Research project J3-1755

Biomarkers of endometriosis: transcriptomics and proteomics approach

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Project group: SICRIS

Project description:

Endometriosis is a female **reproductive disorder**, characterized by the presence of endometrial tissue outside the uterine cavity. **The estimated prevalence** of endometriosis **is 6–10% in reproductive age women** and **30%–50% in women with infertility and/or pain**. Endometriosis is associated with disturbed immune system, aberrant apoptosis, cell adhesion, degradation of extracellular matrix, angiogenesis, cell communication and other pathological processes. The symptoms of endometriosis are non-specific and include debilitating pain and infertility. The gold standard for diagnosis of endometriosis is laparoscopy, a surgical visual inspection of the pelvic organs. As a result of these non-specific symptoms and surgical diagnosis, there is an average delay of 6.7 years between the onset of symptoms and the diagnosis. At present **there are no biomarkers for non-invasive diagnosis of endometriosis**. Different approaches have already been used in the search for blood biomarkers; early research focused mainly on individual proteins, while in the last decades also omics approaches have been introduced into biomarker discovery. Changes in micro RNA and protein profiles have been reported in endometriotic lesions and these alterations can be reflected in blood of these patients.

The aims of the project are: 1) to **identify panels of micro RNAs and proteins** specific for patients with endometriosis and 2) to **develop diagnostic algorithms** for clinical validation and application. These aims will be achieved by the state-of-the-art transcriptomics and proteomics approaches using miRNA and antibody arrays in the discovery phase and low density miRNA arrays and multiplex immunological assays (Luminex®) in the validation phase. **The unique aspect** of the proposal **is evaluation of > 2 500 miRNAs and 900 proteins** in **~400 plasma samples** from patients with and without endometriosis and **development of diagnostic algorithms based on combination of proteomic, metabolomic and clinical data**.

Our case-control study will include patients with endometriosis like symptoms, stratified into case and control groups based on the laparoscopic and histological findings. The targeted transcriptomics and proteomics study will comprise discovery phase to identify new miRNAs and proteins associated with endometriosis on a smaller number of samples and the validation phase where we aim to determine concentrations of these miRNAs and proteins on all case and control samples. Diagnostic algorithms will be constructed using sound bioinformatics and multivariate statistics.

The proposed project will identify novel miRNA molecules and proteins associated with endometriosis and will pinpoint the disturbed molecular processes. The project may thus indirectly contribute to earlier treatment and may also advance understanding of pathophysiology of this intriguing disease, which may in a long term lead to development of new options for treatment. The study will be performed in collaboration with Department of Gynaecology, University Medical Centre Ljubljana, and Centre for Functional Genomics and Biochips at the Faculty of Medicine, Medical University Vienna, Austria and University of Tartu, Estonia.

Bibliography: SICRIS