaging showed a lesion arising from the nerve sheath and was misleading. Endoscopic ultrasound and lower gastrointestinal scopy will be helpful in further characterising the lesion.¹² Routine preoperative biopsy is contraindicated and should be done for solid or heterogenous lesions if no surgical intervention is planned or neoadjuvant therapy is to begin. The transperineal or presacral approach is the ideal route.¹²

Surgery is the main line of management. The approach and the extent of resection depends on location, nature, size of the

lesion and the involvement of the sacrum, pelvic side wall or adjacent viscera.¹³ The three common approaches are anterior (transabdominal) for tumours above S3, posterior (perineal) for lesions below S3 and combined (abdominoperineal) if sacrococcygeal excision is needed.¹ Although small lesions can be easily excised, larger lesions need a multidisciplinary team involving the colorectal surgeon, neurosurgeon, orthopaedic surgeon, vascular surgeon, plastic surgeon as well as medical and radiation oncologists. The lowest sacral nerves S4, S5 and the unilateral S3 nerve can be sacrificed without neurological deficit.¹⁴ Removal of the lower portion of the sacrum (below S3) will not result in pelvic instability.¹⁴ Complication of surgery include bleeding, urinary or faecal incontinence, abscess or fistula. Long term results after excision of a benign tumour are good unless incompletely removed, which results in recurrence. The outcome after excision of a malignant lesion depends on the biology of the tumour and the extent of the resection. The 5 year and 10 year survival rates for sacral chordoma in the NSEER database are 74% and 32%, respectively.¹

In conclusion, presacral tumours, although rare, can present with acute perianal pain without any palpable mass per rectum, mimicking a benign anal pathology. Pelvic imaging is mandatory in a patient with persistent symptoms. A wide variety of lesions can occur in the presacral space and most are congenital. Accurate imaging is needed to assess the location of the lesion, the extent of involvement of the surrounding structures and to decide on the surgical approach. A multidisciplinary team needs to be involved during surgery.

Patient's perspective

I developed sudden pain in the anal region, more so during defecation. I was unable to tolerate the pain. I thought it was due to my previous surgery and was very upset. I was reassured by my doctors and I accepted surgery. After the operation I feel no pain and am very happy.

Learning points

- ▶ Presacral tumours are extremely rare causes of acute perianal pain.
- ▶ Lesions may not be palpable per rectum.
- ▶ Both benign and malignant tumours can occur although congenital lesions are the most common.
- ▶ Preoperative imaging guides the management and approach.
- ▶ Multidisciplinary team work is needed.

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