Breast carcinoma is the most common cancer among women in our country and worldwide. One of the obstacles to successful therapy is a multidrug resistance. It can be caused by different factors, such as overexpression of ABC transporters, or decreased expression of SLC transporters, deregulation of drug metabolizing enzymes, variability of the targets of anticancer drugs, failure of apoptosis or increased capacity of repair genes. The aim of this study was to search for associations of genes of drug transport and metabolism with the prognosis of patients or response to chemotherapy. From the view of preventive medicine, this aim constitutes an important part of both secondary and tertiary prevention of cancer, i.e., discovery of markers enabling optimal therapy selection for each patient, decreasing the risk of disease progression to advanced and resistant stage, and elimination of side effects of chemotherapy.

The expression profile of ATP-binding cassette (ABC) transporters (49 genes), cytochromes P450 (CYPs, 10 genes), aldo-keto reductases (AKRs, 13 genes) and carbonyl reductase 1 was analyzed in the tumor and adjacent non-tumor control tissues in a cohort of neoadjuvantly treated patients. Genes deregulated in tumors compared with control tissues or genes associated with clinical data were assessed in an independent set of pretreatment patients. Protein levels in tumor tissues were confirmed by immunoblotting. Genetic variability was assessed in selected candidate genes.

The vast majority of genes was deregulated in tumors compared with control tissues. A number of significant associations of intratumoral transcript levels with clinical characteristics of the patients were found. Most interestingly, transcript levels of ABCA12, ABCA13, ABCD2, AKR1C1, AKR1C2, and CYP2W1 associated with patients´ response to neoadjuvant therapy. Moreover; AKR1C2, AKR7A3, CYP3A4, and CYP2B6 associated with disease-free survival and ABCC1, ABCC8, AKR7A3, and CYP2B6 associated with the expression of hormonal receptors in both sets. Several non-coding polymorphisms in the functional nucleotide binding domain 1 of *ABCC1* gene significantly associated with disease-free survival of patients. New variants in *ABCC8* and *ABCD2* genes were found using targeted exome sequencing and *in silico* methods predicted their functional effects.

Candidate genes which might be clinically significant were discovered. The genetic variability in some of these genes may explain prognostic or predictive roles in breast carcinoma patients. Potential biomarkers of prognosis and response of patients to chemotherapy found by this study should be further followed to confirm their clinical significance and decipher underlying mechanism of action.