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## Molecular analysis of RET proto-oncogene in patients with Medullary Thyroid Carcinoma

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**Introduction:** Medullary Thyroid Carcinoma (MTC) accounts for 5-10% of Thyroid cancers and derives from the para-follicular C cells of the Thyroid gland. MTC has worse prognosis compare to the most common form of Thyroid cancer, papillary Thyroid carcinoma (PTC) which accounts for 60-80% of Thyroid carcinomas and derives from follicular cells. There are 2 forms of sporadic and hereditary MTC, accounts for 75% and 25% of the cases, respectively. The inherited form is transmitted as autosomal dominant manner and presents either as isolated familial MTC (FMTC) or as a part of syndromic cancer: multiple endocrine neoplasia type 2 (MEN2).

**Methods:** A total of 42 MTC patients from 35 independent families participated in this study. All of the patients were clinically and pathologically diagnosed as MTC. Four families had more than one affected and characterized as FMTC or MEN2 and 31 of cases were sporadic. Genomic DNA was extracted from PBMC. PCR amplification followed by direct DNA sequencing of the RET gene hot spots (exons 10, 11, 13, 14, 15 and 16) was done in all affected. Offspring's of affected were subjected to RET analysis, if the affected parent had pathogenic mutation in RET gene.

**Results & Conclusions:** We identified RET germ line mutation in 14 families (4 familial and 10 apparently sporadic cases), out of 35 families. Interestingly, 16 asymptomatic family members were carried pathogenic RET germ line mutations that directed to total Thyroidectomy. The highest prevalence of mutations was found in exon 11 (9 out of 14/ about 64% of mutations).

### Biography

Mohsen Ghadami completed his MD at Tehran University of Medical Sciences in Iran and his PhD in Medical Genetics at Nagasaki University in Japan. He obtained his Post-doctoral studies in the field of Gene therapy and Stem Cell Therapy at Baylor College of Medicine in Houston, TX and Texas University of Medical Branch, in Galveston, TX, USA. He is Director of Plenary Genetic Center at Tehran University of Medical Sciences and has published more than 20 papers.

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