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Genome-wide detection of loss of heterozygosity and copy number variation in a human lung large cell carcinoma cell line by affymetrix single-nucleotide polymorphism array 500K

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摘要

Background and objective Loss of heterozygosity (LOH) and Copy number copy number variation (CNV) of DNA sequences is a common feature of cancer genomes, which is thought to be linked to tumorigenesis and progression. High-density single-nucleotide polymorphism (SNP) genotyping array are able to provided a genotype and copy number information with genome-wide coverage, which is suitable for the analysis of complex genetic alterations present in cancer. Thus a human lung large cell carcinoma cell line NL9980 was assayed for the global profile of LOH and CNV. Methods Genomic DNA from the cell line was screened for LOH and CNV using Affymetrix GeneChip® Human Mapping array 500K Set. The hybridization intensity data of 500 000 SNP loci was analyzed using Affymetrix proprietary software for genotyping and copy number of each locus, and a genome-wide map of LOH and CNV of the cell line was constructed. Results The SNP call rate of array Nsp I (-262K) was 95.14%, and the rate of Sty I (-238K) was 97.15%. The both call rates of the components of 500K array set were in excess of 93%, the cardinal quality control standard. LOH profiles of the sample were across all chromosomes, and most of CN gains and losses regions were found on chromosomes such as 2, 3, 4, 5, 7, 10, 11, and 18. Conclusion The results have shown that there were complex genetic alterations present in NL9980. And it is possible to achieve high performance outcomes using Affymetrix SNP array 500K to interrogate LOH and CNV in lung cancer genome. This advance of high-resolution with allelic information should substantially improve the ability to further understanding of the genetic basis of lung cancers.

关键词

Lung neoplasms; Single-nucleotide polymorphism; Loss of heterozygosity; Gene copy number; Microarray

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