Title :Resistance to thyroid hormone

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Introduction:

Resistance to thyroid hormone Resistance to thyroid hormone (RTH), first described in 1967 and characterized by elevated circulating TH (T4, T3) and nonsuppressed thyroid-stimulating hormone (TSH) levels together with variable tissue refractoriness to hormone action, is mediated by defects in thyroid hormone receptor β. It is a rare syndrome with usually elevated levels of TSH, T4 and T3. Here we present a case where TSH was normal and was referred for hyperthyroidism , but the unusual clinical picture suggested something more.

Case report:

A 6 year 7 month old boy was referred with diagnosis of thyrotoxicosis. He was born out of non -consanguineous marriage by normal vaginal delivery, pre term baby at 7 months, appropriate for gestational age. There was nohistory of antenatal complications. However gross developmental delay was there and had history of poor scholastic performance. On examination, his height was 100cm, Ht SD Score being -3.6, weight was 15 kg , arm span of 101 cm and an upper segment by lower segment ratio of 1.12. He had mild exophthalmos, goitre, without any bruit. He had tachycardia with a heart rate of 126/min and other systemic examination being normal. His height age was 3y 9m, weight age was 3y 6m and bone age was 3y 6m suggestive of attenuated growth pattern. Thyroid function tests showed TSH - 1.71, T3 – 214 (60 – 200) ng/dland T4 - 12.47(4.5 - 12) mcg/dl . Other laboratory work up for short stature was normal. Clinical features of short stature, developmental delay were suggestive of hypothyroidism whereas tachycardia, exophthalmos and goitre were suggestive of hyperthyroidism. TSH was normal , however T3 and T4Differential diagnoses of drugs like levothyroxine , amiodarone were ruled out by history. Differentials of thyroid hormone transporter defect and resistance to thyroid hormone were considered. To confirm the diagnosis Werner's test was done which showed an elevation in the SHBG levels after 7days of 50mcg T3 orally from 71.4 to 100.7nmol/L. Hence the diagnosis of resistance to thyroid hormone was confirmed. He was started on T3 supplementation, showed a gain of 9.5cm in height which further supported diagnosis of resistance to thyroid hormone.

Conclusions:

Resistance to thyroid hormone is a diagnosis which should be considered even when TSH is normal and not elevated , when clinical features of both hypothyroidism and hyperthyroidism are present. In such scenarios, it is very important to note even the mildest abnormalities of thyroid function tests.