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Determination of EGFR gene somatic mutations in tissues and plasma of patients with advanced non-small cell lung cancer

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Abstract

The presence of activating mutations in the EGFR gene influences cell proliferation, angiogenesis, and increases metastatic ability; it has a significant impact on the choice of medical therapy of non-small cell lung cancer (NSCLC). The use of targeted therapy with tyrosine kinase inhibitors requires performance of appropriate genetic tests. The aim of this study was to design a real-time PCR-based diagnostic kit for fast and cheap of EGFR mutations testing in paraffin blocks and plasma, and kit validation using samples from patients with NSCLC, and also comparative estimation of diagnostic features of real-time PCR with wild type blocking and digital PCR for mutation testing in blood plasma. The study included 156 patients with various types of adenocarcinoma differentiation. It was designed a simple and efficient real-time PCR-based method of detecting L858R activating mutation and del19 deletion in the EGFR gene for DNA isolated from paraffin blocks. Kit for EGFR mutations was validated using 411 samples of paraffin blocks. The proposed system showed high efficiency for DNA testing from paraffin blocks: a concordance with results of testing with theascreen® EGFR RGQ PCR Kit ("Qiagen", Germany) was 100%. It has been shown the possibility of using this test system for the detection of mutations in plasma.

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Keywords

activating mutations in EGFR gene, non-small cell lung cancer, plasma, real-time PCR