Endometriosis: symptoms and diagnosis

Päivi Härkki
GKS
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Epidemiology

- estrogen dependent inflammatory disease
- endometrium-like tissue outside the uterine cavity (Bulun 2009)
- prevalence: 10% of women, 25-50% of women with pain and/or infertility (Giudice and Kao 2004)
- 40% of adolescents with chronic pelvic >6 months had endometriosis in laparoscopy (Vercellini 1989)
- 70% of adolescents with pain not responding to contraceptives have endometriosis in laparoscopy (Laufer 2003)
Life with endometriosis

- nulliparous women with short and heavy menstrual bleeding have increased risk (Vercellini 2004)
- diagnostic delay is 6-9 years (Husby 2003, Ballard 2006)
- 7-fold risk for first-degree relatives (Moen 1993)
- cost estimates of endometriosis (Simoens 2007)
  - annual healthcare cost/patient $2801
  - annual cost of productivity loss/patient $1023
  - $22 billion/year in US
  - costs are higher than with Crohn’s disease or migraine
- great impact on woman’s quality of life (Jones 2004)
Menstrual pain

- **Endometriosis**
  - pain begins several days before bleeding
  - painkillers needed for many days, effect is not sufficient
  - contraceptives ineffective
  - absence from work/school is usual

- **Dysmenorrhea**
  - pain begins together with bleeding
  - painkillers only for 1-2 days, effect is good
  - contraceptives effective
  - no absence from work/school
Other symptoms

- site specific cyclic pain
  - sacrouterine ligament: dyspareunia
  - rectovaginal space: dyspareunia, painful defecation
  - bowel: painful defecation, painful bowel movements
  - bladder: painful micturation

- ovarion cyst/tumour
- infertility
- dysfunctional uterine bleeding
- unusual cyclical symptoms in rare cases
Diagnosis and treatment of endometriosis

- Peritoneal endometriosis
  - underdiagnosed, well treated?
- Endometrioma
  - diagnosis easy, well treated?
- Deep endometriosis
  - underdiagnoses, undertreated
Possible sites of distribution of endometriosis:

- Pelvic peritoneum
- Fallopian tube
- Sigmoid colon
- Ovary
- Surface of uterus
- Myometrium (adenomyosis)
- Uterosacral ligament
- Rectovaginal septum
- Cervix
- Vagina
- Groin
- Vulva and Bartholin's gland
- Perineum
Peritoneal endometriosis

- gynecological examination often normal
- transvaginal ultrasonography (TVS) often normal
- laboratory normal, sometimes Ca-125 elevated
- symptoms most important findings
- empirical medical therapy with contraceptives allowed
- contraceptives non-responding women should undergo laparoscopy
Diagnosis with endometrial biopsy

- endometrium of endometriosis women differs from that of healthy women (Bukub 2009)
- sensory nerve fibers are found in the functional layer of endometrium and in endometriotic lesions of endometriosis patients (Tokyshige 2006, Wang 2009)
- increased levels of sensory nerve fibers were detected in endometrial biopsy of endometriosis patients (A-Jefot et al 2009, Bokor et al 2009)
Laparoscopy
Laparoscopy
Laparoscopy is golden standard of diagnosis

- histology confirms endometriosis of black lesions in 64% and of atypical lesions in 42% of cases (Marchino et al. Fertil Steril 2005)
- endometriosis was confirmed by histology in 54% of the excised lesions (Marchino et al 2005)
- 67% of lesions identified at surgery contained histologic evidence of endometriosis (Stratton et al 2003)
Endometrioma

- often asymptomatic
- easy diagnosis with TVS
- differential diagnosis
  - hemorrhagic cyst, dermoid, mucinous cyst
- CA-125 often elevated (50-200) non-specifically
- unusual finding with papils in elderly woman = suspicion of malignancy
  - HE4 (human epididymal secretory protein E4) elevated in cancer not in endometriosis (Huhtinen 2009)
  - risk of malignancy index / CT
Transvaginal ultrasonography of endometrioma

Round and homogenous cyst
Regular thick cyst walls
Hypoechogenic
Ovarian tissue seen separately
No papils
Might be multinucleated
Endometrioma
Laparoscopy
Rectovaginal endometriosis

- specific symptoms
  - dyspareunia
  - pain at defecation
  - pressure at pelvis

- clinical status important !!
  - blue lesions in vagina
  - painful nodule in posterior fornix

- diagnosis in clinical
pelvic examination

Courtesy of P. Catapano
Rectovaginal endometriosis: status
Rectovaginal endometriosis: status
Accuracy of clinical examination in rectovaginal endometriosis
(Chapron et al. J Am Assoc Gynecol Laparosc 2002)

- Speculum examination revealed 14.4% of rectovaginal endometriosis
- Classical painful, nodules were found in 43% of clinical examinations
- Rectovaginal examination revealed 35% rectovaginal disease and 33% of sacrouterine ligament endometriosis
- BUT: 91% of rectovaginal nodules were found in preoperative clinical examination in Helsinki material 2000-2004 (Tarjanne et al. J Minim Invasive Gynecol 2009)
Rectovaginal endometriosis: MRI
Rectum endometriosis

- severe pain at defecation
- blood in stools
- MRI
- TVS
- sigmoideoscopy
- preoperative diagnosis of invasion
  important!!
Rectum endometriosis: MRI and TVS

- MRI good to diagnose rectal invasion (Bazot 2007)
- MRI diagnoses also lesions in ureter, sigmoid etc
- TVS more difficult but can diagnose rectal invasion (Guerriero 2008, Hudelist 2009)
Deep and superficial endometriosis in sacrouterine ligament
Rectovaginal nodule might be difficult to see in laparoscopy
Rectal endometriosis
Sigmoid endometriosis
Bladder endometriosis

- typically pain at micturition and menstrual pain
- sometimes hematuria
- painful nodule between uterus and bladder in gynecological examination
- nodule can be detected with ultrasonography or MRI
- blue/white protruding lesion in cystoscopy
- hard, white, scarred nodule in bladder in laparoscopy
Diagnosis of bladder endometriosis

TVS

MRI
MRI- bladder endometriosis
Bladder endometriosis
Ureter endometriosis

- mainly asymptomatic because the stricture occurs slowly
- suspect if rectovaginal nodule is >3cm
- hydronephroses is diagnosed with ultrasonoraphy, intravenous urography or MRI
Prevalence of ureteral lesion according to the size of the nodule

Donnez, Fertil Steril 2002

<table>
<thead>
<tr>
<th>size of nodule</th>
<th>nodule (n)</th>
<th>ureter lesion</th>
<th>prevalence</th>
</tr>
</thead>
<tbody>
<tr>
<td>&gt; 3 cm</td>
<td>96</td>
<td>9</td>
<td>9.3%</td>
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<tr>
<td>2-3 cm</td>
<td>97</td>
<td>1</td>
<td>1%</td>
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<tr>
<td>&lt; 2 cm</td>
<td>61</td>
<td>0</td>
<td>0%</td>
</tr>
<tr>
<td>total</td>
<td>254</td>
<td>10</td>
<td>3.9%</td>
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MRI - hydroureter
Conclusion

- Symptoms and clinical examination are most important for diagnosis.
- TVS good for diagnosis of endometrioma and bladder endometriosis.
- Deep endometriosis has site-specific symptoms.
- Diagnosis of rectovaginal endometriosis is clinical.
- MRI good for diagnosis of deep endometriosis.
- Laparoscopy is still the golden standard for diagnosis.
Lopuksi

- Endometrioosin diagnostiikassa paastaan pitkälle tyyppillisten oireiden ja kliinisten statuslöydösten perusteella

- Vaginallisella ultraäänitutkimuksella löytyy endometrioomat ja virtsarakon syvät pesäkkeet

- Endometrioomat ja kaikki syvät pesäkkeet löytyvät hyvin MRI-tutkimuksessa, mutta tutkimusta tarvitaan harvoin

- Suolen syvien pesäkkeiden diagnoistiikassa MRI olisi paras menetelmä, jos pystyttäisiin tunnistamaan potilaat, jotka tarvitsevat kuvauksen

- Rektovaginaalinen endometrioosi on ainoa endometrioosin muoto, joka voidaan varmuudella kliinisesti diagnosoida ilman leikkausta

- Muissa tapauksissa leikkaus on edelleen ainoa tapa tehdä varma diagnoosi