**11. Applicability of polygenic risk scores in endometriosis clinical presentation**

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**Abstract**

**Background:** Risk prediction is an essential part of preventative medicine and in recent years genomic information has become an interesting factor in risk models. Polygenic risk scores (PRS) combine the effect of many genetic variations into a single score which has been shown to have predictive value for many diseases. This study aimed to investigate the association between PRS for endometriosis and the clinical presentation of the disease.

**Methods:** Women with endometriosis (N = 172) were identified at the Department of Gynecology. All participants answered questionnaires regarding sociodemographic factors, lifestyle habits and medical history, registered bowel symptoms on the Visual Analog Scale for Irritable Bowel Syndrome and passed blood samples. DNA was extracted and samples were genotyped, and a PRS was calculated based on previous genome-wide association studies of endometriosis. Inflammatory proteins and TSH receptor antibodies (TRAb) in serum were analyzed.

**Results:** Inverse associations were identified between PRS and spread of endometriosis, involvement of the gastrointestinal tract and hormone treatment. However, significance was lost when calculated as p for trend and the specificity and sensitivity were low. There were no correlations between PRS and TRAb or inflammatory proteins.

**Conclusion:** The findings indicate that specific PRS should be developed to predict clinical presentations in patient with endometriosis.

**Keywords:** Endometriosis; Gastrointestinal symptoms; Genetics; Inflammatory proteins; Polygenic risk score.