RET

Frequent germ line mutations in RET proto-oncogene exons 10 and 11 in hereditary medullary thyroid carcinomas of Iranian patients

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Abstract

Aims: Medullary thyroid carcinoma (MTC) occurs in both sporadic and hereditary forms. The important role of the RET proto-oncogene mutations in MTC development has been well demonstrated. The aim of this study was to determine the prevalence of common mutations in exons 10 and 11 of RET proto-oncogene among Iranian patients with MTC and their family members.

Methods: 76 MTC patients and 21 members of their families were selected voluntarily by call method and then were examined. Genomic DNAs were extracted from peripheral blood samples of participants according to “standard salting out/proteinase K” method and exons 10 and 11 of RET proto-oncogene were proliferated using PCR method. Then, PCR products were exposed to certain limiting enzyme and polymorphism pattern of parts length obtained from limiting enzyme operation on poly acrylic amide gel was analyzed.

Results: By analysis of patterns obtained from RFLP, 13 mutations were detected in exons 10 and 11 of RET proto-oncogene, of which 8 mutations were related to the codon 634 (exon 11) and 5 mutations were related to codon 618 (exon 10). The number of mutations in patients was 12 cases and in their family members was one case.

Conclusion: The prevalence of RET proto-oncogene mutations in exons 10 and 11 among Iranian MTC patients and their family members is relatively high (13.4%).

Keywords: Hereditary MTC, Germline Mutation, RET Proto-Oncogene