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Extraspinal sacrococcygeal myxopapillary ependymoma mimicking pilonidal disease in an adolescent: a case report

Anlan Yu[1,2,3]
First Teaching Hospital of Tianjin University of Traditional Chinese Medicine
Haikuo Wu[1,2]
First Teaching Hospital of Tianjin University of Traditional Chinese Medicine
Li Sun[1,2,3]
First Teaching Hospital of Tianjin University of Traditional Chinese Medicine
Jian Liu[1,2]
First Teaching Hospital of Tianjin University of Traditional Chinese Medicine
Zhiyuan Yang[1,2]
673167456@qq.com

First Teaching Hospital of Tianjin University of Traditional Chinese Medicine

Case Report

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Abstract

Background: Extraspinal sacrococcygeal myxopapillary ependymoma (MPE) can mimic pilonidal disease and present to colorectal surgeons before its neoplastic nature is recognized.

Case presentation: We report a 15-year-old boy with a progressively enlarging natal-cleft mass present for more than 3 years, without recurrent infection, drainage, a midline pit, or a visible sinus opening. Ultrasonography showed a 4.8 × 3.1 × 2.3 cm solid hypoechoic lesion with heterogeneous echoes and appreciable internal vascularity. Pilonidal disease remained the working diagnosis, and preoperative magnetic resonance imaging (MRI) was not performed. Surgery revealed an encapsulated solid tumor without hair, a sinus tract, or an abscess. The lesion was excised en bloc without visible capsular rupture. Histological examination and positivity for glial fibrillary acidic protein and S-100 supported MPE, CNS WHO grade 2. Microscopic margin status was not documented. Postoperative whole-spine MRI identified no intraspinal lesion. At approximately 2 months, the wound had healed and no new local mass or neurological symptoms were detected clinically.

Conclusions: This case highlights the need to reconsider pilonidal disease when clinical findings and ultrasonography are discordant. For colorectal surgeons, further evaluation should clarify the relationship of an atypical mass to the anal sphincter complex, rectum, coccyx, and spinal canal before definitive surgery. An unexpected solid tumor warrants intraoperative reassessment rather than automatic continuation of a sinus-directed procedure. The short follow-up and undocumented margins preclude conclusions about durable oncological control.

Background

Pilonidal disease is a common consideration in adolescents presenting to colorectal and proctological services with a natal-cleft lesion. Diagnosis is generally clinical, but the examination should also address alternative anorectal disorders[1–3]. A persistent, progressively enlarging solid mass without pits, drainage, or recurrent inflammation requires a broader assessment before a pilonidal procedure is selected.

Myxopapillary ependymoma (MPE) usually arises in the conus medullaris, cauda equina, or filum terminale and is classified as CNS WHO grade 2 in the 2021 WHO classification[4]. Extraspinal sacrococcygeal disease is rare; proposed origins include ectopic ependymal tissue or remnants of the coccygeal medullary vestige [5,6]. Published cases document initial misdiagnosis as pilonidal disease [3,7,8].

We describe an adolescent whose solid, vascular natal-cleft mass remained classified clinically as pilonidal disease until surgery. The case illustrates two decision points relevant to colorectal surgeons: reassessing discordant clinical and imaging findings before surgery, and reconsidering the operative strategy when an unexpected tumor is encountered. The report is organized according to the CARE framework [9], with a clinical timeline in Table 1.

Table 1
Clinical timeline

Date or interval	Clinical event	Finding or decision
~ 2023	Patient first noticed a small sacrococcygeal subcutaneous mass	Gradual enlargement; no recurrent infection or drainage
Aug 2025	First superficial ultrasonography	Subcutaneous hypoechoic lesion
Jun 2026	Repeat ultrasonography and surgical assessment	4.8 × 3.1 × 2.3 cm solid heterogeneous lesion with relatively abundant Doppler flow; pilonidal disease still considered
30 Jun 2026	Surgical excision	Encapsulated solid tumor; no hair, sinus, abscess, or granulation tissue; en bloc macroscopic excision
Postoperative period	Histopathology and immunohistochemistry	MPE, CNS WHO grade 2; GFAP(+), S-100(+), Ki-67 < 5%
Postoperative period	Whole-spine 3.0-T MRI	No intraspinal lesion identified; imaging was obtained after resection
~ 2 months	Clinical follow-up	Healed wound; no new palpable mass or neurological symptoms; short clinical follow-up

Case presentation

Patient information and clinical findings

A 15-year-old boy was admitted on 29 June 2026 because of a progressively enlarging sacrococcygeal mass that had been present for more than 3 years. The lesion had initially been a small subcutaneous nodule in the natal cleft and had enlarged gradually. He reported no spontaneous pain, recurrent erythema, ulceration, or purulent discharge. He had no hematochezia, tenesmus, anal discomfort, lumbosacral pain, lower-limb weakness or numbness, or bowel or bladder dysfunction. His previous medical and family histories were unremarkable. Psychosocial history, relevant genetic information, and details of any prior treatment of the lesion were not documented in the available record.

On anorectal surgical examination, an approximately 5 × 5 cm subcutaneous mass was palpable in the natal cleft near the anal margin. The overlying skin was violaceous. The lesion was relatively soft, mobile, and fairly well circumscribed, with mild tenderness. No midline pit, protruding hair, purulent discharge, external fistulous opening, or obvious sinus tract was identified. There was no lumbosacral percussion tenderness, and limb strength and sensation were clinically normal. Routine laboratory assessment showed no obvious evidence of systemic inflammation.

Diagnostic assessment

Superficial ultrasonography had been performed in August 2025 and again in June 2026. Both examinations demonstrated a subcutaneous hypoechoic sacrococcygeal lesion. The June 2026 study measured the lesion at approximately 4.8 × 3.1 × 2.3 cm. It was well demarcated but mildly irregular in shape, with heterogeneous internal echoes. Color Doppler imaging demonstrated relatively abundant internal vascular signals (Fig. 1). The ultrasonographic report favored a pilonidal sinus, while a lipomatous lesion remained in the differential diagnosis.

Because of the patient's age and the location of the lesion in the natal cleft, pilonidal disease remained the leading clinical diagnosis. However, in retrospect, several findings were discordant with that diagnosis: there was no history of recurrent infection or drainage, no visible midline pit or hair, and ultrasonography showed a solid lesion with appreciable vascularity rather than a typical inflammatory sinus process. Preoperative MRI was not obtained. The patient was scheduled for excision of the sacrococcygeal lesion.

Surgical intervention and pathology

Surgical intervention and pathology
On 30 June 2026, surgery was performed under spinal anesthesia. After incision of the skin and subcutaneous tissue, the operative findings differed substantially from the expected appearance of pilonidal disease. A dark-purple, firm, well-encapsulated solid mass was identified. No hair, subcutaneous sinus tract, abscess cavity, or inflammatory granulation tissue was present.

At this point, the operative diagnosis was reconsidered. The surgical team dissected along the capsule and removed the mass en bloc while avoiding visible capsular rupture. Estimated blood loss was approximately 3 mL. The specimen measured 5.2 × 3.2 × 2.7 cm and had a gray-red, soft cut surface. The pathological margin status was not documented in the available report; therefore, the procedure can only be described as macroscopically complete excision rather than histologically margin-negative resection.

Hematoxylin and eosin staining showed epithelioid or spindle-shaped tumor cells arranged around vascular cores in papillary structures, with abundant perivascular myxoid stroma and rare mitotic figures (Fig. 2). Immunohistochemistry showed positivity for S-100 and glial fibrillary acidic protein (GFAP), negativity for cytokeratin, epithelial membrane antigen, and progesterone receptor, and a Ki-67 proliferation index below 5%. The combined morphology and immunophenotype supported a diagnosis of subcutaneous sacrococcygeal MPE, CNS WHO grade 2 [4,6].

Postoperative assessment and outcomes

After the pathological diagnosis, whole-spine 3.0-T MRI was performed to assess for an intraspinal primary lesion or neuraxial involvement. No space-occupying lesion was identified in the cervical, thoracic, or lumbosacral spinal canal. The conus medullaris, filum terminale, and cauda equina showed

no obvious abnormality, and no intraspinal tumor was identified. Because imaging was obtained after resection, the original relationship of the mass to the spinal canal could not be reconstructed fully. These findings supported an extraspinal sacrococcygeal presentation.

The postoperative course was uncomplicated, and the patient was discharged on postoperative day 4. At approximately 2 months of follow-up, the wound had healed well, no new local mass was palpable, and there was no lumbosacral pain, lower-limb sensory or motor abnormality, or bowel or bladder dysfunction. The follow-up period remains too short to establish long-term oncological control. Continued local and neuraxial surveillance is planned; the available record does not specify intervals or a definitive adjuvant-treatment decision.

Discussion

Recognizing an atypical natal cleft mass

The diagnostic difficulty in this case arose from the combination of a familiar location and atypical lesion characteristics. A slowly enlarging mass in the natal cleft suggested pilonidal disease, but the absence of pits, hair, drainage, or recurrent inflammation weakened that interpretation. These absences do not independently exclude pilonidal disease. The more consequential finding was their combination with a solid lesion containing internal Doppler flow. Such discordance should prompt reconsideration of the diagnosis rather than reassurance based on location alone.

Guidance from the American Society of Colon and Rectal Surgeons and the European Society of Coloproctology provides the appropriate colorectal context for examining suspected pilonidal disease and distinguishing other anorectal conditions [1,2]. In this patient, no external fistulous opening or bowel symptoms were reported. Nevertheless, the lesion's precise distance from the anal verge, digital rectal examination findings, and relationship to the sphincter complex were not documented in the available account. These missing observations limit anatomical interpretation; absence of symptoms cannot establish absence of rectal or sphincter involvement.

The differential diagnosis of an atypical sacrococcygeal mass includes developmental cysts, lipomatous or other soft-tissue tumors, and ependymal lesions[3,6–8]. A deeper presacral lesion is a separate anatomical possibility, not an established feature of this case. The documented subcutaneous location distinguishes this presentation from a primary retrorectal tumor.

Imaging and operative planning

MRI can define the extent of a sacrococcygeal lesion and its relationship to adjacent structures, although imaging alone does not establish MPE histology[6,10,11]. In colorectal practice, the clinically useful questions are whether the mass remains superficial, extends into the presacral space, abuts the rectum or anal sphincter complex, involves the coccyx or sacrum, or communicates with the spinal canal.

Experience with retrorectal tumors supports the importance of cross-sectional anatomical assessment for surgical planning, but those deeper tumors are not equivalent to this subcutaneous lesion[11] .

Preoperative MRI would have been reasonable in this case because a roughly 5 cm progressively enlarging solid mass was poorly explained by the working diagnosis. This is a case-based inference, not evidence that all suspected pilonidal lesions need MRI or that Doppler vascularity is specific for MPE. When imaging suggests deep extension or the diagnosis could alter the extent of surgery, multidisciplinary review and a planned approach to tissue diagnosis are preferable to treating the lesion as routine sinus disease. The eventual need for biopsy or primary excision depends on anatomy and the suspected tumor type; this single case cannot resolve that choice.

Intraoperative reassessment and pathological margins

The discovery of an encapsulated solid tumor without a sinus, hair, abscess, or granulation tissue changed the operative problem. Macroscopically intact excision provided a specimen for diagnosis in this patient. However, encountering an unexpected mass does not justify proceeding with excision in every setting. If the lesion is fixed, extends deeply, or cannot be separated safely from the sphincter complex, rectum, bone, or neural structures, stopping for further assessment may be more appropriate. The operative priority is to avoid an unplanned procedure that compromises subsequent tumor treatment or anorectal function.

In spinal MPE, capsular violation has been associated with recurrence, and a systematic review found differences in tumor control between children and adults[12,13]. These observations support careful handling but cannot establish the benefit of a particular operation for pediatric extraspinal MPE. Dissection along a visible capsule does not prove a microscopically clear margin. Here, the absence of reported margin assessment is a substantive limitation, not a minor reporting omission.

Papillary architecture with myxoid stroma and GFAP/S-100 positivity supported the diagnosis[4,6]. The reported negative epithelial membrane antigen stain does not by itself overturn the integrated diagnosis. Specialist pathological review should address diagnostic concordance and, where the specimen permits, inked margins and tumor at the resection surface. Without that information, neither an R0 designation nor a conclusion that further local treatment is unnecessary is justified.

Follow up and limits of the evidence

Postoperative whole-spine MRI found no intraspinal lesion, supporting an extraspinal presentation. This examination does not substitute for assessment of the operative bed or establish the absence of microscopic residual tumor. The available account does not document dedicated postoperative pelvic imaging, brain imaging, systemic staging, or a finalized multidisciplinary decision on further treatment. These are reporting gaps rather than evidence that such assessments were either indicated universally or completed.

Pediatric extraspinal reports describe heterogeneous treatment and recurrence patterns, while delayed local recurrence and distant spread have also been documented[5,6,14]. The recent adult case reported by Kalfoutzou and colleagues illustrates the possibility of late extraneural disease; it cannot quantify risk in an adolescent with a newly excised subcutaneous lesion.[15] The need for re-excision, radiotherapy, additional staging, and the surveillance schedule should therefore be individualized after review of margins, anatomy, and the clinical course.

The strengths of this report are the explicit clinical–ultrasonographic discrepancy and documentation of the intraoperative change in diagnostic reasoning. Its principal limitations are retrospective interpretation, lack of preoperative MRI, undocumented microscopic margins and detailed anorectal anatomy, and approximately 2 months of clinical follow-up. Wound healing and absence of a palpable mass establish an early postoperative outcome only. Longer follow-up should document local examination, relevant imaging, bowel and bladder function, continence symptoms, and neurological status; no normal continence score or durable tumor control can be inferred from the present data.

Conclusions

A solid, vascular natal-cleft mass without typical sinus-related findings warrants reconsideration of pilonidal disease. The colorectal contribution is careful anorectal assessment, appropriate anatomical imaging, and reassessment of an unexpected tumor before further dissection. Macroscopically intact removal should be distinguished from histologically margin-negative resection. This case demonstrates an early uncomplicated recovery, but undocumented margins and short follow-up prevent conclusions about long-term tumor control.

Abbreviations

CNS
central nervous system
GFAP
glial fibrillary acidic protein
MPE
myxopapillary ependymoma
MRI
magnetic resonance imaging
WHO
World Health Organization.

Declarations

Patient perspective

A detailed patient perspective was not available.

Ethics approval and consent to participate

Ethics approval was not required for this individual case report. Consent to participate was not applicable.

Consent for publication

Written informed consent was obtained from the patient's legal guardians for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

Competing interests

The authors declare that they have no competing interests.

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Author Contribution

AY: Conceptualization, investigation, data curation, literature review, writing—original draft. HW: Investigation, literature review, writing—review and editing. LS: Data curation, investigation, literature review. JL: Clinical investigation, writing—review and editing. ZY: Conceptualization, supervision, clinical management, writing—review and editing. All authors contributed to the article and approved the submitted version.

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Availability of data and materials

The original contributions presented in this case report are included in the article. Further inquiries may be directed to the corresponding author, subject to applicable privacy and ethical restrictions.

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Figures

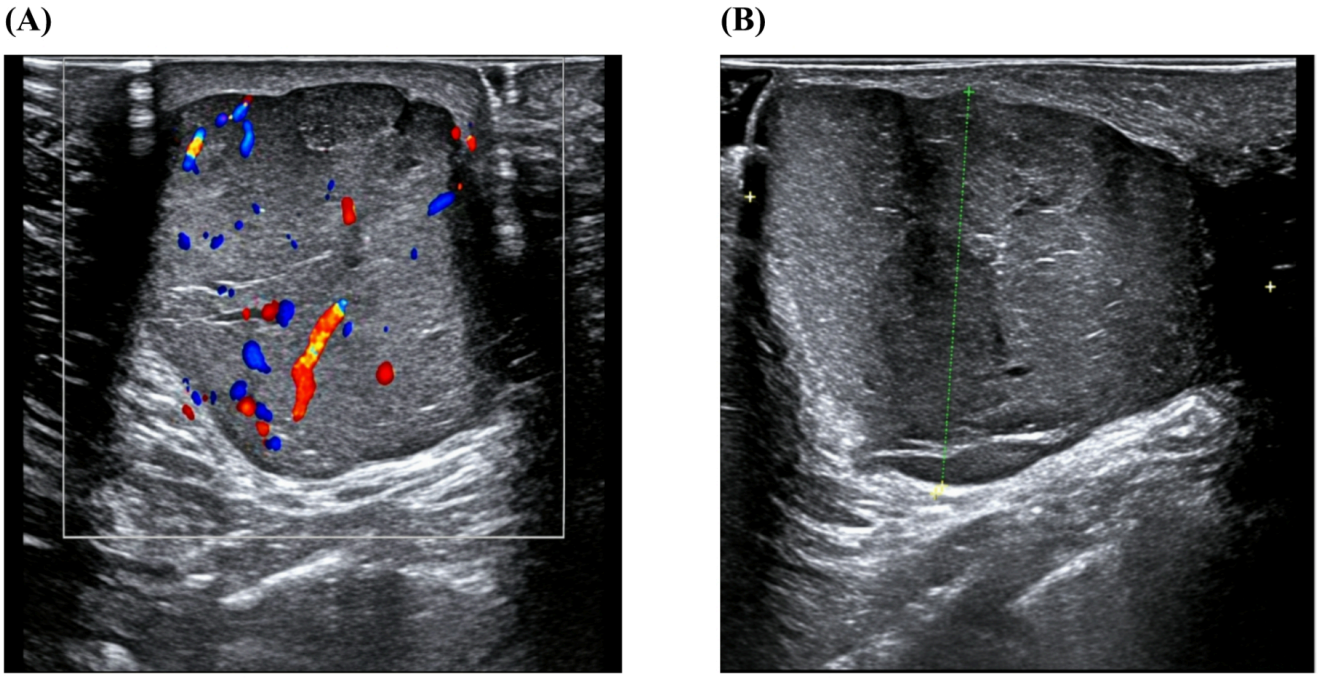


Figure 1

Preoperative ultrasonography of the sacrococcygeal subcutaneous mass. (A) Color Doppler image showing internal vascular signals. (B) Gray-scale image showing a solid hypoechoic lesion with heterogeneous internal echoes. The June 2026 examination reported dimensions of 4.8 × 3.1 × 2.3 cm.

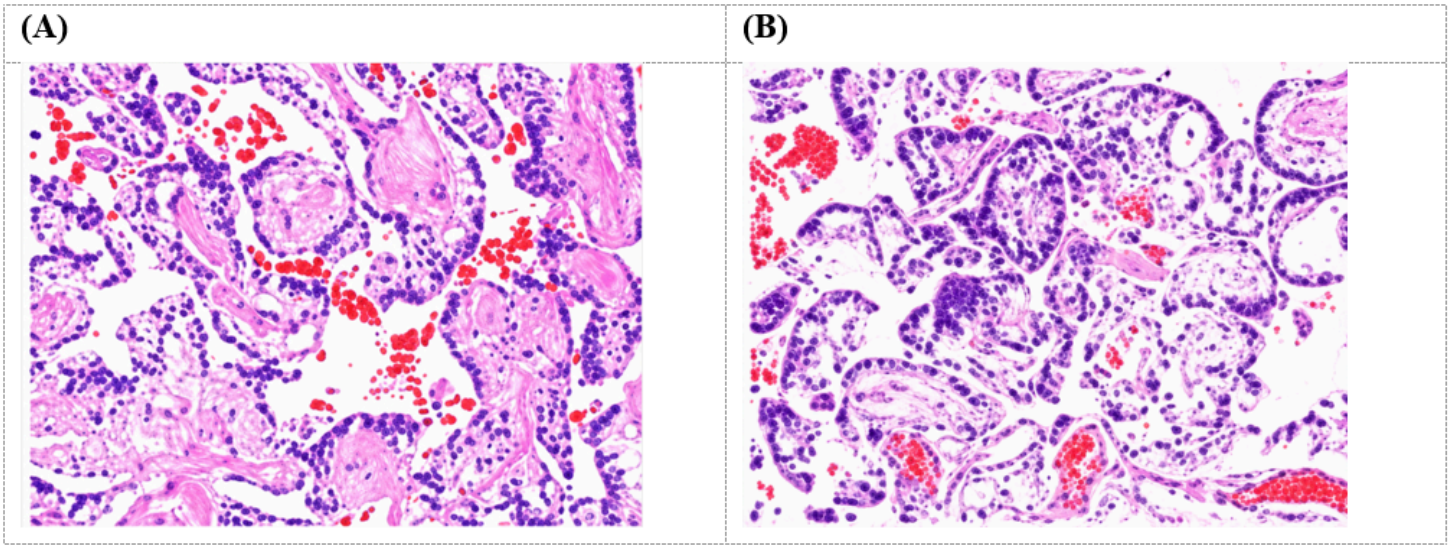


Figure 2

Histopathological features of the resected tumor. (A, B) Hematoxylin and eosin sections show papillary structures with vascular cores and myxoid stroma.

Supplementary Files

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