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REVIEW

- 64 New insights of *Helicobacter pylori* host-pathogen interactions: The triangle of virulence factors, epigenetic modifications and non-coding RNAs
Vaziri F, Tarashi S, Fateh A, Siadat SD

ORIGINAL ARTICLE

Retrospective Cohort Study

- 74 Effect and safety of sorafenib in patients with intermediate hepatocellular carcinoma who received transarterial chemoembolization: A retrospective comparative study
Lei XF, Ke Y, Bao TH, Tang HR, Wu XS, Shi ZT, Lin J, Zhang ZX, Gu H, Wang L

CASE REPORT

- 84 Serum matrix metalloproteinase 3 in detecting remitting seronegative symmetrical synovitis with pitting edema syndrome: A case report
Kenzaka T, Goda K
- 88 Magnetic resonance imaging findings for differential diagnosis of perianal plexiform schwannoma: Case report and review of the literature
Sun XL, Wen K, Xu ZZ, Wang XP
- 94 Asymmetrical traumatic bilateral hip dislocations with hemodynamic instability and an unstable pelvic ring: Case report and review of literature
Huang K, Giddins G, Zhang JF, Lu JW, Wan JM, Zhang PL, Zhu SY

Contents

World Journal of Clinical Cases
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Editorial Board Member of *World Journal of Clinical Cases*, Harunor Rashid, MD, Doctor, Senior Lecturer, Senior Postdoctoral Fellow, National Centre, the Children's Hospital at Westmead, Sydney 2145, NSW, Australia

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Baishideng Publishing Group Inc
7901 Stoneridge Drive, Suite 501, Pleasanton, CA 94588, USA
Telephone: +1-925-2238242
Fax: +1-925-2238243
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Magnetic resonance imaging findings for differential diagnosis of perianal plexiform schwannoma: Case report and review of the literature

Xue-Liang Sun, Ke Wen, Zhi-Zhong Xu, Xiao-Peng Wang

Xue-Liang Sun, Ke Wen, Zhi-Zhong Xu, Xiao-Peng Wang, Department of Colorectal Surgery, the Suzhou Affiliated Hospital of Nanjing University of Chinese Medicine, Suzhou 215000, Jiangsu Province, China

ORCID number: Xue-Liang Sun (0000-0001-8520-6937); Ke Wen (0000-0003-3925-6567); Zhi-Zhong Xu (0000-0002-1725-7190); Xiao-Peng Wang (0000-0002-4747-8197).

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Correspondence to: Xiao-Peng Wang, MD, Professor of Surgery, Chief Doctor, Department of Colorectal Surgery, the

Suzhou Affiliated Hospital of Nanjing University of Chinese Medicine, Yangsu Road No.18, Suzhou 215000, Jiangsu Province, China. wxpeng2004@163.com
Telephone: +86-512-67872805
Fax: +86-512-65225929

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Abstract

Plexiform schwannoma is an extremely rare variant of schwannoma, accounting for approximately 5% of cases. Due to the rarity and lack of typical symptoms, signs and radiological images, a definite diagnosis of plexiform schwannoma may not be made by clinicians prior to biopsy. In the present study, we report the first case (to our knowledge) of perianal plexiform schwannoma arising from the overlapped skin of the ischioanal fossa, and we propose an intratumorally nonenhanced circumferential capsule dividing the tumour into multiple homogeneously enhanced nodules as a magnetic resonance imaging feature to aid in the differential diagnosis of plexiform schwannoma from ancient schwannoma, cavernous haemangioma, liposarcoma and plexiform neurofibroma.

Key words: Plexiform schwannoma; Magnetic resonance imaging; Perianal; Bascom cleft lift; Diagnosis

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Core tip: Plexiform schwannoma is an extremely rare variant of schwannoma, without typical symptoms, signs

and radiological images. Hence, a definite diagnosis of plexiform schwannoma may not be made by clinicians prior to biopsy. In this work, to our knowledge, we report the first case of perianal plexiform schwannoma arising from the overlapped skin of ischioanal fossa, and we propose an intratumorally nonenhanced circumferential capsule dividing the tumour into multiple homogeneously enhanced nodules as an magnetic resonance imaging feature to aid in the differential diagnosis of plexiform schwannoma from common nodular lesions, such as ancient schwannoma, cavernous haemangioma, liposarcoma and plexiform neurofibroma.

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INTRODUCTION

Plexiform schwannoma (PS) is a rare subtype of schwannoma, accounting for approximately 5% of cases^[1]. To date, the literature on PS is all case reports. The differential diagnosis of PS predominantly depends on histopathological examination, which weakens the role of diagnosis according to symptoms, signs and radiological images, and makes a dilemma for planning treatment protocol^[1-3]. Hence, it is difficult for clinicians to make a definite diagnosis of PS. The head, neck, trunk and extremities are common lesion sites, while colon, rectum, penis and clitoris involvements have been documented^[2-5]. To our knowledge, we report the first case of PS located in the perianal region and demonstrate a magnetic resonance imaging (MRI) feature for differential diagnosis in the present paper.

CASE REPORT

A nineteen-year-old male presented with a five-year history of a slowly growing, painless mass on the right ischioanal fossa. Haematochezia and change in bowel habit were not observed. Before referral to our institution, perianal percutaneous colour Doppler ultrasound revealed a solid lesion with abundant blood flow, and then, a diagnosis of haemangioma was made. After admission, physical examination revealed a multinodular cystic-solid mass of 5 cm × 3 cm on the perianal region, which was ductile, elastic, non-tender, movable and smooth (Figure 1A). No fistula tract could be palpated around the mass. Rectal digital examination could not detect any abnormality. No cafe-au-lait spots, axillary freckling or enlarged inguinal lymph nodes were observed. The results of routine laboratory tests and computed tomography of the chest and abdomen were normal. Pelvic MRI showed a well-circumscribed lobulated mass

by a thin complete capsule, which demonstrated a T1-isointense signal and heterogeneously T2-hyperintense signal compared to striated muscle, and homogeneously intense enhancement on contrast-enhanced images (Figure 2A-C). A neurogenic tumour was considered by the radiologist.

Local excision without preoperative biopsy was the treatment protocol because the anal sphincters and gluteus maximus were uninvaded. The operative procedure referenced the Bascom cleft lift technique^[6]. After intraspinal anaesthesia, the patient was placed in the prone jack-knife position. A longitudinal spindle incision was made with a 1 cm surgical margin. After the lesion completely resected, a skin flap 1 cm thick was freed from the right buttock until the wound could be sutured without tension. The defect was restored by suturing the fat cushion with vicryl suture. A negative-pressure drainage tube was put upon the fat cushion. The wound was closed by interrupted suture and then dressed with pressure by using an elastic abdominal bandage.

Histologically, the mass was composed of 10 variably sized nodules that appeared white-grey and were individually encapsulated. Observation in high-power fields confirmed a predominant Antoni type A structure with hypercellularity, Verocay bodies and a rare Antoni type B structure with hypocellularity. Spindle cells of the tumour were shown with elongated nuclei and palisading parallel rows (Figure 3A). Increased vascularity and sporadic haemorrhage were evident. Nuclear atypia and mitotic figures were not evident. Immunostains showed that the tumour cells were diffusely positive for vimentin, S100 protein and CD56 and negative for desmin, smooth muscle actin, CD34 and CD68 (Figure 3B). The Ki-67 labelling index was 3%. A definitive diagnosis of PS was made.

DISCUSSION

Although pelvic MRI has been widely applied for staging and differential diagnosis of perianal neoplasms and thus assists in making treatment protocols, literature regarding the MRI findings of PS is scarce. Hence, a radiological misdiagnosis might be easily made and lead to a misguided treatment strategy. The knowledge of MRI features of PS is required for differential diagnosis.

Compared to striated muscle, PS in our case presented as a T1-isointense signal, which was evidently distinguished from the surrounding tissue by a circumferential hypointense capsule. An intratumoral T2-hypointense capsule divided the tumour into multiple nodules that exhibited a predominant isointense signal area mixed with a minor hyperintense signal area. Contrast-enhanced images showed that the nodules were homogeneously enhanced and encapsulated by a nonenhanced capsule. A target sign characterized by the rim of a T2-hyperintense signal surrounding a central hypointense signal was absent on MR images of PS, which is one of the characteristics of peripheral

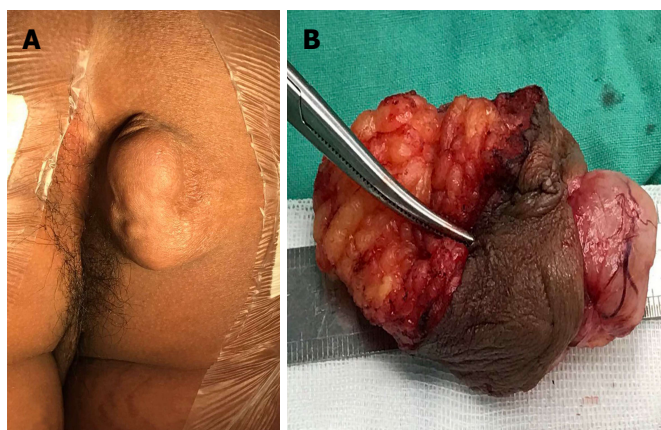


Figure 1 Macrography of plexiform schwannoma. A: A multinodular cystic-solid mass of 5 cm × 3 cm is located on the right ischioanal fossa; B: The resected specimen shows a superficial vessel implanting into the tumour.



Figure 2 Coronal magnetic resonance imaging features. A: T1-weighted images demonstrate a well-defined subcutaneous mass located on the right ischioanal fossa with an isointense signal; B: T2-weighted images demonstrate that the mass is divided into multiple heterogeneously hyperintense nodules by intratumoral capsules; C: T1-weighted images after administration of intravenous contrast demonstrate homogeneously intense enhancement of the nodules, but with unenhanced intratumoral fibrous capsules.

nerve sheath tumours. This was the reason that a histopathologically hypercellular area accompanied by fibrous tissue is predominant while a hypocellular matrix area is rare or even absent, and haemorrhagic foci can be found in some lesions.

To our knowledge, only one previous case report of solitary PS depicted the MRI feature that aids in the radiologic diagnosis. This feature is that both a T1- and a T2-hypointense thin capsule separated the multinodular tumour from the surrounding soft tissue^[7]. However, this feature might not make it easy to differentially diagnose PS from those multinodular lesions with pseudocapsule. In the present paper, we noted a more specific radiological feature: intratumoral smooth capsules that displayed both T1- hypointense and T2-hypointense signals and were not enhanced on contrast-enhanced scan divided the tumour into multiple nodules with heterogeneously T2-hyperintense signal and homogeneous enhancement, which may aid in the radiological diagnosis of this extremely rare perianal tumour that seems no specific nerve of origin.

Perineal ancient schwannoma (AS) has been reported

in few literatures, which is characterized by degenerative changes including cyst formation, haemorrhage and calcification^[8,9]. Both PS and AS as rare variants of schwannoma are encapsulated with a true capsule consisting of the epineurium, that is demonstrated as a T1-hypointense and T2-hypointense rim. However, a slight to intense enhancement of the fibrous capsule may be expected in a few cases of AS, which is differential from PS^[10,11]. AS is divided into a multilocular cystic-solid mass by intratumoural fibrous septa and shows a T1-hypointense and heterogeneously T2-hyperintense signal. An enhanced marginal crescent solid component of AS combining with unenhanced cystic area is the MRI feature for differentiating from homogeneously enhanced multinodular PS^[10].

Morphologically, PS mimicking haemangioma has been depicted in two reports^[12,13]. The dilatancy growth of PS increased vascularity in the overlapped skin and made a superficial vessel implant into the tumour (Figure 1B). Hence, sonographically abundant blood flow led to a misdiagnosis of haemangioma in our clinical practice. Among the three subtypes of haemangioma,

Table 1 magnetic resonance imaging features for differential diagnosis

Item	T1 signal	T2 signal	T2 homogeneity	Enhancement	Enhancement homogeneity	Intratumoral capsule
Plexiform schwannoma	Isointense	Hyperintense	Heterogeneous	Intense	Homogeneous	circumferential fibrous capsule
Ancient schwannoma	Hypointense	Hyperintense	Heterogeneous	Intense	Heterogeneous	Fibrous septa
Liposarcoma						
Well-differentiated	Hyperintense	Hyperintense	Homogeneous	Mild	Homogeneous	Fibrous septa
Myxoid	Isointense	Hyperintense	Homogeneous	Intense	Heterogeneous	Fatty septa
Pleomorphic	Isointense	Hyperintense	Heterogeneous	Intense	Heterogeneous	Fatty septa
Cavernous haemangioma	Hypointense	Hyperintense	Heterogeneous	Intense	Heterogeneous	Fibrous septa
Plexiform neurofibroma	Hypointense	Hyperintense	Heterogeneous	Mild	Heterogeneous	Fibrocollagenous septa

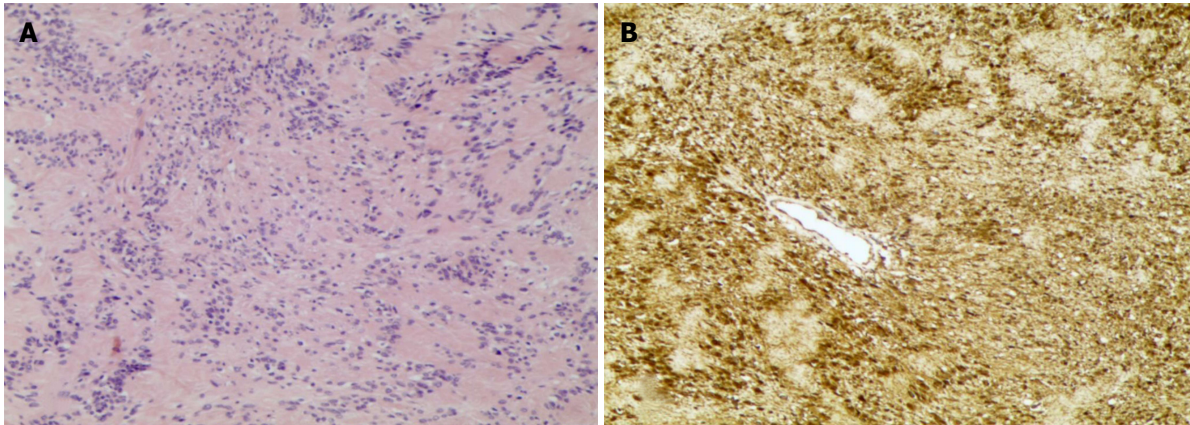


Figure 3 Pathological results. A: Predominant Antoni A areas composed of spindle cells with palisading parallel rows are displayed by haematoxylin and eosin stain; B: Immunostain shows the tumour cells with diffuse immunoreactivity to S100 protein.

capillary haemangioma, cavernous haemangioma and sclerosing haemangioma, giant cavernous haemangioma measuring over 4 cm in diameter presents as a well-defined nodular mass resembling PS. On MRI, giant cavernous haemangioma demonstrates a T1-hypointense and heterogeneously T2-hyperintense signal with liquefaction, thrombus, blood calculus, vascular smooth muscle and discontinuously fibrous septa components^[14]. The vascular flowing-void effect may be seen in some lesions. A heterogeneously nodular peripheral enhancement is the characteristic MRI finding of cavernous haemangioma^[14].

PS on the perianal fat space should be differentiated from lobulated liposarcoma (LPS), which comprises 20% of soft tissue sarcomas and has a low degree of malignancy and potential risk of metastasis^[15]. The World Health Organization divides LPS into 3 broad categories: well-differentiated (containing atypical lipomatous tumours) and dedifferentiated LPS; myxoid LPS (containing round-cell LPS); and pleomorphic LPS^[16]. The presence of visible fat is their common MRI feature. Well-differentiated LPS with a predominant adipose tissue component demonstrates homogeneously T1-hyperintense and T2-hyperintense signals and a hypointense signal on fat-suppressed images, which makes it easy to differentiate PS from LPS^[17]. Dedifferentiated LPS appears as a lipomatous tumour with a prominent soft tissue component, intense heterogeneous enhancement, but no encapsulation^[17].

As PS tends to occur in adolescents and young adults, similar to our case, myxoid LPS also often occurs in young adults and is the mainstay of differential diagnosis^[18,19]. However, clinicians may face a radiologic diagnostic dilemma, as visible fat accounts for < 10% of the tumour volume in < 40% of myxoid LPS cases. Myxoid LPS with high water content demonstrates T1-isointense and homogeneously T2-hyperintense signals with encapsulated margins^[17]. In addition, intratumorally discontinuous fatty septa instead of intact fibrous capsules divide the tumour into a multinodular pattern of heterogeneously intense enhancement, which is the differentiating feature from PS. The absence of encapsulated margins, peritumoral oedema and nodular enhancement correlate with high-grade histology.

Pleomorphic LPS with significant metastatic potential mostly occurs in the deep soft tissue of the extremities in elderly patients^[19]. T1-isotense, heterogeneously T2-hyperintense signals and heterogeneously intense enhancement with intratumoral haemorrhage and necrosis are the MRI manifestations of pleomorphic LPS^[17]. The lack of encapsulated margins and enhanced intratumoral fatty septa are differentiating features from PS.

PS and plexiform neurofibroma (PN) are both neurogenic tumours with multinodular growth patterns. Differential diagnosis is essential due to the potential malignant risk of PN, whose clinical diagnosis may be made according to cafe-au-lait spots or axillary

freckling^[20]. Radiologically, PN demonstrates T1-hypointense and heterogeneously T2-hyperintense signals or typical target sign with a central fibrocollagenous and peripheral myxoid stroma component^[21]. Usually, encapsulated margins are not shown. Mild enhancement is displayed on contrast-enhanced images, corresponding to the absence of flow on colour Doppler ultrasound^[21]. The MRI feature of the tumour orientated longitudinally in the nerve distribution and pierced by the peripheral nerve is more significant for diagnosis, as the target sign may not always be present. The above features make it easy to differentiate PS from PN. Heterogeneous enhancement, peritumoral oedema, intratumoral cystic lesions and maximum diameter of the mass > 5 cm indicate the potential of malignant transformation of PN^[22].

Absence of Verocay bodies and fibrous capsules and a 30%-40% immunopositive rate for S100 are immunohistopathological features of PN. In contrast, PS displays Verocay bodies, fibrous capsules, and diffuse positivity for S100 and vimentin^[2]. Mitotic figures and a Ki-67 labelling index > 5% may be observed once PN transforms into malignancy^[23].

Local excision is the treatment strategy for benign PS, but incomplete resection may cause recurrence^[13]. Preoperative biopsy was not conducted in our practice because local excision is utilized as a total biopsy to thoroughly assess any perianal neoplasm without involvement of the anal sphincter. Postoperatively, a large defect might not be primarily sutured without tension, which could prolong the healing time, increase the infection risk of the perianal wound and form extensive scarring that hinders sitting. The Bascom advancement flap was applied to primarily suture the defect in our patient, which was utilized as an off-midline tension-free repair technique for the sacrococcygeal pilonidal sinus. The Bascom advancement flap provides a shorter learning curve, lower pain scores, lower flap necrosis risk and better quality of life^[24,25].

In conclusion, we have described the first case of perianal PS characterized by an intratumoral non-enhanced capsule dividing the tumour into multiple homogeneously enhanced nodules, which we propose as the radiologically key point to differentially diagnose PS from AS, cavernous haemangioma, LPS and PN (Table 1), all with a capsule or a pseudocapsule. The Bascom advancement flap provides a convenient and safe modality to restore perianal defects.

ARTICLE HIGHLIGHTS

Case characteristics

A nineteen-year-old male presented with a slowly growing, painless, multinodular mass on the right ischioanal fossa.

Clinical diagnosis

Clinical diagnosis was a dilemma due to no characteristic clinical feature observed.

Differential diagnosis

Magnetic resonance images and histopathologic examination were focused on to differentially diagnose plexiform schwannoma from multilocular or multinodular ancient schwannoma, liposarcoma, cavernous haemangioma and plexiform neurofibroma.

Laboratory diagnosis

There was no specific laboratory testing contributing to the diagnosis of plexiform schwannoma.

Imaging diagnosis

Magnetic resonance images showed an intratumoral nonenhanced capsule dividing the tumour into multiple homogeneously enhanced nodules.

Pathological diagnosis

Neoplasm biopsy revealed Verocay bodies, fibrous capsules, and diffuse positivity for S100 and vimentin.

Treatment

Complete resection combined with the Bascom advancement flap to suture a large defect was performed to prevent a recurrence.

Related reports

Plexiform schwannoma commonly located on head, neck, trunk and extremities was definitely diagnosed based on postoperative histopathologic examination, whereas the foci located in the perianal region was rarer and was more difficult for preoperative diagnosis without a specific nerve of origin.

Term explanation

Perianal plexiform schwannoma is an extremely rare variant of schwannoma and is difficultly diagnosed in clinic owing to the absence of a specific nerve of origin and a characteristic clinical manifestation.

Experiences and lessons

Due to a multinodular growth pattern, a suspectable plexiform schwannoma can be preoperatively confirmed according to the magnetic resonance imaging feature of an intratumoral nonenhanced capsule dividing the tumour into multiple homogeneously enhanced nodules, which contributes to plan treatment protocol.

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