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Functional Analysis of Thyroid Peroxidase (TPO) Gene Mutations Detected in Patients with Thyroid Dyshormonogenesis

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Abstract : Purpose: Thyroid peroxidase (TPO) is the key enzyme in the biosynthesis of thyroid hormones. We aimed to identify the spectrum of mutations in the TPO gene leading to hypothyroidism in the population of West Bengal to establish the genetic etiology of the disease. Methods: 200 hypothyroid patients (case) and their corresponding sex and age matched 200 normal individuals (control) were screened depending on their clinical manifestations. Genomic DNA was isolated from peripheral blood samples and TPO gene (Exon 7 to Exon 14) was amplified by PCR. The PCR products were subjected to sequencing to identify mutations. Results: Single nucleotide changes such as Glu 641 Lys, Asp 668 Asn, Thr 725 Pro, Asp 620 Asn, Ser 398 Thr, and Ala 373 Ser were found. Changes in the TPO were assayed in vitro to compare mutant and wild-type activities. Five mutants were enzymatically inactive in the guaiacol and iodide assays. This is a strong indication that the mutations are present at crucial positions of the TPO gene, resulting in inactivated TPO. Key Findings: The results of this study may help to develop a genetic screening protocol for goiter and hypothyroidism in the population of West Bengal.

Keywords : thyroid peroxidase, hypothyroidism, mutation, in vitro assay, transfection
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