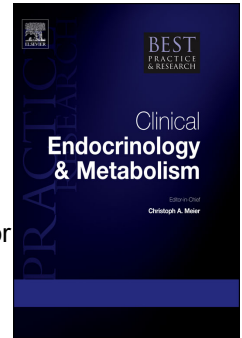


Accepted Manuscript

Resistance to thyrotropin

Helmut Grasberger, M.D., Postdoctoral Researcher, Samuel Refetoff, M.D., Professor
in Medicine, Pediatrics and Committee on Genetics



PII: S1521-690X(17)30010-6

DOI: [10.1016/j.beem.2017.03.004](https://doi.org/10.1016/j.beem.2017.03.004)

Reference: YBEEM 1139

To appear in: *Best Practice & Research Clinical Endocrinology & Metabolism*

Please cite this article as: Grasberger H, Refetoff S, Resistance to thyrotropin, *Best Practice & Research Clinical Endocrinology & Metabolism* (2017), doi: 10.1016/j.beem.2017.03.004.

This is a PDF file of an unedited manuscript that has been accepted for publication. As a service to our customers we are providing this early version of the manuscript. The manuscript will undergo copyediting, typesetting, and review of the resulting proof before it is published in its final form. Please note that during the production process errors may be discovered which could affect the content, and all legal disclaimers that apply to the journal pertain.

Resistance to thyrotropin

Helmut Grasberger, M.D.^a (Postdoctoral Researcher) & Samuel Refetoff, M.D.^b
(Professor in Medicine, Pediatrics and Committee on Genetics)

^a University of Michigan, 6504 MSRB I
1150 West Medical Center Drive
Ann Arbor, MI 48109, USA
Tel: (1-734) 883-7562
FAX: (1-734) 763-2535
e-mail: hgrasber@gmail.com

^b The University of Chicago, MC3090
5841 South Maryland Avenue
Chicago, IL 60637, USA
Tel: (1-773) 702-6939
FAX: (1-773) 702-694
e-mail: refetoff@uchicago.edu

ABSTRACT

Resistance to thyrotropin (RTSH) is broadly defined as reduced sensitivity of thyroid follicle cells to stimulation by biologically active TSH due to genetic defects. Affected individuals have elevated serum TSH in the absence of goiter, with the severity ranging from nongoitrous isolated hyperthyrotropinemia to severe congenital hypothyroidism with thyroid hypoplasia. Conceptually, defects leading to RTSH impair both aspects of TSH-mediated action, namely thyroid hormone synthesis and gland growth. These include inactivating mutations in the genes encoding the TSH receptor and the PAX8 transcription factor. A common third cause has been genetically mapped to a locus on chromosome 15, but the underlying pathophysiology has not yet been elucidated. This review provides a succinct overview of currently defined causes of nonsyndromic RTSH, their differential diagnoses (autoimmune; partial iodine organification defects; syndromic forms of RTSH) and implications for the clinical approach to patients with RTSH.

KEYWORDS

Thyrotropin receptor; TSHR; paired domain; PAX8; mutations; congenital hypothyroidism; subclinical hypothyroidism; hormone resistance

Download English Version:

<https://daneshyari.com/en/article/5586639>

Download Persian Version:

<https://daneshyari.com/article/5586639>

[Daneshyari.com](https://daneshyari.com)