

建立快速高分辨點分析法檢測急性骨髓性白血病 NPM1 突變檢驗方法

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Establishment of rapid high-resolution melting analysis to detect NPM1 mutation in acute myeloid leukemia

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BACKGROUND: Nucleophosmin mutations of exon 12 (NPM1 mutations) represent the most frequent molecular aberration in about 35% of acute myeloid leukaemia (AML) patients. High resolution melting (HRM) is a novel screening method that enables rapid identification of mutation positive DNA samples.

METHODS and MATERIALS: We developed HRM assays to detect NPM1 mutations. Twenty-one AML patients were enrolled. The research project was reviewed and approved by the IRB committee of our hospital (DMR99-IRB-108). All mutations are confirmed with directly sequencing.

RESULTS: Herein, we established molecular diagnosis of NPM1 mutation assays for AML. Analysis of a series of 32 patient specimens revealed 11 positive for NPM1 mutation and 21 negatives. In addition, NPM1 positive mutation is referred to as c.960 ins TCTG (Type I) were 9 patients and c.960 ins GTCG and c.962 ins TCGA heterozygote insertion were 1 patients, respectively.

CONCLUSIONS: This study demonstrates that HRM is a rapid and efficient method of screening NK-AML samples for NPM1 mutations. We expect to set up the molecular diagnostic technique for clinical laboratory service

乳房微鈣化組織檢體之 UBE2C 腫瘤生物標記之分析

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Analysis of UBE2C tumor biomarker in breast microcalcifications tissue

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Breast microcalcification is an important feature of early breast cancer. The data of mammography and histology reveal that 15% of breast calcification could be further defined as breast carcinoma. UBE2C, an ubiquitin conjugation E2 enzyme, is a newly identified biomarker candidate expressed in various solid tumor and cancer cells. It contributes to the process of ubiquitin-mediated proteasome degradation in cell cycle. To evaluate the clinical application of UBE2C in diagnosis of early breast cancer, we measured the UBE2C mRNA expression level from 28 sets of breast biopsy samples; each set contains normal and microcalcification tissue from individual. The quantitative PCR indicated that approximately 70% microcalcification samples with UBE2C mRNA expressed were detected; immunohistochemistry also revealed 68% of microcalcification tissues were UBE2C positive. In addition, UBE2C over expressed breast cancer cells of MCF-7 and MDA-MB-231 were rapid proliferation. Contrarily, reduced cell growth rate and increased cell death were observed in UBE2C knockdown cells. Our data suggest that the UBE2C promoted cell proliferation may contribute to breast ontogenesis. The molecular mechanism of UBE2C in breast cancer will be further investigated.

