Resistance to Thyroid Hormone Caused by Heterozygous Mutation of Thyroid Hormone Receptor B Gene c.G1378A Report of One Chinese Pedigree and Literature Review

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Abstract

A 30-year-old female with clinical manifestations of palpitations and goiter was admitted to our department of endocrinology. Laboratory tests showed elevated thyroid hormone with non-suppressed TSH. Genetic analysis identified heterozygous mutation of the THRB exon10 c.G1378A (p.E460K). The proband's kindreds had the same mutation, but their clinical manifestations were different.

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