Learning from errors

Presacral schwannoma treated as irritable bowel syndrome

Triq Habib,¹ Jamal T Hamdi,² Waleed Hussain,³ Wail Almiadini,⁴ Khaled Hamdi,⁴ Abdul Majid Wani,⁵ Najah Rashid Al Zeyani⁵

¹Department of Surgery, Hera General Hospital, Makkah, Saudi Arabia

²Umm Al-Qura University, Makkah, Saudi Arabia

³Department of Medicine, Hera General Hospital, Makkah, Saudi Arabia

⁴Hera General Hospital, Makkah, Saudi Arabia

⁵Department of Emergency Medicine, Hera General Hospital, Makkah, Saudi Arabia

Correspondence to Abdul Majid Wani, dr_wani_majid@yahoo.co.in

Summary

Presacral tumours represent a heterogeneous group of predominantly benign and occasionally malignant neoplasms. These tumours, though rare, frequently present either incidentally or with vague symptoms. Schwannomas of the presacral region are one variant described as benign tumours of neurogenic origin. The case of a large presacral schwannoma in a 26-year-old man, who was treated for irritable bowel syndrome for 4 years, is presented. The patient presented with intermittent constipation, a feeling of incomplete evacuation of the bowel and vague abdominal discomfort relieved by defecation. The symptomatology worsened and constipation became frequent, and the patient experienced increased urinary frequency. Baseline investigations were normal and ultrasonography of the abdomen revealed a pelvic mass, which on CT scanning was revealed to be a large retrorectal mass. The tumour was resected and histology revealed it to be a schwannoma. This unique case is presented to emphasise that irritable bowel syndrome must be a diagnosis of exclusion, especially if atypical symptoms are present.

BACKGROUND

We decided to submit this case report because of the misdiagnosis of the patient for almost 4 years, and the patient's presentation with constipation and not dorsal root pain, from which the tumour was resected.

CASE PRESENTATION

A 26-year-old Saudi man presented with constipation and increased urinary frequency for a duration of 4 years, beginning with intermittent constipation and vague abdominal pain relieved by defecation. The patient was

Figure 1 CT scan showing the tumour in the presacral area.

Figure 2 Pedunculated tumour as seen during surgery.

Figure 3 The tumour cut open, with yellowish–grey cut surface.

diagnosed as having irritable bowel syndrome in view of a normal physical examination. The patient took symptomatic medicine (mebaverine, simethicone) but the constipation increased in duration, and stool volume and

consistency changed. The patient also reported increased frequency of urination, which prompted one of the doctors to order baseline investigations and ultrasonography of the abdomen. The latter revealed a retrorectal mass. The

Figure 4 Histopathological examination revealing mature spindle cells, stroma and sparse vascularity.

patient was hospitalised, and a digital rectal examination and colonoscopy were normal as were the complete blood counts, serum chemistry and urine analysis. A CT scan revealed a huge homogenous mass, well encapsulated and without invasion of adjacent structures (figures 1 and 2). The patient was scheduled for surgery and a large tumour with intact capsule was resected from its pedicle (figure 3; video 1). The tumour was 10 × 11 cm in dimensions and when cut open was yellowish-grey in colour with few mucoid foci (figure 4). Histopathology revealed features consistent with a benign tumour/schwannoma. The patient was discharged home 1 week later and is doing well on follow-up, with improvement in constipation and without any neurological deficit. The patient is scheduled for annual MRI scans to rule out any recurrence.

Video 1

INVESTIGATIONS

A CT scan was performed.

DIFFERENTIAL DIAGNOSIS

- ▶ Neurofibroma
- ▶ Neurolemmoma
- ▶ Ependymoma
- ▶ Ganglioneuroma
- ▶ Neurofibrosarcoma

TREATMENT

Surgical resection was performed.

OUTCOME AND FOLLOW-UP

The patient improved and is on regular follow-up.

DISCUSSION

Presacral tumours are rare and can be challenging to diagnose and treat. Because the retrorectal space contains multiple embryological remnants derived from various tissues, the tumours that develop in this space are heterogeneous. Lesions are classified as congenital, neurogenic, osseous, inflammatory or miscellaneous. Although treatment depends on diagnosis and anatomic location, most retrorectal lesions will require surgical resection.¹ Symptoms on presentation can be back pain, constipation, neurological symptoms or defecatory dysfunction due to compression, and some cases are detected incidentally.^{2,3} Rectal examination is useful in detecting retrorectal masses.⁴ Misinterpretation of symptoms and diagnosis of irritable bowel syndrome has been reported in many pelvic pathologies,^{5,6} and even on strict adherence to Manning and Rome criteria I and II for case definition of irritable bowel syndrome the percentage of concordance is low.⁷ The diagnosis of irritable bowel syndrome is a diagnosis of exclusion, especially when red flag signs are present. In this case the delay of 4 years from presentation to diagnosis is unacceptable for a mass of such a size, especially when urinary symptoms were also present. Diagnostic difficulty arises in cases that are asymptomatic. Our patient was symptomatic, with urinary symptoms in addition to those of defecation, and even a properly performed digital rectal examination would have picked up the mass.

Retrorectal or presacral masses are classified as per the tissue of origin and pathology (table 1). Neurogenic lesions typically arise from peripheral nerves and represent about 10% of retrorectal tumours. These tumours include neurofibromas and sarcomas, neurilemmomas, ependymomas, schwannomas and ganglioneuromas. Ependymomas are most common. Pain and neurological dysfunction are

Table 1 Classification of retrorectal masses

Classification	Tumour type
Congenital	Developmental cyst
	Epidermoid cyst
	Dermoid cyst
	Teratoma
	Chordoma
	Anterior meningocele
Neurogenic	Rectal duplication
	Adrenal rest tumour
	Neurofibroma
	Neurolemmoma
	Ependymoma
	Ganglioneuroma
Osseous	Neurofibrosarcoma
	Osteoma
	Osteogenic sarcoma
	Sacral bone cyst
	Ewing tumour
	Giant cell tumour
Inflammatory	Chondromyxosarcoma
	Granuloma
	Perineal abscess
	Pelvic abscess
	Fistula
	Chorionic granuloma
Miscellaneous	Metastatic disease
	Lymphangioma
	Desmoid tumour
	Leiomyoma
	Fibrosarcoma
	Endothelioma

often presenting symptoms and are related to the route of the involved nerve. Tumours arising from the sheath of lumbar root four and descending into the pelvis as a pedunculated mass have not been reported in the literature to date. Our patient had no pain related to the nerve root of origin, probably because of support by pelvic structures and no stretching of the root. This was more evident postoperatively as the defecatory dysfunction, which was pressure related, improved, and there was no neurological deficit related to the root of origin. Radical resection is usually required, but overall survival seems to be good.⁸ Presacral schwannomas with degeneration and

calcification have been reported, with favourable outcome after resection.⁹

Therefore, review of symptomatology and appearance of red flag signs in a case of irritable bowel syndrome must be considered with extreme caution and re-evaluation considered at any stage of illness to diagnose a coexisting grave pathology.

Learning points

- Diagnosis of irritable bowel syndrome must be made with caution, especially when atypical symptoms or red flag signs are present, and re-evaluation must be made to rule out any coexisting grave pathology.
- Pedunculated neurogenic tumours arising from the lumbar root and occupying the presacral area are reported here for the first time.
- Proper examination, including digital rectal examination, must be performed to rule out underlying grave pathology.

Competing interests None.

Patient consent Obtained.

REFERENCES

1. Bullard Dunn K. Retrorectal tumors. *Surg Clin North Am* 2010;**90**:163–71.
2. Zerrweck-López C, Quijano-Orvañanos F, Montañez-Ramírez H, et al. Neuroblastoma in the adult. Case report. *Cir Cir* 2009;**77**:397–401.
3. Jao SW, Beart RW Jr, Spencer RJ, et al. Retrorectal tumors. Mayo Clinic experience, 1960–1979. *Dis Colon Rectum* 1985;**28**:644–52.
4. Cerullo G, Marrelli D, Rampone B, et al. Presacral ganglioneuroma: a case report and review of literature. *World J Gastroenterol* 2007;**13**:2129–31.
5. Ferrero S, Camerini G, Ragni N, et al. Endometriosis and irritable bowel syndrome: co-morbidity or misdiagnosis? *BJOG* 2009;**116**:129; author reply 129–30.
6. Wilson HM. Chronic subacute bowel obstruction caused by carcinoid tumour misdiagnosed as irritable bowel syndrome: a case report. *Cases J* 2009;**2**:78.
7. Yale SH, Musana AK, Kieke A, et al. Applying case definition criteria to irritable bowel syndrome. *Clin Med Res* 2008;**6**:9–16.
8. Stewart RJ, Humphreys WG, Parks TG. The presentation and management of presacral tumours. *Br J Surg* 1986;**73**:153–5.
9. Santiago C, Lucha PA. Atypical presentation of a retrorectal ancient schwannoma: a case report and review of the literature. *Mil Med* 2008;**173**:814–16.

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