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Case Report

``Umbilical mass'': a case of primary umbilical endometriosis and literature review

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Background: To report on a case of primary Umbilical Endometriosis (PUE). *Case*: We report a new rare case of PUE in a 45-year-old woman arising in the umbilicus, presenting as three purple-blue firm masses with a smooth surface, and clear borders. The patient presented to our hospital with a complaint of dark-red blood flowing out of the umbilical fossa accompanied by periodic menstrual pain for more than a year. PUE was initially diagnosed based on clinical signs and imaging studies and the mass was widely excised. The postoperative histologic examination of the tissue confirmed the diagnosis of PUE. *Conclusion*: PUE is a rare umbilical disorder. Its diagnosis may be complicated due to clinician lack of knowledge. The possibility of endometriosis must be considered during the evaluation of an umbilical mass despite the absence of any previous surgery, with special attention to menstrual symptoms or bloody discharge.

Keywords

Endometriosis; Umbilical; Umbilical endometriosis; Review; Case report

1. Introduction

Endometriosis is a relatively common benign disease that occurs in 5% to 10% of women of reproductive age and is histopathologically characterized by the presence of functional endometrial glands and stroma outside the uterine cavity [1]. Pelvic pain, dyspareunia, dysmenorrhea, and infertility are the most common clinical symptoms of the endometriosis [2]. Endometriosis is most commonly found in the pelvic cavity, such as the ovaries and fallopian tubes, but can develop in any organ and has been reported in the lungs, liver, pleura, brain and skin [3–5]. It is important to note that deep infiltrating endometriosis (DIE) can also occur in the intestine [5].

Umbilical endometriosis is a rare form of extra-pelvic endometriosis accounting for 0.4 to 4% of all cases of endometriosis [6]. Umbilical endometriosis forms 30% to 40% of all cutaneous endometriosis [7]. Cutaneous endometriosis is typically secondary to surgical scars. PUE is an extremely uncommon occurrence, and its pathogenesis is unclear. Herein we describe a rare case in a 45-year-old female who presented with painful, purple-blue nodules in the umbilicus. The umbilical lesion was removed by local surgical excision. We followed up with the patient for 10 months and found no recurrence.

2. Case presentation

A 45-year-old patient was referred to our hospital with a complaint of dark-red blood flowing out of the umbilical fossa accompanied by periodic menstrual pain for more than a year. On admission, the physical examination did not reveal significant abnormalities. The patient denied a history of dyspareunia, infertility, and chronic pelvic pain. She had no history of any surgery, including cesarean section. She was not taking any form of hormonal therapy. On clinical examination, there were three purple-blue, tender, firm, irreducible nodules of about 0.6 to 0.9 cm in diameter in the center of a widened umbilicus (Fig. 1A). Upon ultrasonographic examination, a poorly shaped hypoechoic mass about 1.1 $\times$ 0.9 $\times$ 0.9 cm in size was detected in the umbilicus. The size of the mass was not influenced by abdominal pressure (Fig. 2). No abnormalities were found in the uterus and the adnexal region. There was no relevant family history or other history of significant diseases. General laboratory investigations, including complete blood count and blood chemistry, were within normal limits. The umbilical lesion was removed by local surgical excision under local anesthesia. Grossly, the tumor was three polyploidy and purple-blue masses with no extension to the fascia or abdominal cavity, and they were limited to the skin and subcutaneous tissue. Histological examination of the surgical specimen showed endometrial glands and stroma in the dermis and hypodermis with margins of healthy resections, confirming a diagnosis of PUE (Fig. 3A–B). Postoperative follow-up showed no abnormalities, and the final scar was hidden beneath the umbilical cord to give the new umbilical a normal appearance (Fig. 1B).

3. Discussion

Cutaneous endometriosis is a rare gynecological disease primarily affecting women of childbearing age. Cutaneous umbilical endometriosis is a rare entity, representing 0.5% to

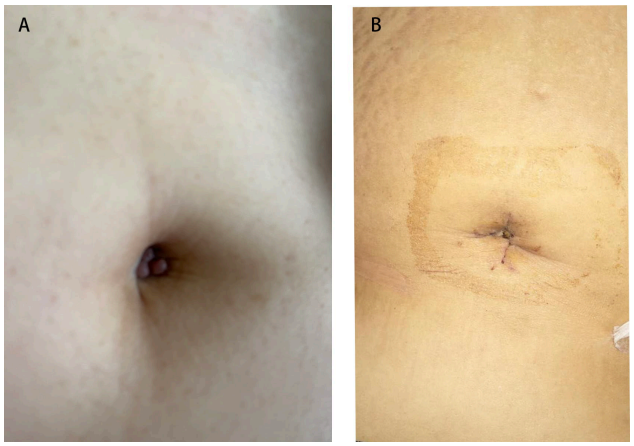

Fig. 1. Comparison of patient's umbilicus before and after surgery. (A) Three purple-blue, firm, irreducible nodules of about 0.6 to 0.9 cm in diameter in the centre of a widened umbilicus. (B) The final scar was hidden beneath the umbilical cord to give the new umbilical a normal appearance.

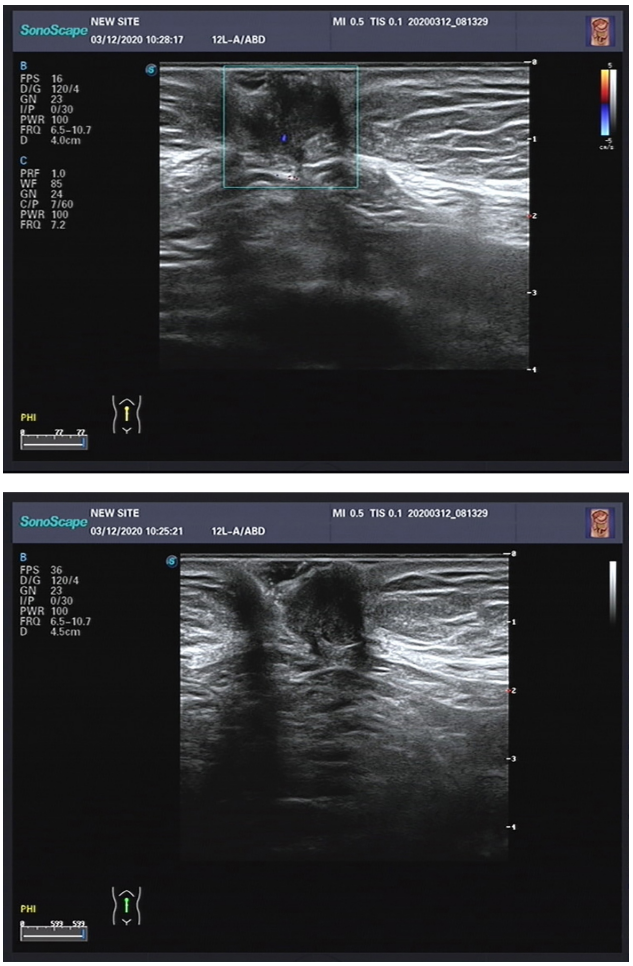

Fig. 2. Umbilical ultrasonography showed a mass of 1.1 × 0.9 × 0.9 cm with heterogenous echogenicity.

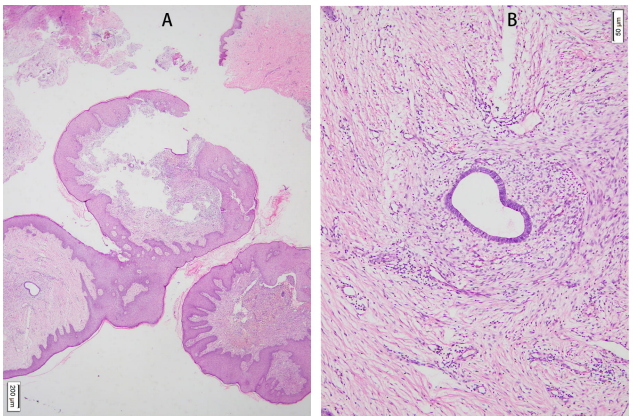

Fig. 3. A & B Specimen showed lesions in the superficial dermis and deep dermis comprising dilated glandular structures, surrounded by cellular endometrial-type stroma (H&E; A: ×4, B: ×10, respectively).

1% of all endometriosis cases [8]. Cutaneous endometriosis is subdivided into primary and secondary types according to the patient's surgical history. PCE is the less common of the two, referring to cases in which the endometriosis develops spontaneously without any history of local surgery. Secondary cutaneous endometriosis often occurs in pregnant women who have undergone abdominal surgery, especially cesarean section and laparoscopic surgery [9, 10].

Many theories have been proposed with regard to the pathogenesis of primary and secondary cutaneous endometriosis. The most commonly accepted mechanisms are lymphatic or vascular migration, cellular metaplasia, and iatrogenic dissemination [10]. It has been postulated that the umbilicus may mimic a physiologic scar and therefore, the endometrial cell can implant in it more easily [8]. The urachal remnant could undergo metaplasia and transform into endometrial like glands forming the primary umbilical endometriosis [11]. Iatrogenic dissemination, when endometrial cells implant in scars after surgery, could be the central etiologic pathogenesis of secondary cutaneous endometriosis, by contrast, primary cutaneous endometriosis may be explained by the theory of vascular or lymphatic migration [9, 10].

As with our case, the umbilicus is the most common location for cutaneous endometriosis, also referred to as Villar's nodule, as first described by Villar in 1886 [12]. PUE is a rare type of endometriosis typically characterized by colored subcutaneous papules or nodules (blue, brown, cutaneous) in the umbilicus, usually between 0.5 and 3 cm in size [8, 13]. Patients frequently complain of cyclical pain, swelling, and even bleeding concomitantly with the menstrual cycle [6]. Although the initial diagnosis for treatment workup is mainly based on clinical symptoms, so far, histological confirmation is the gold standard for diagnosing PUE. The irregular endometrial gland structure with the appearance of spindle cells in the basophilic cytoplasm, accompanied by hypercellular and vascular interstitium, is a characteristic of histological findings [14]. The endometrial glands tend to be immunopositive to estrogen and progesterone receptors and the surrounding stroma tend to be immunopositive for CD-10 [15].

Table 1. Literature review of primary umbilical endometriosis.

Reference	Age (yr)	Presenting symptom	Treatment	Additional therapy	Follow-up
Egin *et al.* [26]	28	Painful swelling in the umbilicus during her menstruation	total resection including umbilicus	None	No recurrent at 19 mon
Rzepecki *et al.* [27]	44	Asymptomatic umbilical lesion	None	None	Not described
Hoopes *et al.* [28]	30	tenderness, bleeding, and pain from the umbilicus	surgical excision	hormonal therapy	Not described
Loh *et al.* [9]	38	swolling with spontaneous bleeding during menstruation	complete wide excision	None	No recurrent at 24 mon
Müller and Flux [13]	44	dark red to brownish coloured discharge from her umbilicus	surgical excision	GnRH analogues	No recurrent
Al-Quorain *et al.* [29]	31	swelling was slowly increasing in size and mild pain	Surgical excision	None	No recurrent at 6 mon
Nellihel *et al.* [30]	16	with painful umbilical lump	surgical exploration of the umbilicus	None	No recurrent at 6 mon
Chew *et al.* [31]	44	umbilical nodule without pain	GnRH analogue and Vissane® (dienogest)	DNG	Nodules narrow
Claas-Quax *et al.* [32]	27	The umbilical cord bleeding	complete umbilical resection	OC	No recurrent
Taniguchi *et al.* [23]	45	painful umbilical mass concomitant with menstruation	complete umbilical resection	DNG and OC	No recurrent at 12 mon
Laferriere and Yheulon [33]	34	umbilical bulge and pain	open umbilical hernia repair	None	Not described
Bonne *et al.* [34]	41	increasing and bleeding umbilical nodules	Biopsy	Levonorgestrel	Not described
Genovese *et al.* [35]	42	painful um-bilical nodule	Umbilical nodule resection	None	Not described
Kydd *et al.* [24]	32	Pruritus, irritation at the lesion	surgical excision	Local herbal tea	Not described
Brătilă *et al.* [36]	46	Asymptomatic umbilical nodule	Surgical excision	None	No recurrent
Benardete-Harari *et al.* [37]	34	swelling and cyclic menstrual bleeding	Surgical excision	None	Not described
Calagna *et al.* [22]	33	Painful, reddish nodule	Surgical excision	None	No recurrent at 6 mon
Present study	45	Dark-red blood flowing out of the umbilical fossa	Surgical excision	None	No recurrent at 9 mon

DNG, Dienogest, a progestin; GnRH, gonadotropin releasing hormone analogues; OC, oral contraceptives.

The differential diagnosis of PUE includes benign and malignant conditions. Differential diagnoses of a recent onset umbilical nodule in a fertile woman includes benign disease such as purulent granulomas, umbilical polyps, dermatofibromas, neurofibromas, and keloid [10, 16]. More importantly, keloid is clinically very similar to umbilical endometriosis. Clinicians should pay particular attention to patients, especially those with a history of surgery or trauma, and presenting symptoms related to the menstrual cycle. If treatment with steroid intralesional injection does not improve the symptoms, umbilical endometriosis should be considered for differential diagnosis [9].

In the meantime, malignant tumors caused by metastasis of visceral carcinoma to the abdomen, such as a Sister Mary Joseph nodule and melanoma, should also be ruled out [8, 10, 13, 16]. Sister Mary Joseph nodule is a metastatic umbilical lesion present in 1–3% of patients with intra-abdominal or pelvic neoplasms. Dermoscopy is a tool that can help narrow the differential diagnosis of umbilical tumors. The dermoscopic features include a polymorphous vascular pattern, a milky red or pink structureless area, and white lines. Basal cell carcinoma can present with similar dermoscopic features as elsewhere in the body. Dermoscopic features of cutaneous endometriosis are usually unspecific; however, the presence of a homogeneous lesion with a bluish blotch/clot in or around the umbilical region requires consideration of cutaneous endometriosis in the differential diagnosis [17].

The gold standard of treatment is extensive resection of the mass with safety margins of at least 1 cm to reduce the risk of recurrence, followed by umbilical plasty [18–21]. Previous publications have reported that PUE can be treated with oral contraceptives, progesterone, Danazol or GnRH analogues to reduce the size of the nodule; relieve patients' symptoms; and minimize damage to the body during the operation. However, overall results from these studies suggest unsatisfactory results [6, 8, 22–25].

In a review of PUE literature published during the period 2015-2020, seventeen studies were identified written in English or French language (Table 1) [9, 13, 22–24, 26–37]. Based on all reports, the mean age of patients with PUE was 36.3 years. Recurrence and malignant alteration were infrequent. In our case, although the patient had clinical symptoms similar to endometriosis such as umbilical cord bleeding and dysmenorrhea a year ago, at the age of 45, the diagnosis of PUE was confirmed by umbilical ultrasound and histopathological examination.

In conclusion, PUE is a rare umbilical disorder. Its diagnosis may be complicated due to clinician lack of knowledge, the possibility of endometriosis must be considered during the evaluation of an umbilical mass despite the absence of previous surgery, paying special attention to menstrual symptoms or bloody discharge. Surgical excision is the treatment of choice to prevent recurrence and to reduce the risk of malignant transformation.

Author contributions

JZP contributed to collecting clinical data, gross photos and wrote the manuscript. YT provided the case and revised the manuscript. LL and XYX contributed to clinical diagnosis and revised the manuscript. XC and YYC contributed to pathological diagnosis.

Ethics approval and consent to participate

The present study was approved by the Scientific and Ethical Committee of the Third People Hospital of Chengdu (NO.2021-S-76). Written informed consent was obtained from the patient for publication of this case report and any accompanying images.

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Conflict of interest

The authors declare no conflict of interest.

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