

Resistance to Thyroid Hormone: Laboratory Support of Diagnosis and Management

Resistance to thyroid hormone (RTH) is characterized by reduced end-organ responsiveness of thyroid hormone relative to circulating hormone levels.¹ In 85% of cases, it is caused by mutation of the thyroid hormone receptor beta gene (*THRB*). Patients are usually identified with elevated levels of free or total T4 and/or T3 in association

with normal or slightly elevated thyroid stimulating hormone (TSH). Clinical and artifactual reasons for hyperthyroxinemia and nonsuppressed TSH must be considered before a diagnosis of RTH can be made (Figure). Tests available for the diagnosis and management of RTH are listed in the Table.

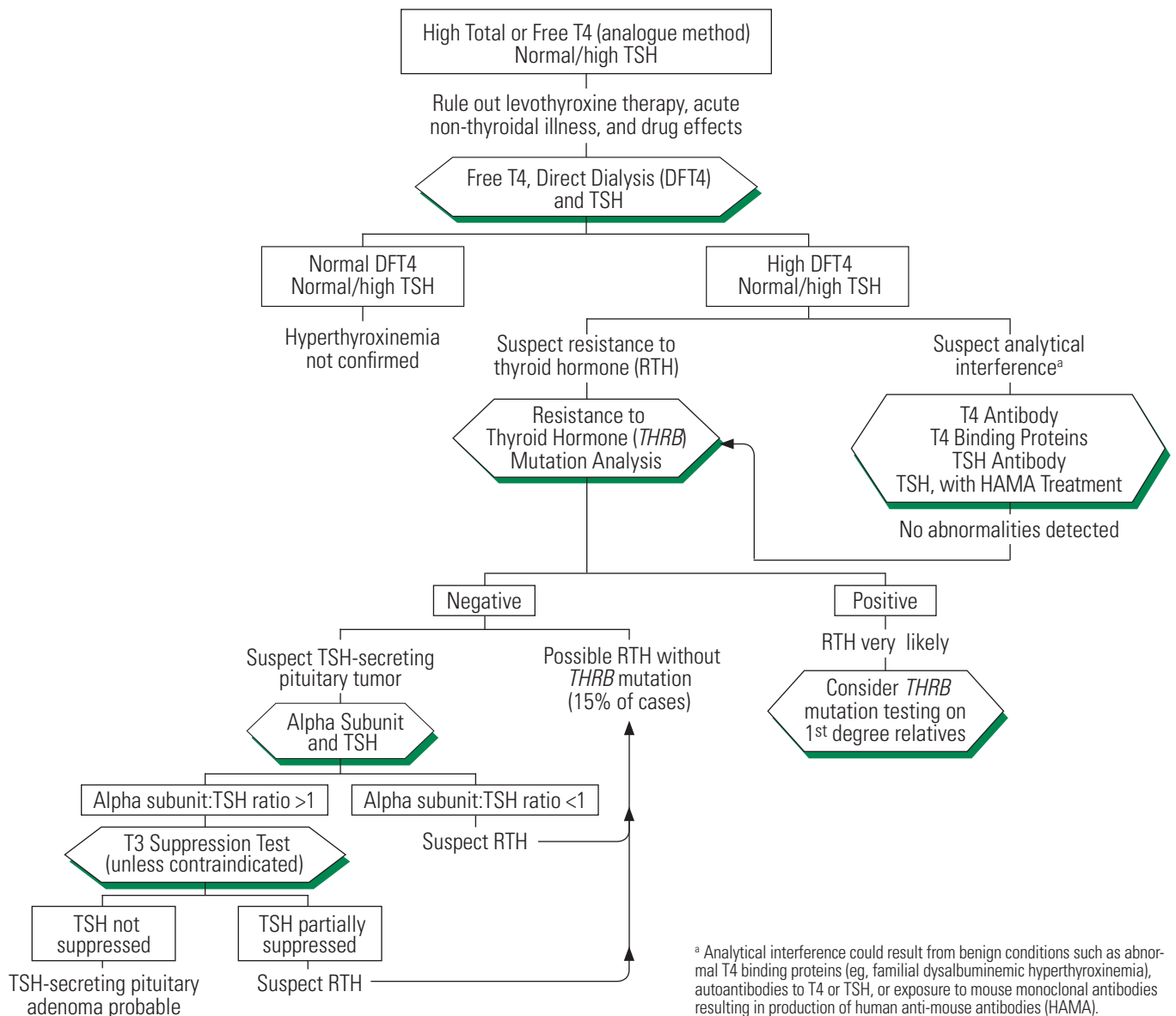


Figure. Differential diagnosis of hyperthyroxinemia and nonsuppressed TSH. Adapted from references 1 and 2.

Table. Laboratory Tests Useful for the Differential Diagnosis and Management of Resistance to Thyroid Hormone

Test Code	Test Name
8658X	Alpha Subunit ^a
16053X	Resistance to Thyroid Hormone (RTH) Mutation Analysis ^{b,c} <i>Detects mutations in the THRB gene.</i>
36574X	T3 (Triiodothyronine) Antibody
36598X	T3, Free, Tracer Dialysis
859X	T3, Total (Triiodothyronine)
36738X	T4 Binding Proteins <i>Includes total T4, total and T4-bound albumin, prealbumin, and thyroxine binding globulin.</i>
36576X	T4 (Thyroxine) Antibody
35167X	T4, Free, Direct Dialysis
36577X	T4, Total, (Thyroxine)
870X	TBG (Thyroxine Binding Globulin)
19577X	TSH Antibody
8660X	TSH, Ultrasensitive
19537X	TSH, with HAMA Treatment

HAMA, human anti-mouse antibodies.

^a This test was developed and its performance characteristics have been determined by Quest Diagnostics Nichols Institute. It has not been cleared or approved by the US Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary. Performance characteristics refer to the analytical performance of the test.

^b This test was developed and its performance characteristics have been determined by Quest Diagnostics Nichols Institute. Performance characteristics refer to the analytical performance of the test.

^c Additional assistance in interpretation of results is available from our Genetic Counselors by calling 1-866-GENE-INFO (1-866-436-3463).

References

1. Refetoff S. Syndromes of reduced sensitivity to thyroid hormone: genetic defects in hormone receptors, cell transporters, and deiodination. *Best Pract Res Clin Endocrinol Metab.* 2007;21:277-305.
2. Beck-Peccoz P, Persani L. TSH-producing adenomas. In: DeGroot LJ, Jameson JL, eds. *Endocrinology*. 5th ed. Philadelphia, PA: Elsevier-Saunders; 2006:475-484.

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