EPIGENOMIC STUDY REVEALS HIGHLY SIMILAR ENDOMETRIAL DNA METHYLATION PROFILES OF WOMEN WITH AND WITHOUT ENDOMETRIOSIS

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Introduction: Altered epigenetic DNA modifications in endometrial cells are thought to be one possible cause for endometriosis development. This assumption is supported by large-scale endometrial DNA methylation studies indicating a distinct DNA methylation profile in women with endometriosis. However, there is evidence that endometrial DNA methylation signature is influenced by the menstrual cycle phase and therefore, the aim of this study was to analyse only secretory phase endometria of patients with endometriosis and healthy women to find genome-wide DNA methylation differences.

Methods: Illumina’s Infinium Human-Methylation450K bead-chips were used to determine the methylation profiles of early-, mid- and late secretory endometria of 24 patients with endometriosis and 17 mid-secretory endometria of healthy women. Differentially methylated regions (DMRs) were identified as regions with more than three consecutive CpG sites (FDR corrected P value 0.05).

Results: No significant differences were detected between the methylation profiles of early- and mid-secretory endometria from healthy women and patients. However, the results showed more than 6700 DMRs when healthy endometria were compared to late secretory endometria of patients. Among endometriosis patients, comparisons of early- and mid-secretory endometria with late secretory endometria revealed 4811 and 7108 DMRs, respectively.

Conclusions: This study provides no evidence of DNA methylation differences in mid-secretory endometria of endometriosis patients compared to healthy women. However, the results of this study indicate that there are remarkable changes in endometrial methylation profile during the secretory phase irrespective of the disease status, and the menstrual cycle day should be considered in order to detect disease-specific alterations.