Umbilical hernia masking primary umbilical endometriosis – a case report

Elvira Brătilă1, Oana-Maria Ionescu2, Dumitru-Cristinel Badiu3, Costin Berceanu4, Simona Vladăreanu5, Doina Mihaela Pop6, Claudia Mehedină5

1Department of Obstetrics, Gynecology and Neonatology, “Carol Davila” University of Medicine and Pharmacy, Bucharest, Romania; 2Department of Obstetrics, Gynecology and Neonatology, “Carol Davila” University of Medicine and Pharmacy, Bucharest, Romania; 3Department of Surgery, “Carol Davila” University of Medicine and Pharmacy, Bucharest, Romania; 4Department of Obstetrics and Gynecology, University of Medicine and Pharmacy of Craiova, Romania; 5Department of Obstetrics, Gynecology and Neonatology, “Carol Davila” University of Medicine and Pharmacy, Bucharest, Romania; 6Department of Pathology, “Carol Davila” University of Medicine and Pharmacy, Bucharest, Romania

Abstract
Endometriosis is a gynecologic condition affecting mainly the pelvic organs. However, extrapelvic endometriosis has been reported in almost all parts of the body. Umbilical endometriosis, either primary or secondary, is uncommon and has a documented neoplastic risk. We present the case of a 46-year-old woman with a large umbilical hernia associating primary umbilical endometriosis discovered during surgery and confirmed later by pathological and immunohistochemical exams. The patient underwent omphalectomy and partial omentum resection, alongside with mesh abdominal wall repair. The patient was informed about the recurrence risk and was asymptomatic at follow-up consults.

Keywords: umbilicus, hernia, endometriosis, surgery, immunohistochemistry.

Introduction
Endometriosis is defined by the presence of functional endometrial glands and stroma outside the uterine cavity [1, 2]. Affecting up to 10% of women of reproductive age [2], the common location for the ectopic endometrial tissue is the pelvic peritoneum [1], but extrapelvic involvement has been described in almost every area of the female body [2, 3]. If umbilical endometrioma is a rare condition, usually following surgical procedures that involve the umbilicus [2, 4], primary umbilical endometriosis (PUE) is an even rarer encounter [4]. In this paper, we present a case of primary umbilical endometriosis, accidentally found during surgery for umbilical hernia.

Case presentation
We report the case of a 46-year-old woman, presenting to the emergency room at the “Nicoale Malaxa” Clinical Emergency Hospital, Bucharest, Romania, with excruciating abdominal pain without signs of peritonitis or bowel obstruction. The local examination revealed a large (8/10 cm in diameter), very painful and irreducible umbilical hernia with red covering skin. General physical examination was unremarkable. Tracing back her medical history, she had two uncomplicated vaginal deliveries, the last one 23 years prior, while for the last five years she experienced irregular menstrual cycles with severe dysmenorrhea. The umbilical hernia was also first time noted three years prior, but its surgical resolution was ignored despite the medical advice. The patient had no history of any prior abdominal or pelvic surgery. The initial diagnosis was incarceration of the omentum in the umbilical hernia sac and surgical management was offered. The abdominal ultrasound noted a 4 cm hypoechoic nodule within the hernia sac (attached to the umbilical ring), with no bowel loops inside and a normal appearance of the uterus and adnexa. Blood tests were within normal ranges. The informed consent of the patient was obtained both for surgical intervention and the communication of particular clinical aspects the case, which the Ethics Committee of the Hospital also approved. Surgery was performed by a multidisciplinary team, including general surgeons and gynecologists, and it revealed a large umbilical hernia sac containing partially necrotized omentum and a 3/4 cm bluish nodule, adherent to the muscular aponeurotic hernia ring, containing a dark fluid – omphalectomy and partial omentum resection, with abdominal wall reconstruction using a polypropylene mesh was practiced. The postoperative evolution was uneventful, the patient being discharged eight days later. Despite the absence of local signs of recurrence and normal plasma levels of CA125 every three months at follow-up consults, the patient was warned about the recurrence risk.

During surgery, the macroscopic aspect of the nodule and the frozen sections raised the suspicion of primary umbilical endometriosis, since careful peritoneal surface inspection showed absence of other macroscopic endometriosis implants and the patient had a personal medical history lacking pelvic surgery.
The surgical specimens were fixed in 10% formalin, processed in paraffin wax, cut in 4-μm thick sections and stained with Hematoxylin and Eosin (HE). Two independent microscopic examinations were performed.

Abdominal wall endometriosis as seen in the histological sections revealed endometrial stroma and glands dispersed under the epidermis and also in deep dermal tissue (Figure 1). Multiple and different foci of endometriosis, are a replica of hormone induced endometrial proliferation, showing hyperplastic glands with abundant collapsed hemorrhagic endometrial stroma in some areas (Figure 2) and cystic glands lined by a monolayer of low cuboidal epithelium surrounded by endometrial stroma, suggestive for inactive endometrium as seen in postmenopausal women (Figure 3). The host dermal tissue in which the endometrial foci were discovered was edematous.

As for the immunohistochemistry, as expected, we noted positive staining for CK7 in the endometrial glands (Figure 4) and for CD10 in the endometrial stroma, ER (estrogen receptor) and PR (progesterone receptor) nuclear positivity in glands and stroma (Figures 5 and 6) and a significant score for Ki-67 (Figure 7) compatible with the proliferative phase of the endometrium but not as high as it is usually seen in carcinoma. The analysis of the hormonal receptors expression of the selected sections have evidenced the presence of ER in a percentage of approximately 100% in both stromal and glandular epithelium level and, respectively, the presence of PR receptors in a percentage of 60% in the glandular epithelium and negative in the stroma. The Ki-67 marker expression has been reported in 10% of cases in the stroma, with a variable expression from one focus to another – epithelial cells active and passive, expression varying from 2 to 40%. The apoptotic Bcl2 gene was highly expressed at the glandular epithelium level (100%), while in the stroma it varied from one focus to another with an expression from negative to minimum positive. The only differential diagnosis in this case would be endometrial carcinoma, which was refuted by histological grounds and also by immunohistochemistry. Malignant transformation of extra-gonadal endometriosis to carcinoma is a rare event but not unheard of, so one should always be suspicious.

Figure 1 – Ectopic foci of endometrial glands and stroma in the cutaneous and subcutaneous tissue. HE staining, ×100.

Figure 2 – Umbilical endometriosis with recent hemorrhage in the endometrial stroma. HE staining, ×100.

Figure 3 – Atrophied and cystic endometrial glands filled with necrotic material surrounded by endometrial stroma next to edematous dermal tissue. HE staining, ×100.

Figure 4 – Immunohistochemical stain for CK7 highlighting the endometrial epithelial cells in the endometriotic foci, ×200.
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Figure 5 – Positive immunohistochemical nuclear staining for estrogen receptors in glandular epithelial cells and stroma strikingly delineating the endometriotic foci. Immunostaining with anti-ER antibody, ×100.

Figure 6 – Positive immunohistochemical nuclear reaction for progesterone receptors in glandular epithelial cells and stroma. Immunostaining with anti-PR antibody, ×200.

Figure 7 – Positive immunohistochemical nuclear staining for the proliferation marker Ki-67 showing an uneven distribution in the glandular epithelium of the endometriotic foci, with indexes similar to those of the proliferative phase of the endometrium. Immunostaining with anti-Ki-67 antibody, ×200.

Discussion

Endometriosis is a benign disease found in reproductive age women, usually affecting the pelvic peritoneum [2, 5]. Umbilical endometriosis (UE), known as Villar’s nodule [6] is quite rare (0.5–1% of all cases of extrapelvic endometriosis) [2] and often occurs on umbilical scars secondary to obstetric or gynecologic procedures involving the umbilicus [1]. Primary umbilical endometriosis is an even rarer phenomenon [4], a 2010 meta-analysis reporting a little more than 120 cases reported worldwide [7].

The rarity of this disorder represents a challenge for the clinicians. The condition often presents itself as a single or a multiple [8] brownish or dark-bluish, painful umbilical swelling [2, 9, 10], varying in size [11]. The slow-growing umbilical tumors [9] can be entirely asymptomatic or associating, in 75% of cases, symptoms like pain, swelling, discharge or bleeding correlated with the menstrual cycle [9, 11]. Pelvic endometriosis symptoms can also be associated [11]. In our case, the patient had very little symptoms, masked by the umbilical hernia.

The differential diagnosis of an umbilical nodule should include abscess, subcutaneous cyst, desmoid tumor, hematoma, lipoma, lymphadenopathy, lymphoma, melanoma, soft tissue sarcoma or metastatic tumors [12]. Furthermore, the association between umbilical endometrioma and umbilical hernia is uncommon, the first condition being misrecognized sometimes during the surgical intervention, causing the recurrence of the painful manifestations after the surgery [13, 14].

Preoperative color Doppler ultrasonography scan [10] determined whether the cystic or solid component of the mass, but this is not a specific feature for abdominal wall endometriosis [4, 15]. Computed tomography (CT) scan or magnetic resonance imaging (MRI) shows the extent of the disease [4]. Endometrioma appear as a solid well circumscribed mass, homogeneously hyperintense in T1-weighted MRI sequences [4, 10, 16], while the signal loss within the lesion seen on T2-weighted images helps differentiate endometriomas from other blood containing lesions [11].

Dermoscopy is used for the differential diagnosis with cutaneous malignant neoplasms (basal cell and squamous cell carcinomas, melanoma) [10, 11, 17].

In 2011, Fernandes et al. recommended the use of fine-needle aspiration cytology (FNAC) for preoperative diagnosis, emphasizing on the cytological features of cutaneous and subcutaneous endometriosis related to cyclic hormonal changes. The cytology smears are generally cellular with epithelial and stromal fragments admixed with hemorrhage and hemosiderin laden macrophages [9].

If FNAC smears are hemorrhagic, the endometriosis diagnose can be missed [9]. In these cases, the microscopic examination of the surgically excised specimen is mandatory. On high-resolution microscopy, although the pathological appearance of the lesion varies according to the phase of the cycle, it is always characterized by the presence of endometrial glands and stroma in the mid or deep dermis. The endometrial glands are formed from tall columnar epithelium with basophilic cytoplasm and basally located nuclei, forming irregular glandular lumina, sometimes with a marked mitotic activity or
metaplastic changes (tubal, oxyphilic, hobnail, mucinous, papillary syncytial). The stroma is usually edematous, composed of spindle cells with metaplastic changes (smooth muscle metaplasia, decidualization, stromal endometriosis, elastosis). Menstrual bleeding into the dermis leads to hemosiderin deposition (which is seen with the Perls stain) and chronic inflammation. Atypical changes include reactive atypia and atypical mitoses [18].

Immunohistochemical examination is carried out for the differential diagnosis with different malignancies. Endometrial epithelial cells express estrogen and progesterone receptors, keratin 7, the human epithelial antigen (BerEP4), Ki-67 and are negative for keratin 20 and calretinin, similar to normal human endometrial epithelium (keratin 7+/keratin 20−). CD10 is used as a marker for stromal cells. Other less specific markers expressed by stromal cells include smooth-muscle actin, von Willebrand factor, caldesmon, desmin, VEGF and COX-2 [18].

The pathogenesis of this disease remains a mystery [1]. While the pathogenesis of secondary endometriosis is easy to explain [8], speculations have been made on the pathogenesis of primary endometriosis [3]. In this regard, several theories have been proposed – the migration of endometrial cells to the umbilicus through the abdominal cavity, the lymphatic system and the umbilical vessels [5] or cellular proliferation of endometrial cells from initial extraperitoneal disease along the urachus [2]. A new pathogenic theory emerged recently, linking the occurrence of endometriosis to environmental factors like dioxins food contamination [19].


The authors declare that they have no conflict of interests.

References

[19] Carvalho BR, Rosa e Silva JC, Rosa e Silva AC, Barbosa Hde F,


Corresponding author
Costin Berceanu, University Lecturer, MD, PhD, Department of Obstetrics and Gynecology, University of Medicine and Pharmacy of Craiova, Emergency University Hospital Craiova, 2 Petru Rareş Street, 200349 Craiova, Romania;
Phone +40722–728 180, e-mail: dr_berceanu@yahoo.com

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