

Abstract

Currently infertility affects 10 to 15 percent of couples. In nearly 50% of all cases male factor contributes to infertility of the couple. Majority of causes of male infertility remains unexplained. For this reason, finding simple and clinically useful tools for improving male infertility diagnostics can be important at present. Based on the analysis of sperm transcriptome, in this diploma thesis we aimed to find genes that show differential expression between normal and pathological sperm, this could provide information about molecular basis of male infertility, moreover, expression profile of such genes in sperm could be employed for noninvasive diagnosis of male infertility. This study was conducted by using 67 sperm samples, including 16 control sperm samples from fertile men. The samples of infertile patients were divided into 3 groups according to morphology and motility using results of standard sperm evaluation according to WHO.

First group included 20 patients diagnosed with astenoteratozoospermia (low sperm motility and abnormal morphology), second group was 15 patients diagnosed with asthenozoospermia (normal morphology and low sperm motility), the third group comprised 16 samples from infertile patients with normal spermiogram. 16 control samples were from fertile men (conception of their partners in less than 12 months before sperm evaluation) with normal spermiogram.

Transcriptome analysis was performed using microarray technology on 24 semen samples from 3 groups of infertile man and one control group (7 astenoteratozoospermic, 5 asthenozoospermic, 6 infertile with normal sperm parameters and 6 fertile controls). From candidate genes with possible differential expression, we confirmed 8 expression profiles using quantitative real-time PCR. Moreover, we confirmed differential expression of one gene, *ARHGAP17*, using the entire sample set of 16 patients with idiopathic infertility with normal spermiogram compared to 16 fertile men.