Hypermobile Ehler Danlos Syndrome: a differential diagnosis for gynecologist

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INTRODUCTION

• Rare heritable disease, characterized by a defect in collagen production
• First described by E. Ehlers and H-A Danlos
• 3 major types of EDS
  - **Hypermobile type** - mobile loose joints, chronic pain, variable skin involvement
  - **Classical type** – skin hyperextensibility, abnormal wound healing, joint hypermobility
  - **Vascular type**, more severe
• Increased propensity to hemorrhage
Patients and methods

- 387 consecutive patients with hypermobile EDS included
- May 2012 - December 2014
- Standardized questionnaire (in presence of MD)
  - EDS symptoms
  - Gynecological symptomatology
  - Pregnancy outcomes
- Gynecological examination systematically performed by a senior gynecologist
- Pelvic sonogram and MRI in case of suspicion of endometriosis
RESULTS

• Diagnosis of EDS made at a mean of 35.2 ± 14.2 years of age
• Mean age of onset of symptoms of EDS : 12.4 ± 11.8 years old

Main symptoms:
• Joint pains, in 96.6% of the patients
• Fatigue, in 92.2%
• Proprioceptive disorders, in 87.2%
• Skin fragility, in 86.1%
• Spontaneous hematoma, in 78.8%
• Joint laxity, in 46.6% of the patients
Gynecological symptoms

- **Bleeding disorders**
  - *Menorrhagia* reported by 77.6% of the patients
  - *Metrorrhagia*, in 22.5% of them

- **Gynecological pain**
  - *Dysmenorrhea*, in 73.1% of them (primary and lasting)
  - Deep *dyspareunia*, in 42.7% of the patients
  - 42.3% described burning, itching, tearing pains on penetration, during and following intercourse.
Endometriosis

- Dysmenorrhea, menorrhagia and deep dyspareunia
  => Highly suggestive of endometriosis
- Based on clinical history (including laparoscopy), clinical examination ± pelvic sonogram and MRI in case of doubt:
  Rate of pelvic endometriosis = 4.9% (as in the general pop)
- Endometriosis may be over-diagnosed in these patients
- Risk of being operated incorrectly, with hemorrhagic complications and healing disorders
Obstetrical outcomes

• **No infertility**: 724 pregnancies in 224 women
• **Miscarriages** occurred in **27.2%** of pregnancies
• Recurrent miscarriages (≥3): **12.5%** of the women

Might suggest adenomyosis. However, the prevalence of adenomyosis
Obstetric and gynecologic challenges in women with Ehlers-Danlos syndrome.


- Gynecological and obstetrical outcomes in patients with EDS
- 775 reproductive-aged women with EDS
- Higher rates of metrorragia (32.9%), dysmenorrhea (92.5%) and dyspareunia (77%)
- Higher rates of infertility (44.1%) and endometriosis (44%) than in the general population.
- However, emailed questionnaire only, with no clinical evaluation of the symptoms reported by patients
- In our study, no difference in rates of infertility and endometriosis was found with the general population
DISCUSSION

Bleeding disorders and dysmenorrhea:
- Progestin only effectives; combined contraceptives
- Tranexamic acid ineffective

Vaginal or deep dyspareunia:
- Topical estrogens twice/week
- Hyaluronic acid
- Benzydamine chlorhydrate after intercourses
- Lidocaine gel just before the intercourse in the most severe cases

• Advices for delivery or surgery (skin fragility and potential bleeding risk):
  - hemostasis++
  - non absorbable suture
CONCLUSION

• Gynecological symptoms are extremely frequent in EDS

• Important to exclude the diagnosis of EDS in women consulting with cardinal symptoms suggesting endometriosis: join and muscular pains, abnormal scars, hyperlaxity…