Abstract

Molecular genetics has become an essential part of diagnostic and prognostic methods in oncology of solid tumors. One representative of human solid tumors was selected for this study - breast carcinoma, which is the most common malignancy among women. Breast carcinomas were investigated by immunohistochemistry (IHC), fluorescence in situ hybridisation (FISH) and comparative genomic hybridization (CGH).

Trastuzumab (Herceptin) is the cornerstone of molecular biology treatment of breast cancer women with HER2/NEU modification. The accurate assessment of HER2 status is therefore critical for identifying of patients who may benefit from trastuzumab-based therapy. HER2/NEU gene amplification and overexpresion were evaluated by IHC and FISH. The first aim of this study was to correlate the test results of IHC and FISH with HER2/NEU gene amplification in breast cancer patients. IHC was performed with the DAKO Cytomation HercepTest and FISH with the PathVysion Probe kit. A total of 213 (10,56 %) of 2017 patients were positive by IHC. Our results indicate that high-level and normal expresion of HER2/NEU status of breast carcinomas can by detected reliably both by IHC and FISH. However, bordeline results, especially those with 2+ IHC, should by interpreted with caution using both IHC and FISH with standardised methodology.

The second aim was to examine the use of CGH to screen breast tumors for copy number changes (DNA deletions and gains). Invasive breast carcinomas are characterized by a complex pattern of chromosomal alterations. We obtained succeful results in 15 out of 17 cases. The most frequent DNA sequence copy number changes were DNA gains on 1q, 4q and 8q and losses on 1p, 16q, 17p, 19 and 22. Hence, the CGH technique represents a specific tool for the whole-genome screening of the chromosomal abnormalities in human solid tumors.