

Background: Uterine fibroid (UF) or leiomyoma is the most frequent benign tumour upon lower genital tract and represents the most frequent indication for hysterectomy. The aetiology remains still unknown. The genetic factors contributing for the development of UF are being intensively investigated. The aim of our study was to look for possible genetic markers which could be used as prognostic tools for evaluation of an increased risk for development of UF.

Methods: The study group enrolled 102 patients diagnosed with UF and 145 healthy controls. Ultrasonographic examination of the pelvis was performed and a single blood sample was taken in all women. Histological verification followed the surgery in the patient group. The principal of the cytokine gene polymorphisms detection is based on PCR reaction with sequence-specific primers.

Results: A large spectrum of Th1/Th2 cytokine gene polymorphisms in patients with uterine fibroid was compared with control group. The frequencies of the majority of tested cytokine gene SNP in the patient cohort were not statistically different from the cytokine SNP in the control group. However, an intriguing association between polymorphisms of the IL-4 gene promotor at positions -590 C/T and -33 C/T, and the risk of leiomyoma was observed. The CC genotype of IL-4 at position -590 and -33 was less frequent in the patient group than in the control group ($p=0.03$). Besides IL-4, we observed different genotype distribution of the gene for TNF- α -308A/G. The frequency of genotype AA was higher in the younger (<35 years) patient group ($p=0.02$).

Conclusion: This study as a first has analysed such a large spectrum of cytokines and their polymorphism using molecular and genetic techniques (PCR-SSP). Our results suggest that certain cytokine gene polymorphisms, especially of the IL-4 and TNF- α genes may be associated with increased risk for development of uterine fibroid. Further investigation will be needed for elucidation of the mechanisms responsible for these associations.