Cell-free DNA has potential as a low-invasive diagnostic marker for early endometriosis

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Background

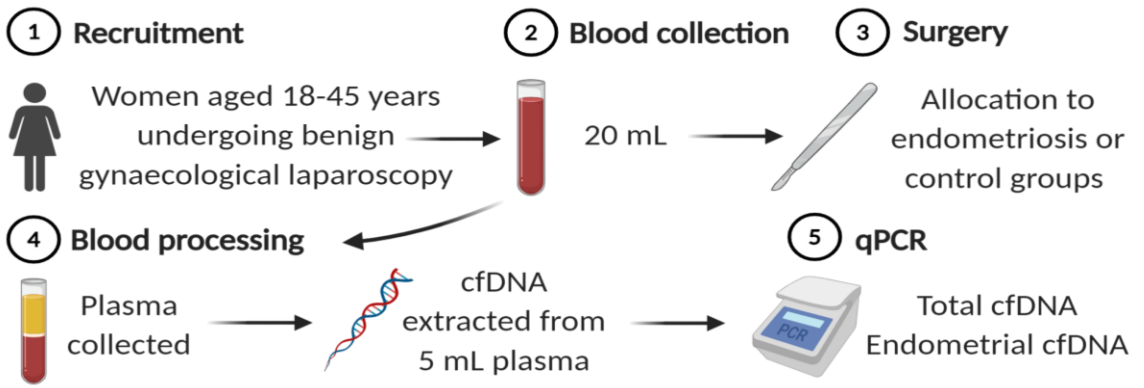
A **non-** or **low-invasive diagnostic test** would decrease the substantial **diagnostic delay** currently reported for endometriosis.[1]

One small retrospective study has suggested that **cell-free DNA** (cfDNA) is **elevated** in the **plasma** of women with endometriosis.[2] Additionally, we propose **endometrial cfDNA** as a **novel biomarker**.

Aim

To evaluate **total** and **endometrial-derived cfDNA** as low-invasive biomarkers for endometriosis in women **with** and **without** laparoscopically-confirmed **endometriosis**.

Methodology



Results

28 women with endometriosis

- **10** (36%) Superficial endometriosis
- **18** (64%) Deep endometriosis

15 controls

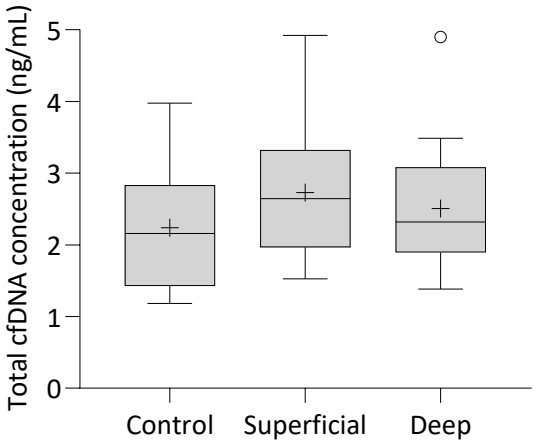
- **8** (53%) Leiomyomata
- **6** (40%) Adenomyosis
- **1** (7%) Appendicitis
- **2** (13%) No pathology

Total cfDNA concentration

Control
2.24 ± 0.89 ng/mL

Endometriosis
2.56 ± 0.92 ng/mL

p=0.274



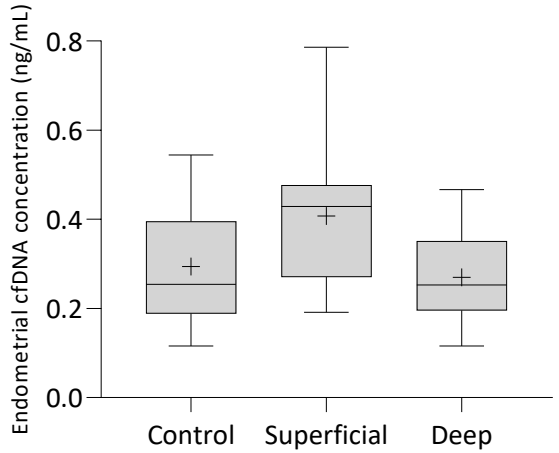
p=0.426

Endometrial cfDNA concentration

Control
0.25 ng/mL (IQR 0.19-0.40)

Endometriosis
0.29 ng/mL (IQR 0.20-0.43)

p=0.593



Significantly higher concentration in superficial vs deep endometriosis (p=0.025), but not compared to controls

Conclusions

The quantification of plasma **endometrial cfDNA** is **feasible**.

The **small sample size** and **population heterogeneity** may have contributed to the **negative findings**.

Next steps

Reinvestigation of **endometrial cfDNA** in a larger cohort consisting of women with **superficial endometriosis** and controls **without coexisting pelvic pathology**.

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References: 1. Nnoaham, K.E., *et al.* Impact of endometriosis on quality of life and work productivity: a multicenter study across ten countries. *Fertil Steril* **96**, 366-373 e368 (2011). 2. Zachariah, R., *et al.* Circulating cell-free DNA as a potential biomarker for minimal and mild endometriosis. *Reproductive BioMedicine Online* **18**, 407-411 (2009).