عنوان مقاله:

Point Mutations in RET Proto-Oncogene Exon ۱۰ in Patients with Medullary Thyroid Carcinoma

محل انتشار:

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خلاصه مقاله:

Background & Aims: Thyroid cancer is the most common endocrine malignancy. Medullary thyroid carcinoma (MTC) is an aggressive malignant tumor arising from parafollicular cells of the thyroid. MTC occurs in hereditary (۲۵%, hMTC) or sporadic (۷۵%, sMTC) forms. The hMTC form has an autosomal dominant inheritance. RET proto-oncogene mutations, especially the ۱۰, ۱۱, and ۱۶ exones, are associated with MTC. The aim of this study was to determine the type and frequency of RET proto-oncogene exon ۱۰ in patients with MTC. Methods: The study participants included ۳۴۷ individuals, including ۲۰۷ patients and ۱۴۰ of their first degree relatives. Genomic DNA was extracted from peripheral leukocytes using salting out/Proteinase K method. All individuals were tested for RET mutations in exon ۱۰ using polymerase chain reaction (PCR)- DNA sequencing method. Results: A total of ۱۴ germline missense RET mutations were identified in cysteine codons ۶۱۱, ۶۱۸, and ۶۲۰ in ۱۱ patients(۱۰ mutation in males, ۴ in females), and ۳ of their first-degree relatives (frequency: ۳.۶%) which were as follows: four C۶۱۱Y (three FMTC, one relative), one C۶۱۸R (FMTC), one C۶۱۸S (sMTC), one C۶۲۰G (sMTC), four C۶۲۰R (one FMTC, three sMTC), and three C۶۲۰F (one FMTC, two relatives). The most predominant mutations in exon ۱۰ in our FMTC and sMTC patients were C۶۱۱Y and C۶۲۰R, respectively. We did not find any mutations in cysteine codon ۶۰۹. Conclusion: In the present study, ۶ different types of

missense mutations were identified in exon ۱۰ of RET in the nonsyndromic form of MTC. Based on the results of this study, mutation detection using DNA sequencing in exons ۱۰, ۱۱, and ۱۶ of RET in patients with MTC and their relatives .is recommended

کلمات کلیدی:

medullary thyroid cancer, RET proto-oncogene, Exon ۱۰, Germline mutation, Iranian population