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

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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.

<table>
<thead>
<tr>
<th>TSH</th>
<th>FT4</th>
<th>FT3</th>
<th>Tg</th>
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<tbody>
<tr>
<td>4.1</td>
<td>2.86</td>
<td>9.3</td>
<td>110</td>
<td>negative</td>
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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 hypopituitarism 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