Thyroid hormone receptor beta (THRB) gene testing in resistance to thyroid hormone (RTH)

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Citation
Thyroid hormone receptor beta (THRB) gene testing in resistance to thyroid hormone (RTH) Lansdale: HAYES, Inc.. Genetic Testing Publication. 2012

Authors' conclusions
Resistance to thyroid hormone (RTH) is an endocrine disorder characterized by reduced tissue responsiveness to circulating thyroid hormone. The condition, which is estimated to occur in approximately 1 in 40,000 individuals, may be diagnosed in patients with elevated levels of the thyroid hormones thyroxine (T4) and triiodothyronine (T3), along with normal to high levels of thyroid-stimulating hormone (TSH). The activity of thyroid hormone, which functions in growth, development, and the regulation of body metabolism, is mediated by thyroid hormone receptor alpha (THRA) and thyroid hormone receptor beta (THRB). RTH most often results from deleterious variants in the THRB gene, which is located on chromosome 3 at band p24.3 and encodes a ligand-dependent transcription factor. In the vast majority of cases, THRB-related RTH is inherited in an autosomal dominant manner. However, autosomal recessive cases have been reported, typically the result of whole gene deletions. Clinically, patients with autosomal dominant RTH may be asymptomatic or may present with a variable combination of hyperthyroid and hypothyroid symptoms, including goiter, tachycardia, hyperactivity, anxiety, insomnia, growth retardation, developmental delay, intellectual disability, delayed bone age, and hearing loss. The clinical manifestations in patients with autosomal recessive RTH are similar but often more severe, with significant intellectual disability and hearing deficits. Most RTH patients are clinically asymptomatic and receive no treatment for the condition. For symptomatic individuals, standard treatments for hyperthyroidism and hypothyroidism are typically ineffective. However, patients may benefit from the treatment of specific symptoms, and some patients with signs of thyrotoxicosis may respond to 3,5,3'-triiodothyroacetic acid (TRIAC), a thyroid hormone analog that may help decrease the secretion and activity of TSH.

Final publication URL
The report may be purchased from: http://www.hayesinc.com/hayes/crd/?crd=14195

Indexing Status
Subject indexing assigned by CRD

MeSH
Thyroid Hormone Receptors betas; Genetic Testing

Language Published
English

Country of organisation
United States

English summary
An English language summary is available.

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AccessionNumber
32013000118

Date abstract record published
28/02/2013