

# A Very Rare Thyroid Hormone Resistance Case Having Heterozygous Mutation in THRB Genes

Mehmet Keskin<sup>1</sup>, Seniha Kiremitci Yilmaz<sup>2</sup>, Emel H. Aytac Kaplan<sup>1</sup>, Murat Karaoglan<sup>1</sup>, Kadri Karaer<sup>3</sup>, Ozlem Keskin<sup>4</sup>

<sup>1</sup> Gaziantep University Faculty of Medicine, Department of Pediatric Endocrinology

<sup>2</sup> Istanbul Haseki Education and Research Hospital

<sup>3</sup> Gaziantep Dr. Ersin Arslan Training and Research Hospital, Medical Genetics

<sup>4</sup> Gaziantep University Faculty of Medicine Children's Department

## INTRODUCTION

Thyroid hormone resistance is a rare autosomal dominant disorder. In the pathogenesis of the disease mutations have been reported in two types of thyroid hormone receptors, called alpha and beta, in mutations, deletions or cofactors required to demonstrate receptor effect in the beta receptor gene. The symptoms vary according to the cases. Here; a thyroid hormone resistance phenomenon is presented which is found incidentally and does not require treatment.

## CASE

A 10-year-old girl was diagnosed with thyroid hormone resistance. Late speech and walking narrative body weight: 25 kg (10p), height: 137.5 cm (25p), system examinations were natural.

TSH	ft4	ft3	Tg	otoantikör
4,1	2,86	9,3	110	negative

In the genetic test for thyroid hormone resistance; THRB **c949G>A (p.A317T) (p.Ala317Thr)** (Heterozygous) mutation was detected.

This mutation was limited in the literature. Your parents' thyroid and genetic tests were normal. In the metabolic parameters; Bone age was in accordance with calendar age, lipids were at normal level. The case is monitored without treatment.

## DISCUSSION

Thyroid hormone resistance is an autosomal dominant disorder. It can be 15% sporadic. There was no evidence of a thyroid hormone resistance in the family of ours. Patients with complete resistance present with symptoms of severe hyperthyroidism. Asymptomatic asymptomatic hypophysis suggests resistance. Only cases with peripheral resistance come with goitre and mild hypothyroid findings. Your presence clinic was not following it. Treatment is directed at the symptom. There are only cases where the clinic is recovering with beta blocker therapy. Surgery may be needed in patients who can not receive medical treatment Our case is now asymptomatic.

## CONCLUSION

In this study; a rarely seen thyroid hormone resistance is presented as a result of abnormal thyroid tests

## References

[J. Qinuo, MD,<sup>a,b</sup> Bao-An Wang, MD,<sup>a</sup> Chen-Zhi Wang, MD,<sup>a</sup> Min Wang,<sup>c</sup> Ju-Ming g-Hua GLu, MD,<sup>a</sup> Zhao-Hui Ly, MD,<sup>a,\\*</sup> and Yi-Ming Mu, MD<sup>a,\\*</sup> \*Medicine \(Baltimore\)\*. 2016 Aug; 95\(33\): e4415. Published online 2016 Aug 19. Thyroid hormone resistance syndrome caused by heterozygous A317T mutation in thyroid hormone receptor  \$\beta\$  gene Report of one Chinese pedigree and review of the literature](#)