Rare somatic mutation of pro-apoptotic BAX and BAK genes in common human cancers

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ABSTRACT

Aims and background. BAX and BAK are both pro-apoptotic Bcl-2 proteins and are essential for the pathway of intrinsic apoptosis. Apoptosis in cancer cells is frequently inactivated by somatic mutations. The aim of the study was to see whether somatic mutations of BAX and BAK genes are characteristics of common human cancers.

Methods. We analyzed somatic mutation of BAX and BAK genes in 47 gastric, 47 colorectal, 47 breast, 47 lung and 47 prostate carcinomas, and 47 acute leukemias by a polymerase chain reaction and single-strand conformation polymorphism assay.

Results. We identified BAX gene mutations in one colon (2.1%) and three gastric (6.4%) cancers. All of the mutations were frameshift mutations in the G8 repeat sequences and were detected in cancers with high microsatellite instability (36.4%). There was no evidence of BAX mutation in the other cancers, nor was somatic mutation of the BAK gene detected in the cancers.

Conclusions. Our data indicate that somatic mutation of BAX and BAK genes are rare in the common cancers (besides the cancers with high microsatellite instability) and suggest that neither BAX nor BAK mutation may causally be implicated in their tumorigenesis.

Key words: apoptosis, BAK, BAX, cancer, mutation.

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