



A Child with Resistance to Thyroid Hormone without Thyroid Hormone Receptor Gene Mutation: A 20-Year Follow-Up

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Résumé en anglais

We report here the 20-year follow-up study of a male subject diagnosed at 15 months of age as a sporadic case of pituitary resistance to thyroid hormone on the combination of clinical hyperthyroidism, elevated serum thyroid hormone (TH) levels and inappropriate thyrotropin (TSH). On d-thyroxine (D-T4) therapy from 30 months of age to 12.5 years, hyperactivity and hyperthyroid signs and symptoms as well as growth abnormalities improved, serum l-thyroxine (L-T4) enantiomer normalized, and basal and stimulated TSH decreased significantly without complete suppression. After 8 years off D-T4, at 20 years of age, clinical status was normal despite persisting high TH levels and inappropriate TSH. Evolution of serum markers of TH action and echocardiography measurements followed up from 15 months to 20 years of age either in basal condition or on triiodothyronine (T3), as well as the sequential determination of bone mineral density suggest differences in the tissue responses to T3: normal in bone with a high remodelling rate, heterogeneity for various hepatic markers, and decreased at heart level. No mutations were found in the coding sequence of TR β 1, TR β 2, TR α 1, RXR γ , SMRT, NCoR1, and NCoA1. In this patient the putative long-term effects of the persisting high bone resorption are unknown.

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