Resistance to Thyroid Hormone

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Resistance to thyroid hormone (RTH) is an inherited syndrome caused by a genetic mutation, which leads to a decreased response of thyroid hormone at the peripheral tissues. It could be a challenge to diagnose because patients can be asymptomatic or present with symptoms of hyperthyroidism. The biochemical hallmark of this syndrome is an elevated FT4, FT3 with a non-suppressed thyroid-stimulating hormone (TSH).

We report a case of thirty-five-year-old male who presented with palpitations. A thyroid uptake scan (using intravenous Tc-99m pertechnetate) was highly suggestive of Graves' disease. THRβ gene showed a heterozygous c.1009A>G, p.(Thr337Ala) variant. The result supports the clinical diagnosis of THR caused by a mutation in the THRβ gene. He was treated with propranolol and Rivaroxaban (Xarelto) 15 mg twice daily, the ECG demonstrated controlled atrial fibrillation.

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