Primary umbilical endometriosis. Case report and discussion on management options

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Article history:
Received 16 August 2013
Received in revised form 22 September 2013
Accepted 1 November 2013
Available online 6 November 2013

Keywords:
Endometriosis
Umbilicus
Treatment

ABSTRACT

INTRODUCTION: We report a recently observed case of primary umbilical endometriosis (UE), with the main aim to discuss the management of this rare condition.

PRESENTATION OF CASE: A 24-year-old woman complained of a painful nodule on her umbilical region, bleeding with her menstrual cycle. Ultrasonography showed a hypoechoic superficial mass in the umbilicus and no signs of intra-abdominal endometriosis. Excision of the nodule under local anesthesia was performed. Histopathological analysis confirmed the diagnosis of umbilical endometriosis. Neither symptoms nor signs of local recurrence have been observed after 24 months.

DISCUSSION: UE should be taken into account in differential diagnosis of umbilical disorders even in young nulliparous women with no typical symptoms of pelvic endometriosis. Although there is a substantial agreement about the necessity of surgery, treatment options are either local excision of the lesion or removal of the whole umbilicus with or without laparoscopic exploration of the peritoneal cavity. The decision should be tailored for the individual patient, taking into consideration the size of the lesion, the duration of symptoms and the presence of possible pelvic endometriosis.

CONCLUSION: Local excision saving the umbilicus may be the treatment of choice in patients with small UE lesions.

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1. Introduction

Endometriosis is defined as the presence of endometrial glands and stroma abnormally located outside the uterine cavity. It is a benign gynecological disorder affecting 10–15% of all women of reproductive age and represents an important cause of infertility.1,2 Although different theories have been postulated in order to elucidate the patho-physiology of this condition, to date none of them has been proven to be to be completely exhaustive. Common locations of endometriosis are the pelvic organs, mostly the ovaries, the Fallopian tubes, the utero-sacral ligaments, the rectovaginal septum and the pelvic peritoneum. Clinical manifestations include pelvic pain arising before and/or after menstruation, menorrhagia, painful intercourse, intestinal and urinary complaints.3,4

Extra-pelvic locations of endometriosis have been described in almost every tissue and organ (i.e. gastro-intestinal apparatus, abdominal organs, skin, diaphragm, lung and even brain).2-5 Umbilical endometriosis (UE), also known as Villar’s nodule from the first physician describing the disease, is a very rare entity, especially in nulliparous women. Due to the rarity of this entity, no guidelines for treatment exist. We report a recently observed case with the main goal being to discuss the treatment options.

2. Presentation of case

A 24-year-old woman, gravida 0, was admitted to outpatient clinics with a seven-month history of umbilical nodule. She stated that the nodule had slowly increased in size and had started to bleeding concomitantly with the menstrual periods in the previous 4 months. Her medical history was unremarkable and she denied symptoms of pelvic endometriosis such as dysmenorrhea, abdominal pain or dyspareunia. She was not taking any oral contraceptives and had regular menstrual cycles. Physical examination revealed a brown, moderately tender nodule of about 1 centimeter in diameter located deep in the umbilical fold (Fig. 1A and B). On the basis of
history and clinical findings, primary umbilical endometriosis was suspected and the patient was asked to return for further examination during her menstrual period, which occurred after one week. At this second look, the umbilical nodule appeared more tender, showing with signs of recent bleeding. An ultrasound confirmed the presence of a hypoechoic mass of 10 mm in the umbilicus, with no blood vessels at Doppler examination. The patient was thus referred to a gynecologist for clinical evaluation, transvaginal and abdominal ultrasonography. No clinical or ultrasonographic signs of endometriosis could be detected. Thus, surgical removal of the umbilical nodule was proposed and the patient was informed about the risk of local recurrence. In May 2011, the patient underwent excision of the nodule, saving the navel, under local anesthesia. The lesion was entirely excised deep to the fascia, together with a rim of macroscopic normal skin of 0.5 cm all around. There was no evidence of connection with the peritoneal cavity and the umbilicus was reconstructed with discontinuous suture using non-absorbable stitches. On gross examination a nodular, tan lesion of 1 cm × 0.8 cm covered by normal skin was appreciable. For the light microscopic examination, the specimen was fixed in 10% buffered formalin and embedded in paraffin. 4 μm-thick sections were stained with hematoxylin and eosin (H&E). Immunohistochemistry was performed using antibodies against Estrogen Receptor, Progesteron Receptor and CD10 (Novocastra, Leica Biosystems, Newcastle, UK). On microscopic examination, histologic sections revealed a glandular proliferation of monolayered endometrial epithelium surrounded by a cytostroma with extravasated erythrocytes (Figs. 2–3A). On immunohistochemistry findings, the epithelial and stromal cells too, showed a nuclear immunoreactivity for ER and PR (Fig. 3C and D); stromal cells expressed cytoplasmic positivity for CD10 (Fig. 3B). All these features were consistent with the diagnosis of umbilical endometriosis.

Postoperative recovery was uneventful. Following the operation, the patient was referred to a gynecologist who prescribed oral contraceptives. After two years of follow-up there were no signs of local relapse nor other clinical and ultrasonographic signs of endometriosis.

3. Discussion

Extrapelvic endometriosis accounts up to 15% of all cases of endometriosis.1,2 In particular, UE has been reported to be around 0.4–4% of all patients with endometriosis and accounts for up to 30–40% of all cases of cutaneous endometriosis.3,5 The latest detailed review of endometriosis externa of the umbilicus found 122 cases published in English between 1966 and 2007. UE is therefore a rare presentation, widely known among gynecologists, but possibly unfamiliar to many other specialists.

The particularity of the case described herein, is that the patient had primary or spontaneous UE, i.e. the presence of ectopic endometrial tissue located in the umbilicus in absence of previous surgery for either gynecological disorders or cesarean section. In the latter cases, UE should be defined as secondary UE, which is more common than primary UE and is probably due to the iatrogenic dissemination and implant of endometrial cells during either laparoscopic or open surgical procedures.4,6 While the pathogenesis of secondary UE seems to be relatively easy to explain, it is harder to clarify the origin of primary UE. In this regard, different hypothesis have been proposed, such as the embrional rest theory of Wolffian or Mullerian remnants, the transplantation theory in which the ectopic endometrial tissue harbors from retrograde menstruation or hematogenous/lymphatic dissemination, or a combination of them.4,5 However, the pathogenesis of primary endometriosis still remains unclear.

As for clinical presentation, the typical symptoms of UE are the presence of a discrete bluish-purple mass in the umbilicus, becoming swollen, painful and bleeding concomitantly with the menstrual cycle. In the review of Victory and co-workers, the mean age at diagnosis was 37.7 years, with the youngest being 23-years old.1 Notably, the patient of the present report was 24-years old.

In patients with UE the clinical picture and physical examination are the mainstay for diagnosis. Differential diagnosis of umbilical nodules includes hernia, benign and malignant skin neoplasms, metastatic adenocarcinoma, inflammatory and infectious lesions,
Interestingly, the majority of patients with UE do not report a history of pelvic endometriosis, as in the patient described in this report. Of note in the literature is the relative long duration of symptoms before clinical presentation, which was about 18 months.

In line with others, we evaluated the umbilical lesion by means of ultrasound, which may also detect connection with the peritoneum or fascia. Magnetic resonance imaging has also been reported to be accurate in the diagnosis of UE, while histopathological diagnosis by means of needle biopsy or fine needle aspiration cytology have been shown to be questionable in the majority of cases.

Histopathological features of UE include the presence of endometrial glands embedded in the stroma with a high cellular and vascular component. The endometrial glands form irregular glandular lumina and the bleeding into the dermis during menstruations leads to extravasated erythrocytes. Immunohistochemistry may help to confirm the diagnosis of UE by showing positivity for both estrogen and progesterone receptors and for antigen CD10, a marker used for stromal cells in endometriosis, as in the case described.

Management of UE is not standardized. In general, medical treatment using progesterone, danazol, norethisterone, and GnRh analogs has not shown reliable results, although some authors have reported some success in relieving symptoms and reducing the size of the endometriotic nodule by using medical hormonal treatment. In the review of Victory et al., almost 70% of patients required surgical treatment. However, consensus about standard surgical management of UE is lacking, probably due to the rarity of the disease. The operative options are:

1. Complete umbilical resection, with or without repair of the underlying fascia and peritoneum
2. Local excision of the endometriotic nodule, sparing the umbilicus

Total removal of the umbilicus is the most frequently performed operation for UE. This option is sometimes required due to the extension and size of the endometriotic nodule, especially in patients with years of UE symptoms. Some of the cases of UE reported in the literature, indeed extensively involved the abdominal wall structures adjacent to the umbilicus such as the umbilical fascia. This radical approach is warranted when UE is associated with large umbilical hernias. Reconstruction of discrete wall defects may require mesh placement and may be linked to poor cosmetic results. Nonetheless, some authors recommend whole umbilical excision, irrespective of the size of the endometrial nodule. Local excision of the endometriotic lesion should be done obtaining an adequate rim of normal tissue all around, in order to avoid local recurrence. In this regard, few studies report on follow-up after excision of UE. The patient in the present report has been followed-up clinically every six months. Neither symptoms nor signs of local recurrence have been observed after 24 months. We decided on conservative treatment based on the small size of the umbilical nodule, the young age of the patient, her desire to keep the anatomical integrity of the umbilical region, and the conviction that local recurrence is rare if adequate margins of resection are achieved. Nonetheless, patients in which simple local excision of UE nodule is proposed, should be fully informed of the risk of relapse. Of note, the patient has been started oral contraceptives after surgery. The protective role of endocrine treatment in preventing recurrences has been underlined.

In patients operated on for UE, laparoscopic exploration has been advocated in order to exclude possible further foci of intra-abdominal endometriosis, since pelvic endometriosis cannot be definitively excluded on transvaginal ultrasound or clinical examination. However, laparoscopic exploration remains debatable in asymptomatic patients and was not performed to our patient since she had no referred complaints typical for pelvic endometriosis. Nevertheless, she was well-informed about the possibility of having coexistence of both umbilical and pelvic disease.
When UE is suspected on the basis of clinical and imaging work-up, surgical treatment is recommended for several reasons. Firstly, the removal of the entire lesion only enables accurate histopathological diagnosis of UE, thus excluding unusual malignant disorders of the umbilicus, such as metastases or skin neoplasms. Furthermore, removal of UE nodule is warranted because malignant transformation of endometriotic lesions, although rare, has been described. Finally, early surgery is more likely to result in good cosmetic outcomes, especially when the size of the UE nodule does not mandate for the removal of the entire umbilical fold.

In our opinion, the case described in this paper contains some points of interest. To our knowledge, the patient is one of the youngest women with primary UE reported in the literature. This reinforces the concept that UE should be taken into account in differential diagnosis of umbilical disorders even in young nulliparous women with no typical symptoms of pelvic endometriosis. Moreover, this case emphasizes the importance of early diagnosis of UE in order to avoid extensive abdominal wall surgery.

4. Conclusion

UE is a rare entity which deserves attention in the differential diagnosis of umbilical disorders. The decision on treatment should be tailored on each single patient, taking into consideration the size of the lesion, the duration of symptoms and the presence of possible associated pelvic endometriosis. Simple excision of the endometrial nodule sparing the umbilicus under local anesthesia may offer definitive management in selected cases, such as young patients with small umbilical UE nodules.

Conflict of interest statement

The authors declare that they have no conflicts of interest.

Funding

None.

Ethical approval

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

Author’s contributions

Alessandro Fancellu performed the operation, designed the research, wrote the paper and obtained consent. Antonio Pinna assisted in writing case and in processing digital images.

Alessandra Manca conducted the pathology report of the specimen and contributed to the manuscript editing.

Giampiero Capobianco was involved in gynecologic management of the patient, and contributed to the literature review and manuscript editing.

Alberto Porcu provided overall supervision and suggestion for the case report.

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