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The associations of polymorphisms of TSH receptor and thyroid hormone receptor genes with L-thyroxine treatment in hypothyroid patients.

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Abstract: **OBJECTIVE:** To investigate the possible association between response to levothyroxine (L-T4) doses in hypothyroid patients and variation in thyroid stimulating hormone receptor (TSHR) gene and thyroid hormone receptor (THR?) gene. **DESIGN:** This cross-sectional correlation study included 228 patients with primary hypothyroidism who were using L-T4 replacement therapy. Thyroid function test was performed using standard techniques. Genotyping of rs939348 of the THR? gene, and rs2268458 and rs2239610 of the TSHR gene was performed using the polymerase chain reaction-based restriction fragment length polymorphism assay (PCR-RFLP). Patient history of illness, medication and compliance data were collected using the patients' medical files. **RESULTS:** The THR? rs939348 polymorphism was associated with L-T4 replacement doses in hypothyroid patients and in central obesity. No significant correlation was detected between the examined SNPs to TSHR and L-T4 doses or the different clinical and biochemical parameters. Finally, L-T4 dose was associated with lower BMI, waist circumference and TSH, and higher free T4 (fT4) among hypothyroid patients. **CONCLUSIONS:** Whereas the two tested TSHR polymorphisms were not associated with the dose of T4, the THR? rs939348 polymorphism was associated with L-T4 dose and central obesity among hypothyroid patients. T4 dose was also associated with multiple beneficial effects among hypothyroid patients.