TWO PATIENTS WITH RESISTANCE TO THYROID HORMONES

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OBJECTIVE: Resistance to thyroid hormone (RTH) is an inherited syndrome characterized by reduced sensitivity of target tissues to thyroid hormone. We describe the clinical, biochemical data and mutation analysis of two patients and their families with (RTH).

METHOD: We conducted clinical studies and genetic analysis of these two patients and their families. Genomic DNA extracted from peripheric blood sample. Whole gene sequence analysis performed.

RESULTS: First patient referred to pediatric endocrinology department for hyperthroidism associated with supraventricular tachycardia and thyroid hormone levels consistent with RTH. We found heterozygous c.962A>G mutation in THRB gene (Figure-1). The mother and siblings of this patient had no mutation in this gene. We could not evaluate this patient's father. Second patient admitted for hyperactivity and referred for the abnormalities in thyroid function tests. We found heterozygous c.1378G>A mutation in THRB gene in this patient and his father and brother (Figure-2).
There were no goiter in any of the two patients and their family members

	Free T4 (ng/dL) (0,7-1,48)	FreeT3 (pg/mL) (1,71-3,71)	TSH (µIU/mL) (3,5-4,94)	CILINICAL PRESENTATION	MUTATION
PATIENT 1	3,36	8,15	1,94	Supraventricular tachycardia	c.962A>G
PATIENT 2	2,2	6,34	3,23	Attention deficit hyperactivity disorder	c.1378G>A
PATIENT 3 (Sibling of the Patient 2)	2,08	7,23	2,85	Attention deficit hyperactivity disorder	c.1378G>A

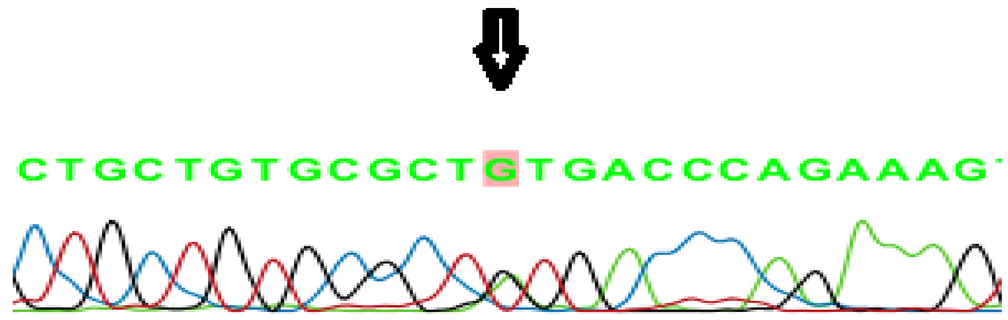

Figure-1: Heterozygous c.962 A>G mutation in THRB gene

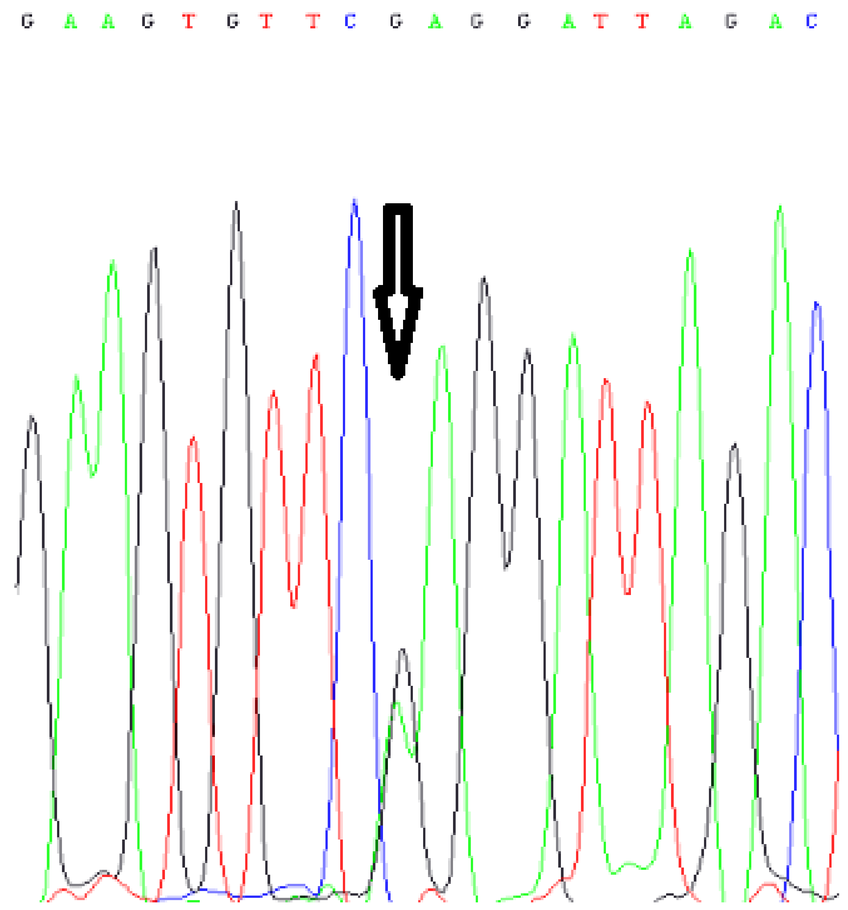

Figure-2: Heterozygous c.1378 G>A mutation in THRB gene

DISCUSSION AND CONCLUSION : Goiter, hyperactivity and tachycardia are the most common clinical features in the patients with RTH syndrome . Diagnosis of RTH depends on the characteristic elevations in thyroid hormone and the exclusion of other causes of hyperthyroxinemia. When RTH is suspected, the diagnosis should be confirmed by direct sequencing of the THRB gene to identify mutations.

There is no conflict of interest

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