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IMPORTANCE OF NOVEL SEQUENCE ALTERATIONS IN THE *FHIT* GENE ON FORMATION OF BREAST CANCER

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Aims and background: **The character, role and impact of *FHIT* gene alterations, for which recent studies have shown that the gene has a role in the early stage of carcinogenesis in breast cancer, are still unclear. Thus, the current study evaluated *FHIT* gene mutations from breast tissue of women with malignant and benign breast disease and to elucidate the frequency and type of mutations in this gene.**

Patients and methods: **Mutations in exons 5-9 of the *FHIT* gene were screened using the intronic primer pairs in 83 breast (67 malignant and 16 benign) tissue samples by single-strand conformational polymorphism and sequencing analysis.**

Results: ***FHIT* mutations were detected in 13 of the 67 malignant cases (19.4%) and 2 of the 16 benign cases (12.5%). Four different sequence variants were determined: two novel frame shift mutations (codon 90 insA, codon 146 delT), one intronic novel mutation (IVS8 -17 insA), and one previously identified silent transition type alteration (codon 88 C to T). In addition, determination of this silent alteration caused formation of new exonic splicing enhancer (ESE) motifs on mutated sequences by using the ESEfinder program.**

Conclusions: **Our data contribute significantly to that currently known about the presence of *FHIT* gene mutations on the formation of breast cancer.**

Key words: benign breast diseases, exonic splicing enhancers, *FHIT* gene, malignant breast disease, novel mutations, single-strand conformational polymorphism.

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