The clinical features and management of perineal endometriosis with anal sphincter involvement: a clinical analysis of 31 cases

Na Chen¹, Lan Zhu¹,*, Jinghe Lang¹, Zhufeng Liu¹, Dawei Sun¹, Jinhua Leng¹, Qingbo Fan¹, Hui Zhang², and Quancai Cui²

¹Department of Obstetrics and Gynecology, Peking Union Medical College (PUMC) Hospital, No. 1 Shuaifuyuan Wangfujing, Dongcheng District, Beijing 100730, P.R. China ²Department of Pathology, Peking Union Medical College (PUMC) Hospital, Beijing 100730, P.R. China

*Correspondence address. Tel: +8610 13911714696; Fax: +8610 65124875; E-mail: zhulan.julie@gmail.com

Submitted on October 5, 2011; resubmitted on December 21, 2011; accepted on February 7, 2012

BACKGROUND: The aim of this study was to investigate the appropriate measures for diagnosing and treating perineal endometriosis (PEM) with anal sphincter involvement.

METHODS: Between January 1992 and April 2011, the clinical features, diagnosis and management of 31 patients who were diagnosed with PEM with anal sphincter involvement at the Peking Union Medical College Hospital were retrospectively analyzed using their clinical records. A range of 6–78 months of outpatient follow-up after surgery were conducted for these 31 patients but was extended by telephone interviews with 29 patients conducted in December 2011.

RESULTS: All 31 patients had a history of vaginal delivery. The level of serum CA125 was elevated in only 2 (6.5%) cases. All cases received surgical treatment, which included narrow excision (NE, close to the edge of the endometrioma) with primary sphincteroplasty (PSp) for 30 cases and incomplete excision (IE) for 1 case. Of the 30 cases in the NE group, 20 (66.7%) received hormone therapy preoperatively. Up until December 2011, there was one recurrence (3.6%) of PEM in the NE group. PEM relapse occurred in the IE patient 6 years after the initial IE surgery. Perineal abscesses were found in one patient post-operatively. No complaint of dyspareunia and no fecal incontinence episodes were observed during follow-up.

CONCLUSIONS: Based on our own experience, NE and PSp may be indicated for the treatment of PEM with anal sphincter involvement.

Key words: perineal endometriosis / anal sphincter / diagnosis / therapy / recurrence

Introduction

Endometriosis (EM), which is defined as the presence of endometrial tissues outside of the uterine cavity, is one of the most common diseases in women of reproductive age. In general, endometriosis can be divided into pelvic and extrapelvic sites. Endometriosis is commonest in the pelvis. It is very occasionally found in extra-pelvic sites such as the gastrointestinal tract, pulmonary structures, the urinary system, abdominal wall, perineum or vagina, skin and even the central nervous system. Perineal endometriosis (PEM) is the occurrence in the perineum of endometrial glands and stroma that respond to the hormone variations in the cycle. When islands of endometrial tissues invade the sphincteric muscular tissue, PEM with anal sphincter involvement occurs. Since 1923, case studies of PEM with anal sphincter involvement, a condition that has a low prevalence, have been published in the obstetric and gynecologic literature (Schickele, 1923). Using PubMed to search for case reports of this condition since 1957, we found only 13 cases in 11 different case reports. As a relatively rare disease, the diagnosis and treatment of the disease need to be further studied. The Peking Union Medical College (PUMC) Hospital is the referral center for complex obstetric and gynecologic medical problems in China, and as a result, patients with complicated and unusual endometrioses are likely to be referred to our hospital. This retrospective study shows 31 cases of PEM with anal sphincter involvement that received surgical treatment between January 1992 and April 2011 in PUMC Hospital. Through following analysis of the clinical features, diagnosis, treatment and prognosis of the 31 cases, we put forward our understanding of the diagnosis and treatment of the disease.

© The Author 2012. Published by Oxford University Press on behalf of the European Society of Human Reproduction and Embryology. All rights reserved.
For Permissions, please email: journals.permissions@oup.com
Materials and Methods

A computer search of pathology records between January 1992 and April 2011 revealed that 31 patients with PEM with anal sphincter involvement received surgical treatment at PUMC Hospital. Their medical records, which included medical history, physical examination (the gynecological and rectal examinations were included), serum levels of CA125 for each patient, surgical records and preoperative and/or post-operative medical treatment, were studied retrospectively. All patients were followed up clinically from 6 to 78 months after the surgery. Outpatient records including whether there were post-operative complications or PEM recurrence were studied retrospectively. A telephone interview was conducted to 29 patients in December 2011, 2 patients (6.5%) were lost to follow-up.

Results

Between January 1992 and April 2011, 17,263 women received surgical treatment for endometriosis at PUMC Hospital. Of these women, 64 (0.37%) had PEM; of whom, 31 (0.18%) had PEM with anal sphincter involvement. The mean age of these 31 cases at the time of surgery was 33.4 (range, 26–43) years. All cases of PEM with anal sphincter involvement had a history of vaginal delivery. In 20 (64.5%) of these patients, endometriotic lesions occurred at the episiotomy site. For the remaining 11 (35.5%) cases, the lesions occurred at the site of perineal laceration.

The mean number of pregnancies in these cases was 2, with a range from 1 to 4, while the mean delivery was 1, with a range from 1 to 2. The median latent period (months from delivery to the women reporting perineal pain or nodule) of these 31 cases was 36 months, with a range from 1 to 204 (95% confidence interval: 23–56) months. All cases had cyclical perineal pain, which was progressive and correlated with their menstrual cycles.

A thorough physical examination, which included a bimanual gynecologic examination, a recto-vaginal abdominal examination and a digital rectal examination (DRE), was performed on each case. The examination revealed a hard perineal nodule associated with the anal sphincter, which corresponded to the episiotomy scar or perineal laceration scar.

The mean serum level of cancer antigen 125 (CA125) was 23.3 U/ml for all cases except 4 (36.9, 38.2, 106 and 122.4 U/ml). Two of these four cases had concomitant ovarian endometrioma. In other words, the serum level of CA125 was elevated in only two (6.5%) cases because of simple PEM with anal sphincter involvement.

All these 31 cases received pelvic ultrasonography, 2 (6.5%) cases had concomitant ovarian cysts which turned out to be ovarian endometriomas in the laparoscopic ovarian cystectomy. Perineal ultrasonography has been used to make the diagnosis of PEM at PUMC Hospital since 2003, and 5 of the 31 cases received this examination. The ultrasound showed an irregular hypoechoic mass at the perineal site of the episiotomy (or in the perianal area), which contained a rounded or oval anechoic area within it. For some patients with PEM, the examination revealed a heterogeneous mass containing cystic anechoic and/or hyperechoic areas. To date, endoanal ultrasonography has not been performed in our hospital due to technical reasons.

Surgical treatment was performed for all 31 cases. Complete narrow excision (NE) was performed on 30 (96.8%) cases, during which a circumferential and deep surgical margin of 0.3–0.5-cm outside the edge of perianal endometrioma was resected. A part of anal sphincter (the largest excised part of anal sphincter was halved) was excised during surgery. Primary anal sphincter repair was performed using the apposition technique in 29 cases and overlapping technique in 1 case. The perineal wound was closed intermittently by 4–0 silk sutures and the stitches were removed 1 week later. Incomplete excision was performed for one (3.2%) case because her perianal endometrioma was too large to excise completely. The average diameter of the excised perianal endometriomas was 2.7 (range: 1.5–4) cm. The diameters of the excised endometriomas of 28 cases of them ranged from 1.5 to 3 cm. For the remaining three patients, the diameters of the excised lesions ranged from 3 to 4 cm.

In the NE group, 10 cases received hormone therapy [including GnRH-agonist (GnRH-a), Nemestran and Depot medroxyprogesterone acetate (DMPA)] both pre- and post-operatively for 1–6 months. In one case GnRH-a was given preoperatively for 3 months, and a Mirena IUD was placed intraoperatively. Nine cases received only GnRH-a for 3–5 months preoperatively. Two cases received only GnRH-a for 4 months post-operatively. The remaining eight patients did not receive any hormone therapy either before or after the surgery. For the patient who received IE, DMPA was given to her both pre- and post-operatively for 3–6 months.

In all 31 cases histopathology revealed islands of endometriomas in perirectal fat and muscle tissues. Endometrial glands appeared throughout the fibro-fatty and external sphincteric muscle tissues (skeleton muscle tissues). Skeleton muscle tissues were disrupted by endometrial glands in some patients. Stromal surrounded most of the glands, but this was seldom uniform or complete [hematoxylin and eosin (H&E), ×100]. All cases were followed up clinically to determine whether there were complications or PEM recurrence. The follow-up period varied from 6 to 78 months and the average follow-up time was 18 months. However, from 18 November 2011 to 10 December 2011 telephone interviews were conducted to 29 cases (two cases were lost to follow-up and the loss ratio of follow-up was 6.5%).
cases had good sphincter control with no episodes of incontinence during follow-up. No complaint of dyspareunia was observed during follow-up. Healing of the perineal wound was satisfactory in 30 (96.8%) cases but was not satisfactory in 1 (3.2%) case. Perineal abscesses were found in that patient 7 days after the surgery. After antimicrobial treatment and drainage, the patient achieved satisfactory healing of the perineal wound. One recurrence of PEM occurred in the NE group 12 months after her surgery on 4 August 2004. The patient just received GnRH-a for 3 months pre-operatively. Failed to visit our clinic and opposed to the possibility of a second surgery for her recurrence, she took oral contraceptives (OCs) periodically by a local doctor’s advice for the past 6 years and the OCs worked well on her. Her perineal pain relieved and her perineal lesion was the size of a peanut till now. Relapse of PEM occurred in the IE patient 6 years after the IE surgery. As the patient was nearing menopause (42 years old) and had been diagnosed with adenomyosis accompanied with severe dysmenorrhea when the PEM recurred, laparoscopic hysterectomy and bilateral salpingo-oophorectomy were performed. The post-operative course was uneventful, and the PEM diminished gradually.

**Discussion**

Endometriosis is one of the most common diseases in women of reproductive age. Endometriotic lesions can occur anywhere in the body, including Caesarean section scars and the episiotomy site. The first reported case of perianal endometriosis was in 1923 (Schickele, 1923). PEM with anal sphincter involvement is extremely rare, and we found only 13 cases of PEM with anal sphincter involvement reported in the literature until now.

According to our retrospective study of the 31 cases of PEM with anal sphincter involvement in our hospital and the literature review we performed, the early diagnosis and treatment of PEM with anal sphincter involvement is important for the prevention of progressive involvement of surrounding tissues (especially the anal sphincter), thus decreasing the risk of post-operative fecal incontinence. A detailed medical history is of great significance for the diagnosis. Three facts should be considered when taking a history: (i) reproductive age; (ii) previous vaginal delivery and (iii) palpable firm nodules near a scar accompanied by cyclic pain and swelling during menses. If these three criteria are met, the diagnostic accuracy of PEM is 100% (Zhu et al., 2009).

In nearly half of all patients with PEM at PUMC Hospital, the endometrioma eroded into the anal sphincter. Thus, it is imperative that further examination (including DRE and endoanal ultrasonography) should be used to confirm whether the anal sphincter is involved in a patient with PEM. Physical examination (including DRE) could provide extremely important additional clues. The mass is generally hard, frequently adjacent to an existing episiotomy scar or previous site of tearing or injury and involved with a part of the anal sphincter. All 31 cases of PEM with anal sphincter involvement at PUMCH met the above-mentioned criteria on history and physical examination. According to the literature, preoperative endoanal ultrasonography is a reliable technique for visualizing perianal endometriosis and for diagnosing anal sphincter involvement. The ultrasonographic features of the lesion are similar to those observed with perineal ultrasonography. Advantage of endoanal ultrasonography over perineal ultrasonography is that it can reveal the involvement of the anal sphincter more clearly. In addition, it enables the surgeon to determine the operative approach and to explain the possible complications of sphincteroplasty to the patient (Bacher et al., 1999; Watanabe et al., 2003; Toyonaga et al., 2006). Till now, endoanal ultrasonography has not been performed yet in our hospital due to technical reasons. Later patients suspected of PEM will receive endoanal ultrasonography examination to evaluate the involvement of anal sphincters in our hospital.

The value of CA125 for diagnosing endometriosis has been found to be slight, with sensitivity described in a meta-analysis (Spaczynski and Duleba, 2003) as ranging between 13 and 36%, though an additional single study found a sensitivity value of 61%. In our study, serum CA125 levels were elevated (>35 U/ml) in two (6.5%) cases due to simple PEM with anal sphincter involvement (of those four cases with elevated serum CA125 levels, two also had ovarian endometrioma). Pelvic examination and ultrasonography should be performed when serum CA125 levels are elevated in patients with PEM with anal sphincter involvement to exclude ovarian endometrioma.

Surgical excision of the endometriotic lesions is the preferred treatment for PEM with anal sphincter involvement. A PubMed search showed only 13 cases of the condition in 11 different case reports since 1957 (Prince and Abrams, 1957; Beischer, 1966; Gordon et al., 1976; Hambrick et al., 1979; Sayfan et al., 1991; Bacher et al., 1999; Dougherty and Hull, 2000; Kanellos et al., 2001; Hernandez-Magro et al., 2002; Barisic et al., 2006; Toyonaga et al., 2006). In the 13 cases of PEM with anal sphincter involvement in the literature, wide excision (WE) and primary sphincteroplasty were performed in 6 cases, NE was performed in 5 cases, IE was performed in 1 case and spontaneous regression after a subsequent delivery was registered in 1 case. In cases where NE was performed, there were no complications associated with this procedure; however, in two cases, recurrence developed and required subsequent hormone therapy (Prince and Abrams, 1957; Gordon et al., 1976). In the group where WE was performed, there were no complications, no incontinence and no recurrence during a variable follow-up period (range of 6–78 months). Based on these studies, WE and primary sphincteroplasty (PSp) are recommended as the best treatment for PEM with anal sphincter involvement (Sayfan et al., 1991; Dougherty and Hull, 2000; Kanellos et al., 2001; Hernandez-Magro et al., 2002; Barisic et al., 2006; Toyonaga et al., 2006). Some authors have suggested that in younger patients, WE with PSp may be optimal, obviating the need for additional therapy. In older patients who were closer to menopause, narrow or incomplete excision with subsequent hormonal therapy could be advantageous (when endometriosis tends to regress) to lessen the risk of incontinence with sphincter resection (Dougherty and Hull, 2000).

In our study, NE and PSp were conducted in 30 (96.8%) patients, while IE was performed in 1 (3.2%) patient because her perineal endometrioma was too large to excise completely. Of these 31 cases, preoperative hormone therapy was prescribed for 21 (67.7%) cases and not prescribed for the rest 10 (32.3%) cases. As they all have the same good outcome, we deem that there is no difference if pre-operative medical therapy is given or not. However, we still recommend the use of preoperative hormone therapy and the reasons are as follows: preoperative hormone therapy could reduce the size of the endometrioma and make the boundaries of these lesions clearer, thus facilitating the complete excision of the lesions and reducing damage to surrounding tissues. Pathological examination of the
resected lesions showed gland atrophy and stroma edema, thus providing evidence for the effectiveness of hormone therapy.

Based on our own experience, NE and PSp may be indicated for the treatment of PEM with anal sphincter involvement.

**Supplementary data**

Supplementary data are available at http://humrep.oxfordjournals.org/.

**Acknowledgements**

The authors gratefully acknowledge help from San Francisco Edit to polish the English in this paper.

**Authors’ roles**

N.C. wrote the first draft and revised the manuscript with L.Z. and J.L., L.Z., Z.L., D.S., J.L. and Q.F. did surgical treatment and outpatient clinic visits for the 31 cases during January 1992 and April 2011 in PUMC Hospital. N.C. and L.Z. reviewed and analysed all the case data of the 31 cases. The specimens were examined pathologically by H.Z. and Q.C.

**Funding**

No external funding was either sought or obtained for this study.

**Conflict of interest**

None declared.

**References**


