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Thyroid Hormone Resistance With a Novel Mutation

Abstract:

Syndrome of thyroid hormone resistance (THR) is a rare inherited condition characterized by a reduced responsiveness of the tissues to thyroid hormone. The syndrome is caused primarily by mutations in the thyroid hormone receptor beta (THRB) gene, leading to impaired hormone receptor function. It is a diagnosis of exclusion and often leads to delays in establishing the diagnosis. Management is usually conservative, as over-treating can be unnecessary and potentially detrimental. Our case report aims to highlight the changes in thyroid function tests and the subtle presenting symptoms of this disease so that clinicians are more mindful of this rare condition. It brings to attention the importance of follow-up to monitor the lab values and reach an accurate diagnosis. We also report a novel mutation identified in the THRB gene.

Keywords: hyperthyroidism, thyroid disorder, thyroid function test, thyroid hormone receptor mutation, thyroid, nodule size, thyroid-stimulating hormone (TSH)