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A case series of thyroid hormone resistance

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Thyroid hormone resistance is a rare condition where there is an impaired sensitivity of target tissues to thyroid hormone. This leads to a situation where both the thyroid hormone levels and the thyroid stimulating hormone (TSH) are raised, as TSH is not suppressed as would normally be expected. Incidence of this condition is around 1 in 40,000 live births. TR-beta gene mutation is the most common cause of thyroid hormone resistance. Clinical manifestations are dependent on the type of mutant thyroid hormone receptor and each target tissue's predominant thyroid hormone receptor expression. Affected patients can present with a range of hyperthyroid or hypothyroid signs and symptoms. Majority of the cases described so far have an autosomal dominant inheritance. In this case series, we describe two young children and their father who have thyroid hormone resistance. They all possess a TR beta- gene defect secondary to a heterozygous mutation. Due to the widely variable signs and symptoms and the non-typical trend of laboratory markers in thyroid hormone resistance, it can be difficult for the clinician to diagnose if one is not familiar with it. Thus, this entity should be taken into consideration when one encounters a patient with elevated serum FT4, unsuppressed TSH and decreased serum T4/T3 ratio.