Editorial

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www.imedpub.com

Insights in Biomedicine
ISSN 2572-5610

2021
Vol.6 No.12:56

# Genetic Defect in Thyroid Hormonogenesis -1

Chih-Chang Chu*

Department of Medicine, Florida State University, USA

**Received:** December 18, 2021; **Accepted:** December 23, 2021; **Published:** December 28, 2021

***Corresponding author:**
Dr. Chih-Chang Chu

cc62@cornell.edu

Department of Medicine, Florida State University, USA.

**Citation:** Chu CC (2021) Genetic Defect in Thyroid Hormonogenesis-1. Insights Biomed Vol.6 No.12: 56

## Editorial

Thyroid dyshormonogenesis is a condition in which one or more steps of thyroid hormone synthesis are disrupted, and it accounts for 10%–15% of congenital hypothyroidism (CH). SLC5A5 (NIS), SCL26A4 (PDS), TG, TPO, DUOX2, DUOXA2, and IYD are seven genes linked to thyroid dyshormonogenesis (DHEAL1). CH might be permanent or temporary, depending on the underlying cause. Autosomal recessive inheritance is the most common; however autosomal dominant inheritance is also possible. The molecular foundation, clinical presentation, and genetic diagnosis of CH caused by thyroid dyshormonogenesis are described in this review, with a focus on the advantages of targeted exome sequencing as an updated diagnostic technique.

If left untreated, congenital hypothyroidism is the most prevalent metabolic disease among newborns, resulting in significant neurodevelopmental damage and infertility. Congenital hypothyroidism is mainly sporadic, although up to 2% of thyroid dysgenesis is familial, and organification abnormalities induce congenital hypothyroidism that is recessively inherited. The candidate genes associated with this genetically heterogeneous disorder form two main groups: those causing thyroid gland dysgenesis and those causing dyshormonogenesis. Genes associated with thyroid gland dysgenesis include the TSH receptor in non-syndromic congenital hypothyroidism, and Gsα and the thyroid transcription factors (TTF-1, TTF-2, and Pax-8), associated with different complex syndromes that include congenital hypothyroidism. Among those causing dyshormonogenesis, the thyroid peroxidase and thyroglobulin genes were initially described, and more recently PDS (Pendred syndrome), NIS (sodium iodide symporter), and THOX2.

Thyroid hemiagenesis (THA) is an uncommon congenital condition marked by the loss of one of the thyroid lobes. Specific clinical advice is missing, especially for asymptomatic patients, because the pathophysiology and clinical relevance of this abnormality are unknown. Thyroid hemiagenesis (THA) is an uncommon congenital condition marked by the loss of one of the thyroid lobes. The presence of this abnormality is frequently undetected and discovered by chance (i.e., during screening tests or diagnostic procedures performed for other problems). Specific clinical advice are missing, especially for asymptomatic patients, because the pathophysiology and clinical relevance of this abnormality are unknown. The goal of this review is to outline the genetic elements that contribute to its aetiology and development, as well as to investigate known epidemiologic factors and associated thyroid and nonthyroidal illnesses.

Thyroid embryogenesis is a complicated and poorly understood process. The mechanisms directing the thyroid primordium's descent and lobulation are yet unknown. During embryogenesis, the thyroid gland is the first endocrine organ to develop. By the 20$^{th}$ day after conception, the thyroid anlage is normally visible. Thyroid precursor cells form as endodermal epithelium, which lines the bottom of the primordial pharynx between the first and second pharyngeal arch, proliferates. A strong trophic influence of the surrounding endoderm on thyrocyte proliferation has been observed in the laboratory. As a result, a lack of stimulation from the neighbouring endoderm could lead to a decrease in thyroid tissue bulk, stifling the thyroid primordium's growth. The thyroid anlage is a part of the thyroid gland.

 