Background: Endometriosis is a complex disease defined as the presence of endometrial tissue outside the uterine cavity. Although it afflicts approximately 10% of the women in reproductive age, there are no reliable diagnostic biomarkers and a surgical procedure is still required to make a definitive diagnosis. The nature of this disease is heterogeneous and comprises peritoneal, deep infiltrating and ovarian endometriosis (OE) that represent different entities with different pathogenesis.

Approach: In the search for novel biomarkers of OE, we selected 152 genes from the GeneLogic database based on results of genome-wide expression analysis of OE, ovarian and endometrial tissue and performed low-density array analysis in 11 OE and 9 control endometrium samples. Among differentially expressed genes biglycan was selected for further analyses. We evaluated biglycan serum and peritoneal fluid concentrations (ELISA, 55 OE and 38 controls) as well as the protein levels (Western blotting, 55 tissue samples of OE and different controls) in women with OE and in control women.

Key Findings: We identified 78 genes as differentially expressed. Functional annotation showed that 25 and 22 genes are associated with the biological terms "secreted" and "extracellular region", respectively. Biglycan was detected in ectopic endometrium of OE patients and ovarian benign cysts tissue but not in normal eutopic endometrium of healthy women. Biglycan concentrations were significantly increased in peritoneal but not in serum samples of OE patients.

Implications & Conclusion: Our systematic biomarker study revealed differential expression of several genes that encode potential novel biomarkers and have not previously been associated with OE. Biglycan is a new biomarker candidate and should be further studied.