

A Rare Endocrine Causes Of Tachycardia: Refetoff Syndrome

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Introduction:

Thyroid hormone resistance (Refetoff syndrome) is a syndrome that caused by the decrease in susceptibility to thyroid hormone in end-organs. The incidence was reported as one in 40,000 live births. Mutations in thyroid hormone receptors play role in the pathogenesis. Typical laboratory findings are elevated T3, T4 levels and normal or mildly elevated TSH levels. Same mutations may lead to different symptoms in different patients because of different end organ expressions for a mutation. There are different therapy strategies like thyroid hormone replacement for patients with hypothyroidism or antithyroid treatment for patients with hyperthyroidism. However only propranolol therapy may be sufficient for patients who has only tachycardia compliant. We aimed to report a case with Refetoff Syndrome, tachycardia caused by thyroid hormone resistance ameliorated with propranolol treatment.

Case Report

Nine years old boy admitted to hospital with palpitation. In physical examination, he had tachycardia with 126/minute heart rate. Other physical examination findings were normal. His electrocardiogram revealed sinus tachycardia. No other rhythm problem was seen in 24 hour electrocardiogram monitoring. In laboratory examination, free T4 level was 4,27 ng/dl (0,96-1,77) and TSH was 2,87 μ IU/ml(0,7-5,97). Thyroid auto antibodies were negative and thyroid ultrasonography was normal. Laboratory findings indicated thyroid hormone resistance. Genetic analysis was performed. c.926> G mutation which is responsible for Y321C amino acid was detected as heterozygous positive. He was given propranolol therapy: 2 mg/kg/day and after one month, in the second control he didn't have any tachycardia compliant.

Discussion

The sensitivity of peripheral tissues to thyroid hormone is different. While some patients have hyperthyroidism or hypothyroidism signs, some of them are asymptomatic. So that, there is no certain consensus about treatment. Propranolol takes control adrenergic symptoms and decrease T4 to T3 conversion by inhibiting 5-deiodination path. Propranolol treatment only by itself may be sufficient for the treatment of a this rare disease called Refetoff syndrome.